CURRICULUM VITAE
NICOLA LONGO MD PhD

NAME: **Nicola Longo** Updated 12-Sep-2022

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CITIZENSHIP: **USA**

II. EDUCATION AND POST-GRADUATE TRAINING

Institution	*Date*	*Degree*
Liceo Scientifico Statale *Guglielmo Marconi*, Parma, Italy	July 1976	Maturita' Scientifica
Univ. Parma Medical School, Italy	**November 1982**	**MD (9/1976-10/Nov/1982)**
Univ. Parma, Italy (Chair GG Guidotti MD)	November 1988	PhD Molecular Biology and Pathology
Post-Doctoral Fellow (Louis J Elsas MD)	**1985-87**	**Medical Genetics/Pediatrics Emory Univ., Atlanta, GA**
Post-Doctoral Fellow (Gian C Gazzola MD)	1988-1989	Istituto Patologia Generale University of Parma, Italy
Pediatrics Internship	**7-9/1993, 1-3/1994 7-9/ 1994, 1-3/1995**	**Emory University**
Residency in Clinical Genetics	7/1/93 – 6/30/96	Emory University
Fellowship in Clinical Biochemical Genetics	**6/23/97–11/30/98**	**Emory University**

III. PROFESSIONAL EXPERIENCE

Title	*Institution*	*Date*
Assistant Professor	Emory University (Medical Genetics/Pediatrics)	1989-1998
	Emory University (Physiology)	1989-2001
Director of Research	Emory University (Medical Genetics/Pediatrics)	1997-2001
Associate Professor	Emory University (Medical Genetics/Pediatrics)	1998-2001
Associate Professor	University of Utah (Medical Genetics/Pediatrics)	2001-2003
Director, Metabolic Service	University of Utah (Medical Genetics/Pediatrics)	2001-present
Associate Professor	University of Utah (Pathology)	2001-2006
Professor (Tenured)	University of Utah (Medical Genetics/Pediatrics)	2003-present
Professor (Adjunct)	University of Utah (Pathology)	2006-present
Director, Clinical Biochemical Genetics Fellowship Program	University of Utah (Medical Genetics/Pediatrics)	2004-2019
Chief, Division of Medical Genetics, University of Utah (Pediatrics)		2007-present
Director, Residency Program Medical Genetics, University of Utah		2014-2015
Associate Director, Residency Program Medical Genetics, University of Utah		2015-Present
Professor (Adjunct) University of Utah (Nutrition and Integrative Physiology)		2019-present
Co-Director, ABMG Clinical Biochemical Genetics Training Program		2019-Present
Director, ABMG Medical Biochemical Genetics Training Program		2020-Present

Attending Physician (Medical Genetics)

Children's Healthcare of Atlanta at Egleston Hospital	3/1997-8/2001
Emory University Hospital	3/1997-8/2001
Crawford Long Hospital	3/1997-8/2001
Grady Memorial Hospital, Atlanta GA	11/1997-8/2001
Primary Children's Medical Center	9/2001-Present
100 Mario Capecchi Dr, Salt Lake City, UT 84113	
University of Utah School of Medicine	9/2001-Present
30 N 1900 E, Salt Lake City, UT, 84132	
Cure 4 the Kids Foundation (Monthly Genetics Clinic)	2011-Present
One Breakthrough Way, Las Vegas, NV 89135	
Alaska Native Medical Center (quarterly genetics clinics)	2020-Present
4315 Diplomacy Drive, Anchorage, AK 99508	

LICENSURES AND BOARDS

State	***Number***	***Year***
Italy	1466	1/1983
ECFMG (USA)	04697926	07/15/1992
FLEX (USA)	570427026	12/1993
Georgia, USA	040144 (inactive)	06/08/1995
Utah, USA	4916705-1205	09/12/2001
Nevada, USA	12808	7/11/2008
Wyoming, USA	8948A	09/11/2011
Alaska, USA	150326	01/07/2020
DEA	BL4639413	2001 (UT)
DEA	FL5013747	04-02-2018 (NV)
DEA	FL9102221	03/02/2020 (AK)
American Board of Medical Genetics		
Clinical Genetics	96117	09/27/96 rec. 2005, 12/4/09, MOC
Clinical Biochemical Genetics	99095	09/01/1999 rec. 12/4/2009, MOC

CLINICAL SERVICE CONTRIBUTIONS

Consultant, Emory Genetics DNA Diagnostic Laboratory,	1989-1994
Consultant, Emory Genetics Biochemical Genetics Laboratory	1994-1998
Assistant Director, Biochemical Genetics Laboratory	1998-2001
Emory Genetics Laboratories, Atlanta GA	
Consultant, ARUP Laboratories, Biochemical Genetics Laboratory	2001-2002
Co-Director, ARUP Biochemical Genetics & Newborn Screening Laboratory	2003-2019
Co-Director, ARUP Biochemical Genetics Laboratory	2019-Present
ARUP Laboratories at the University of Utah, Salt Lake City UT 84108	

SCIENTIFIC REVIEW COMMITTEE - STUDY SECTIONS

-Telethon, Italy (1996-2000)
-CDC Nutritional Biochemistry special emphasis panel: Innovative technology development grant for the detection and monitoring of diabetic hypoglycemia (1999), Member
-NIH-NIDDK Special Emphasis Panel - Diabetes Research Centers (2001,2002)-Ad Hoc Member
-NIH-NIDDK General Medicine B Study Section (2002), Ad Hoc Member
-Italian Research Ministry, Reviewer, 2003
-NIH-NIDDK Special Emphasis Panel (ZDK1-GRB8-M1) Small Grants in Digestive Diseases and Nutrition (2004-2006), Member

-AHA Western Regional Study Section, Member, 2004-2008.
-NIH-NICHD SEP Novel Technologies in Newborn Screening, 2006
-Cineca (Consortium of Italian Universities, Research Institutions, and the Italian Ministry of Education) 2004-2008, reviewer
-NIH-NIDDK, Rare Disease Network, Data Safety and Motoring Board, 2006-2009
-NIH-NIDDK SEP ZRG1 MDCN-J (91) S Molecular and Cellular Neuroscience, 2007
-NIH-NICHD SEP ZHD1-DRG-A (01) Newborn Screening: Translational Research Network Coordinating Review, 2007, 2008, 2009
-NIH-NIDDK ZRG1 ETTN-G (02) SEP Molecular Neurogenetics, 2008.
-AHA Western Regional Study Section, Co-Chair, 2009
-ACMG Newborn Screening National Coordinating Center Work Group, 2008-2012
-NIH-NIDDK 2009/05 ZRG1 HOP-Y (50) R Rare Diseases Clinical Research Consortia, 2009
-AHA Western Regional Study Section, Chair, 2010.
-AHA National Study Section, Vice-Chair, 2011-2012
-NIH- Center for Scientific Review: Therapeutic Approaches to Genetic Diseases (TAG), Study Section Member, 07/2011-06/2017
-NIH-NICHD, Rare Disease Network, Data Safety and Motoring Board, 2010-2011
-Poland Science Foundation, 2012, Reviewer
-NIH-NICHD, Chair, Rare Disease Network, Data Safety and Motoring Board, 2011-2014
-NIH-TRND, CincY as a Treatment for Creatine Transporter Deficiency, Advisory Board Member 2012-Present
-Wellcome Trust, UK, Reviewer, 2015
-NIH-NICHD, Chair, Data Safety and Motoring Board, Rare Disease Network, 2014-2019
-Reviewer-Site visitor: NIH/NIHGRI, Bethesda, MD 6-7 June 2016
-ERA-Net for Research Programmes on Rare Diseases (European Union), Reviewer, 2017
-Swiss National Science Foundation, Reviewer, 2017
-Galactosemia Foundation, Reviewer 2017
-NIH-NICHD, Chair, Study Section, Newborn Screening Translational Research Network (NBSTRN) 13 Aug 2018
-Japan Agency for Medical Research and Development (AMED), 2018, reviewer
-The Netherlands Organisation for Health Research and Development, Reviewer, 2018
-King Abdullah International Medical Research Center (KAIMRC), reviewer, 2014, 2018
-NIH BST IRG Small Business: Biomaterials, Delivery, and nanotechnology Study Section, 5 Nov 2018
-Telethon Rare Disorders, Italy, Reviewer 2019
-NIH-NIAID, Chair, Resource Related Research Projects, Study Section 20 Oct 2021
-Swiss National Science Foundation, Reviewer, 2021
NIH-NIAID, Chair, Resource Related Research Projects, Study Section 20 May 2022

EDITORIAL EXPERIENCE - MANUSCRIPT REVIEWER FOR SCIENTIFIC JOURNALS

-Archives of Biochemistry and Biophysics
-American Journal of the Medical Sciences
-American Journal of Physiology
-Biochimica et Biophysica Acta
-Diabetes
-FEBS Letters
-Human Genetics
-Journal of Clinical Endocrinology and Metabolism
-Journal of Clinical Investigation
-Journal of Inherited Metabolic Disorders
-Journal of Pediatric Endocrinology and Metabolism
-American Journal of Medical Genetics
-American Journal of Human Genetics
-Biochemical Pharmacology
-Brain
-FASEB Journal
-Genetics in Medicine
-Human Mutation
-Journal of Biological Chemistry
-Pediatric Research
-Pediatrics
-Journal of Pediatrics

-Journal of Pharmacology and Experimental Therapeutics
-Molecular Genetics and Metabolism
-New England Journal of Medicine
-Proceedings of the National Academy of Sciences USA
-Journal of Neurochemistry
-Molecular and Cellular Biochemistry
-PloS Genetics

RESEARCH GRANTS, CONTRACTS, AND AWARDS

ACTIVE

U54HD100982-01 Cary Harding project PI, Nicola Longo Local PI 09/16/2019 – 08/31/2024 NIH
Hyperphenylalaninemia Disorders Consortium of the Rare Disease Clinical Research Network
U54HD100982-01 (Nicola Longo) 09/16/2019 – 08/31/2024 NIH
Hyperphenylalaninemia Disorders Consortium of the Rare Disease Clinical Research NetworkPilot/Feasibility Core
The objective of this project is to define the natural history and devise new therapies for the hyperphenylalaninemias.
As PI of the feasibility core, I need to solicit and select novel project applications in the field of hyperphenylalaninemia.

5R13HD062129-08 (Longo) 7/14/2009-01/31/2023 NIH/NIDDK/NICHD/NINDS
Society for Inherited Metabolic Disorders Annual Meeting
This grant support attendance of young investigators to the SIMD annual meeting

UFDSP00011718 (Stacpoole, Longo site PI) 06/01/2017-05/31/2022 NIH/NICHD
Phase 3 Trial of DCA in PDC Deficiency IND 028,625 The objective of this study is to evaluate the efficacy of dichloroacetae in teh treatment of pyruvate dehydrogenase deficiency**.**

09/01/2008 – 12/31/2023 (Longo N PI) PROJECT : 50300913 - LYSOSOMAL STORAGE
Genzyme Corp/ Sanofi Aventis
Registry for patients with lysosomal storage disorders

07/01/2002 – 12/31/2025 (Longo N PI) 50301676 - PKUDOS- PKU DEMOGRAPHICS, OUTCOMES, AND SAFETY REGISTRY BioMarin Pharmaceutial Corp
Registry for patients with phenylketonuria

12/14/18 - 12/31/22 Atb200-03 Pompe disease
Principal Investigator(s): Nicola Longo
Direct Costs: $519,926 Total Costs: $622,805
Role: Principal Investigator
09/26/18 - 12/31/22 Pb-102-F51 Fabry Disease
Principal Investigator(s): Nicola Longo
Direct Costs: $200,592 Total Costs: $245,119
Role: Principal Investigator
07/01/18 - 06/30/23 Metabolic Program Support
Principal Investigator(s): Nicola Longo
Yearly Direct Costs: $138,075 Total Costs: $138,075
Utah Department of Health
Role: Principal Investigator
06/25/18 - 12/31/23 Mrna-3704-P001 Mma Methylmalonic acidemia
Principal Investigator(s): Nicola Longo
Direct Costs: $31,462 Total Costs: $42,757
Moderna Therapeutics
Role: Principal Investigator
03/26/18 - 12/31/22 Pbd-001 Cnsa-001 PTPS deficiency
Principal Investigator(s): Nicola Longo
Direct Costs: $78,364 Total Costs: $98,815
Role: Principal Investigator
03/16/18 - 12/30/22 Poms-003 Amicus Pompe

Principal Investigator(s): Nicola Longo
Direct Costs: $109,397 Total Costs: $143,544
Role: Principal Investigator

02/12/18 - 12/31/21 Msc12790/Agluo7710 Pompe Pk
Principal Investigator(s): Nicola Longo
Direct Costs: $33,331 Total Costs: $42,867
Role: Principal Investigator

12/01/17 - 12/31/21 Spimm-301 Mitochondrial disorders
Principal Investigator(s): Nicola Longo
Direct Costs: $257,751 Total Costs: $321,750
Stealth BioTherapeutics Inc.
Role: Principal Investigator

11/27/17 - 12/31/21 Hpn-100-021 Urea cycle disorders
Principal Investigator(s): Nicola Longo
Direct Costs: $178,522 Total Costs: $221,434
Role: Principal Investigator

06/15/17 - 12/31/20 Caeb1102-101ª Arginase deficiency
Principal Investigator(s): Nicola Longo
Direct Costs: $142,135 Total Costs: $175,328
Aeglea BioTherapeutics
Role: Principal Investigator

05/24/17 - 12/31/20 Spimm-300 Mitochondrial disorders
Principal Investigator(s): Nicola Longo
Direct Costs: $23,338 Total Costs: $31,491
Role: Principal Investigator

05/06/17 - 12/31/20 Pb-102-F50 Brigt Fabry
Principal Investigator(s): Nicola Longo
Direct Costs: $288,818 Total Costs: $352,837
Role: Principal Investigator

03/21/17 - 12/31/21 Lum-001-C-01 Creatine transporter deficiency (basic science)
Principal Investigator(s): Nicola Longo
Direct Costs: $77,591 Total Costs: $101,402
Role: Principal Investigator

03/13/17 - 12/31/21 Neogaa - Efc14028 Pompe
Principal Investigator(s): Nicola Longo
Direct Costs: $265,446 Total Costs: $323,400
Role: Principal Investigator

12/05/16 - 12/31/20 Balance Fabry
Principal Investigator(s): Nicola Longo
Direct Costs: $452,825 Total Costs: $599,964
Protalix Biotherapeutics
Role: Principal Investigator

08/15/16 - 12/30/22 B3031002 Tali Registry Gaucher
Principal Investigator(s): Nicola Longo
Direct Costs: $12,563 Total Costs: $17,149
Pfizer Inc.
Role: Principal Investigator

02/05/16 - 02/05/21 Pompe Subregistry Lts13930
Principal Investigator(s): Nicola Longo
Direct Costs: $15,261 Total Costs: $20,251
Role: Principal Investigator

07/01/14 - 12/31/21 701-301 Pompe Bmrn
Principal Investigator(s): Nicola Longo
Direct Costs: $267,853 Total Costs: $303,062
Role: Principal Investigator

03/15/14 - 12/31/24 Thrive Ucd Registry
Principal Investigator(s): Nicola Longo
Direct Costs: $121,900 Total Costs: $161,761
Hyperion Therapeutics, Inc.
Role: Principal Investigator

12/01/12 - 12/31/23 Gos Gaucher
Principal Investigator(s): Nicola Longo
Direct Costs: $36,182 Total Costs: $48,013
PharmaNet, Inc.
Role: Principal Investigator

Past Grants

02/07/19 - 02/28/20 Synlogic Pku Mutation Consult
Principal Investigator(s): Nicola Longo
Direct Costs: $18,315 Total Costs: $25,000
Synlogic
Role: Principal Investigator

02/04/19 - 02/27/19 Horizon Consulting Nl
Principal Investigator(s): Nicola Longo
Direct Costs: $2,954 Total Costs: $4,032
Role: Principal Investigator

12/03/18 - 12/31/19 Mma Phase1-2 Study
Principal Investigator(s): Nicola Longo
Direct Costs: $3,909 Total Costs: $5,336
Moderna Therapeutics
Role: Principal Investigator

11/29/18 - 12/31/19 Biomarin Sow #20
Principal Investigator(s): Nicola Longo
Direct Costs: $1,505 Total Costs: $2,054
Role: Principal Investigator

11/01/18 - 08/01/19 Bmrn Japan Ad Board Pegvalias
Principal Investigator(s): Nicola Longo
Direct Costs: $3,663 Total Costs: $5,000
Role: Principal Investigator

09/20/18 - 03/19/19 Pb-102-F60
Principal Investigator(s): Nicola Longo
Direct Costs: $1,055,177 Total Costs: $1,280,179
Role: Principal Investigator

09/18/18 - 09/30/19 Biomarin Sow #18
Principal Investigator(s): Nicola Longo
Direct Costs: $5,273 Total Costs: $7,198
Role: Principal Investigator

08/01/18 - 08/01/19 Biomarin Sow #17
Principal Investigator(s): Nicola Longo
Direct Costs: $5,614 Total Costs: $7,663
Role: Principal Investigator

07/25/18 - 07/31/19 Coa Nl Scientific Board
Principal Investigator(s): Nicola Longo
Direct Costs: $1,172 Total Costs: $1,600
CoA Therapeutics
Role: Principal Investigator

06/27/18 - 11/04/18 Bmrn Sow#16
Principal Investigator(s): Nicola Longo
Direct Costs: $1,172 Total Costs: $1,600
Role: Principal Investigator

06/01/18 - 06/30/18 Leadiant Biosciences Nl Consu
Principal Investigator(s): Nicola Longo
Direct Costs: $1,099 Total Costs: $1,500
Leadiant Biosciences, Inc.
Role: Principal Investigator

04/26/18 - 11/04/18 Bmrn Sow#15
Principal Investigator(s): Nicola Longo
Direct Costs: $1,465 Total Costs: $2,000
Role: Principal Investigator

04/26/18 - 11/04/18 Advisor Agreement Brain Pku
Principal Investigator(s): Nicola Longo
Direct Costs: $3,663 Total Costs: $5,000
Role: Principal Investigator
03/14/18 - 12/31/18 Evidera Longo Consulting
Principal Investigator(s): Nicola Longo
Direct Costs: $1,099 Total Costs: $1,500
Evidera
Role: Principal Investigator
03/08/18 - 03/31/19 Nl Reneo Consulting Agreement
Principal Investigator(s): Nicola Longo
Direct Costs: $1,099 Total Costs: $1,500
Reneo Pharmaceuticals, Inc.
Role: Principal Investigator
02/27/18 - 12/31/18 Synlogic Advisory Longo Ucd
Principal Investigator(s): Nicola Longo
Direct Costs: $18,315 Total Costs: $25,000
Synlogic
Role: Principal Investigator
02/08/18 - 12/31/18 Ovid Ov101-15-001 Stars
Principal Investigator(s): Nicola Longo
Direct Costs: $2,198 Total Costs: $3,000
Ovid Therapeutics
Role: Principal Investigator
02/05/18 - 02/04/19 Logicbio Consulting Nl
Principal Investigator(s): Nicola Longo
Direct Costs: $43,956 Total Costs: $60,000
LogicBio Therapeutics, Inc.
Role: Principal Investigator
12/22/17 - 12/31/18 Bmrn Sow#13 Longo
Principal Investigator(s): Nicola Longo
Direct Costs: $411 Total Costs: $561
Role: Principal Investigator
11/01/17 - 05/31/18 Bmrn Sow #12 Texas Mtg
Principal Investigator(s): Nicola Longo
Direct Costs: $2,198 Total Costs: $3,000
Role: Principal Investigator
08/21/17 - 12/31/17 Bmrn Sow #11 Pegvaliase Recom
Principal Investigator(s): Nicola Longo
Direct Costs: $1,924 Total Costs: $2,626
Role: Principal Investigator
08/10/17 - 08/01/18 Nestec Consultant Nl
Principal Investigator(s): Nicola Longo
Direct Costs: $2,562 Total Costs: $3,497
Nestec S.A.
Role: Principal Investigator
08/01/17 - 12/31/17 Bmrn Sow #12 Peg Advisory Boar
Principal Investigator(s): Nicola Longo
Direct Costs: $274 Total Costs: $374
Role: Principal Investigator
07/19/17 - 07/19/19 Moderna Consulting Agreement
Principal Investigator(s): Nicola Longo
Direct Costs: $8,789 Total Costs: $11,997
Moderna Therapeutics
Role: Principal Investigator
07/10/17 - 12/31/18 Sow 11 - Ny Oct 2017
Principal Investigator(s): Nicola Longo
Direct Costs: $3,121 Total Costs: $4,260
Role: Principal Investigator
03/31/17 - 03/31/19 Consultingagreement Orphantech

Principal Investigator(s): Nicola Longo
Direct Costs: $1,829 Total Costs: $2,497
Orphan Technologies
Role: Principal Investigator

03/31/17 - 06/30/17 Biomarin Sow #9 - Peg Mtg P2
Principal Investigator(s): Nicola Longo
Direct Costs: $1,099 Total Costs: $1,500
Role: Principal Investigator

03/14/17 - 03/14/20 Hemoshear Consulting Agreement
Principal Investigator(s): Nicola Longo
Direct Costs: $47,471 Total Costs: $64,798
HemoShear Therapeutics, LLC
Role: Principal Investigator

01/13/17 - 01/13/18 La Jolla Pharm Consulting Agre
Principal Investigator(s): Nicola Longo
Direct Costs: $17,948 Total Costs: $24,499
La Jolla Pharmaceutical Company
Role: Principal Investigator

11/01/16 - 12/31/17 Pku Burden
Principal Investigator(s): Nicola Longo
Direct Costs: $16,895 Total Costs: $23,062
Role: Principal Investigator

10/07/16 - 10/07/18 Pkudos Consulting Sow #8
Principal Investigator(s): Nicola Longo
Direct Costs: $14,640 Total Costs: $19,983
Role: Principal Investigator

09/23/16 - 12/22/16 Pegvaliase Treatment Algorithm
Principal Investigator(s): Nicola Longo
Direct Costs: $1,130 Total Costs: $1,500
Role: Principal Investigator

08/05/16 - 12/31/19 Rp-103-Mito-002
Principal Investigator(s): Nicola Longo
Direct Costs: $61,402 Total Costs: $74,994
Role: Principal Investigator

07/15/16 - 12/31/16 Ravicti Advisory Board
Principal Investigator(s): Nicola Longo
Direct Costs: $732 Total Costs: $1,000
Horizon Pharma plc
Role: Principal Investigator

06/01/16 - 05/31/18 Bmrn Sow #10 Jismd
Principal Investigator(s): Nicola Longo
Direct Costs: $12,800 Total Costs: $17,472
Role: Principal Investigator

06/01/16 - 05/31/17 Dca In Pdc Deficiency
Principal Investigator(s): Nicola Longo
Direct Costs: $5,199 Total Costs: $7,746
Role: Principal Investigator

06/01/16 - 12/30/16 Registry 2016 Nl
Principal Investigator(s): Nicola Longo
Direct Costs: $1,808 Total Costs: $2,399
Role: Principal Investigator

05/17/16 - 05/17/18 Censa Pharma Consulting
Principal Investigator(s): Nicola Longo
Direct Costs: $6,795 Total Costs: $9,275
Censa Pharmaceuticals Inc.
Role: Principal Investigator

04/29/16 - 12/31/16 Mitobridge Consulting Faod
Principal Investigator(s): Nicola Longo
Direct Costs: $3,765 Total Costs: $4,996

Mitobridge, Inc.
Role: Principal Investigator
04/01/16 - 12/31/19 Ux007-Cl202
Principal Investigator(s): Nicola Longo
Direct Costs: $53,809 Total Costs: $66,658
Role: Principal Investigator
03/07/16 - 12/30/17 Vts301 Phase 2b/3 Npc
Principal Investigator(s): Nicola Longo
Direct Costs: $4,943 Total Costs: $6,747
Role: Principal Investigator
03/01/16 - 12/31/19 Retrophin Ctx 018ctxx15001
Principal Investigator(s): Nicola Longo
Direct Costs: $29,204 Total Costs: $38,190
Retrophin, Inc.
Role: Principal Investigator
11/10/15 - 11/10/18 Biomarin Sow6 Kuvan
Principal Investigator(s): Nicola Longo
Direct Costs: $15,072 Total Costs: $20,000
Role: Principal Investigator
11/03/15 - 12/31/17 Lumos Creatine Lab Support
Principal Investigator(s): Nicola Longo
Direct Costs: $30,143 Total Costs: $40,000
Lumos Pharma, Inc.
Role: Principal Investigator
10/27/15 - 10/26/17 Peg-Pal P3 Steering Sow#4
Principal Investigator(s): Nicola Longo
Direct Costs: $22,606 Total Costs: $30,000
Role: Principal Investigator
10/27/15 - 01/26/16 Pegvaliase Japan Mtg Nov 15
Principal Investigator(s): Nicola Longo
Direct Costs: $1,846 Total Costs: $2,450
Role: Principal Investigator
06/30/15 - 12/31/18 Hpn-100-009
Principal Investigator(s): Nicola Longo
Direct Costs: $168,924 Total Costs: $203,916
Hyperion Therapeutics, Inc.
Role: Principal Investigator
06/30/15 - 12/31/17 165-303
Principal Investigator(s): Nicola Longo
Direct Costs: $14,995 Total Costs: $19,643
Role: Principal Investigator
06/30/15 - 12/29/17 Mito-001
Principal Investigator(s): Nicola Longo
Direct Costs: $76,438 Total Costs: $96,366
Raptor Pharmaceutical Corp.
Role: Principal Investigator
11/18/14 - 11/18/17 Dimension Scientific Advisory
Principal Investigator(s): Nicola Longo
Direct Costs: $22,600 Total Costs: $29,990
Dimension Therapeutics
Role: Principal Investigator
10/03/14 - 10/02/15 Codexis Pku Consulting Agreeme
Principal Investigator(s): Nicola Longo
Direct Costs: $2,260 Total Costs: $3,000
Codexis, Inc.
Role: Principal Investigator
06/01/14 - 12/31/14 Mgm116041 - Gsk Fabry
Principal Investigator(s): Nicola Longo
Direct Costs: $7,784 Total Costs: $10,329
GlaxoSmithKline

Role: Principal Investigator
05/16/14 - 07/06/14 Consulting Agreement Lee Pharm
Principal Investigator(s): Nicola Longo
Direct Costs: $3,605 Total Costs: $4,784
Lee's Pharmaceutical Holdings Limited
Role: Principal Investigator
04/01/14 - 12/31/18 Msc12711/Agal19110 Fabry Map
Principal Investigator(s): Nicola Longo
Direct Costs: $13,120 Total Costs: $16,527
Role: Principal Investigator
02/20/14 - 12/31/17 Ux007-Cl201
Principal Investigator(s): Nicola Longo
Direct Costs: $178,038 Total Costs: $213,358
Ultragenyx Pharmaceutical Inc.
Role: Principal Investigator
11/18/13 - 12/31/19 165-302
Principal Investigator(s): Nicola Longo
Direct Costs: $1,025,526 Total Costs: $1,279,032
Role: Principal Investigator
04/15/13 - 04/14/14 Pfizer Gaucher Advisory 22apr
Principal Investigator(s): Nicola Longo
Direct Costs: $2,261 Total Costs: $3,000
Pfizer Inc.
Role: Principal Investigator
03/05/13 - 03/04/16 Pkudos Advisory Board
Principal Investigator(s): Nicola Longo
Direct Costs: $22,607 Total Costs: $30,000
Role: Principal Investigator
11/04/12 - 11/03/14 Peg-Pal Phase 3 Steering Committee
Principal Investigator(s): Nicola Longo
Direct Costs: $15,072 Total Costs: $20,000
Role: Principal Investigator
10/31/12 - 11/01/13 Hyperion Ucd National Advisory
Principal Investigator(s): Nicola Longo
Direct Costs: $1,884 Total Costs: $2,500
Hyperion Therapeutics, Inc.
Role: Principal Investigator
08/01/12 - 02/28/15 Live Cell Assay
Principal Investigator(s): Nicola Longo
Direct Costs: $248,681 Total Costs: $330,000
Marker Gene Technologies, Inc.
Role: Co-Principal Investigator
07/14/12 - 02/28/15 Simd Annual Meeting
Principal Investigator(s): Nicola Longo
Direct Costs: $40,000 Total Costs: $40,000
Eunice Kennedy Shriver National Institute of Child Health and Human Development
Role: Principal Investigator
06/27/12 - 12/31/13 Pact
Principal Investigator(s): Nicola Longo
Direct Costs: $15,000 Total Costs: $19,905
Symbiotix, Inc.
Role: Principal Investigator
05/04/12 - 12/31/16 Biomarin 165-301
Principal Investigator(s): Nicola Longo
Direct Costs: $301,580 Total Costs: $374,936
Role: Principal Investigator
04/01/12 - 12/31/14 Biomarin 205-165
Principal Investigator(s): Nicola Longo
Direct Costs: $163,306 Total Costs: $197,237

BioMarin Pharmaceutical, Inc.
Role: Principal Investigator
07/19/11 - 07/31/12 Bpms-1001
Principal Investigator(s): Nicola Longo
Direct Costs: $5,014 Total Costs: $6,654
Hantel Technologies, Inc.
Role: Principal Investigator
07/01/11 - 06/30/12 Biomarin Oard Bh4 Autism
Principal Investigator(s): Nicola Longo
Direct Costs: $1,507 Total Costs: $2,000
BioMarin Pharmaceutical, Inc.
Role: Principal Investigator
07/01/11 - 06/30/12 Genzyme Fellowship Support
Principal Investigator(s): Nicola Longo
Direct Costs: $60,000 Total Costs: $60,000
Genzyme Corporation
Role: Principal Investigator
06/01/11 - 12/31/13 Hgt-Rep-084
Principal Investigator(s): Nicola Longo
Direct Costs: $88,022 Total Costs: $102,152
Shire Human Genetic Therapies
Role: Principal Investigator
03/01/11 - 12/31/12 Pal-004
Principal Investigator(s): Nicola Longo
Direct Costs: $60,796 Total Costs: $75,503
BioMarin Pharmaceutical, Inc.
Role: Principal Investigator
02/28/11 - 02/27/14 Consulting Agreement Pku-016
Principal Investigator(s): Nicola Longo
Direct Costs: $33,911 Total Costs: $45,000
BioMarin Pharmaceutical, Inc.
Role: Principal Investigator
02/11/11 - 02/10/12 Hyperion Advisory Mtg
Principal Investigator(s): Nicola Longo
Direct Costs: $1,356 Total Costs: $1,800
ScienceVision LLC
Role: Principal Investigator
02/01/11 - 06/30/11 Food Frequency Questionnaire
Principal Investigator(s): Nicola Longo
Direct Costs: $35,000 Total Costs: $35,000
BioMarin Pharmaceutical, Inc.
Role: Principal Investigator
01/01/11 - 12/31/16 Encore (Gzgd02607)
Principal Investigator(s): Nicola Longo
Direct Costs: $55,995 Total Costs: $64,044
Genzyme Corporation
Role: Principal Investigator
01/01/11 - 12/31/15 Hgt-Hit-046: Hunter It
Principal Investigator(s): Nicola Longo
Direct Costs: $109,320 Total Costs: $133,296
Shire Human Genetic Therapies
Role: Principal Investigator
12/08/10 - 12/07/11 Nutricia Consulting Agreement
Principal Investigator(s): Nicola Longo
Direct Costs: $1,507 Total Costs: $2,000
Nutricia North America, Inc.
Role: Principal Investigator
07/01/10 - 05/31/13 Pb-06-004
Principal Investigator(s): Nicola Longo
Direct Costs: $33,741 Total Costs: $40,621

Target Research Associates, Inc
Role: Principal Investigator

05/30/10 - 06/01/13 Consultant Agreement
Principal Investigator(s): Nicola Longo
Direct Costs: $7,084 Total Costs: $9,400
Pfizer Inc.
Role: Principal Investigator

05/01/10 - 12/31/10 Cz-011-01 - Mri Study
Principal Investigator(s): Nicola Longo
Direct Costs: $200 Total Costs: $200
Genzyme Corporation
Role: Principal Investigator

04/01/10 - 12/31/14 Gzgd03109-Edge
Principal Investigator(s): Nicola Longo
Direct Costs: $80,291 Total Costs: $92,217
Genzyme Corporation
Role: Principal Investigator

02/01/10 - 12/23/10 Adapt
Principal Investigator(s): Nicola Longo
Direct Costs: $32,027 Total Costs: $42,500
Symbiotix, Inc.
Role: Principal Investigator

01/01/10 - 12/31/13 At1001-011
Principal Investigator(s): Nicola Longo
Direct Costs: $43,960 Total Costs: $52,831
Amicus Therapeutics, Inc.
Role: Principal Investigator

10/15/09 - 12/31/14 Hpn-100-007
Principal Investigator(s): Nicola Longo
Direct Costs: $202,476 Total Costs: $253,196
Hyperion Therapeutics, Inc.
Role: Principal Investigator

07/14/09 - 02/28/11 Simd Annual Meeting
Principal Investigator(s): Nicola Longo
Direct Costs: $29,000 Total Costs: $29,000
National Institutes of Health
Role: Principal Investigator

04/13/09 - 12/31/17 Pku-015
Principal Investigator(s): Nicola Longo
Direct Costs: $110,043 Total Costs: $140,439
BioMarin Pharmaceutical, Inc.
Role: Principal Investigator

01/15/09 - 01/30/19 Mps Vi Csp
Principal Investigator(s): Nicola Longo
Direct Costs: $3,781 Total Costs: $5,017
Role: Principal Investigator

10/01/08 - 05/31/11 Carnitine Transporter
Principal Investigator(s): Nicola Longo
Direct Costs: $86,340 Total Costs: $129,942
National Institute of Diabetes and Digestive and Kidney Diseases
Role: Principal Investigator

09/01/08 - 12/31/13 Pkudos Registry
Principal Investigator(s): Nicola Longo
Direct Costs: $2,504 Total Costs: $3,323
BioMarin Pharmaceutical, Inc.
Role: Principal Investigator

09/01/08 - 08/31/11 Shire 044 Ga-Gcb
Principal Investigator(s): Nicola Longo
Direct Costs: $26,421 Total Costs: $33,138

Quintiles Transnational Corporation
Role: Principal Investigator

07/01/08 - 12/31/14 Myozyme Iti
Principal Investigator(s): Nicola Longo
Direct Costs: $134,987 Total Costs: $159,687
Genzyme Corporation
Role: Principal Investigator

02/28/08 - 12/31/19 Pal-001
Principal Investigator(s): Nicola Longo
Direct Costs: $2,713,500 Total Costs: $3,506,998
BioMarin Pharmaceutical, Inc.
Role: Principal Investigator

08/15/07 - 06/30/09 Anaplerotic Therapy In Propionic Acidemia
Principal Investigator(s): Nicola Longo
Direct Costs: $272,500 Total Costs: $408,613
National Institutes of Health
Role: Principal Investigator

07/20/07 - 12/31/11 Mtap
Principal Investigator(s): Nicola Longo
Direct Costs: $9,412 Total Costs: $12,000
Genzyme Corporation
Role: Principal Investigator

06/01/07 - 05/30/17 Hunter Outcome Survey
Principal Investigator(s): Nicola Longo
Direct Costs: $5,882 Total Costs: $7,500
Shire Human Genetic Therapies
Role: Principal Investigator

06/01/07 - 12/31/09 Pku-Seap-001
Principal Investigator(s): Nicola Longo
Direct Costs: $7,795 Total Costs: $9,502
BioMarin Pharmaceutical, Inc.
Role: Principal Investigator

05/01/07 - 12/31/09 Tkt034 (Shire)
Principal Investigator(s): Nicola Longo
Direct Costs: $30,745 Total Costs: $37,227
Quintiles Transnational Corporation
Role: Principal Investigator

03/01/06 - 05/31/13 NIH R01 DK 53824 Carnitine Transporter in Human Disease
Principal Investigator(s): Nicola Longo
Direct Costs: $919,680 Total Costs: $1,371,577
National Institute of Diabetes and Digestive and Kidney Diseases
Role: Principal Investigator

01/01/06 - 12/31/06 Newborn Screening Surveillance Program
Principal Investigator(s): Nicola Longo
Direct Costs: $27,273 Total Costs: $30,000
Mountain States Genetic Foundation
Role: Principal Investigator

05/01/05 - 08/31/06 Non-Invasive Blood Phenylalanine Monitor.
Principal Investigator(s): Nicola Longo
Direct Costs: $17,584 Total Costs: $22,420
Aciont Inc.
Role: Principal Investigator

09/01/04 - 01/31/07 Expanded Access Use Of Recombinant Human Acid Alpha-Gluc
Principal Investigator(s): Nicola Longo
Direct Costs: $15,150 Total Costs: $16,550
Genzyme Corporation
Role: Principal Investigator

07/01/04 - 06/30/06 0455086Y Carnitine in Heart Disease.
Principal Investigator(s): Nicola Longo
Direct Costs: $70,000 Total Costs: $70,000

Role: Principal Investigator
07/01/04 - 06/30/06 Phase 4...Study...Cerezyme Infusions Every 4 Weeks Vs Ev
Principal Investigator(s): Nicola Longo
Direct Costs: $60,133 Total Costs: $60,133
Genzyme Corporation
Role: Principal Investigator
01/01/04 - 12/31/06 NIH-NIDDK R43DK HD60308
Non-invasive phenylalanine monitor
Direct Costs: $100,000 Total Costs: $100,000
National Institute of Diabetes and Digestive and Kidney Diseases
Role: Co-Investigator
09/01/03 - 08/31/05 OPEN-LAPEL...PHARMACOKINETICS...RECOMBINANT HUMAN ACID A
Principal Investigator(s): Nicola Longo
Direct Costs: $84,613 Total Costs: $84,613
Genzyme Corporation
Role: Primary Investigator
04/01/02 - 03/31/03 The Insulin Receptor in Human Growth. Funding Incentive Seed Grant.
Principal Investigator(s): Nicola Longo
Direct Costs: $31,800 Total Costs: $31,800
University of Utah Research Foundation
Role: Principal Investigator
09/01/01 - 05/31/05 THE CARNITINE TRANSPORTER IN HUMAN DISEASE.
Principal Investigator(s): Nicola Longo
Direct Costs: $758,950 Total Costs: $758,950
National Institute of Diabetes and Digestive and Kidney Diseases
Role: Primary Investigator
09/01/00 - 08/31/01 Dean's Clinical Investigator Award
Principal Investigator(s): Nicola Longo
Direct Costs: $50,000 Total Costs: $50,000
Emory University School of Medicine
Role: Principal Investigator
02/01/00 - 01/31/01 Insulin regulation of Cell Growth. Emory University Research Committee
Principal Investigator(s): Nicola Longo
Direct Costs: $30,000 Total Costs: $30,000
Emory University School of Medicine
Role: Principal Investigator
06/01/99 - 05/31/00 Inhibition of carnitine transport by novel pharmaceutical compounds
Principal Investigator(s): Nicola Longo
Direct Costs: $1,350 Total Costs: $1,350
Merck & Co., Inc.
Role: Principal Investigator
04/01/99 - 03/31/00 Carnitine transporters in human disease. Emory Children's Center
Principal Investigator(s): Nicola Longo
Direct Costs: $25,000 Total Costs: $25,000
Emory University School of Medicine
Role: Principal Investigator
09/01/96 - 08/01/97 Genotype-Phenotype correlation in lysyl hydroxylase deficiency (Ehlers-Danlos Syndrome type VI) Research Committee
Direct Costs: $15,000 Total Costs: $15,000
Emory University
Role: Co-Investigator
07/01/96 - 06/30/97 Genes controlling human growth
Principal Investigator(s): Nicola Longo
Direct Costs: $67,000 Total Costs: $67,000
Genentech Foundation For Growth and Development
Role: Principal Investigator
09/01/95 - 02/28/01 NIH R29 DK 48742
Regulation of ion fluxes by the human insulin receptor
Principal Investigator(s): Nicola Longo

Direct Costs: $348,711 Total Costs: $348,711
National Institute of Diabetes and Digestive and Kidney Diseases
Role: Principal Investigator

07/01/93 - 06/30/94 IRG 182-A
The insulin receptor in tumor growth
Principal Investigator(s): Nicola Longo
Direct Costs: $9,000 Total Costs: $9,000
American Cancer Society
Role: Principal Investigator

07/01/92 - 06/30/93 The insulin receptor in inherited insulin resistance. Emory-Egleston CRC
Principal Investigator(s): Nicola Longo
Direct Costs: $23,000 Total Costs: $23,000
Emory University School of Medicine
Role: Principal Investigator

07/01/91 - 06/30/94 Insulin effect on the membrane potential of cells with genetically altered insulin receptors
North Atlantic Treaty Organization
Role: Co-Investigator

07/01/90 - 06/30/91 Insulin and growth factors interactions in the regulation of glucose transporter gene expression. Research Committee
Principal Investigator(s): Nicola Longo
Direct Costs: $4,000 Total Costs: $4,000
Emory University School of Medicine
Role: Principal Investigator

07/01/89 - 06/30/91 Molecular basis for hormonal regulation of glucose transport in human fibroblasts. Emory-Egleston CRC
Principal Investigator(s): Nicola Longo
Direct Costs: $23,000 Total Costs: $23,000
Emory University School of Medicine
Role: Principal Investigator

07/01/88 - 06/30/92 NIH R01-DK 40362
Insulin receptors in heritable insulin-resistance
National Institute of Diabetes and Digestive and Kidney Diseases
Role: Co-Investigator

07/01/86 - 06/30/87 Emory-Egleston CRC
Molecular biology of the insulin receptor
Principal Investigator(s): Nicola Longo
Direct Costs: $10,000 Total Costs: $10,000
Emory University School of Medicine
Role: Principal Investigator

Past Contracts

Contract (Longo) 02/20/14 - 12/31/17 Ultragenyx Pharmaceutical Inc.
OPEN-LABEL PHASE 2 STUDY TO ASSESS SAFETY & CLINICAL EFFECTS OF UX007ON LONG-CHAIN FATTY ACID OXIDATION DISORDERS (LC-FAOD) Ux007-CI201. The objective of tis trial is to determine efficacy of triheptanoin in teh treatment of patients with long-chain fatty acid disorders

Contract (Longo) 06/30/2015-12/31/2018 HORIZON THERAPEUTICS INC
HPN-009 OPEN LABEL STUDY OF SAFETY EFFICACY & PK OF GLYCEROL PHENYLBUTYRATE (GPB; RAVICTI) IN SUBJECTS UNDER TWO With UREA CYCLE DISORDERS. The objective of this trial is to establish effecacy and safety of glycerol phenylbutyrate in patinets with urea cycle defects younger than 2 years of age.

Contract (Longo) 03-15-2014-12-31-2024 Horizon Pharma/United biosource
THRIVE HPN-100-014 UCD REGISTRY The goal of this research is to study the natural history of patients with urea cycle defects treated with glycerol phenylbutyrate

Contract (Longo) 03/02/2016-12/31/2019 RETROPHIN INC
PREVALENCE OF CEREBROTENDINOUS XANTHOMATOSIS (CTX) IN PT POPULATIONS DIAGNOSED W/EARLY-ONSET IDIOPATHIC BILATERAL CATARACTS. The objective of this trial is to determine the prevalence of cerebrotendinous xanthomatosis in patients with bilateral cataracts.

Contract (Longo) 09/01/2008-12/31/2023 BioMarin Pharmaceutical Inc.
PKUDOS- PKU DEMOGRAPHICS, OUTCOMES, AND SAFETY REGISTRY The goal of this research is to study the natural history of patients with phenylketonuria, receiving or not sapropterin.

Contract (Longo) 04/13/2009-12/31/2017 BioMarin Pharmaceutical Inc.
EFFECT OF KUVAN ON NEUROCOGNITIVE FUNCTION OF BLOOD PHENYLALANINE CONCENTRATIONS, SAFETY & POPULATION PHARMACOKINETICS IN PHENYLKETONURIC PATIENTS The goal of this research is to define neuropsychological outcome in patients with phenylketonuria receiving sapropterin

Contract (Longo) 2/01/2008-8/31/2019 BioMarin Pharmaceutical Inc.
PEG-PAL-003, 165-301, 205-165 PHASE 1-3 CLINICAL TRIAL OF rAvPAL-PEG IN PHENYLKETONURIA. The goal of this research is to define whether patients with phenylketonuria respond to therapy with injectable recombinant phenylalanine ammonia lyase (PEGylated).

Contract (Longo) 01/01/2014-01/30/2019 BioMarin Pharmaceutical Inc **MPS VI CLINICAL SURVEILLANCE PROGRAM** The goal of this research is to study the natural history of patients with mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)

Contract (Longo) 02/05/2016-02/15/2021 Genzyme Corp
SUB-REGISTRY OF ANAPHYLAXIS/SEVERE ALLERGIC REACTIONS & SEVERE CUTANEOUS/SYSTEMIC IMMUNE-MEDIATED REACTIONS W/GLUCOSIDASE ALFA the objective of this study is to obtain all immune related reactions in patients with Pompe disease while receiving enzyme replacement therapy.

Contract (Longo) 01/01/2002-12/31/2020 Genzyme Therapeutics Corporation
Lysosomal storage disorder registry. The goal of this research is to study the natural history of patients with lysosomal storage disorders (Gaucher, Fabry, MPS-1, and Pompe disease).

Contract (Longo) 12/01/2012-12/01/2018 Shire/Pharmanet
GAUCHER DISEASE OUTCOME SURVEY (GOS) The goal of this research is to study the natural history of patients with Gaucher disease

Contract (Longo) 12/05/2016-12/31/2020 Protalix Ltd
RANDOM D-B ACTIVE CONTROL STUDY OF PRX-102 VS AGALSIDASE BETA ON RENAL FUNCTION IN FABRY DISEASE PREVIOUSLY TREATED W/ AGALSID This study will test a new type of enzyme replacement therapy in patients with Fabry disease.

Contract (Longo) 1/1/06-12/31/18 Utah Department of Health
Metabolic screening. This project involves the organization of the newborn screening program in the state of Utah and nutritional follow-up by patients identified through screening.

Contract (Longo) 10/1/08-6/30/2019 Nevada Department of Health
Metabolic screening. This project involves the organization of the newborn screening program in the state of Nevada and nutritional follow-up by patients identified through screening.

Contract (Longo) 1/1/2012-06/30/2017 Wyoming Department of Health
Genetic Services. This project involves the provision of genetic services in the state of Wyoming.

Contract (Longo) 1/1/2011-12/31/2018 Children's Specialty Center of Nevada
Genetic Services. This project involves the provision of genetic services to patients in Nevada

Grant-in-Aid 0455086Y (Longo) 07/01/2004-06/30/2007 American Heart Association **Carnitine in heart disease** The goal of this project is to determine whether cardiomyopathy is associated with heterozygous mutations in the carnitine transporter gene.

R43DK HD60308 (Ghanem) 09/01/2002-8/31/2006 NIH/NIDDK
Non-Invasive Blood Phenylalanine Monitor. The goal of this project is to develop a non-invasive device to monitor phenylalanine levels in patients with phenylketonuria.

Contract (Longo) 06/01/2005-5/31/2007 Genzyme Corporation
Multicenter study. Comparison of Q2 versus Q4 weeks treatment in Gaucher disease
In this study, two treatment modalities (every 2 or 4 weeks) for Gaucher disease are compared.

Grant (Longo) 1/1/06-6/30/07
Newborn screening surveillance program HRSA/Mountain States Genetics Network
The goal of this project is to develop a comprehensive plan for population-based outcome surveillance and longitudinally tracking of people identified through genetic screening programs.

Contract (Longo) 11/01/2004-12/31/2009 BioMarin Pharmaceutical Inc.
BMRN-PKU-001 to PKU-008. Multicenter Study to Evaluate the Response to and Safety of Phenoptin™ Treatment in Subjects with Phenylketonuria. The goal of this research is to define whether patients with phenylketonuria respond to tetrahydrobiopterin (BH4).

Contract (Longo) 07/01/2007-12/31/2010 Genzyme Therapeutics Corporation
MYOZYME TEMPORARY ACCESS PROGRAM. The goal of this research is to study the effect of anew form of enzyme replacement therapy for Pompe disease.

Contract (Longo) 01/01/2007-12/31/2010 Shire Therapeutics
A MULITCENTER OPEN LABEL STUDY OF GENE-ACTIVATED HUMAN GLUCOCEREBROSIDASE (GA-GCB) ENZYME REPLACEMENT THERAPY IN PATIENTS WITH GAUCHER DISEASE
The goal of this research is to study the effect of anew form of enzyme replacement therapy for Gaucher disease.

R21 DK 077415-A1 (Longo) 9/01/2007-8/31/2010 NIH/NIDDK/NICHD
Anaplerotic therapy in propionic acidemia
The goal of this grant is to determine whether supplementation with glutamine and ornithine alpha ketoglutarate improves the clinical outcome of patients with propionic acidemia.

1U50DD000483-01 (Botto) 9/01/2008-8/31/2011
Centers for Disease Control and Prevention **Utah Newborn Metabolic Surveillance**
The goal of this project is to establish long-term follow-up for metabolic disorders identified by newborn screening.

Contract (Longo) 07/01/2010-05/31/2012 Target research associates (Pfizer and Protalix)
PLANT CELL EXPRESSED RECOMBINANT HUMAN GLUCOCEREBROSIDASE (PRGCD) IN GAUCHER DISEASE W/ENZYME REPLACEMENT THERAPY. The goal of this research is to determine the efficacy of a new form of enzyme replacement therapy to treat patients with Gaucher disease.

5-R01 DK 53824 (Longo) 6/1/2000-5/31/2013
NIH/NIDDK **The carnitine transporter in human disease** This project has initially characterized mutations in the carnitine transporter gene causing primary carnitine deficiency. The project then characterized genes modifying the clinical presentation of primary carnitine deficiency.

3-R01 DK053824-08S1 (Longo) 6/1/2008-5/31/2010 Minority Supplement Award

Contract (Longo) 01/01/2010-12/31/2013 Amicus Therapeutics Inc.
SAFETY & PHARMACODYNAMICS OF AT1001 IN PATIENTS W/FABRY DISEASE & AT1001-RESPONSIVE GLA MUTATIONS The goal of this research is to determine the efficacy of a new oral drug acting as a chaperone to treat patients with Fabry disease and specific mutations in the alpha galactosidase gene.

Contract (Longo) 06/01/2011-12/31/2013
REPLAGAL ENZYME REPLACEMENT THERAPY IN CHILDREN W FABRY DISEASE NAIVE TO ENZYME REPLACEMENT THERAPY HGT-REP-084 The goal of this research is to determine the efficacy of a new form of enzyme replacement therapy to treat children with Fabry disease.

Contract (Longo) 10/15/2009-12/31/2014 Hyperion Therapeutics .
HPN-100-007 to 011: PHASE 3, OPEN-LABEL STUDY OF SAFETY OF HPN-100 FOR LONG-TERM TREATMENT OF UREA CYCLE DISORDERS (TREAT UCD) The goal of this research is to determine the efficacy of a new drug to treat hyperammonemia in patients with urea cycle defects.

Contract (Longo) 04/01/2010-12/31/2014 Genzyme Therapeutics Corporation

ONCE DAILY VS TWICE DAILY DOSING OF GENZ-112638 IN PATIENTS W/ GAUCHER DISEASE TYPE 1 W/CLINICAL STABILITY AFTER 2X DAILY DOSE. The goal of this research is to determine the efficacy of a new oral drug acting as a substrate-inhibitor to treat patients with Gaucher disease.

2 R44 NS073225-03A1 (Naleway) 03/01/2012-02/28/2016 NIH/NINDS
Live-Cell Assays for Lysosomal Enzyme Activity. The objective of the application is to develop new targeted fluorogenic substrates capable of measuring lysosomal enzyme activity in living cells and tissues. (Role: Consultant)

5R13HD062129-04 (Longo) 7/14/2009-2/28/2016 NIH/NIDDK/NICHD/NINDS
Society for Inherited Metabolic Disorders Annual Meeting
This grant support attendance of young investigators to the SIMD annual meeting

IV. HONORS AND AWARDS

M.D. Summa cum Laude (1982)
Battioni Award for Cancer Research, Parma, Italy (1988)
NIH FIRST Award (1995)
Dean's Clinical Investigator Award, Emory University (2000)
President, Society for Inherited Metabolic Disorders SIMD (2013-2015),
University of Utah Catalyst Investigator (2015)
Visiting Professor, Children's Hospital of Los Angeles (2017)

Others
Compassionate Doctor Award 2011, 2012
America's top Physician 2012-
Patients' Choice Physician 2012-
Castle Connolly Award 2014-

V. ADMINISTRATIVE EXPERIENCE

Emory University School of Medicine

-Chairman, Faculty Committee for Medical Student Research	2000-2001
-Member, Faculty Committee for Medical Student Research	1994-2000
-Member, Human Investigations Committee (3 years)	1997-1999
-Member (ad hoc), Faculty Committee on Appointments and Promotions	1999

Emory University Department of Pediatrics

-Member, Research Committee	1991-2001

Emory University Division of Medical Genetics

-Director of Research	1997-2001

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-Director of Metabolic Services	2001-present
-Co-Director, ARUP Biochemical Genetics Laboratory	2002-present
-Retention, Promotion and Tenure Committee, Dept. Pediatrics	2006-present
-Chief, Division of Medical Genetics	2007-Present

VIII. MEMBERSHIP IN PROFESSIONAL SOCIETIES

-Ordine dei Medici di Parma, Italy (Parma Medical Association) 1983-Present
-American Medical Association 2005-Present
-American Society of Human Genetics (ASHG), 1994-Present
-Society for Inherited Metabolic Disorders (SIMD), Board of Directors, 1994-2017, Membership Chair 2007-2009, Program Chair 2009-2011, President-Elect 2011-2013, President 2013-2015; Past-President 2015-2017.
-Society for the Study of Inborn Errors of Metabolism (SSIEM), 1999-present
-Fellow, American College of Medical Genetics, 1997-Present

IX. TEACHING RESPONSABILITIES

EMORY UNIVERSITY

Responsibility **Course** **Dates**

Small group seminar facilitator **Human and Molecular Genetics** 1989-93
(***MEDI-545 IBS 505***) 1995-2001
Teach to a small group (7-10) of medical and graduate students that meet weekly during this annual course. Duties include attendance to a preparatory lecture before each class (15 hours/year).

Medical Genetics Faculty **Pediatrics** 1989-93, 1995-2001
Teaching to House staff and Medical students during their Pediatrics/Medical Genetics rotation (3 months per year). Medical students are followed in clinic every Tuesday. Regular student examinations.

Voluntary Faculty **Medical Biochemistry (Medi 515/ IBS 528)** 1998-2001
Provide clinical correlations for first year medical students. (4 hours/year, 120 students)

Faculty/Speaker **Seminars in Medical Genetics** (***IBS-782***) 1989-2001
Organize and attend weekly seminars for one month/year and give lectures twice a year.

Faculty. **Research in Medical Genetics** (Elective, Second Year Medical School) 1997-2001
Follow individual students for 4 hours/week during the Spring semester. Teaching includes exposure to laboratory techniques and patients with genetic diseases relevant to the focused research.

Member, **Graduate Division of Biological and Biomedical** 1996-2001
Sciences, Program in ***Cell and Developmental Biology***. Thesis committees.

Co-Director. **Molecular genetic bases of inherited metabolic diseases (IBS 737)** 2000-2001
This course is given in the fall semester to graduate students of the programs in Biochemistry, Genetics, Public Health, and Nutrition. 50 hours per year, 5-8 students/year.

Voluntary Faculty **Allied Health Biochemistry (BAHS 501)** 1997-1999
Lectures on inborn errors of metabolism (3 hours/year, 80 students)

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Medical Genetics Faculty **Pediatrics** **2001-Present**
Teaching to House staff and Medical students during their Pediatrics/Medical Genetics rotation (3 months per year). Medical students are followed in clinic every Monday and Tuesday. Three noon lectures per year are given to the house staff.

Faculty **Human Genetics for Graduate and Medical Students** **2001-Present**
Lectures on medical genetics topics (metabolic disorders, gene therapy) are given to graduate students.

Course Director **HGEN/PATH 7380 Biochemical Genetics** (Fall semester) **2006-Present**
Required course for Genetic Counselors, Fellows in Biochemical and Clinical Genetics, Optional Course for Graduate students in Biochemistry, Human Genetics, Pathology & Laboratory Medicine

Program Faculty **Graduate Program in Genetic Counseling** **2006-Present**
Participates in the education of Genetic Counselor Students at the University of Utah during their rotation in Biochemical and Clinical Genetics.

SUPERVISORY TEACHING

Candidate	**Degree/Activity**	**Date**
EMORY UNIVERSITY		
Lorri D. Griffin MS	MS in Physiology	1987-90

T. Reid Fotion MD	Clinical Post-Doc	1987-90
David M. Byers PhD	Sabbatical	Jan-June 1995
Daniel Giannella-Neto MD PhD	Sabbatical	July-Nov 1996
Fernando Scaglia MD	Clinical Post-Doc	1995-1998
Shelley A. Smith	Medical Student	1997 Summer
Jessica C. Chiang	Undergraduate Student	1997-1998
Michelle A. Kelly MD	Resident	1999
Telly A. Meadows	Medical Student	1999 Summer
Paul B. Pruett	Medical Student	2001 Summer
Misa Graff	Graduate student (Nutrition)	May-July 2001

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Andrea Bartuli MD,	Sabbatical,	June-Aug 2006,
Director, Rare Diseases Program, Ospedale Bambino Gesu', Rome, Italy.		
Pietro Strisciuglio MD,	Sabbatical,	6-7/2008, 9-10/2012
Professor of Pediatrics, University of Naples, II Facolta' di Medicina, Italy.		
Giuseppe Zampino MD,	Sabbatical,	1-4/2010,
Professor/ Catholic University, Rome, Italy		
Nathaniel Miller	Medical Student	2003 Summer
Virginia Beltrama	Medical Student/Univ Milan, Italy	1-2/2010
Valentina Rovelli	Medical Student/Univ Milan, Italy	6-8/2010
Gerarda Cappuccio MD	Resident/Univ. Naples, Italy	8-10/2010
Anna Giulia Cimatti MD	Resident/Univ. Bologna, Italy	10-12/2010
Federica Bedetti MD	Resident/Univ. Bologna, Italy	11/2010-02/2011
Francesca Ciuffini	Medical Student/Univ Milan, Italy	2-5/2011
Anna Cereda MD,	Resident/Univ. Milano, Italy	4-7/2011
Federica Tamburrino MD,	Resident, Univ. Bologna, Italy	5-7/2011
Marta Frigeni	Medical Student/Univ Milan, Italy	2-4/2012, 8-10/2013
Beatrice Crippa	Medical Student/Univ Milan, Italy	2-4/2012, 8-9/2013
Chiara Leoni MD,	Resident, Univ. Rome, Italy	7/2012-7/2013
Angela Peron MD,	Resident, Univ. Milano, Italy	09/2013-08/2014, 01/2016-
Daniela Anderson MD,	Medical Student	2016-2017

Mentee Name and Degree	Time Period as a Mentee	Position While Mentee	Project Title(s)	Capacity of Mentoring	Current Position (if known)
Cristina Amat di San Filippo PhD	2001-2009	Post-Doc	Carnitine Transport	Mentor	Senior Research Associate, University of Utah
Roberta Melis PhD	2001-2004	Post-doc	Insulin receptor signaling	Mentor	Scientist, ARUP Laboratories
Kristin Bryant MS	2002-2004	Nurse Practitioner Student	Newborn screening	Mentor	Nurse Practitioner, Primary Children Hospital
Heather R. Filipowicz	2003-2005	Master Nutrition Student	Propionic Acidemia	Mentor	Dietitian, Intermountain Healthcare, Utah
Larissa Furtado MD	2006 -2008	Fellow, Biochemical Genetics	Metabolic control and psychometric functions in phenylketonuria	Research Mentor, Clinical supervisor.	Assistant Professor of Pathology, University of Utah
Nicholas James Stracensky	08/2006-08/2008	Master Nutrition Student	Propionic Acidemia	Mentor	Medical Student, University of Utah
Lucia Santoro MD	09/2007-03/2008	Resident, University of Ancona, Italy	Definition of genetic syndromes	Research and clinical supervisor	Assistant Professor of Pediatrics, University of Ancona, Italy
Lorenzo Botto MD	2007-2008	Fellow, Biochemical Genetics	Epidemiology of metabolic disorders	Research Mentor, Clinical supervisor	Professor of Pediatrics, University of Utah

Uzochi Chimdinma Ndukwe Erlingsson MD	2008-2011	Post-doc	Carnitine and fatty acid oxidation	Research Mentor	Research Scientist, Broad Institute, Boston, MA
Krista Viau MS	2008-2015	Public Health PhD Student	Psychometric outcome in phenylketonuria	Research and clinical Mentor	Metabolic Dietitian, Boston Children Hospital
JoDell E. Whittington, PhD	2010-2012	Fellow, Biochemical Genetics	Newborn screening in phenylketonuria	Research Mentor, Clinical supervisor	Medical Director, Quest Diagnostic, Chantilly, VA
Francesco Iacobazzi MS, PhD	2011	Pharmacology PhD Student	Fatty acid oxidation	Research mentor	Program Manager, Merck Serono, Bari, Italy
Tatiana Yuzyuk PhD	2010-2013	Fellow, Biochemical Genetics	Creatine deficiency syndromes	Mentor, Clinical supervisor	Medical Director, ARUP laboratories, Salt Lake City, UT
Amy Calhoun MD	2009-2012	Fellow, clinical Genetics	Definition of genetic syndromes	Mentor, Clinical supervisor	Assistant Professor, University of Iowa
Eyby Leon MD	2010-2012	Fellow, Clinical and Biochemical Genetics	Natural history of lysosomal storage disorders	Mentor, Clinical supervisor	Assistant Professor, Children's National Medical Center
Leisa Price	08/2011-05/2013	Nutrition MS Student	Propionic acidemia	Mentor	Dietitian, Logan, UT
Elisa Ballarini MD	05/2014-11/2014	Pediatric Resident, University of Bologna, Italy	Multiple AcylCoA Dehydrogenase deficiency	Mentor	Pediatrician, Brescia, Italy
Hunter Underhill MD PhD	2014-2015	Fellow, Clinical and Biochemical Genetics	Brain imaging in Lysosomal storage disorders	Mentor, Clinical supervisor	Associate Professor, University of Utah
Marta Frigeni MD	2014-2017	Post-Doctoral Fellow	Carnitine transport and fatty acid oxidation	Mentor	Resident, Pediatrics/Medical Genetics, Houston, TX
Filippo Ingoglia	2016	PhD Student	Carnitine transport	Mentor	Fellow, Biochemical Genetics, University of Utah
Bijina Balakrishnan PhD	07/2015-	Post-Doctoral Fellow	Carnitine transport	Mentor	
Leon Chong, PhD	07/2016-08/2018	Fellow, Biochemical Genetics	Arginase deficiency	Mentor	Medical Director, LabCorp, NC
Francesca Manzoni MD	04/2017-12/2017 and 01/2019-05/2019	Neurology Resident, University of Milan, Italy	PTPS Deficiency	Mentor	Attending Neurologist, Padua University Hospital, Italy
Valentina Rovelli MD	01/2018-06/2018	Pediatrics Resident, University of Milan, Italy	VLCAD Deficiency	Mentor	Attending Physician, San Paolo Hospital, Milan, Italy
Chelsea Norman	2017-2019	Nutrition MS Student	Phenylketonuria therapies	Mentor	Associate Scientific Director at Lockwood
Katherine J. Anderson, MD	2018-2020	Genetics Resident	Urea Cycle disorders	Mentor	Assistant Professor and Chief of Medical Genetics, Univ Vermont
Amy Nicole Lebedoff, MD	2018-2020	Genetics Resident	Trisomy 18	Mentor	Assistant Professor, Children's Minnesota
Filippo Ingoglia, PhD	07/2018-06/2021	Fellow, Biochemical	Creatine deficiency and urea cycle	Mentor	Assistant Professor of Pathology, Utah

		Genetics			
Aneta Kaczmarczyk PhD	07/2019-06/2021	Fellow, Biochemical Genetics	Glycosaminoglycans in mucopolysaccharidoses	Mentor	
Laura Duque Lasio MD	07/2020-06/2022	Fellow, Biochemical Genetics	Glycosaminoglycans in mucopolysaccharidoses	Mentor	

RESEARCH FOCUS

-***Molecular genetics of inherited insulin-resistant syndromes***. The objective of this research is to understand how mutations in the insulin receptor gene affect human growth and metabolism by modifying insulin regulation of membrane transport and mitogenesis.

-***Molecular basis of carnitine deficiency***. The objective of this research is to identify genes involved in carnitine transport and metabolism and define mutations in these genes in patients with inherited fatty acid oxidation defects. Different transporters for carnitine are analyzed at the functional and molecular level in patients with carnitine deficiency and related disorders. Proteins interacting with the carnitine transporter are identified using the yeast two-hybrid screen. Variations in their sequence are sought in patients with carnitine deficiency, but no mutations in the carnitine transporter gene.

-Therapy of metabolic disorders. The objective of this research is to develop and test new therapies for patients with inborn errors of metabolism. The approaches followed include use of vitamin/cofactor supplements, novel drugs capable of bypassing metabolic blocks, infusion of recombinant enzymes is used in patients with storage disorders, and therapy with nutritional supplements to replace products of defective biochemical reactions.

-***Expanded newborn screening program***. The objective of this research is to define parameters to follow the long-term outcome of patients identified presymptomatically by expanded newborn screening programs.

***-Brain creatine deficiency syndromes*.** The objective of this research is to determine the mechanism by which lack of creatine impairs brain function and to devise new therapies to prevent intellectual disability in affected patients.

PUBLICATIONS OF DR. NICOLA LONGO

PEER-REVIEWED

1.Dall'Asta V, Gazzola GC, Franchi-Gazzola R, Bussolati O, **Longo N**, Guidotti GG (1983) Pathways of L-glutamic acid transport in cultured human fibroblasts. ***J Biol Chem*** *258*: 6371-6379.

2.Gazzola GC, Dall'Asta V, Franchi-Gazzola R, Bussolati O, **Longo N**, Guidotti GG (1984) Post-translational control by carrier availability of amino acid transport in fetal human fibroblasts. ***Biochem Biophys Res Commun*** *120*: 172-178.

3.Gazzola GC, Bussolati O, **Longo N**, Dall'Asta V, Franchi-Gazzola R, Guidotti GG (1984) Effect of *in vitro* ageing on the transport of neutral amino acids in human fibroblasts. in **Cellular Ageing** (Sauer HW ed), ***Monogr Devl Biol*** (Karger, Basel) *17*: 234-244.

4.Dall'Asta V, Franchi-Gazzola R, Bussolati O, **Longo N**, Gazzola GC (1984) Recognition properties of Na^+-dependent amino acid transport systems in cultured human fibroblasts. ***Ital J Biochem*** *33*: 208A-209A.

5.**Longo N**, Franchi-Gazzola R, Bussolati O, Dall'Asta V, Foà PP, Guidotti GG, Gazzola GC (1985) Effect of insulin on the activity of amino acid transport systems in cultured human fibroblasts. ***Biochim Biophys Acta*** *844*: 216-223.

6.Bussolati O, Laris PC, **Longo N**, Dall'Asta V, Franchi-Gazzola R, Guidotti GG, Gazzola GC (1986) Effect of extracellular potassium on amino acid transport and membrane potential in fetal human fibroblasts. ***Biochim Biophys Acta*** *854*: 240-250.

7.Dall'Asta V, Gazzola GC, **Longo N**, Bussolati O, Franchi-Gazzola R, Guidotti GG (1986) Perturbation of Na^+ and K^+ gradients in human fibroblasts incubated in unsupplemented saline solutions. ***Biochim Biophys Acta*** *860*: 1-8.

8.Dall'Asta V, Rebecchi F, **Longo N**, Bussolati O, Gazzola GC (1986) Serum dependent changes of intracellular Na^+ and K^+ concentrations in cultured human fibroblasts. ***Cell Biol Int Rep*** *10*(3): 156.

9.Bussolati O, Laris PC, Squassoni E, Ferrari MG, **Longo N**, Dall'Asta V, Guidotti GG (1986) Na,K-ATPase-mediated hyperpolarization stimulates amino acid transport in cultured human fibroblasts. ***Cell Biol Int Rep*** *10*(3): 157.

10.Elsas LJ, **Longo N** (1987) Impaired insulin binding and excess glucose transport in fibroblasts from a patient with leprechaunism. ***Enzyme*** *38*: 184-193.

11.Endo F, Nagata N, Priest JH, **Longo N**, Elsas LJ (1987) Structural analysis of normal and mutant insulin receptors in fibroblasts cultured from families with leprechaunism. ***Am J Hum Genet*** *41*: 402-417.

12.Bussolati O, Laris PG, Nucci FA, Dall'Asta V, **Longo N**, Guidotti GG, Gazzola GC (1987) The dependence of L-arginine accumulation on the membrane potential in cultured human fibroblasts. ***Am J Physiol*** (Cell 22) *253*: C391-C397.

13.**Longo N**, Griffin LD, Elsas LJ (1988) Influx and efflux of 3-O-methyl-D-glucose by cultured human fibroblasts. ***Am J Physiol*** (Cell 23) *254*: C628-C633.

14.**Longo N**, Franchi-Gazzola R, Bussolati O, Dall'Asta V, Nucci FA, Gazzola GC (1988) Amino acid and sugar transport in mouse 3T3 cells expressing activated *ras* and *neu* oncogenes. ***Ann NY Acad Sci*** *551*: 374-377.

15.**Longo N**, Franchi-Gazzola R, Bussolati O, Dall'Asta V, Nucci FA, Spisni A, Gazzola GC (1988) Glycine transport by cultured human fibroblasts. ***Biochem Biophys Res Commun*** *152*: 617-622.

16.**Longo N**, Griffin LD, Shuster RC, Langley S, Elsas LJ (1989) Increased glucose transport by human fibroblasts with a heritable defect in insulin binding. ***Metabolism*** *38*: 690-697.

17.**Longo N**, Fotion TR, Langley S, Elsas LJ (1989) Restriction fragment length polymorphisms of the insulin receptor gene in families with insulin resistance and leprechaunism. ***Am J Med Sci*** *298*: 366-370.

18.**Longo N**, Shuster RC, Griffin LD, Elsas LJ (1990) Insulin-receptor autophosphorylation and kinase activity are constitutively increased in fibroblasts cultured from a patient with heritable insulin resistance. ***Biochem Biophys Res Commun*** *167*: 1229-1234.

19.**Longo N**, Griffin LD, Elsas LJ (1991) A simple method for the evaluation of Rb^+ influx and Na,K-pump stochiometry in adherent cells. ***Am J Physiol*** (Cell 29) *260*: C1341-C1346.

20.**Longo N**, Griffin LD, Langley SD, Elsas LJ (1992) Glucose transport by cultured human fibroblasts: regulation by phorbol esters and insulin. ***Biochim Biophys Acta*** *1104*: 24-30.

21.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1992) Reduced mRNA and a nonsense mutation in the insulin receptor gene produce heritable severe insulin resistance. ***Am J Hum Genet*** *50*: 998-1007.

22.**Longo N**, Shuster RC, Griffin LD, Langley SD, Elsas LJ (1992) Activation of insulin receptor signaling by a single amino acid substitution in the transmembrane domain. ***J Biol Chem*** *267*: 12416-12419.

23.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1993) Activation of glucose transport by a natural mutation in the human insulin receptor. ***Proc Natl Acad Sci USA*** 90: 60-64.

24.**Longo N** (1993) Defective receptors for Platelet-Derived Growth Factor AA in human fibroblasts with mutant insulin receptors. ***Biochem Biophys Res Commun*** 197: 812-817.

25.**Longo N**, Singh R, Griffin LD, Langley SD, Parks JS, Elsas LJ (1994) Impaired growth in Rabson-Mendenhall syndrome: Lack of effect of growth hormone and Insulin-like Growth Factor I. ***J Clin Endocrinol Metab*** 79: 799-805.

26.Ekstrand AJ, **Longo N**, Hamid ML, Olson JJ, Collins VP, James CD (1994) Functional characterization of an EGF receptor with a truncated extracellular domain expressed in glioblastomas with EGFR gene amplification. ***Oncogene*** 9: 2313-2320.

27.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1995) Two mutations in the insulin receptor gene of a patient with leprechaunism: Application to prenatal diagnosis. ***J Clin Endocrinol Metab*** 80: 1496-1501.

28.Ekstrand AJ, Liu L, He J, Hamid M, **Longo N**, Collins VP, James CD (1995) Altered subcellular location of an activated and tumour-associated EGF receptor. ***Oncogene*** 10: 1455-1460.

29.**Longo N**, Langley SD, Still MJ, Elsas LJ (1995) Prenatal analysis of the insulin receptor gene in a family with leprechaunism. ***Prenatal Diagnosis*** 15: 1070-1074.

30.**Longo N** (1996) Insulin stimulates the Na,K-ATPase and the Na/K/Cl cotransporter of human fibroblasts. ***Biochim Biophys Acta*** 1381: 28-44.

31.**Longo N**, Langley SD, Still MJ, Elsas LJ (1996) Mutational analysis of the insulin receptor: Arginine 86 plays an essential role in insulin binding and stimulation of glucose transport. ***Exp Clin Endocrinol Diabetes*** 104: 78-80.

32.Pasquali M, Still MJ, Vales T, Rosen RI, Evinger JD, Dembure PP, **Longo N**, Elsas LJ (1997) Abnormal formation of collagen cross-links in skin fibroblasts cultured from patients with Ehlers-Danlos syndrome type VI. ***Proc Assoc Am Physicians*** 109: 33-41.

33.**Longo N**, Langley SD, Still M (1998) Role of arginine 86 of the insulin receptor in insulin binding and activation of glucose transport. ***Biochim Biophys Acta*** 1402:86-94.

34.Scaglia F, Wang Y, Singh RH, Dembure PP, Pasquali M, Fernhoff PM, **Longo N** (1998) Defective urinary carnitine transport in heterozygotes for primary carnitine deficiency. ***Genet Med*** 1: 34-39.

35. Scaglia F, Wang Y, **Longo N** (1999) Functional characterization of the carnitine transporter defective in primary carnitine deficiency. ***Arch Biochem Biophys*** 364: 99-106**.**

36. Wang Y, Ye J, Ganapathy V, **Longo N** (1999) Mutations in the organic cation/carnitine transporter OCTN2 in primary carnitine deficiency. ***Proc Natl Acad Sci USA*** 96: 2356-2360.

37. **Longo N**, Wang Y, Pasquali M (1999) Progressive decline in insulin levels in Rabson-Mendenhall syndrome. ***J Clin Endocrinol Metab*** 84: 2623-2629.

38. Wang Y, Kelly MA, Cowan TM, **Longo N** (2000) A missense mutation in the OCTN2 gene associated with residual carnitine transport activity. ***Hum Mutat*** 15: 238-245. PubMed PMID: 10679939

39. Melendez PA, **Longo N**, Jimenez B, Cadilla CL (2000) Insulin-induced gene 33 mRNA expression in Chinese hamster ovary cells is insulin receptor dependent. ***J Cell Biochem*** 77: 432-444.

40. Wang Y, Meadows TA, **Longo N** (2000) Abnormal sodium stimulation of carnitine transport in primary carnitine deficiency. ***J Biol Chem*** 275: 20782-20786. PubMed PMID: 10783384

41. Wang Y, Taroni F, Garavaglia B, **Longo N** (2000) Functional Analysis of Mutations in the OCTN2 Transporter Causing Primary Carnitine Deficiency: Lack of Genotype-Phenotype Correlation. ***Hum Mutat*** 16: 401-407. PubMed PMID: 11058897.

42. **Longo N**, Scaglia F, Wang Y (2001) Insulin increases the turnover rate of Na,K-ATPase in human fibroblasts. ***Am J Physiol*** (Cell) 280: C912-C919.

43. **Longo N**, Singh R, Elsas LJ (2001) Decreased half-life of Insulin-like Growth Factor-I in Rabson-Mendenhall syndrome. ***J Inherit Metab Dis*** 24: 546-550.

44.Wang Y, Korman SH, Ye J, Gargus JJ, Gutman A, Taroni F, Garavaglia B, **Longo N** (2001) Phenotype and Genotype Variation in Primary Carnitine Deficiency. ***Genet Med*** 3: 387-392. PubMed PMID: 11715001

45.**Longo N**, Wang Y, Smith SA, Langley SD, DiMeglio LA, Giannella-Neto D (2002) Genotype-phenotype correlation in inherited severe insulin-resistance. ***Human Molec Genet*** *11:* 1465-1475**.**

46.Melis R, Pruett P, Wang Y, **Longo N** (2003) Gene expression in human cells with mutant insulin receptors. ***Biochem Biophys Res Commun*** 307: 1013-1020.

47. Amat di San Filippo C, Wang Y, **Longo N** (2003) Functional domains in the carnitine transporter OCTN2 defective in primary carnitine deficiency. ***J Biol Chem*** 278: 47776-47784. Epub 2003 Sep 23. PubMed PMID: 14506273

48.Iacobazzi V, Pasquali M, Singh R, Matern D, Rinaldo P, Amat di San Filippo C, Palmieri F, **Longo N** (2004) Response to Therapy in Carnitine/Acylcarnitine Translocase (CACT) Deficiency due to a Novel Missense Mutation. ***Am J Med Genet*** 126A(2):150-155. PubMed PMID: 15057979.

49.Amat di San Filippo C, **Longo N** (2004) Tyrosine residues affecting sodium stimulation of carnitine transport in the OCTN2 carnitine/organic cation transporter. ***J Biol Chem*** 279: 7247-7253. Epub 2003 Dec 9. PubMed PMID: 14665638.

50. Stevenson DA, Brothman AR, Carey JC, Chen Z, Dent KM, Bale JF Jr, **Longo N** (2004) 6q subtelomeric deletion: is there a recognizable syndrome? ***Clin Dysmorphol*** 13(2):103-106.

51. McKinney JT, **Longo N**, Hahn SH, Matern D, Rinaldo P, Strauss AW, Dobrowolski SF (2004) Rapid, comprehensive screening of the human medium chain acyl-CoA dehydrogenase gene. ***Mol Genet Metab*** 82:112-120.

52. Stevenson DA, Brothman AR, Chen Z, Bayrak-Toydemir P, **Longo N** (2004) Uniparental disomy of chromosome 14: confirmation of a clinically-recognizable phenotype. ***Am J Med Genet*** 130A:88-91.

53. **Longo N,** Fukao T, Singh R, Pasquali M, Barrios RG, Kondo N, Gibson KM (2004) Succinyl-CoA:3-Keto Acid Transferase (SCOT) Deficiency in a New Patient Homozygous for an R217X mutation. ***J Inherit Metab Dis*** 27(5): 691-692.

54. Makhseed N, Vallance HD, Potter M, Waters PJ, Wong LTK, Lillquist Y, Pasquali M, Amat di San Filippo C, **Longo N** (2004) Carnitine transporter defect due to a novel mutation in the *SLC22A5* gene presenting with peripheral neuropathy. ***J Inherit Metab Dis*** 27(6): 778-780. PubMed PMID: 15617188.

55. Dobrowolski SF, McKinney JT, Amat di San Filippo C, Sim KG, Wilcken B, **Longo N** (2005) Validation of dye-binding/high-resolution thermal denaturation for the identification of mutations in the *SLC22A5* gene. ***Hum Mutat*** 25(3):306-313. PubMed PMID: 15714519.

56. Garavelli L, Cerruti-Mainardi P, Virdis R, Pedori S, Pastore G, Godi M, Provera S, Rauch A, Zweier C, Zollino M, Banchini G, **Longo N**, Neri G, Bernasconi S (2005) Genitourinary Anomalies in Mowat-Wilson Syndrome with deletion/mutation in the Zinc Finger Homeo Box 1B Gene (ZFHX1B): Report of 3 Italian Cases with Hypospadias and Review. ***Hormone Res*** 63(4):187-92.

57. Park HS, Jin DK, Shin SM, Jang MK, **Longo N**, Park JW, Bae DS, Bae YS (2005) Impaired Generation of Reactive Oxygen Species in Leprechaunism through Downregulation of Nox4. ***Diabetes*** 54(11): 3175-81.

58. Filipowicz HR, Ernst SL, Ashurst CL, Pasquali M, **Longo N** (2006) Metabolic changes associated with hyperammonemia in patients with propionic acidemia. ***Molec Genet Metab*** 88(2): 123-130.

59. Amat di San Filippo C, Pasquali M, **Longo N** (2006) Pharmacological rescue of carnitine transport in primary carnitine deficiency. ***Hum Mutat*** 27(6): 513-23. PubMed PMID: 16652335.

60. Dobson CM, Gradinger A, **Longo N**, Wu X, Leclerc D, Lerner-Ellis J, Lemieux M, Belair C, Watkins D, Rosenblatt DS, Gravel RA (2006) Homozygous nonsense mutation in the *MCEE* gene and siRNA suppression of methylmalonyl-CoA epimerase expression: A novel cause of mild methylmalonic aciduria. ***Molec Genet Metab*** 88: 327-333.

61. Toydemir RM, Brassington AE, Bayrak-Toydemir P, Krakowiak PA, Jorde LB, Whitby FG, **Longo N**, Viskochil DH, Carey JC, Bamshad MJ (2006) A novel mutation in *FGFR3* causes camptodactyly, tall stature, and hearing loss (CATSHL) syndrome. ***Am J Hum Genet*** 79: 935-941.

62. Schimmenti LA, Crombez EA, Schwahn BC, Heese BA , Wood TC, Schroer RJ, Bentler K, Cederbaum S, Sarafoglou K, McCann M, Rinaldo P, Matern D, Amat di San Filippo C, Pasquali M, Berry SA, **Longo N** (2007) Expanded newborn screening identifies maternal primary carnitine deficiency. ***Molec Genet Metab*** 90(4):441-5. PubMed PMID: 17126586.

63. Calderon FR, Nelson L, Dobrowolski P, Sinitsyna I, Phansalkar A, **Longo N**, Pasquali M, Mao R (2007) Combination of enzyme analysis, allele-specific PCR and sequencing to detect mutations in the GALT gene. ***J Inherit Metab Dis*** 30(5):818 (online)

64. **Longo N**, Li SK, Yan G, Kochambilli RP, Papangkorn K, Berglund D, Ghanem A-H, Ashurst CL, Ernst SL, Pasquali M, Higuchi WI (2007) Noninvasive Measurement of Phenylalanine by Iontophoretic Extraction in Patients with Phenylketonuria. ***J Inherit Metab Dis*** 30(6):910-5.

65. Dimmock DP, Zhang Q, Dionisi-Vici C, Carrozzo R, Shieh J, Truong C, Schmitt E, Sifry-Platt M, Lucioli S, Santorelli FM, Ficicioglu CH, Rodriguez M, Wierenga K, Enns GM, Arch EM, **Longo N**, Lipson MH, Vallance H, Craigen WJ, Scaglia F, Wong L-J (2008) Clinical and Molecular Features of Mitochondrial DNA Depletion due to Mutations in Deoxyguanosine Kinase. ***Hum Mutat*** 29(2):330-1

66. Arnold GL, Koeberl DD, Matern D, Barshop B, Braverman N, Burton B, Cederbaum S, Fiegenbaum A, Garganta C, Gibson J, Goodman SI, Harding C, Kahler S, Kronn D, **Longo N** (2008) A Delphi-based consensus clinical practice protocol for the diagnosis and management of 3-methylcrotonyl CoA carboxylase deficiency. ***Mol Genet Metab*** 93(4):363-70

67. **Longo N,** Schrijver I, Vogel H, Pique LM, Cowan TM, Pasquali M, Steinberg GK, Hedlund GL, Ernst SL, Gallagher RC, Enns GM (2008) Progressive cerebral vascular degeneration with mitochondrial encephalopathy. ***Am J Med Genet* A** 146(3):361-7.

68. Amat di San Filippo C, Taylor MR, Mestroni L, Botto LD, **Longo N** (2008) Cardiomyopathy and carnitine deficiency. ***Mol Genet Metab*** 94(2):162-6. PubMed PMID: 18337137; PubMed Central PMCID: PMC2430214

69. Lee P, Treacy EP, Crombez E, Wasserstein M, Waber L, Wolff J, Wendel U, Dorenbaum A, Bebchuk J, Christ-Schmidt H, Seashore M, Giovannini M, Burton BK, Morris AA; Sapropterin Research Group (Clarke L, Feigenbaum A, Abadie V, Dobbelaere D, Feillet F, Hennermann J, Trefz F, Meli C, Milanowski A, Chakrapani A, Cleary M, Baker J, Bergoffen J, Grange D, Harding C, Harmatz P, Koch R, Levy H, Lipson L, **Longo N**, Randolph L, Vockley G, Whitley C.) (2008) Safety and efficacy of 22 weeks of treatment with sapropterin dihydrochloride in patients with phenylketonuria. ***Am J Med Genet A*** 146A(22):2851-9.

70. Trefz FK, Burton BK, **Longo N**, Casanova MM, Gruskin DJ, Dorenbaum A, Kakkis ED, Crombez EA, Grange DK, Harmatz P, Lipson MH, Milanowski A, Randolph LM, Vockley J, Whitley CB, Wolff JA, Bebchuk J, Christ-Schmidt H, Hennermann JB; Sapropterin Study Group. (2009) Efficacy of Sapropterin Dihydrochloride in Increasing Phenylalanine Tolerance in Children with Phenylketonuria: A Phase III, Randomized, Double-blind, Placebo-controlled Study. ***J Pediatrics*** 154(5):700-7

71. Arnold GL, VanHove J, Freedenberg D, Strauss A, **Longo N,** Burton B, Garganta C, Ficicioglu C, Cederbaum S, Harding C, Boles RG, Matern D, Fiegenbaum A (2009) A Delphi Clinical Practice Protocol for the Management of Very Long Chain Acyl-CoA Dehydrogenase Deficiency. ***Mol Genet Metab*** 96(3):85-90).

72. Botkin JR, Anderson R, Staes C, **Longo N** (2009) Developing a National Registry for Conditions Identifiable Through Newborn Screening. ***Genet Med*** 11(3): 176-182

73. Schwarz E, Liu A, Randall H, Haslip C, Keune F, Murray M, **Longo N**, Pasquali M (2009) Use of Steroid Profiling by UPLC-MS/MS as a Second Tier Test in Newborn Screening for Congenital Adrenal Hyperplasia: the Utah experience. ***Ped Res*** 66(2):230-5.

74. Ardon O, Amat di San Filippo C, Salomons GS, **Longo N**. 2010. Creatine transporter deficiency in two half-brothers. ***Am J Med Genet*** 152A(8):1979-83.

75. Salek J, Byrne J, Box T, **Longo N**, Sussman N. 2010. Recurrent liver failure in a 25-year-old female. ***Liver Transplantation*** 16:1049-1053.

76. Kronn D, Mofidi S, Braverman N, Harris K; Diagnostics Guidelines Work Group (Adams D, Arnold G, Berry S, Brooks S, Desposito F, Diaz G, Ficiogliu C, Gallagher R, Galvin-Parton P, Garganta C, Gartzke M, Greene C, Levy P, Levy-Fisch J, Lichter-Konecki U, Lim-Melia E, **Longo N**, Martin M, Matern D, McCurdy K, Pasquali M, Pass K, Rapaport R, Singh R, Speiser P, Stein V, Suchy S, and Wilson W) . 2010. Diagnostic guidelines for newborns who screen positive in newborn screening. ***Genet Med***. 12(12 Suppl):S251-5.

77. Wright EL, Van Hove JL, Thomas J; Mountain States Metabolic Consortium (Aleck K, Bernstein L, Ernst S, Freedenberg D, Freehauf C, Gallagher R, Gibson J, Heidenreich R, Kaye C, Leonard C, **Longo N**, Pasquali M, Root S). 2010. Mountain states genetics regional collaborative center's metabolic newborn screening long-term follow-up study: a collaborative multi-site approach to newborn screening outcomes research. ***Genet Med***. 12(12 Suppl):S228-41.

78. Amat di San Filippo C, Ardon O, **Longo N**. 2011. Glycosylation of the OCTN2 carnitine transporter: Study of natural mutations identified in patients with primary carnitine deficiency. ***Biochim Biophys Acta*** 1812(3): 312-320. Epub 2010 Nov 29. PubMed PMID: 21126579; PubMed Central PMCID: PMC3026072.

79. Beaudoin DE, **Longo N**, Logan RA, Jones JP, Mitchell JA. 2011. Using Information Prescriptions to Refer Patients with Metabolic Conditions to the Genetics Home Reference Web Site. ***Journal Med Library Assoc*** 99(1): 70-76.

80. Viau KS, Wengreen HJ, Ernst SL, Cantor NL, Furtado LV, **Longo N**. 2011. Correlation of age-specific phenylalanine levels with intellectual outcome in patients with phenylketonuria. ***J Inherit Metab Dis*** 34(4):963-71. PMID: 21556836

81. De Biase I, Champaigne NL, Schroer R, Pollard LM, **Longo N**, Wood TC. 2012. Primary carnitine deficiency presents atypically with long QT syndrome: A case report. ***J Inherit Metab Dis*** 34: 1242-1243 JIMD Rep. 2012;2:87-90. doi: 10.1007/8904_2011_52. Epub 2011 Sep 6. PubMed PMID: 23430858; PubMed Central PMCID: PMC3509843

82. Rose EC, Amat di San Filippo C, Erlingsson UCN, Ardon O, Pasquali M, **Longo N**. 2012. Genotype-phenotype correlation in primary carnitine deficiency. ***Hum Mutat*** 33: 118-123. doi: 10.1002/humu.21607. Epub 2011 Oct 11. PubMed PMID: 21922592; PubMed Central PMCID: PMC3240685.

83. Jumbo-Lucioni PP, Garber K, Kiel J, Baric I, Berry GT, Bosch A, Burlina A, Chiesa A, Couce Pico ML, Estrada SC, Henderson H, Leslie N, **Longo N**, Morris AAM, Ramirez-Farias C, Scheweitzer-Krantz S, Silao CLT, Vela-Amieva M, Waisbren S, Fridovich-Keil JL. 2012. Diversity of approaches to classic galactosemia around the world: a comparison of diagnosis, intervention, and outcomes. ***J Inherit Metab Dis*** 35(6):1037-49. PMID: 22450714

84. Viau K, Ernst SL, Vanzo RJ, Botto LD, Pasquali M, **Longo N**. 2012. Glutaric acidemia type I: Outcomes before and after newborn screening. ***Molec Genet Metab*** 106 (2012) 430–438. PMID: 22728054

85. Coelho D, Kim JC, Miousse IR, Fung S, du Moulin M, Buers I, Suormala T, Burda P, Frapolli M, Stucki M, Nürnberg P, Thiele H, Robenek H, Höhne W, **Longo N**, Pasquali M, Mengel E, Watkins D, Shoubridge EA, Majewski J, Rosenblatt DS, Fowler B, Rutsch F, Baumgartner MR. Mutations in ABCD4 cause a new inborn error of vitamin B12 metabolism. ***Nature Genetics*** 2012, Oct; 44(10):1152-5. PMID: 22922874

86. Mokhtarani M, Diaz GA, Rhead W, Lichter-Konecki U, Bartley J, Feigenbaum A, **Longo N**, Berquist W, Berry SA, Gallagher R, Bartholomew D, Harding CO, Korson MS, McCandless SE, Smith W, Vockley J, Bart S, Kronn D, Zori R, Cederbaum S, Dorrani N, Merritt JL 2nd, Sreenath-Nagamani S, Summar M, Lemons C, Dickinson K, Coakley DF, Moors TL, Lee B, Scharschmidt BF. 2012. Urinary phenylacetylglutamine as dosing biomarker for patients with urea cycle disorders. ***Mol Genet Metab***. Nov:107(3):308-14. PMID: 22958974

87. Diaz GA, Krivitzky LS, Mokhtarani M, Rhead W, Bartley J, Feigenbaum A, **Longo N**, Berquist W, Berry SA, Gallagher R, Lichter-Konecki U, Bartholomew D, Harding CO, Cederbaum S, McCandless SE, Smith W, Vockley G, Bart SA, Korson MS, Kronn D, Zori R, Merritt JL, Sreenath-Nagamani S, Mauney J, Lemons C, Dickinson K, Moors TL, Coakley DF, Scharschmidt BF, Lee B. Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. 2013. ***Hepatology*** Jun;57(6):2171-9. doi: 10.1002/hep.26058. Epub 2013 Jan 3. PMID: 22961727

88. Burton BK, Leviton L, Vespa H, Coon H, **Longo N**, Lundy BD; Johnson M, Angelino A, Hamosh A, Bilder D. 2013. A Diversified Approach for PKU Treatment: Routine Screening Yields High Incidence of Psychiatric Distress in Phenylketonuria Clinics. ***Mol Genet Metab*** Jan 108(1):8-12. PMID: 23266195

89. Bilder D, Burton BK, Coon H, Leviton L, Ashworth J, Lundy B, Vespa H, **Longo N.** 2013. Psychiatric Symptoms in Adults with Phenylketonuria. ***Mol Genet Metab***, Mar; 108(3):155-60. doi: 10.1016/j.ymgme.2012.12.006. PMID: 23339767

90. Zimran A, Pastores GM, Tylki-Szymanska A, Hughes DA, Elstein D, Mardach R, Eng C, Smith L, Heisel-Kurth M, Charrow J, Harmatz P, Fernhoff P, Rhead W, **Longo N**, Giraldo P, Ruiz JA, Zahrieh D, Crombez E, Grabowski GA. 2013. Safety and efficacy of velaglucerase alfa in Gaucher disease type 1 patients previously treated with imiglucerase. ***Am J Hematol***. Mar;88(3):172-8. doi: 10.1002/ajh.23383. PMID: 23339116

91. van de Kamp JM, Betsalel OT, Mercimek-Mahmutoglu S, Abulhoul L, Grünewald S, Anselm I, Azzouz H, Bratkovic D, de Brouwer A, Hamel B, Kleefstra T, Yntema H, Campistol J, Vilaseca MA, Cheillan D, D'Hooghe M, Diogo L, Garcia P, Valongo C, Fonseca M, Frints S, Wilcken B, von der Haar S, Meijers-Heijboer HE, Hofstede F, Johnson D, Kant SG, Lion-Francois L, Pitelet G, **Longo N**, Maat-Kievit JA, Monteiro JP, Munnich A, Muntau AC, Nassogne MC, Osaka H, Ounap K, Pinard JM, Quijano-Roy S, Poggenburg I, Poplawski N, Abdul-Rahman O, Ribes A, Arias A, Yaplito-Lee J, Schulze A, Schwartz CE, Schwenger S, Soares G, Sznajer Y, Valayannopoulos V, Van Esch H, Waltz S, Wamelink MM, Pouwels PJ, Errami A, van der Knaap MS, Jakobs C, Mancini GM, Salomons GS.2013. Phenotype and genotype in 101 males with X-linked creatine transporter deficiency. ***J Med Genet*** 2013 Jul;50(7):463-72. doi: 10.1136/jmedgenet-2013-101658. Epub 2013 May 3. PMID: 23644449

92. Pasquali M, **Longo N**. Response to Chen et Al.: carnitine uptake defect (primary carnitine deficiency): risk in genotype-phenotype correlation. ***Hum Mutat***. 2013 Apr;34(4):656. doi: 10.1002/humu.22285. PubMed PMID: 23520116.

93. Erlingsson UCN, Iacobazzi F, Liu A, Ardon O, Pasquali M, **Longo N**. 2013. The effect of valinomycin in fibroblasts from patients with fatty acid oxidation disorders. ***Biochem Biophys Res Commun*** Aug 9;437(4):637-41. doi: 10.1016/j.bbrc.2013.07.020. Epub 2013 Jul 16. PMID: 23867825.

94. **Longo N,** Harding CO, Burton BK, Grange DK, Vockley J, Wasserstein M, Rice GM, Dorenbaum A, Neuenburg JK, Musson DG, Gu Z, Sile S**.**. 2014. Single-dose, subcutaneous recombinant phenylalanine ammonia lyase conjugated with polyethylene glycol in adult patients with phenylketonuria: an open-label, multicentre, phase 1 dose-escalation trial. ***The Lancet*** 2014 Apr 14. pii: S0140-6736(13)61841-3, PMID: 24743000

95. Viau K, Ernst SL, Pasquali M, Botto LD, Hedlund GL, **Longo N.** 2013. Evidence-based treatment of Guanidinoacetate Methyltransferase (GAMT) deficiency. ***Mol Genet Metab*** Nov;110(3):255-62. doi: 10.1016/j.ymgme.2013.08.020. PMID: 24071436.

96. Mokhtarani M, Diaz GA, Rhead W, Berry SA, Lichter-Konecki U, Feigenbaum A, Schulze A, **Longo N**, Bartley J, Berquisti W, Gallagher R, Smith W, McCandless S, Harding C, Rockey DC,

Vierling JM, Mantry P, Ghabril P, Brown RS, Dickinson K, Moors T, Norris C, Coakley D, Milikien A, Nagamani SC, LeMons C, Lee B, Scharschmidt BF (2013) Elevated phenylacetic acid levels correlate with adverse events in healthy adults but not in patients with urea cycle disorders or hepatic encephalopathy and can be predicted based on the plasma PAA to PAGN ratio. ***Mol Genet Metab*** 2013 Dec;110(4):446-53. doi: 10.1016/j.ymgme.2013.09.017. Epub 2013 Oct 8.
PMID: 24144944.

97. Pasquali M, Schwarz E, Jensen M, Yuzyuk T, DeBiase I, Randall H, **Longo N**. 2014. Feasibility of newborn screening for guanidinoacetate methyltransferase (GAMT) deficiency. ***J Inherit Metab Dis****,* 2014 Mar;37(2):231-6. doi: 10.1007/s10545-013-9662-7. Epub 2013 Nov 26. PMID: 24276113

98. Stockler-Ipsiroglu S, van Karnebeek C, **Longo N**, Korenke GC, Mercimek-Mahmutoglu S, Marquart I, Barshop B, Grolik C, Schlune A, Angle B, Araújo HC, Coskun T, Diogo L, Geraghty M, Haliloglu G, Konstantopoulou V, Leuzzi V, Levtova A, Mackenzie J, Maranda B, Mhanni AA, Mitchell G, Morris A, Newlove T, Renaud D, Scaglia F, Valayannopoulos V, van Spronsen FJ, Verbruggen KT, Yuskiv N, Nyhan W, Schulze A. 2014. Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring. ***Mol Genet Metab***. 111(1):16-25. doi: 10.1016/j.ymgme.2013.10.018. Epub 2013 Nov 7. PMID: 24268530

99. Ardon O, Procter M, Tvrdik T, **Longo N**, Mao R (2014) Sequencing Analysis of Insulin Receptor Defects and Detection of Two Novel Mutations in INSR Gene. ***Mol Genet Metab Reports*** 1 (2014) 71–84.

100. **Longo N**, Siriwardena K, Feigenbaum A, Dimmock D, Burton BK, Stockler S, Waisbren S, Lang W, Jurecki E, Zhang C, Prasad S. Long-Term Developmental Progression in Infants and Young Children Taking Sapropterin for Phenylketonuria: A Two-Year Analysis of Safety and Efficacy. ***Genet Med*** 2015 May;17(5):365-73. doi: 10.1038/gim.2014.109. Epub 2014 Sep 18. PMID: 25232857

101. Lee B, Diaz GA, Rhead W, Lichter-Konecki U, Feigenbaum A, Berry SA, Le Mons C, Bartley JA, **Longo N**, Nagamani SC, Berquist W, Gallagher R, Bartholomew D, Harding CO, Korson MS, McCandless SE, Smith W, Cederbaum S, Wong D, Merritt JL 2nd, Schulze A, Vockley G, Kronn D, Zori R, Summar M, Milikien DA, Marino M, Coakley DF, Mokhtarani M, Scharschmidt BF. Blood ammonia and glutamine as predictors of hyperammonemic crises in patients with urea cycle disorder. ***Genet Med***. 2015 Jul;17(7):561-8. doi: 10.1038/gim.2014.148. Epub 2014 Dec 11. PMID: 25503497

102. Crippa BL, Leon E, Calhoun A, Lowichik A, Pasquali M, **Longo N** (2015) Biochemical abnormalities in Pearson Syndrome. ***Am J Med Genet A*** 167(3):621-8. doi: 10.1002/ajmg.a.36939. PMID: 25691415

103. De Biase I, Liu A, Yuzyuk T, **Longo N**, Pasquali M (2015) Quantitative amino acids analysis by liquid chromatography –tandem mass spectrometry: Implications for the diagnosis of argininosuccinic aciduria. ***Clin Chim Acta*** 2015 Mar 10;442:73-4. doi: 10.1016/j.cca.2015.01.008. Epub 2015 Jan 15. No abstract available. PMID: 25598409

104. **Longo N**, Arnold GL, Pridjian G, Enns GM, Ficicioglu C, Parker S, Cohen-Pfeffer JL; Phenylketonuria Demographics, Outcomes and Safety (PKUDOS) registry. ***Mol Genet Metab***. 2015 Apr;114(4):557-63. doi: 10.1016/j.ymgme.2015.02.003. Epub 2015 Feb 16. PMID: 25724073

105. Smith L, Rhead W, Charrow J, Shankar SP, Bavdekar A, **Longo N**, Mardach R, Harmatz P, Hangartner T, Lee HM, Crombez E, Pastores GM. Long-term velaglucerase alfa treatment in children with Gaucher disease type 1 naïve to enzyme replacement therapy or previously treated with imiglucerase. ***Mol Genet Metab***. 2015 Jun 1. pii: S1096-7192(15)30017-2. doi: 10.1016/j.ymgme. 2015.05.012 PMID: 26043810

106. Lukacs V, Mathur J, Mao R, Bayrak-Toydemir P, Procter M, Cahalan SM, Kim HJ, Bandell M, **Longo N**, Day RW, Stevenson DA, Patapoutian A, Krock BL. 2015. Impaired PIEZO1 function in patients with a novel autosomal recessive congenital lymphatic dysplasia***. Nature Commun***. 2015 Sep 21;6:8329. doi: 10.1038/ncomms9329.PMID: 26387913

107. Nagamani SC, Diaz GA, Rhead W,, Berry SA, Le Mons C, Lichter-Konecki U, Bartley J, Feigenbaum A, Schulze A, **Longo N**, Berquist W, Gallagher R, Bartholomew D, Harding CO, Korson

MS, McCandless SE, Smith W, Vockley J, Kronn D, Zori R, Cederbaum S, Lawrence Merritt J 2nd, Wong D, Coakley DF, Scharschmidt BF, Dickinson K, Marino M, Lee BH, Mokhtarani M. Self-reported treatment-associated symptoms among patients with urea cycle disorders participating in glycerol phenylbutyrate clinical trials. ***Mol Genet Metab***. 2015 Sep-Oct;116(1-2):29-34. doi: 10.1016/j.ymgme.2015.08.002. Epub 2015 Aug 5. PMID: 26296711

108. Lee B, Diaz GA, Rhead W, Lichter-Konecki U, Feigenbaum A, Berry SA, Le Mons C, Bartley J, **Longo N**, Nagamani SC, Berquist W, Gallagher RC, Harding CO, McCandless SE, Smith W, Schulze A, Marino M, Rowell R, Coakley DF, Mokhtarani M, Scharschmidt BF. Glutamine and hyperammonemic crises in patients with urea cycle disorders. ***Mol Genet Metab***. 2016 Jan;117(1):27-32. doi: 10.1016/j.ymgme.2015.11.005. Epub 2015 Nov 11. PMID: 26586473

109. Hook D, Diaz GA, Lee B, Bartley J, **Longo N**, Berquist W, Le Mons C, Rudolph-Angelich I, Porter M, Scharschmidt BF, Mokhtarani M. Protein and calorie intakes in adult and pediatric subjects with urea cycle disorders participating in clinical trials of glycerol phenylbutyrate. ***Mol Genet Metab Rep*** 2016 Jan 21;6:34-40. doi: 10.1016/j.ymgmr.2015.11.002. eCollection 2016 Mar.PMID: 27014577

110. Yuzyuk T, Liu A, Thomas A, Wilson JE, De Biase I, **Longo N**, Pasquali M. A novel method for simultaneous quantification of alpha-aminoadipic semialdehyde/piperideine-6-carboxylate and pipecolic acid in plasma and urine. ***J Chromatogr B Analyt Technol Biomed Life Sci***. 2016 Mar 3;1017-1018:145-152. doi: 10.1016/j.jchromb.2016.02.043 PMID: 26970849

111. Frigeni M, Iacobazzi F, Yin X, **Longo N**. Wide tolerance to amino acids substitutions in the OCTN1 ergothioneine transporter. ***Biochim Biophys Acta*** 2016 Mar 16;1860(6):1334-1342. doi: 10.1016/j.bbagen.2016.03.021. PMID: 26994919

112. Thurm A, Himelstein D, D'Souza P, Rennert O, Jiang S, Olatunji D, **Longo N**, Pasquali M, Swedo S, Salomons GS, Carrillo N. Creatine Transporter Deficiency: Screening of Males with Neurodevelopmental Disorders and Neurocognitive Characterization of a Case. ***J Dev Behav Pediatr***. 2016 May;37(4):322-6. doi: 10.1097/DBP.0000000000000299.PMID: 27096572

113. De Biase I, Viau KS, Liu A, Yuzyuk T, Botto LD, Pasquali M, **Longo N**. Diagnosis, Treatment, and Clinical Outcome of Patients with Mitochondrial Trifunctional Protein/Long-Chain 3-Hydroxy Acyl-CoA Dehydrogenase Deficiency. ***JIMD Rep***. 2016 Apr 28. PMID: 27117294

114. Goker-Alpan O, **Longo N,** McDonald M, Shankar SP, Schiffmann R, Chang P, Shen Y, Pano A. An open-label clinical trial of agalsidase alfa enzyme replacement therapy in children with Fabry disease who are naïve to enzyme replacement therapy. ***Drug Des Devel Ther***. 2016 May 25;10:1771-81. doi: 10.2147/DDDT.S102761. eCollection 2016. PMID: 27307708

115. Yuzyuk T, Thomas A, Viau K, Liu A, De Biase I, Botto LD, Pasquali M, **Longo N**. Effect of dietary lysine restriction and arginine supplementation in two patients with pyridoxine-dependent epilepsy. ***Mol Genet Metab***. 2016 Jul;118(3):167-72. doi: 10.1016/j.ymgme.2016.04.015. Epub 2016 May 8. PMID: 27324284

116. Ardon O, Procter M, Mao R, **Longo N**, Landau YE, Shilon-Hadass A, Gabis LV, Hoffmann C, Tzadok M, Heimer G, Sada S, Ben-Zeev B, Anikster Y. Creatine transporter deficiency: Novel mutations and functional studies. ***Mol Genet Metab Rep***. 2016 Jun 30;8:20-3. doi: 10.1016/j.ymgmr.2016.06.005. eCollection 2016 Sep. PMID: 27408820

117. Cetinkaya A, Xiong JR, Vargel İ, Kösemehmetoğlu K, Canter Hİ, Gerdan ÖF, **Longo N**, Alzahrani A, Camps MP, Taskiran EZ, Laupheimer S, Botto LD, Paramalingam E, Gormez Z, Uz E, Yuksel B, Ruacan Ş, Sağıroğlu MŞ, Takahashi T, Reversade B, Akarsu NA. Loss-of-Function Mutations in *ELMO2* Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling. ***Am J Human Genet*** 2016 Aug 4;99(2):299-317. doi: 10.1016/j.ajhg.2016.06.008. Epub 2016 Jul 28. PMID: 27476657

118. Germain DP, Hughes DA, Nicholls K, Bichet DG, Giugliani R, Wilcox WR, Feliciani C, Shankar SP, Ezgu F, Amartino H, Bratkovic D, Feldt-Rasmussen U, Nedd K, Sharaf El Din U, Lourenco CM, Banikazemi M, Charrow J, Dasouki M, Finegold D, Giraldo P, Goker-Alpan O, **Longo N**, Scott CR, Torra R, Tuffaha A, Jovanovic A, Waldek S, Packman S, Ludington E, Viereck C, Kirk J, Yu J,

Benjamin ER, Johnson F, Lockhart DJ, Skuban N, Castelli J, Barth J, Barlow C, Schiffmann R. Treatment of Fabry's Disease with the Pharmacologic Chaperone Migalastat. ***N Engl J Med***. 2016 Aug 11;375(6):545-55. doi: 10.1056/NEJMoa1510198. PMID: 27509102

119. Merrill ST, Nelson GR, **Longo N**, Bonkowsky JL. Cytotoxic edema and diffusion restriction as an early pathoradiologic marker in Canavan disease: case report and review of the literature. ***Orphanet J Rare Dis.*** 2016 Dec 7;11(1):169. PMID: 27927234

120. Sharer JD, Bodamer O, **Longo N**, Tortorelli S, Wamelink MM, Young S. Laboratory diagnosis of creatine deficiency syndromes: a technical standard and guideline of the American College of Medical Genetics and Genomics. ***Genet Med***. 2017 Feb;19(2):256-263. doi: 10.1038/gim.2016.203. PMID: 28055022

121. Vockley J, Burton B, Berry GT, **Longo N**, Phillips J, Sanchez-Valle A, Tanpaiboon P, Grunewald S, Murphy E, Humphrey R, Mayhew J, Bowden A, Zhang L, Cataldo J, Marsden DL, Kakkis E. UX007 for the treatment of long chain-fatty acid oxidation disorders: Safety and efficacy in children and adults following 24weeks of treatment. ***Mol Genet Metab***. 2017 Apr;120(4):370-377. doi: 10.1016/j.ymgme.2017.02.005. Epub 2017 Feb 7. PMID: 28189603

122. **Longo N**, Price LB, Gappmaier E, Cantor NL, Ernst SL, Bailey C, Pasquali M. Anaplerotic therapy in propionic acidemia. ***Mol Genet Metab***. 2017 Sep;122(1-2):51-59. doi: 10.1016/j.ymgme.2017.07.003. Epub 2017 Jul 12. PMID: 28712602

123. Frigeni M, Balakrishnan B, Yin X, Calderon FRO, Mao R, Pasquali M, **Longo N**. Functional and Molecular Studies in Primary Carnitine Deficiency. ***Hum Mutat.*** 2017 Dec;38(12):1684-1699. doi: 10.1002/humu.23315. Epub 2017 Sep 14.PMID: 28841266

124. Simcox J, Geoghegan G, Maschek JA, Bensard CL, Pasquali M, Miao R, Lee S, Jiang L, Huck I, Kershaw EE, Donato AJ, Apte U, **Longo N**, Rutter J, Schreiber R, Zechner R, Cox J, Villanueva CJ. Global Analysis of Plasma Lipids Identifies Liver-Derived Acylcarnitines as a Fuel Source for Brown Fat Thermogenesis. ***Cell Metab***. 2017 Sep 5;26(3):509-522.e6. doi: 10.1016/j.cmet.2017.08.006. PMID: 28877455

125. Berry SA, **Longo N**, Diaz GA, McCandless SE, Smith WE, Harding CO, Zori R, Ficicioglu C, Lichter-Konecki U, Robinson B, Vockley J. Safety and efficacy of glycerol phenylbutyrate for management of urea cycle disorders in patients aged 2months to 2years. ***Mol Genet Metab***. 2017 Nov;122(3):46-53. doi: 10.1016/j.ymgme.2017.09.002. Epub 2017 Sep 8. PMID: 28916119

126. Yuzyuk T, Viau K, Andrews A, Pasquali M, **Longo N**. Biochemical changes and clinical outcomes in 34 patients with classic galactosemia. ***J Inherit Metab Dis***. 2018 Mar;41(2):197-208. doi: 10.1007/s10545-018-0136-9. Epub 2018 Jan 19.PMID: 29350350127.

127. Charrow J, Fraga C, Gu X, Ida H, **Longo N**, Lukina E, Nonino A, Gaemers SJM, Jouvin MH, Li J, Wu Y, Xue Y, Peterschmitt MJ. Once- versus twice-daily dosing of eliglustat in adults with Gaucher disease type 1: The Phase 3, randomized, double-blind EDGE trial. ***Mol Genet Metab***. 2018 Mar;123(3):347-356. doi: 10.1016/j.ymgme.2017.12.001. Epub 2018 Jan 4.PMID: 29358012

128. Harding CO, Amato RS, Stuy M, **Longo N**, Burton BK, Posner J, Weng HH, Merilainen M, Gu Z, Jiang J, Vockley J; PRISM-2 Investigators. Pegvaliase for the treatment of phenylketonuria: A pivotal, double-blind randomized discontinuation Phase 3 clinical trial. ***Mol Genet Metab***. 2018 May;124(1):20-26. doi: 10.1016/j.ymgme.2018.03.003. Epub 2018 Mar 18. PMID: 29628378

129. Carlston CM, Ferdinandusse S, Hobert JA, Mao R, **Longo N.** Extrapolation of Variant Phase in Mitochondrial Short-Chain Enoyl-CoA Hydratase (ECHS1) Deficiency. ***JIMD Rep***. 2019;43:103-109. doi: 10.1007/8904_2018_111. Epub 2018 Jun 20. PMID: 29923089

130. **Longo N**, Zori R, Wasserstein MP, Vockley J, Burton BK, Decker C, Li M, Lau K, Jiang J, Larimore K, Thomas JA. Long-term safety and efficacy of pegvaliase for the treatment of phenylketonuria in adults: combined phase 2 outcomes through PAL-003 extension study. ***Orphanet J Rare Dis***. 2018 Jul 4;13(1):108. doi: 10.1186/s13023-018-0858-7. PMID: 29973227

131. Vockley J, Burton B, Berry GT, **Longo N**, Phillips J, Sanchez-Valle A, Tanpaiboon P, Grunewald S, Murphy E, Bowden A, Chen W, Chen CY, Cataldo J, Marsden D, Kakkis E. Results from a 78-week, single-arm, open-label Phase 2 study to evaluate UX007 in pediatric and adult patients with

severe long-chain fatty acid oxidation disorders (LC-FAOD). ***J Inherit Metab Dis.*** 2019 Jan;42(1):169-177. doi: 10.1002/jimd.12038. PMID: 29987492

132. Zori R, Thomas JA, Shur N, Rizzo WB, Decker C, Rosen O, Li M, Schweighardt B, Larimore K, **Longo N**. Induction, titration, and maintenance dosing regimen in a phase 2 study of pegvaliase for control of blood phenylalanine in adults with phenylketonuria. ***Mol Genet Metab***. 2018 Nov;125(3):217-227. doi: 10.1016/j.ymgme.2018.06.010. Epub 2018 Aug 23. PMID: 30146451

133. Yuzyuk T, Balakrishnan B, Schwarz EL, De Biase I, Hobert J, **Longo N**, Mao R, Lai K, Pasquali M. Effect of genotype on galactose-1-phosphate in classic galactosemia patients. ***Mol Genet Metab***. 2018 Nov;125(3):258-265. doi: 10.1016/j.ymgme.2018.08.012. Epub 2018 Aug 23. PMID: 30172461

134. Berry SA, Vockley J, Vinks AA, Dong M, Diaz GA, McCandless SE, Smith WE, Harding CO, Zori R, Ficicioglu C, Lichter-Konecki U, Perdok R, Robinson B, Holt RJ, **Longo N**. Pharmacokinetics of glycerol phenylbutyrate in pediatric patients 2 months to 2 years of age with urea cycle disorders. ***Mol Genet Metab***. 2018 Nov;125(3):251-257. doi: 10.1016/j.ymgme.2018.09.001. Epub 2018 Sep 4. PMID: 30217721

135. Bennett EE, Hummel K, Smith AG, **Longo N.** Acute Presentation and Management of the Encephalopathic Child With an Undiagnosed Inborn Error of Metabolism. ***J Emerg Med***. 2019 Jan;56(1):e5-e8. doi: 10.1016/j.jemermed.2018.09.037. Epub 2018 Nov 9. PMID: 30420308

136. **Longo N**, Dimmock D, Levy H, Viau K, Bausell H, Bilder DA, Burton B, Gross C, Northrup H, Rohr F, Sacharow S, Sanchez-Valle A, Stuy M, Thomas J, Vockley J, Zori R, Harding CO. Evidence- and consensus-based recommendations for the use of pegvaliase in adults with phenylketonuria. ***Genet Med***. 2019 Aug;21(8):1851-1867. doi: 10.1038/s41436-018-0403-z. Epub 2018 Dec 14. PMID: 30546086

137. Vockley J, Burton B, Berry GT, **Longo N**, Phillips J, Sanchez-Valle A, Tanpaiboon P, Grunewald S, Murphy E, Bowden A, Chen W, Chen CY, Cataldo J, Marsden D, Kakkis E. Results from a 78-week, single-arm, open-label phase 2 study to evaluate UX007 in pediatric and adult patients with severe long-chain fatty acid oxidation disorders (LC-FAOD). ***J Inherit Metab Dis***. 2019 Jan;42(1):169-177. doi: 10.1002/jimd.12038. PMID: 30740733

138. Ng BG, Sosicka P, Agadi S, Almannai M, Bacino CA, Barone R, Botto LD, Burton JE, Carlston C, Chung BH, Cohen JS, Coman D, Dipple KM, Dorrani N, Dobyns WB, Elias AF, Epstein L, Gahl WA, Garozzo D, Hammer TB, Haven J, Héron D, Herzog M, Hoganson GE, Hunter JM, Jain M, Juusola J, Lakhani S, Lee H, Lee J, Lewis K, **Longo N**, Lourenço CM, Mak CCY, McKnight D, Mendelsohn BA, Mignot C, Mirzaa G, Mitchell W, Muhle H, Nelson SF, Olczak M, Palmer CGS, Partikian A, Patterson MC, Pierson TM, Quinonez SC, Regan BM, Ross ME, Guillen Sacoto MJ, Scaglia F, Scheffer IE, Segal D, Singhal NS, Striano P, Sturiale L, Symonds JD, Tang S, Vilain E, Willis M, Wolfe LA, Yang H, Yano S, Undiagnosed Disease Network, Powis Z, Suchy SF, Rosenfeld JA, Edmondson AC, Grunewald S, Freeze HH. SLC35A2-CDG: Functional Characterization, Expanded Molecular, Clinical, and Biochemical Phenotypes of 30 Unreported Individuals. ***Hum Mutat***. 2019 Jul;40(7):908-925. doi: 10.1002/humu.23731. Epub 2019 Apr 24.PMID: 30817854

139. Smith N**, Longo N**, Levert K, Hyland K, Blau N. Phase I clinical evaluation of CNSA-001 (sepiapterin), a novel pharmacological treatment for phenylketonuria and tetrahydrobiopterin deficiencies, in healthy volunteers. ***Mol Genet Metab***. 2019 Apr;126(4):406-412. doi: 10.1016/j.ymgme.2019.02.001. Epub 2019 Feb 10. PMID: 30922814

140. Rovelli V, Manzoni F, Viau K, Pasquali M, **Longo N**. Clinical and biochemical outcome of patients with very long-chain acyl-CoA dehydrogenase deficiency. ***Mol Genet Metab***. 2019 May;127(1):64-73. doi: 10.1016/j.ymgme.2019.04.001. Epub 2019 Apr 16 PMID: 31031081

141. Diaz GA, Schulze A, **Longo N,** Rhead W, Feigenbaum A, Wong D, Merritt JL 2nd, Berquist W, Gallagher RC, Bartholomew D, McCandless SE, Smith WE, Harding CO, Zori R, Lichter-Konecki U, Vockley J, Canavan C, Vescio T, Holt RJ, Berry SA. Long-term safety and efficacy of glycerol phenylbutyrate for the management of urea cycle disorder patients. ***Mol Genet Metab***. 2019 Aug;127(4):336-345. doi: 10.1016/j.ymgme.2019.07.004. Epub 2019 Jul 10 PMID: 31326288

142. Hausmann O, Daha M, **Longo N**, Knol E, Müller I, Northrup H, Brockow K. Pegvaliase: Immunological profile and recommendations for the clinical management of hypersensitivity reactions in patients with phenylketonuria treated with this enzyme substitution therapy. ***Mol Genet Metab***. 2019 Sep - Oct;128(1-2):84-91. doi: 10.1016/j.ymgme.2019.05.006. Epub 2019 Jun 17.PMID: 31375398

143. Smith N, **Longo N**, Levert K, Hyland K, Blau N. Exploratory study of the effect of one week of orally administered CNSA-001 (sepiapterin) on CNS levels of tetrahydrobiopterin, dihydrobiopterin and monoamine neurotransmitter metabolites in healthy volunteers. ***Mol Genet Metab Rep***. 2019 Aug 12;21:100500. doi: 10.1016/j.ymgmr.2019.100500. eCollection 2019 Dec. PMID: 31453106

144. SriBhashyam S, Marsh K, Quartel A, Weng HH, Gershman A, **Longo N**, Thomas J, Zori R. A benefit-risk analysis of pegvaliase for the treatment of phenylketonuria: A study of patients' preferences. ***Mol Genet Metab Rep***. 2019 Aug 30;21:100507. doi: 10.1016/j.ymgmr.2019.100507. eCollection 2019 Dec. PMID: 31497506

145. Anderson DR, Viau K, Botto LD, Pasquali M, **Longo N.** Clinical and biochemical outcomes of patients with medium-chain acyl-CoA dehydrogenase deficiency. ***Mol Genet Metab***. 2020 Jan;129(1):13-19. doi: 10.1016/j.ymgme.2019.11.006. Epub 2019 Nov 25. PMID: 31836396

146. Gorshkov K, Wang AQ, Sun W, Fisher E, Frigeni M, Singleton M, Thorne N, Class B, Huang W, **Longo N**, Do MT, Ottinger EA, Xu X, Zheng W. Phosphocyclocreatine is the dominant form of cyclocreatine in control and creatine transporter deficiency patient fibroblasts. ***Pharmacol Res Perspect***. 2019 Dec;7(6):e00525. doi: 10.1002/prp2.525.PMID: 31859463

147. De Biase I, Gherasim C, La'ulu SL, Asamoah A, **Longo N**, Yuzyuk T. Laboratory evaluation of homocysteine remethylation disorders and classic homocystinuria: Long-term follow-up using a cohort of 123 patients. ***Clin Chim Acta***. 2020 Oct;509:126-134. doi: 10.1016/j.cca.2020.06.014. Epub 2020 Jun 10. PMID: 32533987

148. Burton BK, **Longo N**, Vockley J, Grange DK, Harding CO, Decker C, Li M, Lau K, Rosen O, Larimore K, Thomas J; PAL-002 and PAL-004 Investigators. Pegvaliase for the treatment of phenylketonuria: Results of the phase 2 dose-finding studies with long-term follow-up. ***Mol Genet Metab***. 2 2020 Aug;130(4):239-246. doi: 10.1016/j.ymgme.2020.06.006. Epub 2020 Jun 16. PMID: 32593547

149. Flores Daboub JA, Grimmer JF, Frigerio A, Wooderchak-Donahue W, Arnold R, Szymanski J, **Longo N**, Bayrak-Toydemir P. Parkes Weber syndrome associated with two somatic pathogenic variants in RASA1. ***Cold Spring Harb Mol Case Stud***. 2020 Aug 25;6(4):a005256. doi: 10.1101/mcs.a005256. Print 2020 Aug. PMID: 32843429

150. Vockley J, Burton B, Berry G, **Longo N**, Phillips J, Sanchez-Valle A, Chapman K, Tanpaiboon P, Grunewald S, Murphy E, Lu X, Cataldo J. Effects of Triheptanoin (UX007) in Patients with Long-chain Fatty Acid Oxidation Disorders (LC-FAOD): Results from an Open-Label, Long-Term Extension Study. ***J Inherit Metab Dis***. 2021 Jan;44(1):253-263. doi: 10.1002/jimd.12313. Epub 2020 Sep 14. PMID: 32885845

151. Coughlin CR 2nd, Tseng LA, Abdenur JE, Ashmore C, Boemer F, Bok LA, Boyer M, Buhas D, Clayton PT, Das A, Dekker H, Evangeliou A, Feillet F, Footitt EJ, Gospe SM Jr, Hartmann H, Kara M, Kristensen E, Lee J, Lilje R, **Longo N**, Lunsing RJ, Mills P, Papadopoulou MT, Pearl PL, Piazzon F, Plecko B, Saini AG, Santra S, Sjarif DR, Stockler-Ipsiroglu S, Striano P, Van Hove JLK, Verhoeven-Duif NM, Wijburg FA, Zuberi SM, van Karnebeek CDM. Consensus guidelines for the diagnosis and management of pyridoxine-dependent epilepsy due to α-aminoadipic semialdehyde dehydrogenase deficiency. ***J Inherit Metab Dis.*** 2021 Jan;44(1):178-192. doi: 10.1002/jimd.12332. Epub 2020 Dec 1. PMID: 33200442

152. Manzoni F, Salvatici E, Burlina A, Andrews A, Pasquali M, **Longo N**. Retrospective analysis of 19 patients with 6-Pyruvoyl Tetrahydropterin Synthase Deficiency: Prolactin levels inversely correlate with growth. ***Mol Genet Metab***. 2020 Dec;131(4):380-389. doi: 10.1016/j.ymgme.2020.11.004. Epub 2020 Nov 18. PMID: 33234470

153. Bryant L, Li D, Cox SG, Marchione D, Joiner EF, Wilson K, Janssen K, Lee P, March ME, Nair D, Sherr E, Fregeau B, Wierenga KJ, Wadley A, Mancini GMS, Powell-Hamilton N, van de Kamp J,

Grebe T, Dean J, Ross A, Crawford HP, Powis Z, Cho MT, Willing MC, Manwaring L, Schot R, Nava C, Afenjar A, Lessel D, Wagner M, Klopstock T, Winkelmann J, Catarino CB, Retterer K, Schuette JL, Innis JW, Pizzino A, Lüttgen S, Denecke J, Strom TM, Monaghan KG; DDD Study, Yuan ZF, Dubbs H, Bend R, Lee JA, Lyons MJ, Hoefele J, Günthner R, Reutter H, Keren B, Radtke K, Sherbini O, Mrokse C, Helbig KL, Odent S, Cogne B, Mercier S, Bezieau S, Besnard T, Kury S, Redon R, Reinson K, Wojcik MH, Õunap K, Ilves P, Innes AM, Kernohan KD; Care4Rare Canada Consortium, Costain G, Meyn MS, Chitayat D, Zackai E, Lehman A, Kitson H; CAUSES Study, Martin MG, Martinez-Agosto JA; Undiagnosed Diseases Network, Nelson SF, Palmer CGS, Papp JC, Parker NH, Sinsheimer JS, Vilain E, Wan J, Yoon AJ, Zheng A, Brimble E, Ferrero GB, Radio FC, Carli D, Barresi S, Brusco A, Tartaglia M, Thomas JM, Umana L, Weiss MM, Gotway G, Stuurman KE, Thompson ML, McWalter K, Stumpel CTRM, Stevens SJC, Stegmann APA, Tveten K, Vøllo A, Prescott T, Fagerberg C, Laulund LW, Larsen MJ, Byler M, Lebel RR, Hurst AC, Dean J, Schrier Vergano SA, Norman J, Mercimek-Andrews S, Neira J, Van Allen MI, **Longo N**, Sellars E, Louie RJ, Cathey SS, Brokamp E, Heron D, Snyder M, Vanderver A, Simon C, de la Cruz X, Padilla N, Crump JG, Chung W, Garcia B, Hakonarson HH, Bhoj EJ. Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. ***Sci Adv.*** 2020 Dec 2;6(49):eabc9207. doi: 10.1126/sciadv.abc9207. Print 2020 Dec. PMID: 33268356

154. **Longo N**, Diaz GA, Lichter-Konecki U, Schulze A, Inbar-Feigenberg M, Conway RL, Bannick AA, McCandless SE, Zori R, Hainline B, Ah Mew N, Canavan C, Vescio T, Kok T, Porter MH, Berry SA. Glycerol phenylbutyrate efficacy and safety from an open label study in pediatric patients under 2 months of age with urea cycle disorders. ***Mol Genet Metab***. 2021 Jan;132(1):19-26. doi: 10.1016/j.ymgme.2020.12.002. Epub 2020 Dec 23. PMID: 33388234

155. Waisbren S, Burton BK, Feigenbaum A, Konczal LL, Lilienstein J, McCandless SE, Rowell R, Sanchez-Valle A, Whitehall KB, **Longo N**. Long-term preservation of intellectual functioning in sapropterin-treated infants and young children with phenylketonuria: A seven-year analysis. ***Mol Genet Metab***. 2021 Feb;132(2):119-127. doi: 10.1016/j.ymgme.2021.01.001. Epub 2021 Jan 13. PMID: 33485801

156. Vockley J, **Longo N**, Madden M, Dwyer L, Mu Y, Chen CY, Cataldo J. Dietary management and major clinical events in patients with long-chain fatty acid oxidation disorders enrolled in a phase 2 triheptanoin study. ***Clin Nutr ESPEN***. 2021 Feb;41:293-298. doi: 10.1016/j.clnesp.2020.11.018. Epub 2020 Dec 25. PMID: 33487279

157. Qi Y, Patel G, Henshaw J, Gupta S, Olbertz J, Larimore K, Harding CO, Merilainen M, Zori R, **Longo N**, Burton BK, Li M, Gu Z, Zoog SJ, Weng HH, Schweighardt B. Pharmacokinetic, pharmacodynamic, and immunogenic rationale for optimal dosing of pegvaliase, a PEGylated bacterial enzyme, in adult patients with phenylketonuria. ***Clin Transl Sci***. 2021 Sep;14(5):1894-1905. doi: 10.1111/cts.13043. PMID: 34057292

158. Hart K, Rohrwasser A, Wallis H, Golsan H, Shao J, Anderson T, Wang X, Szabo-Fresnais N, Morrissey M, Kay DM, Wojcik M, Galvin-Parton PA, **Longo N**, Caggana M, Pasquali M. Prospective identification by neonatal screening of patients with guanidinoacetate methyltransferase deficiency. ***Mol Genet Metab***. 2021 Sep-Oct;134(1-2):60-64. doi: 10.1016/j.ymgme.2021.07.012. PMID: 34389248

159. Ingoglia F, Chong JL, Pasquali M, **Longo N**. Creatine metabolism in patients with urea cycle disorders. ***Mol Genet Metab Rep.*** 2021 Aug 23;29:100791. doi: 10.1016/j.ymgmr.2021.100791. PMID: 34471603; PMCID: PMC8387902.

160. Häberle J, Moore MB, Haskins N, Rüfenacht V, Rokicki D, Rubio-Gozalbo E, Tuchman M, **Longo N**, Yandell M, Andrews A, AhMew N, Caldovic L. Non-coding sequence variants define a novel regulatory element in the first intron of the N-acetylglutamate synthase gene. ***Hum Mutat.*** 2021 Dec;42(12):1624-1636. doi: 10.1002/humu.24281. Epub ahead of print. PMID: 34510628.

161. Burton BK, Skalicky A, Baerwald C, Bilder DA, Harding CO, Ilan AB, Jurecki E, **Longo N**, Madden DT, Sivri HS, Wilcox G, Thomas J, Delaney K. A non-interventional observational study to identify and validate clinical outcome assessments for adults with phenylketonuria for use in clinical

trials. ***Mol Genet Metab Rep***. 2021 Nov 9;29:100810. doi: 10.1016/j.ymgmr.2021.100810. PMID: 34815941; PMCID: PMC8591457.

162. Bilder DA, Arnold GL, Dimmock D, Grant ML, Janzen D, **Longo N**, Nguyen-Driver M, Jurecki E, Merilainen M, Amato G, Waisbren S. Improved attention linked to sustained phenylalanine reduction in adults with early-treated phenylketonuria. ***Am J Med Genet A***. 2021 Nov 26. doi: 10.1002/ajmg.a.62574. Epub ahead of print. PMID: 34826353

163. Palmquist R, Jenkins SM, Bentley D, Miller C, Mao R, Meibos B, Bayrak-Toydemir P, Tvrdik T, Nadauld LD, Bleyl SB, Chowdhury S, Ostrander B, Flores-Daboub J, **Longo N**, Tristani-Firouzi M, Hobbs C, Bonkowsky JL, Brunelli L. Evaluating use of changing technologies for rapid next-generation sequencing in pediatrics. ***Pediatr Res***. 2022 Feb 3. doi: 10.1038/s41390-022-01965-5. Epub ahead of print. PMID: 35115709.

164. Tseng LA, Abdenur JE, Andrews A, Aziz VG, Bok LA, Boyer M, Buhas D, Hartmann H, Footitt EJ, Grønborg S, Janssen MCH, **Longo N**, Lunsing RJ, MacKenzie AE, Wijburg FA, Gospe SM Jr, Coughlin CR 2nd, van Karnebeek CDM. Timing of therapy and neurodevelopmental outcomes in 18 families with pyridoxine-dependent epilepsy. ***Mol Genet Metab***. 2022 Apr;135(4):350-356. doi: 10.1016/j.ymgme.2022.02.005. Epub 2022 Feb 17. PMID: 35279367.

165. Tseng LA, Hoytema van Konijnenburg EM, **Longo N**, Andrews A, van Wegberg A, Coene KL, Coughlin CR 2nd, van Karnebeek CD. Clinical Reasoning: Pediatric Seizures of Unknown Cause. ***Neurology***. 2022 Jun 14;98(24):1023-1028. doi: 10.1212/WNL.0000000000200711. Epub 2022 Apr 25. PMID: 35470136.

166. Han ST, Anderson KJ, Bjornsson HT, **Longo N**, Valle D. A promoter variant in the OTC gene associated with late and variable age of onset hyperammonemia. ***J Inherit Metab Dis***. 2022 May 23. doi: 10.1002/jimd.12524. Epub ahead of print. PMID: 35605046.

167. Ventura P, Sardh E, **Longo N**, Balwani M, Plutzky J, Gouya L, Phillips J, Rhyee S, Fanelli MJ, Sweetser MT, Petrides PE. Hyperhomocysteinemia in acute hepatic porphyria (AHP) and implications for treatment with givosiran. ***Expert Rev Gastroenterol Hepatol***. 2022 Aug 5. doi: 10.1080/17474124.2022.2110469. Epub ahead of print. PMID: 35929959.

168. Coughlin CR 2nd, Tseng LA, Bok LA, Hartmann H, Footitt E, Striano P, Tabarki BM, Lunsing RJ, Stockler-Ipsiroglu S, Gordon S, Van Hove JLK, Abdenur JE, Boyer M, **Longo N**, Andrews A, Janssen MC, van Wegberg A, Prasad C, Prasad AN, Lamb MM, Wijburg FA, Gospe SM, van Karnebeek C; International PDE Consortium. Association Between Lysine Reduction Therapies and Cognitive Outcomes in Patients With Pyridoxine-Dependent Epilepsy. ***Neurolog***y. 2022 Aug 25:10.1212/WNL.0000000000201222. doi: 10.1212/WNL.0000000000201222. Epub ahead of print. PMID: 36008148.

169. Burton BK, Hermida Á, Bélanger-Quintana A, Bell H, Bjoraker KJ, Christ SE, Grant ML, Harding CO, Huijbregts SCJ, **Longo N**, McNutt MC 2nd, Nguyen-Driver MD, Santos Pessoa AL, Rocha JC, Sacharow S, Sanchez-Valle A, Sivri HS, Vockley J, Walterfang M, Whittle S, Muntau AC. Management of early treated adolescents and young adults with phenylketonuria: Development of international consensus recommendations using a modified Delphi approach. ***Mol Genet Metab.*** 2022 Jul 29;137(1-2):114-126. doi: 10.1016/j.ymgme.2022.07.012. Epub ahead of print. PMID: 36027720.

PUBLICATIONS AS MEMBER OF THE UNDIAGNOSED DISEASES NETWORK AND OTHER NETWORKS

1. Nicoli ER, Weston MR, Hackbarth M, Becerril A, Larson A, Zein WM, Baker PR 2nd, Burke JD, Dorward H, Davids M, Huang Y, Adams DR, Zerfas PM, Chen D, Markello TC, Toro C, Wood T, Elliott G, Vu M; Undiagnosed Diseases Network, Zheng W, Garrett LJ, Tifft CJ, Gahl WA, Day-Salvatore DL, Mindell JA, Malicdan MCV. Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. ***Am J Hum Genet.*** 2019 Jun 6;104(6):1127-1138. doi: 10.1016/j.ajhg.2019.04.008. Epub 2019 May 30. PMID: 31155284

2. Kanca O, Andrews JC, Lee PT, Patel C, Braddock SR, Slavotinek AM, Cohen JS, Gubbels CS, Aldinger KA, Williams J, Indaram M, Fatemi A, Yu TW, Agrawal PB, Vezina G, Simons C, Crawford J, Lau CC; Undiagnosed Diseases Network, Chung WK, Markello TC, Dobyns WB, Adams DR, Gahl WA, Wangler MF, Yamamoto S, Bellen HJ, Malicdan MCV. De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. ***Am J Hum Genet.*** 2019 Sep 5;105(3):672-674. doi: 10.1016/j.ajhg.2019.07.017. PMID: 31491411

3. Kanca O, Andrews JC, Lee PT, Patel C, Braddock SR, Slavotinek AM, Cohen JS, Gubbels CS, Aldinger KA, Williams J, Indaram M, Fatemi A, Yu TW, Agrawal PB, Vezina G, Simons C, Crawford J, Lau CC; Undiagnosed Diseases Network, Chung WK, Markello TC, Dobyns WB, Adams DR, Gahl WA, Wangler MF, Yamamoto S, Bellen HJ, Malicdan MCV. De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. ***Am J Hum Genet.*** 2019 Aug 1;105(2):413-424. doi: 10.1016/j.ajhg.2019.06.014. Epub 2019 Jul 18. PMID: 31327508
4. Accogli A, Calabretta S, St-Onge J, Boudrahem-Addour N, Dionne-Laporte A, Joset P, Azzarello-Burri S, Rauch A, Krier J, Fieg E, Pallais JC; Undiagnosed Diseases Network, McConkie-Rosell A, McDonald M, Freedman SF, Rivière JB, Lafond-Lapalme J, Simpson BN, Hopkin RJ, Trimouille A, Van-Gils J, Begtrup A, McWalter K, Delphine H, Keren B, Genevieve D, Argilli E, Sherr EH, Severino M, Rouleau GA, Yam PT, Charron F, Srour M. De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Collosum, Axon, Cardiac, Ocular, and Genital Defects. ***Am J Hum Genet***. 2019 Oct 3;105(4):854-868. doi: 10.1016/j.ajhg.2019.09.005. PMID: 31585109
5. Holt JM, Wilk B, Birch CL, Brown DM, Gajapathy M, Moss AC, Sosonkina N, Wilk MA, Anderson JA, Harris JM, Kelly JM, Shaterferdosian F, Uno-Antonison AE, Weborg A; Undiagnosed Diseases Network, Worthey EA. VarSight: prioritizing clinically reported variants with binary classification algorithms. ***BMC Bioinformatics***. 2019 Oct 15;20(1):496. doi: 10.1186/s12859-019-3026-8. PMID: 31615419
6. Mao D, Reuter CM, Ruzhnikov MRZ, Beck AE, Farrow EG, Emrick LT, Rosenfeld JA, Mackenzie KM, Robak L, Wheeler MT, Burrage LC, Jain M, Liu P, Calame D, Küry S, Sillesen M, Schmitz-Abe K, Tonduti D, Spaccini L, Iascone M, Genetti CA, Koenig MK, Graf M, Tran A, Alejandro M; Undiagnosed Diseases Network, Lee BH, Thiffault I, Agrawal PB, Bernstein JA, Bellen HJ, Chao HT. De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. ***Am J Hum Genet.*** 2020 Apr 2;106(4):570-583. doi: 10.1016/j.ajhg.2020.02.016. Epub 2020 Mar 19. PMID: 32197074
7. Schneeberger PE, Kortüm F, Korenke GC, Alawi M, Santer R, Woidy M, Buhas D, Fox S, Juusola J, Alfadhel M, Webb BD, Coci EG, Abou Jamra R, Siekmeyer M, Biskup S, Heller C, Maier EM, Javaher-Haghighi P, Bedeschi MF, Ajmone PF, Iascone M, Peeters H, Ballon K, Jaeken J, Rodríguez Alonso A, Palomares-Bralo M, Santos-Simarro F, Meuwissen MEC, Beysen D, Kooy RF, Houlden H, Murphy D, Doosti M, Karimiani EG, Mojarrad M, Maroofian R, Noskova L, Kmoch S, Honzik T, Cope H, Sanchez-Valle A; Undiagnosed Diseases Network, Gelb BD, Kurth I, Hempel M, Kutsche K. Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. ***Brain***. 2020 Aug 1;143(8):2437-2453. doi: 10.1093/brain/awaa204. PMID: 32761064
8. Schoch K, Esteves C, Bican A, Spillmann R, Cope H, McConkie-Rosell A, Walley N, Fernandez L, Kohler JN, Bonner D, Reuter C, Stong N, Mulvihill JJ, Novacic D, Wolfe L, Abdelbaki A, Toro C, Tifft C, Malicdan M, Gahl W, Liu P, Newman J, Goldstein DB, Hom J, Sampson J, Wheeler MT; Undiagnosed Diseases Network, Cogan J, Bernstein JA, Adams DR, McCray AT, Shashi V. Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. **Genet Med.** 2020 Oct 23. doi: 10.1038/s41436-020-00984-z. Online ahead of print. PMID: 33093671
9. Studwell CM, Kelley EG; Undiagnosed Diseases Network, Sinsheimer JS, Palmer CGS, LeBlanc K. Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. ***J Genet Couns.*** 2020 Oct 27. doi: 10.1002/jgc4.1329. Online ahead of print. PMID: 33108040
10. Meissner LE, Macnamara EF, D'Souza P, Yang J, Vezina G; Undiagnosed Diseases Network, Ferreira CR, Zein WM, Tifft CJ, Adams DR. DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature. ***Mol Genet Genomic Med***. 2020 Nov 7:e1544. doi: 10.1002/mgg3.1544. Online ahead of print. PMID: 33159716
11. Ferdinandusse S, McWalter K, Te Brinke H, IJlst L, Mooijer PM, Ruiter JPN, van Lint AEM, Pras-Raves M, Wever E, Millan F, Guillen Sacoto MJ, Begtrup A, Tarnopolsky M, Brady L, Ladda RL, Sell SL, Nowak CB, Douglas J, Tian C, Ulm E, Perlman S, Drack AV, Chong K, Martin N, Brault J, Brokamp E, Toro C, Gahl WA, Macnamara EF, Wolfe L; Undiagnosed Diseases Network, Waisfisz Q, Zwijnenburg PJG, Ziegler A, Barth M, Smith R, Ellingwood S, Gaebler-Spira D, Bakhtiari S, Kruer MC, van Kampen AHC, Wanders RJA, Waterham HR, Cassiman D, Vaz FM. An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. ***Genet Med.*** 2020 Nov 26. doi: 10.1038/s41436-020-01027-3. Online ahead of print. PMID: 33239752
12. Kobren SN, Baldridge D, Velinder M, Krier JB, LeBlanc K, Esteves C, Pusey BN, Züchner S, Blue E, Lee H, Huang A, Bastarache L, Bican A, Cogan J, Marwaha S, Alkelai A, Murdock DR, Liu P, Wegner DJ, Paul AJ; Undiagnosed Diseases Network, Sunyaev SR, Kohane IS. Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. ***Genet Med***. 2021 Feb 12. doi: 10.1038/s41436-020-01084-8. Online ahead of print. PMID: 33580225
13. Marbach F, Stoyanov G, Erger F, Stratakis CA, Settas N, London E, Rosenfeld JA, Torti E, Haldeman-Englert C, Sklirou E, Kessler E, Ceulemans S, Nelson SF, Martinez-Agosto JA, Palmer CGS, Signer RH; Undiagnosed Diseases Network, Andrews MV, Grange DK, Willaert R, Person R, Telegrafi A, Sievers A, Laugsch M, Theiß S, Cheng Y, Lichtarge O, Katsonis P, Stocco A, Schaaf CP. Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. ***Genet Med***. 2021 Apr 8. doi: 10.1038/s41436-021-01152-7. Online ahead of print. PMID: 33833410
14. Cope H, Barseghyan H, Bhattacharya S, Fu Y, Hoppman N, Marcou C, Walley N, Rehder C, Deak K, Alkelai A; Undiagnosed Diseases Network, Vilain E, Shashi V. Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. ***Mol Genet Genomic Med***. 2021 May 6:e1665. doi: 10.1002/mgg3.1665. Online ahead of print. PMID: 33955715
15. Sturrock BRH, Macnamara EF, McGuire P, Kruk S, Yang I, Murphy J; Undiagnosed Diseases Network, Tifft CJ, Gordon-Lipkin E. Progressive cerebellar atrophy in a patient with complex II and III deficiency and a novel deleterious variant in SDHA: A Counseling Conundrum. ***Mol Genet Genomic Med.*** 2021 May 7:e1692. doi: 10.1002/mgg3.1692. Online ahead of print. PMID: 33960148
16. Baldridge D, Wangler MF, Bowman AN, Yamamoto S; Undiagnosed Diseases Network, Schedl T, Pak SC, Postlethwait JH, Shin J, Solnica-Krezel L, Bellen HJ, Westerfield M. Model organisms contribute to diagnosis and discovery in the undiagnosed diseases network: current state and a future vision. ***Orphanet J Rare Dis.*** 2021 May 7;16(1):206. doi: 10.1186/s13023-021-01839-9. PMID: 33962631
17. Panneerselvam S, Wang J, Zhu W, Dai H, Pappas JG, Rabin R, Low KJ, Rosenfeld JA, Emrick L, Xiao R, Xia F, Yang Y, Eng CM, Anderson A, Chau V, Soler-Alfonso C, Streff H, Lalani SR, Mercimek-Andrews S; Undiagnosed Diseases Network; DDD Study, Bi W. PPP3CA truncating variants clustered in the regulatory domain cause early-onset refractory epilepsy. ***Clin Genet***. 2021 May 8. doi: 10.1111/cge.13979. Online ahead of print. PMID: 33963760
18. Kobren SN, Baldridge D, Velinder M, Krier JB, LeBlanc K, Esteves C, Pusey BN, Züchner S, Blue E, Lee H, Huang A, Bastarache L, Bican A, Cogan J, Marwaha S, Alkelai A, Murdock DR, Liu P, Wegner DJ, Paul AJ; Undiagnosed Diseases Network, Sunyaev SR, Kohane IS. Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. ***Genet Med***. 2021 Jun;23(6):1075-1085. doi: 10.1038/s41436-020-01084-8. Epub 2021 Feb 12. PMID: 33580225

Nicola Longo MD PhD

19. Ravenscroft TA, Phillips JB, Fieg E, Bajikar SS, Peirce J, Wegner J, Luna AA, Fox EJ, Yan YL, Rosenfeld JA, Zirin J, Kanca O; Undiagnosed Diseases Network, Benke PJ, Cameron ES, Strehlow V, Platzer K, Jamra RA, Klöckner C, Osmond M, Licata T, Rojas S, Dyment D, Chong JSC, Lincoln S, Stoler JM, Postlethwait JH, Wangler MF, Yamamoto S, Krier J, Westerfield M, Bellen HJ. Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. ***Genet Med. 2021*** Jun 10. doi: 10.1038/s41436-021-01216-8. Online ahead of print. PMID: 34113007
20. Baldridge D, Wangler MF, Bowman AN, Yamamoto S; Undiagnosed Diseases Network, Schedl T, Pak SC, Postlethwait JH, Shin J, Solnica-Krezel L, Bellen HJ, Westerfield M. Model organisms contribute to diagnosis and discovery in the undiagnosed diseases network: current state and a future vision. ***Orphanet J Rare Dis.*** 2021 May 7;16(1):206. doi: 10.1186/s13023-021-01839-9. PMID: 33962631
21. McConkie-Rosell A, Schoch K, Sullivan J, Spillmann RC, Cope H, Tan QK, Palmer CGS; Undiagnosed Disease Network, Hooper SR, Shashi V. Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples. ***J Genet Couns.*** 2021 Jun 11. doi: 10.1002/jgc4.1451. Online ahead of print. PMID: 34115423
22. Rodan LH, Spillmann RC, Kurata HT, Lamothe SM, Maghera J, Jamra RA, Alkelai A, Antonarakis SE, Atallah I, Bar-Yosef O, Bilan F, Bjorgo K, Blanc X, Van Bogaert P, Bolkier Y, Burrage LC, Christ BU, Granadillo JL, Dickson P, Donald KA, Dubourg C, Eliyahu A, Emrick L, Engleman K, Gonfiantini MV, Good JM, Kalser J, Kloeckner C, Lachmeijer G, Macchiaiolo M, Nicita F, Odent S, O'Heir E, Ortiz-Gonzalez X, Pacio-Miguez M, Palomares-Bralo M, Pena L, Platzer K, Quinodoz M, Ranza E, Rosenfeld JA, Roulet-Perez E, Santani A, Santos-Simarro F, Pode-Shakked B, Skraban C, Slaugh R, Superti-Furga A, Thiffault I, van Jaabrsveld RH, Vincent M, Wang HG, Zacher P; Undiagnosed Diseases Network, Rush E, Pitt G, Au PYB, Shashi V. Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. ***Genet Med***. 2021 Jun 23. doi: 10.1038/s41436-021-01232-8. Online ahead of print. PMID: 34163037
23. Brokamp E, Koziura ME, Phillips JA 3rd, Tang LA, Cogan JD, Rives LC, Robertson AK, Duncan L, Bican A, Peterson JF, Newman JH, Hamid R, Bastarache L; Undiagnosed Diseases Network. One is the loneliest number: genotypic matchmaking using the electronic health record. ***Genet Med***. 2021 Jul 6. doi: 10.1038/s41436-021-01179-w. Online ahead of print. PMID: 34230636
24. Kohler JN, Kelley EG, Boyd BM, Sillari CH, Marwaha S; Undiagnosed Diseases Network, Wheeler MT.Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation.***J Genet Couns***. 2021 Aug 10. doi: 10.1002/jgc4.1493. Online ahead of print. PMID: 34374469
25. van Wegberg AMJ, Trefz F, Gizewska M, Ahmed S, Chabraoui L, Zaki MS, Maillot F, van Spronsen FJ; Study Group on Missed PKU and Missed to Follow-Up. Undiagnosed Phenylketonuria Can Exist Everywhere: Results From an International Survey. ***J Pediatr***. 2021 Dec;239:231-234.e2. doi: 10.1016/j.jpeds.2021.08.070. Epub 2021 Aug 30. PMID: 34474089.
26. Diaz-Manera J, Kishnani PS, Kushlaf H, Ladha S, Mozaffar T, Straub V, Toscano A, van der Ploeg AT, Berger KI, Clemens PR, Chien YH, Day JW, Illarioshkin S, Roberts M, Attarian S, Borges JL, Bouhour F, Choi YC, Erdem-Ozdamar S, Goker-Alpan O, Kostera-Pruszczyk A, Haack KA, Hug C, Huynh-Ba O, Johnson J, Thibault N, Zhou T, Dimachkie MM, Schoser B; COMET Investigator Group. Safety and efficacy of avalglucosidase alfa versus alglucosidase alfa in patients with late-onset Pompe disease (COMET): a phase 3, randomised, multicentre trial. ***Lancet Neuro***l. 2021 Dec;20(12):1012-1026. doi: 10.1016/S1474-4422(21)00241-6. PMID: 34800399.
27. Schoser B, Roberts M, Byrne BJ, Sitaraman S, Jiang H, Laforêt P, Toscano A, Castelli J, Díaz-Manera J, Goldman M, van der Ploeg AT, Bratkovic D, Kuchipudi S, Mozaffar T, Kishnani PS; PROPEL Study Group. Safety and efficacy of cipaglucosidase alfa plus miglustat versus alglucosidase alfa plus placebo in late-onset Pompe disease (PROPEL): an international, randomised, double-blind, parallel-group, phase 3 trial. ***Lancet Neurol***. 2021 Dec;20(12):1027-1037. doi: 10.1016/S1474-4422(21)00331-8. PMID: 34800400.
28. Borja N, Bivona S, Peart LS, Johnson B, Gonzalez J, Barbouth D, Moore H, Guo S; Undiagnosed Disease Network, Bademci G, Tekin M. Genome sequencing reveals novel noncoding variants in PLA2G6 and LMNB1 causing progressive neurologic disease. Mol Genet Genomic Med. 2022 Mar 5:e1892. doi: 10.1002/mgg3.1892. Epub ahead of print. PMID: 35247231. Shankar SP, Grimsrud K, Lanoue L, Egense A, Willis B, Hörberg J, AlAbdi L, Mayer K, Ütkür K, Monaghan KG, Krier J, Stoler J, Alnemer M, Shankar PR, Schaffrath R, Alkuraya FS, Brinkmann U, Eriksson LA, Lloyd K, Rauen KA; Undiagnosed Diseases Network. A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. Genet Med. 2022 Apr 27:S1098-3600(22)00704-3. doi: 10.1016/j.gim.2022.03.014. Epub ahead of print. PMID: 35482014.
29. Barish S, Senturk M, Schoch K, Minogue AL, Lopergolo D, Fallerini C, Harland J, Seemann JH, Stong N, Kranz PG, Kansagra S, Mikati MA, Jasien J, El-Dairi M, Galluzzi P; Undiagnosed Diseases Network, Ariani F, Renieri A, Mari F, Wangler MF, Arur S, Jiang YH, Yamamoto S, Shashi V, Bellen HJ. The microRNA processor DROSHA is a candidate gene for a severe progressive neurological disorder. Hum Mol Genet. 2022 Aug 25;31(17):2934-2950. doi: 10.1093/hmg/ddac085. PMID: 35405010; PMCID: PMC9433733.
30. Shankar SP, Grimsrud K, Lanoue L, Egense A, Willis B, Hörberg J, AlAbdi L, Mayer K, Ütkür K, Monaghan KG, Krier J, Stoler J, Alnemer M, Shankar PR, Schaffrath R, Alkuraya FS, Brinkmann U, Eriksson LA, Lloyd K, Rauen KA; Undiagnosed Diseases Network. A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. Genet Med. 2022 Jul;24(7):1567-1582. doi: 10.1016/j.gim.2022.03.014. Epub 2022 Apr 28. PMID: 35482014; PMCID: PMC9426662.
31. Kozycki CT, Kodati S, Huryn L, Wang H, Warner BM, Jani P, Hammoud D, Abu-Asab MS, Jittayasothorn Y, Mattapallil MJ, Tsai WL, Ullah E, Zhou P, Tian X, Soldatos A, Moutsopoulos N, Kao-Hsieh M, Heller T, Cowen EW, Lee CR, Toro C, Kalsi S, Khavandgar Z, Baer A, Beach M, Long Priel D, Nehrebecky M, Rosenzweig S, Romeo T, Deuitch N, Brenchley L, Pelayo E, Zein W, Sen N, Yang AH, Farley G, Sweetser DA, Briere L, Yang J, de Oliveira Poswar F, Schwartz IVD, Silva Alves T, Dusser P, Koné-Paut I, Touitou I, Titah SM, van Hagen PM, van Wijck RTA, van der Spek PJ, Yano H, Benneche A, Apalset EM, Jansson RW, Caspi RR, Kuhns DB, Gadina M, Takada H, Ida H, Nishikomori R, Verrecchia E, Sangiorgi E, Manna R, Brooks BP, Sobrin L, Hufnagel RB, Beck D, Shao F, Ombrello AK, Aksentijevich I, Kastner DL; Undiagnosed Diseases Network. Gain-of-function mutations in ALPK1 cause an NF-κB-mediated autoinflammatory disease: functional assessment, clinical phenotyping and disease course of patients with ROSAH syndrome. Ann Rheum Dis. 2022 Jul 22:annrheumdis-2022-222629. doi: 10.1136/annrheumdis-2022-222629. Epub ahead of print. PMID: 35868845.

REVIEWS

1.Elsas LJ, **Longo N**, Fotion TR, Langley S (1988) Comparison of the insulin receptor gene and insulin binding in families with severe insulin resistance. ***Trans Ass Am Physicians***, *CI*: 137-148.

2.Elsas LJ, **Longo N**, Langley S, Griffin LD, Shuster RC (1989) Molecular genetics of severe insulin resistance. ***Yale J Biol Med*** *62*: 533-547.

Nicola Longo MD PhD

3.**Longo N**, Bell GI, Shuster RC, Griffin LD, Langley SD, Elsas LJ (1990) Human fibroblasts express the insulin-responsive glucose transporter (GLUT4). ***Trans Ass Am Physicians*** *CIII*: 202-213.

4.Elsas LJ, **Longo N** (1992) Glucose transporters. ***Annu Rev Medicine*** *43*: 377-393.

5.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1992) Mutations in the insulin receptor and their effect on glucose transport. ***Trans Ass Am Physicians*** *CV*: 204-213.

6.Elsas LJ, **Longo N** (1995) Glucose transporters: Human disorders and insulin receptor regulation. ***Int Pediatrics*** 10: 57-68.

7. **Longo N,** Elsas LJ (1998) Human glucose transporters. **Adv Pediatrics** 45: 293-313.

8.Scaglia F, **Longo N** (1999) Primary and secondary alterations of neonatal carnitine metabolism. ***Semin Perinatol*** 23: 152-161.

9**. Longo N** (2003) Mitochondrial Encephalopathy. **Neurologic Clinics** 21: 817-831

10. Bryant KG, Horns KM, **Longo N**, Schiefelbein J (2004) A primer on newborn screening. ***Adv Neonatal Care*** 4(5): 306-17.

11. **N Longo** (2006) Inborn errors of metabolism: New challenges with expanded newborn screning programs. ***Am J Med Genet C Semin Med Genet*** 142(2):61-3.

12.Pasquali M, Monsen G, Richardson L, Alston M, **Longo N** (2006) Biochemical findings in common inborn errors of metabolism. ***Am J Med Genet C Semin Med Genet*** 142(2):64-76

13. **Longo N**, Amat di San Filippo C, Pasquali M (2006) Disorders of carnitine transport and the carnitine cycle. ***Am J Med Genet C Semin Med Genet*** 142(2):77-85. PubMed PMID: 16602102; PubMed Central PMCID:PMC2557099

14. Hedlund GL, **Longo N**, Pasquali M (2006) Glutaric acidemia type 1. ***Am J Med Genet C Semin Med Genet*** 142(2):86-94.

15**. Longo N** (2009) Disorders of biopterin metabolism. ***J Inherit Metab Dis*** 32(3):333-42.

16. Pasquali M, **Longo N**. 2011. Newborn screening and inborn errors of metabolism. ***Am J Med Genet C Semin Med Genet*** 157(1):1-2.

17. **Longo N**, Ardon O, Vanzo R, Schwartz E, Pasquali M. 2011. Disorders of creatine transport and metabolism. ***Am J Med Genet C Semin Med Genet*** 57(1):72-8

18. **Longo N**. I difetti del trasporto di carnitina. (2011) ***Riv Ital Med Perinatale*** 11: 10-11

19. Greene CL, **Longo N**. 2014. National Institutes of Health (NIH) review of evidence in phenylalanine hydroxylase deficiency (phenylketonuria) and recommendations/guidelines for therapy from the American College of Medical Genetics (ACMG) and Genetics Metabolic Dietitians International (GMDI). ***Mol Genet Metab.*** 014 Jun;112(2):85-6. doi: 10.1016/j.ymgme.2014.03.005. Epub 2014 Mar 25. PMID: 24745849

20. Blau N, **Longo N** (2015) Alternative therapies to address the unmet medical needs of patients with phenylketonuria. ***Expert Opin Pharmacother*** 2015 Apr;16(6):791-800. doi: 10.1517/14656566. 2015.1013030. Epub 2015 Feb 7.PMID: 25660215

21. **Longo N**, Frigeni M, Pasquali M. Carnitine transport and fatty acid oxidation. ***Biochim Biophys Acta***. 2016 Oct;1863(10):2422-35. doi: 10.1016/j.bbamcr.2016.01.023. PMID: 26828774

22. **Longo N**. Primary Carnitine Deficiency and Newborn Screening for Disorders of the Carnitine Cycle. ***Ann Nutr Metab***. 2016 Dec 9;68 Suppl 3:5-9. [Epub ahead of print] PMID: 27931018

23. Winter S, Buist NR, **Longo N**, Armenian SH, Lopaschuk G, Wasilewska A. Round Table Discussion. ***Ann Nutr Metab***. 2016 Dec 9;68 Suppl 3:21-23 PMID: 27931031

24. Holt RJ, **Longo N** (2017) Glycerol phenylbutyrate for the maintenance treatment of patients with deficiencies in enzymes of the urea cycle. ***Expert Opinion on Orphan Drugs***, 5:12, 999-1010, DOI: 10.1080/21678707.2017.1405807

25. Steuerwald U, Lund AM, Rasmussen J, Janzen N, Hougaard DM, **Longo N** (2017) Neonatal Screening for Primary Carnitine Deficiency: Lessons Learned from the Faroe Islands. ***Int. J. Neonatal Screen***. 2017, 3, 1:1-10.

26. Häberle J, Chakrapani A, Ah Mew N, **Longo N**. Hyperammonaemia in classic organic acidaemias: a review of the literature and two case histories. ***Orphanet J Rare Dis***. 2018 Dec 6;13(1):219. doi: 10.1186/s13023-018-0963-7. Review. PMID: 30522498

27. van Spronsen FJ, Blau N, Harding C, Burlina A, **Longo N**, Bosch AM. Phenylketonuria. ***Nat Rev Dis Primers***. 2021 May 20;7(1):36. doi: 10.1038/s41572-021-00267-0. PMID: 34017006

28. Hagen-Lillevik S, Rushing JS, Appiah L, **Longo N**, Andrews A, Lai K, Johnson J. Pathophysiology and management of classic galactosemic primary ovarian insufficiency. ***Reprod Fertil***. 2021 Jun 25;2(3):R67-R84. doi: 10.1530/RAF-21-0014. PMID: 35118398; PMCID: PMC8788619.

29. **Longo N**, Sass JO, Jurecka A, Vockley J. Biomarkers for drug development in propionic and methylmalonic acidemias. ***J Inherit Metab Dis***. 2022 Jan 17. doi: 10.1002/jimd.12478. Epub ahead of print. PMID: 35038174.

30. Weinreb NJ, Goker-Alpan O, Kishnani PS, **Longo N**, Burrow TA, Bernat JA, Gupta P, Henderson N, Pedro H, Prada CE, Vats D, Pathak RR, Wright E, Ficicioglu C. The diagnosis and management of Gaucher disease in pediatric patients: Where do we go from here? ***Mol Genet Metab.*** 2022 Mar 9:S1096-7192(22)00152-4. doi: 10.1016/j.ymgme.2022.03.001. Epub ahead of print. PMID: 35367141.

BOOK CHAPTERS

1.Gazzola GC, Dall'Asta V, Franchi-Gazzola R, Bussolati O, **Longo N**, Guidotti GG (1985) Amino acid transport in normal and neoplastic cultured human fibroblasts. in **Cell Membranes and Cancer** (Galeotti T, Cittadini A, Neri G, Papa S, Smets LA, eds), Elsevier Science Publishers, Amsterdam, The Netherlands, 169-174.

2.Dall'Asta V, Bussolati O, **Longo N**, Gazzola GC (1988) The energization of uphill amino acid transport in cultured human fibroblasts. in **Molecular Basis of Biomembrane Transport** (Palmieri F, Quagliariello E, eds), Elsevier Science Publishers, Amsterdam, The Netherlands, 99-108.

3.Bussolati O, **Longo N**, Dall'Asta V, Gazzola GC, Guidotti GG (1990) The regulation of amino acid transport in senescent cultured human fibroblasts. In **Protein Metabolism in Aging** (Segal HL, Rothstein M, Bergamini E, Eds), Wiley-Liss, New York, 245-254.

4.Elsas LJ, **Longo N**, Rosenberg LE (1997) Inherited disorders of amino acid metabolism and storage. In ***Harrison's*** **Principles of Internal Medicine**, 14th Edition, Fauci AS, Braunwald E, Isselbacher KJ, Wilson JD, Martin JB, Kasper DL, Hauser SL, Longo DL (eds), chapter **349**, 2194-2203.

5.Elsas LJ, **Longo N**, Rosenberg LE (1997) Inherited defects of membrane transport. In ***Harrison's*** **Principles of Internal Medicine**, 14th Edition, Fauci AS, Braunwald E, Isselbacher KJ, Wilson JD, Martin JB, Kasper DL, Hauser SL, Longo DL (eds), chapter **350**, 2203-2208.

6.**Longo N** (2001) Inherited disorders of amino acid metabolism and storage. In ***Harrison's*** **Principles of Internal Medicine**, 15th Edition, Braunwald E, Fauci AS, Kasper DL, Hauser SL, Longo DL, Jameson JL (eds), Chapter 352: 2301-2309.

7.**Longo N** (2001) Inherited defects of membrane transport. In ***Harrison's*** **Principles of Internal Medicine**, 15th Edition, Braunwald E, Fauci AS, Kasper DL, Hauser SL, Longo DL, Jameson JL (eds), Chapter 353: 2309-2316.

8.**Longo N** (2003) Leprechaunism. In ***The NORD Guide to Rare Disorders***, Gruson ES (ed), Lippincott, Williams and Wilkins Inc., Philadelphia, 215-216.

9.**Longo N**, Amat di San Filippo C, Pasquali M (2003) The OCTN2 carnitine transporter and fatty acid oxidation. In **Membrane Transport Diseases: The Molecular Basis of Inherited Transport Defects** (Broer S, Wagner CA, Eds), Kluwer Academic/ Plenum Publishers, New York, Chapter 11, 161-174.

10.**Longo N** (2004) Inherited disorders of amino acid metabolism presenting in adults. In ***Harrison's*** **Principles of Internal Medicine**, 16th Edition, Kasper DL, Braunwald E, Fauci AS, Hauser SL, Longo DL, Jameson JL (eds) 2331-2334.

11.**Longo N** (2004) Inherited defects of membrane transport. In ***Harrison's* Principles of Internal Medicine**, 16th Edition, Kasper DL, Braunwald E, Fauci AS, Hauser SL, Longo DL, Jameson JL (eds), 2335-2337.

12. **Longo N**, Amat di San Filippo C, Schwarz EL, Pasquali M. (2009) Primary carnitine deficiency. Encyclopedia of Molecular Mechanisms of Disease, Springer, Heidelberg, Germany, 282-283.

13. **Longo N** (2008) Inherited disorders of amino acid metabolism presenting in adults. In ***Harrison's* Principles of Internal Medicine**, 17th Edition, Fauci AS, Kasper DL, Braunwald E, Hauser SL, Longo DL, Jameson JL, Loscalzo J (eds), Chapter 358: 2470-2474.

14. **Longo N** (2008) Inherited defects of membrane transport. In ***Harrison's* Principles of Internal Medicine**, 17th Edition, Fauci AS, Kasper DL, Braunwald E, Hauser SL, Longo DL, Jameson JL, Loscalzo J (eds), Chapter 359: 2474-2476.

15. Stanley CA, Bennett MJ, **Longo N** (2012) Plasma membrane carnitine transporter defect. In **The Online Metabolic & Molecular Bases of Inherited Diseases (OMMBID)**, Valle D, Beaudet AL, Vogelstein B, Kinzler KW, Antonarakis SE, Ballabio A (eds), Chapter 101.1 (Nov 2012), http://dx.doi.org/10.1036/ommbid.297.

16. Pasquali M, **Longo N**. Newborn Screening and Inborn Errors of Metabolism. Chapter 57. In: Burtis CA, Ashwood ER, Bruns DE, eds. ***Tietz Textbook of Clinical Chemistry and Molecular Diagnostics.*** 5th ed. St. Louis: Saunders, 2012, 2045-2082

17. **Longo N** (2011) Inherited disorders of amino acid metabolism presenting in adults. In ***Harrison's* Principles of Internal Medicine**, 18th Edition, Longo DL Fauci AS, Kasper DL, Hauser SL, Jameson JL, Loscalzo J (eds), 18th ed, Chapter 364: 3214-3219.

18. **Longo N** (2011) Inherited defects of membrane transport. In ***Harrison's* Principles of Internal Medicine**, 18th Edition, Longo DL Fauci AS, Kasper DL, Hauser SL, , Jameson JL, Loscalzo J (eds), Chapter 365: 3220-3222.

19. Pasquali M, **Longo N** (2012) Newborn screening for metabolic disorders. In ***Molecular Genetics and Personalized Medicine*,** Best DH, Swensen JJ (eds), Springer Science and Business media LLC, 163-198.

20. Pasquali M**, Longo N**. 2015. Newborn Screening and Inborn Errors of Metabolism. In Tietz Fundamentals of Clinical Chemistry, Burtis CA, Bruns DE, eds, 7th edition, Chapter 45:870-884.

21. Pasquali M**, Longo N**. 2014. Amino Acids. In Physician's guide to the diagnosis, treatment, and follow-up of inherited metabolic diseases. Blau N, Duran M, Gibson KM, Dionisi-Vici C, eds, Springer-Verlag Berlin Heidelberg, Chapter 49, 749-760

22. **Longo N** (2015) Inherited disorders of amino acid metabolism presenting in adults. In ***Harrison's* Principles of Internal Medicine**, 19th Edition, Dennis Kasper, Anthony Fauci, Stephen Hauser, Dan Longo, J. Larry Jameson, Joseph Loscalzo (eds), Chapter 434e.

23. **Longo N** (2015) Inherited defects of membrane transport. In ***Harrison's* Principles of Internal Medicine**, 19th Edition Dennis Kasper, Anthony Fauci, Stephen Hauser, Dan Longo, J. Larry Jameson, Joseph Loscalzo (eds), Chapter 435e.

24. Pasquali M, **Longo N**. Newborn Screening and Inborn Errors of Metabolism. In: Burtis CA, Ashwood ER, Bruns DE, eds. ***Tietz Textbook of Clinical Chemistry and Molecular Diagnostics***. 6th ed. St. Louis: Saunders, 2017, Chapter 70

25. Vockley J, **Longo N**, Andresen BS, Bennett MJ. Mitochondrial fatty acid oxidation defects. Chapter 9. In: Sarafoglou K, ***Pediatric Endocrinology and Inborn Errors of Metabolism: 2nd Edition***, McGraw-Hill Professional, 2017. 51-70

26. **Longo N** (2018) Inherited disorders of amino acid metabolism presenting in adults. In ***Harrison's* Principles of Internal Medicine**, 20th Edition, J. Larry Jameson, Anthony Fauci, Dennis Kasper, Stephen Hauser, Dan L. Longo, J, Joseph Loscalzo (eds), Chapter 413. 3015-3020

27. **Longo N** (2018) Inherited defects of membrane transport. In ***Harrison's* Principles of Internal Medicine**, 20th Edition J. Larry Jameson, Anthony Fauci, Dennis Kasper, Stephen Hauser, Dan L. Longo, J, Joseph Loscalzo (eds), Chapter 414. 3020-3023

28. Pasquali M, **Longo N**. 2020. Newborn Screening and Inborn Errors of Metabolism. In ***Tietz Fundamentals of Clinical Chemistry***, Rifai N, Horvath AR, Wittwer CT, eds. 8th edition, 882-897.

29. Pasquali M, **Longo N.** 2020. Amino Acids. In ***Physician's guide to the diagnosis, treatment, and follow-up of inherited metabolic diseases***. Blau N, Duran M, Gibson KM, Dionisi-Vici C, eds, Springer-Verlag Berlin Heidelberg, in press.

30. **Longo N**. 2021. Enzyme substitution therapy in PKU. In: ***Phenylketonuria and BH4 Deficiencies***, Nenad Blau (ed), UNI-MED Verlag AG, D-=28323, Bremen, 82-87. ISBN 978-3-8374-1601-5, pp. 82-87.

31. Pasquali, M., **Longo, N.** (2022). Amino Acids. In: Blau, N., Dionisi Vici, C., Ferreira, C.R., Vianey-Saban, C., van Karnebeek, C.D.M. (eds) ***Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases***. Springer, Cham. https://doi.org/10.1007/978-3-030-67727-5_3, Chapter 3, 41-50.

32. **Longo N.** (2022) Inherited Disorders of Amino Acid Metabolism in Adults. In: Loscalzo J, Fauci A, Kasper D, Hauser S, Longo D, Jameson J. eds. ***Harrison's Principles of Internal Medicine 21***. McGraw Hill; 2022. https://accessmedicine.mhmedical.com/content.aspx?bookid=3095&sectionid=265447517, Chapter 420. 3015-3020

33. **Longo N** (2022) Inherited defects of membrane transport. In: Loscalzo J, Fauci A, Kasper D, Hauser S, Longo D, Jameson J. eds. ***Harrison's Principles of Internal Medicine 21***. McGraw Hill; 2022. https://accessmedicine.mhmedical.com/content.aspx?bookid=3095&sectionid=265447545, Chapter 421. 3020-3023

34. **Longo, N**., Lai, K. (2022). Gene Therapy for Inherited Metabolic Diseases. In: Bernstein, L.E., Rohr, F., van Calcar, S. (eds) ***Nutrition Management of Inherited Metabolic Diseases***. Springer, Cham. https://doi.org/10.1007/978-3-030-94510-7_8, 97-109.

35. **Longo, N**., Andrews, A., Rohr, F. (2022). Medical and Nutrition Management of Phenylketonuria: Pegvaliase. In: Bernstein, L.E., Rohr, F., van Calcar, S. (eds) ***Nutrition Management of Inherited Metabolic Diseases.*** Springer, Cham. https://doi.org/10.1007/978-3-030-94510-7_11, 153-167

36. Pasquali M, Longo N. 2022. Newborn Screening and Inborn Errors of Metabolism. In ***Tietz Textbook of Laboratory Medicine.*** Rifai N, Chiu RWK, Young I, Burnham C-AD, Wittwer CT, eds. 7th edition, ISBN-13: 978-0323775724, ISBN-10: 0323775721, Chapter 60.

BOOK REVIEWS/COMMENTARIES/LETTERS TO THE EDITOR

1. **Longo N** (2003) A clinical guide to inherited metabolic disorders. JTR Clarke, Cambridge University Press. **Am J Med Genet**. 120A: 588.
2. **Longo N** (2011) Carnitina: Perche'? (2011) **Riv Ital Med Perinatale** 11: 5
3. Greene CL, **Longo N.** 2014. National Institutes of Health (NIH) review of evidence in phenylalanine hydroxylase deficiency (phenylketonuria) and recommendations/guidelines for therapy from the American College of Medical Genetics (ACMG) and Genetics Metabolic Dietitians International (GMDI). **Mol Genet Metab**. 2014 Mar 25. pii: S1096-7192(14)00093-6. doi: 10.1016/j.ymgme.2014.03.005. PMID: 24745849

PUBLISHED ABSTRACTS

1.Guidotti GG, Dall'Asta V, Franchi-Gazzola R, **Longo N**, Gazzola GC (1981) The regulation of the transport of neutral amino acids across the cell membrane. A working model. **Federation Proceedings** *40*: 570.

2.Dall'Asta V, Franchi-Gazzola R, Bussolati O, **Longo N**, Gazzola GC (1981) Studio cinetico del trasporto di acido glutamico in fibroblasti umani in coltura. Atti XXVII Congresso Nazionale Società Italiana di Biochimica, Parma 14-16 ottobre 1981, 493- 494.

3.Bussolati O, **Longo N**, Spagnolo C, Gazzola GC, Passeri M (1981) Il trasporto di aminoacidi in fibroblasti umani in coltura: influenze dell'invecchiamento *in vivo* e *in vitro*. **Giornale di Gerontologia** *29*: 581-582.

4.Bussolati O, **Longo N**, Dall'Asta V, Franchi-Gazzola R, Gazzola GC (1982) Changes in amino acid transport activity during *in vitro* aging of human fibroblasts. International Congress "**Ageing and Environmental Factors**", Ancona 5-11 settembre 1982, Session 23.

5.**Longo N**, Franchi-Gazzola R, Dall'Asta V, Bussolati O, Gazzola GC (1983) Trasporto di membrana di L-cistina in fibroblasti umani in coltura: aspetti cinetici e regolativi. Atti II Congresso Nazionale Associazione di Biologia Cellulare e del Differenziamento, Bressanone 2-4 maggio 1983, 88.

6.Laris PC, Bussolati O, **Longo N**, Gazzola GC, Guidotti GG (1983) L'influenza del potenziale di membrana sull'attività dei sistemi di trasporto per gli aminoacidi nei fibroblasti umani fetali. Atti VI Congresso Società Italiana di Biofisica Pura e Applicata, Camogli 6-8 ottobre 1983, 82-84.

Nicola Longo MD PhD

7.Dall'Asta V, **Longo N**, Bussolati O, Franchi-Gazzola R, Gazzola GC (1983) Trasporto accoppiato di sodio e aminoacidi in fibroblasti umani in coltura. Atti VI Congresso Società Italiana di Biofisica Pura e Applicata, Camogli 6-8 ottobre 1983, 63-65.

8.Dall'Asta V, Gazzola GC, **Longo N**, Bussolati O, Franchi-Gazzola R, Guidotti GG (1984) Controllo della espressione della regolazione adattativa del trasporto di aminoacidi in fibroblasti umani in coltura. Atti XVII Congresso Società Italiana di Patologia, Firenze 11-13 aprile 1984, 56.

9.Guidotti GG, Franchi-Gazzola R, Dall'Asta V, Bussolati O, **Longo N**, Gazzola GC (1984) The regulation of amino acid transport in human fibroblasts. **Fed Proc** 43: 1814 (Abs 2319).

10.Guidotti GG, Laris PC, Bussolati O, **Longo N**, Dall'Asta V, Franchi-Gazzola R, Gazzola GC (1984) Influence of membrane potential on the activity of amino acid transport systems in human fibroblasts. **Fed Proc** *43*: 1777 (Abs 2109).

11.Gazzola GC, Dall'Asta V, Franchi-Gazzola R, Bussolati O, **Longo N**, Guidotti GG (1984) Coupled transport of sodium and amino acids in cultured human fibroblasts. **Fed Proc** *43*: 1777 (Abs 2108).

12.**Longo N**, Franchi-Gazzola R, Bussolati O, Dall'Asta V, Gazzola GC (1984) Regolazione insulinica del trasporto di aminoacidi in fibroblasti umani in coltura. Atti XXX Congresso Nazionale Società Italiana di Biochimica, Lacco Ameno d'Ischia 7-10 ottobre 1984, 173.

13.Dall'Asta V, Franchi-Gazzola R, **Longo N**, Bussolati O, Gazzola GC, Guidotti GG (1984) Attività modulatrice di frazioni sieriche sulla velocità di trasporto degli aminoacidi in fibroblasti umani in coltura. Atti XXX Congresso Nazionale Società Italiana di Biochimica, Lacco Ameno d'Ischia 7-10 ottobre 1984, 172.

14.Bussolati O, Laris PC, **Longo N**, Gazzola GC, Dall'Asta V, Franchi-Gazzola R, Guidotti GG (1984) Impiego del rapporto di distribuzione del catione lipofilico TPP nello studio del potenziale di membrana in fibroblasti umani. Atti I Convegno Nazionale di Bioenergetica e Biofisica Cellulare, Parma 11-13 ottobre 1984.

15.Dall'Asta V, Franchi-Gazzola R, Bussolati O, **Longo N**, Gazzola GC (1984) Meccanismi di controllo del trasporto di aminoacidi in fibroblasti umani in coltura. Atti Convegno Congiunto ABCD- AGI-SIBBM, Siena 21-25 ottobre 1984, 138.

16.Bussolati O, Franchi-Gazzola R, Dall'Asta V, Gazzola GC, **Longo N**, Guidotti GG (1985) Effetto dell'invecchiamento sul trasporto di L-prolina in fibroblasti umani in coltura. Atti XXI Congresso Nazionale Società Italiana di Biochimica, Rimini 16-19 settembre 1985, 219.

17.**Longo N**, Spisni A, Franchi-Gazzola R, Bussolati O, Dall'Asta V, Gazzola GC (1985) La struttura molecolare della glicina giustifica la reattività dell'aminoacido con il Sistema A di trasporto nei fibroblasti umani. Atti XXXI Congresso Nazionale Società Italiana di Biochimica, Rimini 16-19 settembre 1985, 220.

18.Dall'Asta V, Rebecchi F, **Longo N**, Bussolati O, Gazzola GC, (1985) L'incubazione di fibroblasti umani in soluzione salina di Earle altera marcatamente le concentrazioni ioniche intracellulari. Atti IV Convegno Associazione di Biologia Cellulare e del Differenziamento, Torino 15-18 ottobre 1985, 181.

19.Bussolati O, Laris PC, Squassoni E, Ferrari MG, **Longo N**, Gazzola GC (1985) L'iperpolarizzazione mediata dalla Na,K- ATPasi ouabaino-sensibile stimola il trasporto di aminoacidi nei fibroblasti umani in coltura. Atti IV Convegno Associazione di Biologia Cellulare e del Differenziamento, Torino 15-18 ottobre 1985, 177.

20.**Longo N**, Gazzola GC, Elsas LJ (1986) Insulin regulation of membrane transport by normal and insulin-receptor mutant human fibroblasts. **Clin Res** *34*(1): 242A.

21.**Longo N**, Nagata N, Danner D, Priest J, Elsas LJ (1986) Insulin binding and stimulation of hexose transport by normal and receptor-defective human fibroblasts. **Fed Proc** *45*: 1836 (Abs 2074).

22.Elsas LJ, Nagata N, **Longo N**, Danner D, Priest J (1986) Normal and mutant human insulin receptors in cultured dermal fibroblasts. **Clin Res** *34*(2): 652A.

23.Elsas LJ, Nagata Y, **Longo N**, Priest JH (1986) Analysis of normal and mutant human insulin receptors. Proceedings VII International Congress of Human Genetics, Berlin 22-26 September 1986, 426.

24.Dall'Asta V, Gazzola GC, Bussolati O, **Longo N**, Nucci FA, Guidotti GG (1986) Perturbation of Na^+ and K^+ gradients in human fibroblasts incubated in saline solutions. Protective effect of selected amino acids. **Eur J Cell Biol** *42* (Suppl 16): 13 (Abs 39).

25.Bussolati O, Nucci FA, Zermani R, Dall'Asta V, **Longo N**, Gazzola GC (1986) The transport of neutral amino acids in cultured human keratinocytes. **Eur J Cell Biol** *42* (Suppl 16): 14 (Abs 40).

26.Elsas LJ, **Longo N**, Priest JH (1986) Analysis of normal and mutant human insulin receptors. **Am J Hum Genet** *39*: A8 (Abs 017).

27.Dall'Asta V, Gazzola GC, Bussolati O, **Longo N**, Nucci FA, Guidotti GG (1986) Ruolo della Na,K-ATPasi e di un canale furosemide-sensibile nelle modificazioni dei gradienti ionici indotte da incubazioni in soluzioni saline. Atti XVIII Congresso Società Italiana di Patologia, Roma 14-16 maggio 1986, 287-290.

28.**Longo N**, Elsas LJ, Franchi-Gazzola R, Dall'Asta V, Bussolati O, Gazzola GC (1986) Effetto dell'insulina sul trasporto di esosi e di aminoacidi. Differenze tra cellule normali ed insulino-resistenti. Atti XVIII Congresso Società Italiana di Patologia, Roma 14-16 maggio 1986, 299-302.

29.Bussolati O, Nucci FA, Laris PC, Gazzola GC, Dall'Asta V, **Longo N**, Guidotti GG (1986) Rapporto tra potenziale di membrana e accumulo degli aminoacidi cationici nei fibroblasti umani in coltura. Atti XVIII Congresso Società Italiana di Patologia, Roma 14-16 maggio 1986, 303-305.

30.**Longo N**, Elsas LJ (1987) Restriction fragment length polymorphism for the alpha subunit of the insulin receptor gene in inherited severe insulin resistance. **Clin Res** *35*(3): 624A.

31.**Longo N**, Griffin LD, Priest JH, Elsas LJ (1987) The regulation of 3-O-methyl-D-glucose transport by cultured human fibroblasts. **Fed Proc** *46*: 1982 (Abs 331).

32.Robertson DG, **Longo N**, Newby FD, DiGirolamo M, Merril AH, Lambeth JD (1987) D-Sphingosine inhibits insulin-stimulated hexose transport and glucose oxidation in isolated adipocytes. Proceedings 69E Annual Meeting of the Endocrine Society, Indianapolis (IN), June 10-12, 39 (Abs 71).

33.Elsas LJ, **Longo N** (1987) RFLPs for the *alpha* subunit of the insulin receptor gene in inherited severe insulin resistance. **Am J Hum Genet** *41*: A96 (Abs 281).

34.Fotion TR, **Longo N**, Langley S, Griffin L, Elsas LJ (1988) Insulin binding and insulin receptor gene analysis in heritable insulin resistance. **Clin Res** 36(1): 58A.

35.Gazzola GC, **Longo N**, Franchi-Gazzola R, Bussolati O, Dall'Asta V (1988) Regolazione insulinica del trasporto di aminoacidi in cellule umane in coltura. Congress Italian Society of Diabetology, Venezia Lido 22-25 Maggio 1988, CIC Edizioni Internazionali, 27-32.

36.Elsas LJ, Fotion TR, Langley S, **Longo N** (1988) Comparison of the insulin receptor gene and insulin binding in families with severe insulin resistance. **Clin Res** 36(3): 622A.

37.Bussolati O, Dall'Asta V, Franchi-Gazzola R, **Longo N**, Nucci FA, Rotoli BM, Gazzola GC (1988) Amino acid transport during in vitro aging of cultured human fibroblasts. Atti XV Congresso Nazionale Gruppo Italiano di Bioenergetica e Biomembrane, Trieste 17-18 giugno 1988.

38.Bussolati O, Rotoli BM, Nucci FA, **Longo N**, Dall'Asta V, Gazzola GC (1988) Elettrogenicità del trasporto di aminoacidi anionici nei fibroblasti umani in coltura. Atti 34E Congresso Società Italiana di Biochimica, Padova 2-4 ottobre 1988, 314.

Nicola Longo MD PhD

39.**Longo N**, Franchi-Gazzola R, Bussolati O, Dall'Asta V, Nucci FA, Gazzola GC (1988) Nutrient transport in mouse fibroblasts transfected by **ras** and **neu** oncogenes. **Eur J Cell Biol** 47 (Suppl 23): 27 (Abs 71).

40.**Longo N**, Bussolati O, Nucci FA, Rotoli BM, Dall'Asta V, Guidotti GG (1988) Caratteristiche colturali e trasporto di aminoacidi in cellule ottenute da espianti di tumore desmoide umano. Atti 19E Congresso Società Italiana di Patologia, Palermo 19-22 ottobre 1988, 278-281.

41.Bussolati O, **Longo N**, Nucci FA, Dall'Asta V, Franchi-Gazzola R, Gazzola GC (1988) Alterazioni del trasporto di aminoacidi associate all'invecchiamento in vitro di fibroblasti umani. Atti 19E Congresso Società Italiana di Patologia, Palermo 19- 22 ottobre 1988, 535-538.

42.Langley S, Griffin L, Shuster R, **Longo N**, Fotion T, Elsas LJ (1988) Heritable severe insulin resistance: Characterization of a new mutant insulin receptor. **Am J Hum Genet** 43: A190 (Abs 0759).

43.**Longo N**, Visigalli R, Franchi-Gazzola R, Dall'Asta V, Bussolati O, Guidotti GG, Gazzola GC (1989) Inhibition of aspartate transport in *ras*-transformed or phorbol ester treated NIH 3T3 cells. **European Journal of Cell Biology** *49* (Suppl 28) : 33 (abs 93)

44.Langley S, **Longo N**, Griffin L, Shuster R, Ashley C, Elsas LJ (1989) Heterogeneity in Leprechaunism: Two additional families with different cellular phenotype. **Am J Hum Genet** 45: A203 (Abs 0797).

45.**Longo N**, Bell GI, Shuster RC, Griffin LD, Langley SD, Elsas LJ (1990) Human fibroblasts express the insulin-responsive glucose transporter (GLUT4). **Clin Res** 38(2): 436A.

46.**Longo N**, Shuster RC, Griffin LD, Langley SD, Elsas LJ (1990) Insulin-receptor autophosphorylation and kinase activity are constitutively increased in fibroblasts cultured from a patient with leprechaunism. Proceedings of the 72E Annual Meeting of the Endocrine Society, Atlanta, GA, June 20-23, 45 (Abs 84).

47.**Longo N**, Shuster RC, Griffin LD, Langley SD, Elsas LJ (1990) Activation of the insulin-receptor kinase modifies glucose transporter gene expression in fibroblasts cultured from a patient with leprechaunism. Proceedings of the 5E International Congress of Inborn Errors of Metabolism, Pacific Grove, CA, 1-5 June 1990, W16.3.

48.Elsas LJ, **Longo N** (1990) Molecular genetics of severe insulin resistance. Proceedings of the 5E International Congress of Inborn Errors of Metabolism, Pacific Grove, CA, 1-5 June 1990, W16.4.

49.Elsas LJ, **Longo N**, Langley SD, Shuster RC, Griffin LD (1990) A mutation in the insulin receptor gene produces decreased insulin binding with constitutive increase in receptor autophosphorylation and kinase activity. **American Journal of Human Genetics** *47*: A 154 (604).

50.**Longo N**, Shuster RC, Griffin L, Elsas LJ (1991) Relationship between insulin binding and stimulation of glucose transport by CHO cells overexpressing insulin receptors. **FASEB J** 5: A1228 (4973).

51.**Longo N**, Langley SD, Griffin LD, Shuster RC, Elsas LJ (1991) Leprechaunism: a syndrome of severe insulin resistance caused by heterogeneous mutations in the insulin receptor gene. **American Journal of Human Genetics** 49: 101 (Abs 485).

52.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1992) A single amino acid substitution activates the insulin receptor in a patient with leprechaunism. Abstract Book, Annual Meeting of the Society for Inherited Metabolic Disorders, Pine Mountain, GA, March 20-23,1992, Poster 14.

53.**Longo N**, Shuster RC, Griffin LD, Langley SD, Elsas LJ (1992) Activation of insulin receptor signaling by a single amino acid substitution in the transmembrane domain. **Clinical Research** 40(2): 321A.

54.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1992) Homozygosity for a natural mutation in the insulin receptor *alpha* subunit activates glucose transport. **Clinical Research** 40(2): 329A.

55.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1992) Dissociation of the metabolic and mitogenic effects of the human insulin receptor by a mutation in its extracellular domain. **Am J Hum Genetics** 51 (Suppl): A172 (Abs 674).

56.**Longo N** (1992) Difetti del recettore dell'insulina. **Pathologica** 85: 28-30.

57.**Longo N**, Singh R, Griffin LD, Langley SD, Elsas LJ (1993) Insulin receptor signaling is required for rhGH to stimulate growth and IGF-I production. Proceedings of the 75E Annual Meeting of the Endocrine Society, Las Vegas, NV, June 9-12, 235 (Abs 737).

58.Ekstrand AJ, **Longo N**, Collins VP, James CD (1993) Functional analysis of an EGF receptor with a truncation in the extracellular domain that is frequently produced in glioblastomas with EGFR gene amplification. **J Neuro Oncology** 15(Suppl): S8.

59.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1993) Identification of two novel mutations in the insulin receptor gene of a patient with leprechaunism: application to prenatal diagnosis. **Am J Hum Genet** 53: Abs 922.

60.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1994) The insulin receptor tolerates substitution of arginine 86 with alanine and glycine, but not with proline. **Clin Res** 42(2): 209A.

61.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1994) Prenatal diagnosis in leprechaunism: two novel mutations in the insulin receptor gene. Proceedings VI International Congress Inborn Errors of Metabolism, Milano, Italy, May 27-31, 1994, W10.5, p 95.

62.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1994) Two null alleles for the insulin receptor and a concomitant defect of the EGF receptor in a severe form of leprechaunism.**Am J Hum Genet** 55: A229 (Abs 1338).

63.**Longo N**, Langley SD, Elsas LJ (1996) A severe form of leprechaunism caused by two null alleles for the insulin receptor.**J Invest Medicine** 3: 259A.

64.Pasquali M, Still MJ, Evinger JD, Dembure PP, **Longo N**, Elsas LJ (1996) Effect of vitamin C on pyridinium cross-link formation in skin fibroblasts from patients with Ehlers-Danlos syndrome type VI. **J Invest Medicine** 3: 226A.

65.Coleman KB, **Longo N**, Wieser J, Neu R, Gauthier T, Blackston RD, Saxe D (1996) Trisomy 5 mosaicism detected prenatally with a severely affected liveborn. **Am J Hum Genet** 59: A114 (Abs 630).

66.Scaglia F, Wang Y, Singh RH, Dembure PP, Pasquali M, Evinger JD, Bagnasco S, Fernhoff PM, **Longo N** (1997) Characterization of the carnitine transporter defective in primary carnitine deficiency. 4E Annual Meeting of the American College of Medical Genetics, Fort Lauderdale, FL (USA), February 28-March 2, 1997, A14 (Abs 27).

67.**Longo N**, Giannella-Neto D, Langley SD, Wang Y, Scaglia F, Elsas LJ (1997) Genotype-phenotype correlation in patients with inherited insulin-resistant syndromes. 7th International Congress of Inborn Errors of Metabolism. p. 80 (Abs O42).

68.Pasquali M, **Longo N**, Evinger JD, Dembure PP, Byers P, Elsas LJ (1997) Two abnormal patterns of pyridinium cross-link formation in skin fibroblasts of patients with Ehlers-Danlos syndrome type VI. 7th International Congress of Inborn Errors of Metabolism. p. 81 (Abs O43).

69.**Longo N**, Scaglia F, Wang Y, Singh RH, Dembure PP, Pasquali M, Evinger JD, Bagnasco S, Fernhoff P (1997) Physiological characterization of the carnitine transporter responsible for primary carnitine deficiency. 7th International Congress of Inborn Errors of Metabolism. p. 219 (Abs O106).

70.Scaglia F, Wang Y, Singh RH, Dembure PP, Pasquali M, Evinger JD, Bagnasco S, Fernhoff P, **Longo N** (1997) Functional characterization of the carnitine transporter responsible for primary carnitine deficiency. **Am J Hum Genet** 61: A261 (Abs 1517).

71.**Longo N**, Wang Y, Smith S, Langley SD, Scaglia F (1998) Genotype-phenotype correlation in patients with inherited insulin resistant syndromes. **Am J Hum Genet** 63: A24 (Abs 124).

72.Pasquali M, Ye J, Byers P, Elsas LJ, **Longo N** (1998) Pyridinium cross-links and lysyl hydroxylase (PLOD1) mRNA in Ehers-Danlos syndrome type VI. **Am J Hum Genet** 63: A272 (Abs 1568).

Nicola Longo MD PhD

73.Wang Y, Ye J, Ganapathy V, **Longo N** (1999) Mutations in the organic cation/carnitine transporter OCTN2 in primary carnitine deficiency. Annual Meeting of the Society for Inherited Metabolic Disorders, Lake Lanier Islands, GA, March 12-15, P40

74.Pasquali M, Ye J, Byers P, Elsas LJ, **Longo N** (1999) Pyridinium cross-links and lysyl hydroxylase (PLOD1) mRNA in Ehlers-Danlos syndrome type VI. Annual Meeting of the Society for Inherited Metabolic Disorders, Lake Lanier Islands, GA, March 12-15, P29

75.Wang Y, Taroni F, Garavaglia B, Cowan T, Ye J, **Longo N** (1999) Missense mutations in the organic cation transporter OCTN2 in patients with primary carnitine deficiency. **Am J Hum Genet** 65: A432 (Abs 2450)

76.DiMeglio LA, Smith S, Wang, Y, **Longo N**, Freidenberg GR (1999) Novel insulin receptor mutations responsible for severe insulin resistance with progressive growth failure. **Pediatric Research** 45(4): 88A.

77.**Longo N**, Meadows T, Wang Y (2000) Impaired sodium stimulation of carnitine transport in primary carnitine deficiency. **J Inherit Metab Dis** 23 (Suppl 1): 117 (Abs 233-O).

78.**Longo N**, Wang Y, Taroni F, Garavaglia B (2000) Functional analysis of mutations in the OCTN2 transporter causing primary carnitine deficiency: Lack of genotype-phenotype correlation. **J Inherit Metab Dis** 23 (Suppl 1): 118 (Abs 235-O).

79.Blackston RD, Coleman K, **Longo N**, May K (2001) Should subtelomeric FISH be done in all cases of deletion 1p36? **Genet Med** 3: 259 (Abs 95)

80.May KM, **Longo N** (2001) Interstitial microdeletion of 21q. **Am J Hum Genet** 69: 318 (Abs 791)

81.Canton CS, Garcia R, Pasquali M, Singh RH, Gibson KM, **Longo N** (2001) Succinyl CoA-3-keto transferase (SCOT) deficiency in a Panamenian patient. **Am J Hum Genet** 69: 479 (Abs 1738)

82.Pasquali M, Hobert JA, Liu A, **Longo N**, Singh RH (2003) Stability of sodium phenylbutyrate in combination with common liquid and solid food. **J Inherit Metab Dis** 26 (Suppl 2): 78 (Abs 156-P)

83. **Longo N**, Amat di San Filippo C (2003) Identification of functional residues in the OCTN2 carnitine transporter defective in primary carnitine deficiency. **J Inherit Metab Dis** 26 (Suppl 2): 88 (Abs 175-O)

84. Dobrowolski SF, McKinney JT, Amat di San Filippo C, **Longo N** (2003) Mutational screening in primary carnitine deficiency using LCGreen dye and high resolution melting analysis. **J Inherit Metab Dis** 26 (Suppl 2): 188 (Abs 375-O)

85. Dobrowolski SF, McKinney JT, **Longo N** (2003) Comprehensive homogeneous analysis of the MCAD gene. **J Inherit Metab Dis** 26 (Suppl 2): 188 (Abs 376-O)

86. Makhseed N, Vallance HD, Potter M, Waters PJ, Wong LTK, Lillquist Y, Amat di San Filippo C, **Longo N** (2003) Clinical heterogeneity in carnitine uptake disorder. **J Inherit Metab Dis** 26 (Suppl 2): 95 (Abs 189-P)

87.Toydemir R, **Longo N**, Brassington A, Bayrak-Toydemir P, Krakowiak P, Jorde LB, Bamshad M (2003) A new syndrome caused by a novel loss-of-function mutation in FGFR3. **Am J Hum Genet** 73 (Suppl): 171 (Abs 46)

88. Stevenson DA, Dent KM, Carey JC, Chen Z, **Longo N** (2003) 6q subtelomeric deletion: case report and review of the literature. **Am J Hum Genet** 73(Suppl): 280 (Abs 648)

89. **Longo N**, Miller N, Amat di San Filippo C (2003) Abnormal localization of OCTN2 carnitine transporters in primary carnitine deficiency. **Am J Hum Genet** 73 (Suppl): 446 (Abs 1617)

90. Schwartz E, **Longo N**, Pasquali M (2003) Effect of vitamin C on collagen and lysyl hydroxylase RNA in fibroblasts from patients with Ehlers-Danlos syndrome type VI. **Am J Hum Genet** 73 (Suppl): 447 (Abs 1623)

91. **Longo N**, Amat di San Filippo C (2004) Tyrosine residues affecting sodium stimulation of carnitine transport in the OCTN2 carnitine/organic cation transporter. **Mol Genet Metab** 81: 176 (Abs 47)

92. Amat di San Filippo C, **Longo N** (2005) Pharmacological rescue of mutant OCTN2 transporters in primary carnitine deficiency. **Mol Genet Metab** 84: 202 (Abs 1).

93. Ernst SL, Filipowicz HR, Ashurst CL, Pasquali M, Longo N (2005) Metabolic changes associated with hyperammonemia in patients with propionic acidemia. **Mol Genet Metab** 84: 2228-229 (Abs 48).

94. **Longo N,** Amat di San Filippo C (2005) Partial rescue of cytoplasm-retained OCTN2 carnitine transporters in primary carnitine deficiency. J Inherit Metab Dis 28(Suppl 1): 111 (Abs 220-P).

95. Berry SA, Bentler K, McCann M, **Longo N**, Pasquali M, Rinaldo P, Matern D, Schimmenti LA (2005) Expanded Newborn Screening identifies maternal primary carnitine deficiency. **Am J Hum Genet** 30: (Abs 64-O)

96. **Longo N**, Schrijver I, Vogel H, Pique LM, Cowan TM, Pasquali M, Hedlund GL, Ernst SE, Gallagher RC, Enns GM (2005) Moyamoya syndrome in a patient with concomitant mitochondrial and fatty acid oxidation disorders. **Am J Hum Genet** 259: Abs 1363-P.

97. **Longo N,** Amat di San Filippo C (2006) Glycosylation affects membrane maturation of the OCTN2 carnitine/organic cation transporter. **Acta BioMed** 77(suppl 3): 73-74.

98. Singh RH, Acosta PB, Kennedy MJ, **Longo N**, Elsas LJ (2006) Potassium retention in patients treated for argininosuccinate lyase deficiency. **J Inherit Metab Dis** 29 (suppl 1): 48 (Abs O-8-4)(Platform)

99. Crombez EA, Schimmenti LA, Chang E, Bentler K, Cederbaum SD, Berry SA, **Longo N** (2006) Maternal primary carnitine deficiency identified by newborn screening. **J Inherit Metab Dis** 29 (suppl 1): 55 (Abs O-14-5)(Platform)

100. Dimmock DP, Zhang Q, Shieh J, Chou P-C, Truong C, Schmitt E, Sifry-Platt M, Ficicioglu CH, Enns GM, Arch EM, **Longo N**, Lipson MH, Craigen WJ, Wong L-J (2007) Clinical features and molecular genetics of deoxyguanosine kinase deficiency. ACMG Annual Clinical Gentics Meeting, Nashville TN March 21-25,2007, 95 (Abs 29).

101. **Longo N** (2007) The challenge for the long-term surveillance of metabolic disorders detected by newborn screening. ACMG Annual Clinical Gentics Meeting, Nashville TN March 21-25,2007, 162 (Abs 282) (Invited)

102. Dimmock DP, Zhang Q, Shieh J, Chou P-C, Truong C, Schmitt E, Sifry-Platt M, Ficicioglu CH, Enns GM, Arch EM, **Longo N**, Lipson MH, Craigen WJ, Wong L-J (2007) The clinical features and molecular genetics of deoxyguanosine kinase deficiency. **Mol Genet Metab** 90: 250 (Abs 18).

103. **Longo N**, Amat di San Filippo C (2007) Glycosylation affects membrane maturation of the OCTN2 carnitine transporter. **Mol Genet Metab** 90: 257 (Abs 41).

104. Cassanello M, Caruso U, Cerone R, Schiaffino MC, **Longo N**, Pasquali M (2007) Primary carnitine deficiency in asymptomatic adult female diagnosed by low free carnitine in the newborn. **J Inherit Metab Dis** 30 (Suppl 1): 4 (Abs 016-P).

105. Trefz F, Burton B, **Longo N**, Levy H, Bebchuck J, Christ-Schmidt H, Martinez-Pardo M, Gruskin D, Dorenbaum A, Hennermann JB (2007) PKU 006: The effect of sapropterin dihydrochloride (tetrahydrobiopterin or 6R-BH4) treatment on phenylalanine (Phe) tolerance in children with phenylketonuria controlled on a Phe-restricted diet. **J Inherit Metab Dis** 30 (Suppl 1): 17 (Abs 065-P).

106. Dimmock DP, Zhang Q, Shieh J, Chou P-C, Truong C, Schmitt E, Sifry-Platt M, Ficicioglu CH, Enns GM, Arch EM, **Longo N**, Lipson MH, Craigen WJ, Wong L-J (2007) Deoxyguanosine kinase deficiency: Two diseases or one? **J Inherit Metab Dis** 30 (Suppl 1): 79 (Abs 313-P).

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107. Lloyd MG, Guymon R, Jungerberg R, **Longo N**, Pasquali M (2007) Renal carnitine reabsorption in primary and secondary carnitine deficiency. Annual Meeting of the American Society of Human Genetics, October 24-27, 2007, San Diego, California 295 (Abs 1470/W)

108. Wasserstein M, Burton B, Grange D, Harding C, Lipson M, **Longo N**, Waber l, Whitley C, Wolff J, Bebchuk J, Dorenbaum A, Vockley G (2007) Dose-related effect of sapropterin dihydrochloride (sapropterin) on blood phenylalanine (Phe) in patients with phenylketonuria (PKU). Annual Meeting of the American Society of Human Genetics, October 24-27, 2007, San Diego, California 425 (Abs 2231/W)

109. Amat di San Filippo C, Taylor MRG, Mestroni L, Botto LD, **Longo N** (2007) Heterozygous mutations in the carnitine transporter gene SLC22A5 are not associated with cardiomyopathy. Annual Meeting of the American Society of Human Genetics, October 24-27, 2007, San Diego, California 351 (Abs 1804/T).

110. Dimmock D, Dionisi-Vici C, Zhang Q, Shieh J, Truong C, Schmitt E, Sifry-Platt M, Carrozzo R, Lucioli S, Ficicioglu C, Enns G, Arch E, **Longo N**, Lipson M, Vallance H, Scaglia F, Wong L-J (2007) Clinical and Molecular Features of Mitochondrial DNA depletion due to Mutations in Deoxyguanosine Kinase. Annual Meeting of the American Society of Human Genetics, October 24-27, 2007, San Diego, California 302 (Abs 1510/T)

111. Furtado LV, Cantor Nl, Ernst SL, Fulton JB, **Longo N** (2007) Correlation of phenylalanine levels with intellectual outcome and executive functioning in patients with phenylketonuria. Annual Meeting of the American Society of Human Genetics, October 24-27, 2007, San Diego, California 431 (Abs 2267/T).

112. Viskochil D, Ashurst C, Hung I, Carey J, Bleyl S, **Longo N** (2007) Idursulfase Replacement Therapy in 2 Infants with Hunter Syndrome. Annual Meeting of the American Society of Human Genetics, October 24-27, 2007, San Diego, California 431 (Abs 2268/T)

113. Calderon FRO, Schwarz L, Amat di San Filippo C, Pasquali M, **Longo N**, Mao R (2007) Molecular Diagnosis of Primary Carnitine Deficiency. Annual Meeting of the American Society of Human Genetics, October 24-27, 2007, San Diego, California 307 (Abs 1541/F).

114. Gruskin D, Dorenbaum A, Bebchuk J, **Longo N** (2007) Sapropterin dihydrochloride (sapropterin) increases phenylalanine (Phe) tolerance in children with phenylketonuria (PKU) maintained on a Phe-restricted diet. Annual Meeting of the American Society of Human Genetics, October 24-27, 2007, San Diego, California 88 (Abs 269) (Platform)

115. Arnold GL, Koeberl DD, Barshop BA, Burton BK, Cederbaum S, Feigenbaum A, Harding CO, Kronn D, Matern D, Gibson JB, Garganta CL, Braverman N, **Longo N**, Kahler SG, the 3-MCC working group (2007) Clinical practice protocols for 3-methylcrotonyl CoA carboxylase (3-MCC) deficiency. Annual Meeting of the American Society of Human Genetics, October 24-27, 2007, San Diego, California 89 (Abs 273) (Platform)

116. **Longo N**, Amat di San Filippo C, Ardon O (2008) Effect of glycosylation on activity and membrane maturation of the OCTN2 carnitine transporter. **Mol Genet Metab** 93: 255 (Abs 57).

117. Holbrook R, Staes C, **Longo N**, Botkin J, Anderson R, Mitchell J (2007) Development of requirements and a pilot registry for long-term follow-up of children with heritable conditions. AMIA Annu Symp Proc. 2007 Oct 11:977.

118. **Longo N**, Abbott M-A, Yatsenko A, Dimmock D, Wong L-JC (2008) The spectrum of BCSIL related conditions. Annual Meeting of the American Society of Human Genetics, November 11-15, 2008, Philadelphia, PA (Abs 497) (Poster)

119. Arnold GL, Matern D, VanHove J, Freedenberg D, **Longo N**, Burton B, Garganta C, Ficicioglu C, Cederbaum C, Harding C, Boles R, Feigenbaum A, Chakraborty P, Strauss A (2008) A Delphi-based Consensus Clinical Practice Protocol for Very Long Chain Acyl-CoA Dehydrogenase (VLCAD)Deficiency. Annual Meeting of the American Society of Human Genetics, November 11-15, 2008, Philadelphia, PA (Abs 804) (Poster)

120. Ardon O, Amat di San Filippo C, Salomons GS, **Longo N** (2009) Creatine transporter deficiency in two half-brothers. Annual Meeting American College of Medical Genetics, Tampa, FL, 25-29 March 2009, p. 117 (Abstract 83)(Poster).

121. **Longo N** (2009) Sugar, small molecules, and cardiomyopathy. Annual Meeting American College of Medical Genetics, Tampa, FL, 25-29 March 2009, p. 200 (Abstract 409)(Platform).

122. **Longo N**, Amat di San Filippo C, Vanzo R, Botto LD, Pasquali M (2009) Partial carnitine transporter deficiency in a patient with carnitine palmitoyltransferase-2 deficiency. 11th International Congress of Inborn errors of Metabolism, San Diego CA, 29 Aug-2 Sep, 2009, Molec Genet Metab 98: 50 (Abs 306).

123. Vanzo R, Ernst SL, Botto LD, Pasquali M, **Longo N** (2009) Glutaric acidemia type I in Utah. 11th International Congress of Inborn errors of Metabolism, San Diego CA, 29 Aug-2 Sep, 2009, Molec Genet Metab 98: 104 (Abs 541).

124. Pasquali M, Haslip C, Keune F, Randall H, **Longo N** (2009) Changes with age of collection in the concentration of amino acids and acylcarnitines in blood spots from newborns. 11th International Congress of Inborn errors of Metabolism, San Diego CA, 29 Aug-2 Sep, 2009, Molec Genet Metab 98: 106 (Abs 549).

125. K. Damjanovich, T. Caine, L. Schwarz, M. Pasquali, **N. Longo**, P. Bayrak-Toydemir (2009) Mutation Analysis of PLOD1 gene in Ehlers-Danlos Syndrome Type VI. 59th Annual Meeting of the American Society of Human Genetics, October 20-24, 2009, Honolulu, HI (Abs 364) (Poster)

126. O. Ardon, **N. Longo** (2009) Identification of proteins interacting with the OCTN2 transporter responsible for primary carnitine deficiency. 59th Annual Meeting of the American Society of Human Genetics, October 20-24, 2009, Honolulu, HI (Abs 3007) (Poster)

127. U. C. N. Erlingsson, A. Liu, O. Ardon, M. Pasquali, **N. Longo** (2009) ACYLCARNITINE PROFILING AND FATTY ACID TOXICITY IN FIBROBLASTS FROM PATIENTS WITH FATTY ACID OXIDATION DEFECTS. 59th Annual Meeting of the American Society of Human Genetics, October 20-24, 2009, Honolulu, HI (Abs 3030) (Poster)

128. T. Lewis, **N. Longo**, E. Lyon (2009) A Custom Microarray for Fatty Acid Oxidation Disorders. 59th Annual Meeting of the American Society of Human Genetics, October 20-24, 2009, Honolulu, HI (Abs 3032) (Poster)

129. M. Pasquali, C. Haslip, **N. Longo**, F. Keune, H. Randall (2009) Amino acids and acylcarnitines concentration in blood spots from low birth weight infants. 59th Annual Meeting of the American Society of Human Genetics, October 20-24, 2009, Honolulu, HI (Abs 3096) (Poster)

130. **Longo N**, Harding CO, Burton BK, Grange DK, Vockley J, Wasserstein M, Rice G, Wolff JA, Whitley CB, Neuenburg JK. Phenylalanine ammonia lyase (PAL) enzyme substitution therapy for phenylketonuria (PKU). Serono Symposia, 2nd European Phenylketonuria Group (EPG) Symposium, Advances and Challenges in PKU, Munich, Germany, 22-23 January 2010, Abstract L1 (Plenary session).

131. Erlingsson UCN, Ardon O, Liu A, Pasquali M, **Longo N** (2010) Fatty acid toxicity in fibroblasts from patients with very long-chain acyl CoA dehydrogenase deficiency. Mol Genet Metab 99: 213 (Abs 22).

132. Openshaw A, Procter M, **Longo N**, Mao R, Pasquali M (2010) Correlation between biochemical phenotype and molecular genotype in profound and partial biotinidase deficiency. Mol Genet Metab 99: 229-230 (Abs 59).

133. Schwarz E, Guymon R, Sakashita C, Bunker A, Roberts W, **Longo N**, Pasquali M (2010) Evaluation of guanidinoacetate and creatine in plasma and urine: variation with age. Mol Genet Metab 99: 232 (Abs 65).

134. Vanzo R, Ernst SL, Botto LD, Pasquali M, **Longo N** (2010) Guanidinoacetate methyltransferase deficiency in Utah. Mol Genet Metab 99: 235 (Abs 72).

135. **Longo N**, Pasquali M (2010) Disorders of the carnitine cycle. 2010 ACMG Annual Clinical Genetics Meeting, Albuquerque, NM, March 24-28, 2010, Abstract 428 (Invited talk)

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136. Grabowski G, Pastores G, Mardach R, Eng C, Smith L, Fernhoff P, Charrow J, Heisel-Kurth M, **Longo N**, Rhead W, Harmatz P, Mehta A, Giraldo P, Tylki-Szymanska A, Zimran A, Cohn GM, Crombez E (2010) Baseline characteristics of patients with Gaucher disease type 1 switching from imiglucerase to velaglucerase alfa in clinical trial and real-world settings. 2010 ACMG Annual Clinical Genetics Meeting, Albuquerque, NM, March 24-28, 2010, Abstract 189 (Poster)

137. Liu A, Bunker A, Roberts W, **Longo N**, Pasquali M (2010) Evaluation of very long chain fatty acids by UPLC-MS/MS in a normal pediatric population. J Inherit Metab Dis 33 (Suppl 1): S97 (Abs 289-P)

138. **Longo N**, Pasquali M (2011) Carnitine and carnitine palmitoyl transferase II deficiency. SIMD Annual Meeting, February 27–March 2, 2011 Asilomar Conference Center Pacific Grove, CA, Mol Genet Metab 102: 259..

139. Burton BK, **Longo N**, Arnold GL, Enns GM, Andersson HC, Mofidi S, Peck D, Prado BC, Pridjian1 G, Waisbren S, White D for all PKUDOS participating centers (2011) Baseline characteristics of PKU patients enrolled in the PKUDOS registry. SIMD Annual Meeting, February 27–March 2, 2011 Asilomar Conference Center Pacific Grove, CA, Mol Genet Metab 102: 273 (Abs 17).

140. Diaz GA, Bartley J, **Longo N**, Berquist W, Feigenbaum A, Gallagher R, Rhead W, Bartholomew D, Harding CO, Korson MS, Lichter-Konecki1U, Berry SA, Smith W, McCandless SE, Vockley J, Bart1 S, Kronn D, Zori R, Sreenath-Nagamani S, Summar M, Jomphe C, Beliveau1 M, Mauney J, Dickinson K, Mokhtarani M, Coakley D, Scharschmidt BF, Lee B (2011) Phase 3 blinded, randomized, crossover comparison of sodium phenylbutyrate (NaPBA) and glycerol phenylbutyrate (GPB): Ammonia (NH3) control in adults with urea cycle disorders (UCDS). SIMD Annual Meeting, February 27–March 2, 2011 Asilomar Conference Center Pacific Grove, CA, Mol Genet Metab 102: 276-277 (Abs 25).

141. Rose EC, Amat di San Filippo C, Erlingsson UCN, Ardon O, Pasquali M, **Longo N** (2011) Genotype–phenotype correlation in primary carnitine deficiency. SIMD Annual Meeting, February 27–March 2, 2011 Asilomar Conference Center Pacific Grove, CA, Mol Genet Metab 102: 311 (Abs 90).

142. **Longo N**, Feigenbaum A, Dimmock D, Stockler S, Workman K, Waisbren S, Sapropterin Research Group (2011) Interim report of study PKU-015: A phase 3b study of sapropterin dihydrochloride (Kuvan) in young children with phenylketonuria. 2011 ACMG Annual Clinical Genetics Meeting, Vancouver BC, Canada, March 16-20, 2011, Abstract 96 (Poster).

143. A Zimran, G M Pastores, A Tylki-Szymanska, D Hughes, D Elstein, R Mardach, C Eng, L Smith, M Heisel-Kurth, J Charrow, P Harmatz, P Fernhoff, W Rhead, **N Longo**, P Giraldo, D Zahrieh, E Crombez, G Grabowski (2011) Efficacy and safety of velaglucerase alpha in patients with type I Gaucher disease transitioned from Imiglucerase: TKT034 and extension 2-year results. 2011 ACMG Annual Clinical Genetics Meeting, Vancouver BC, Canada, March 16-20, 2011, Abstract 232 (Poster).

144. Macchiaiolo M, **Longo N**, Gagliardi G, Pilati M, Nicolò R, Grandin A, Bartuli A (2011) Clinical and biochemical experience with LDL-apheresis in homozygous familial hypercholesterolemia. 2011 ACMG Annual Clinical Genetics Meeting, Vancouver BC, Canada, March 16-20, 2011, Abstract 244 (Poster).

145. **Longo N** (2011) Enzyme Substitution Therapy and Other New Treatments for Phenylketonuria. Pediatric Academic Societies/Asian Society for Pediatric Research Annual Meeting, Denver CO, April 30-May 3, 2011, ASPR Symposium, Invited Plenary Lecture, p. 42-34 (Abs)

146. Burton BK, Leviton L, Vespa H, Bilder D, Lundy B, Coon H, **Longo N**. 2011. Routine screening yelds high incidence of psychiatric distress in phenylketonuria (PKU) clinics. SSIEM Symposium, Geneva, Switzerland, August 30-September 2, 2011, J Inherit Metab Dis 34 (suppl 3): S97 (P-058).

147. Zimran A, Pastores GM, Tylki-Szymanska A, Hughes DA, Elstein D, Mardach R, Eng C, Smith L, Heisel-Kurth M, Charrow J, Harmatz P, Fernhoff P, Rhead W, **Longo N**, Giraldo P, Ruiz JA, Zahrieh D, Crombez E, Grabowski GA. 2011. Efficacy of Velaglucerase Alfa in Patients with Type I Gaucher Disease (GD1) Transitioned from Type 1 Patients Previously Treated With Imiglucerase: Phase II/III Trial TKT034 and Extension 2-year results. SSIEM Symposium, Geneva, Switzerland, August 30-September 2, 2011, J Inherit Metab Dis 34 (suppl 3): S224 (P-433).

148. Zimran A, Pastores GM, Tylki-Szymanska A, Hughes DA, Elstein D, Mardach R, Eng C, Smith L, Heisel-Kurth M, Charrow J, Harmatz P, Fernhoff P, Rhead W, **Longo N**, Giraldo P, Ruiz JA, Zahrieh D, Crombez E, Grabowski GA. 2011. 2-Year Safety and Tolerability of Velaglucerase Alfa Enzyme Replacement Therapy (ERT) in Patients with Type I Gaucher Disease (GD1) Switching from Imiglucerase. SSIEM Symposium, Geneva, Switzerland, August 30-September 2, 2011, J Inherit Metab Dis 34 (suppl 3): S225 (P-434).

149. Kim JC, Coelho D, Miousse IR, Fung S, du Moulin M, Buers I, Suormala T, Stucki M, Nürnberg P, Thiele H, **Longo N**, Pasquali M, Mengel E, Watkins D, Shoubridge EA, Rutsch F, Majewski J, Baumgartner M, Fowler B, Rosenblatt DS. Mutations in ABCD4 cause a new inborn error of vitamin B_{12} metabolism. 61st Annual Meeting of the American Society of Human Genetics, 11-15 October 2011, Montreal, CANADA (Page 26, Abs 69) (Platform)

150. Zimran A, Pastores GM, Tylki-Szymanska A, Hughes DA, Elstein D, Mardach R, Eng C, Smith L, Heisel-Kurth M, Charrow J, Harmatz P, Fernhoff P, Rhead W, **Longo N**, Giraldo P, Zahrieh D, Crombez E, Grabowski GA. 2011. Changes in Biomarkers in patients with type 1 Gaucher Disease (GD1) transitioned from imiglucerase to velaglucerase alfa: cumulative 2-year results from the phase II/III trial TKT 034 and extension. 61st Annual Meeting of the American Society of Human Genetics, 11-15 October 2011, Montreal, CANADA (Page 977, Abs 1457F) (Poster)

151. Grange DK, Arnold GA, Burton BK, Enns GM, **Longo N**, Mofidi S, Peck DA, Pridjian G, White DA, Gravance C. SAPROPTERIN TREATMENT FOR PHENYLKETONURIA IN THE PKUDOS REGISTRY: CHILDREN FOUR YEARS OLD AND YOUNGER. 2012 ACMG Annual Clinical Genetics Meeting, Charlotte, NC, 27-31 March 2012, Abstract 77.

151b. Hillman R, Peck DA, Arnold GA, Burton BK, Enns GM, **Longo N**, Mofidi S, Pridjian G, Waisbren S, White DA, Gravance C. Sapropterin Use in Pregnant Phenylketonuria Patient: The PKU MOMS Sub-registry. 2012 ACMG Annual Clinical Genetics Meeting, Charlotte, NC, 27-31 March 2012, Abstract 80.

152. Jurecki E, **Longo N**, Burton BK, Dimmock D, Feigenbaum A, Potter M, Stockler S, Siriwardena K, Lang W, Kim S, Prasad S. Safety and Efficacy of Sapropterin in Children aged 0 to 6 years with Phenylketonuria. Preliminary Findings from a Long Term Open Label Study. 2012 ACMG Annual Clinical Genetics Meeting, Charlotte, NC, 27-31 March 2012, Abstract 85.

152b. **Longo N**, Burton BK, Harding CO, Grange DK, Vockley G, Wasserstein M, Adams DJ, Thomas JA, Gu K, Walling J, Sile S. A Phase 2, Open-Label, Dose-Finding and the Long Term Extension Study to Evaluate the Safety, Efficacy and Tolerability of Multiple Subcutaneous Doses of rAvPAL-PEG in Patients With Phenylketonuria. 2012 ACMG Annual Clinical Genetics Meeting, Charlotte, NC, 27-31 March 2012, Abstract 269.

153. Hillman R, Peck D, Arnold GL, Burton BK, Enns GM, Longo N, Mofidi S, Pridjian G, Waisbren S, White DA, Gravance C. Sapropterin Use in Pregnant Phenylketonuria Patient: The PKU MOMS Sub-registry. 2012 SIMD Annual Meeting, Charlotte, NC, 31 March-3 April 2012, Abstract 47, Mol Genet Metab 105: 324

154a.Burton BK, Arnold GL, Enns GM, Grange DK, **Longo N,** Mofidi S, Peck D, Pridjian G, White D, Gravance C . Sapropterin treatment for phenylketonuria in the PKUDOS registry: children 4 years and younger. 2012 SIMD Annual Meeting, Charlotte, NC, 31 March-3 April 2012, Abstract 17, Mol Genet Metab 105: 307

154b. Leon EL, Bailey C, Warnock AA, Botto LD, **Longo N**. Clinical progression of patients with Fabry disease in Utah. 2012 SIMD Annual Meeting, Charlotte, NC, 31 March-3 April 2012, Abstract 63, Mol Genet Metab 105: 333

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155a. **Longo N**, Burton BK, Harding CO, Hillman R, Grange DK, Vockley J, Wasserstein M, Thomas J, Goodin K, Hostutler R, Gu K, Walling J, Sile S. Hypophenylalaninemia in adult phenylketonuria patients treated with rAvPAL-PEG. 2012 SIMD Annual Meeting, Charlotte, NC, 31 March-3 April 2012, Abstract 67, Mol Genet Metab 105: 335

155b. O. Ardon, X. Yin, N. Longo, A. Shilon-Hadass, G. Gabis, C. Hoffman, M. Tzadok, G. Heimer, S. Sadan, B. Ben-Zeev, Y. Anikster. Creatine transporter deficiency: novel mutations and functional studies. American Society of Human Genetics 62nd Annual Meeting, November 6-10, 2012 San Francisco, California, 75: Abs 660T

156. Procter M, O. Ardon, A. Carey, H. Amiri, N. Longo, R. Mao. Insulin Resistance: Development of a clinical assay for INSR mutation detection. American Society of Human Genetics 62nd Annual Meeting, November 6-10, 2012 San Francisco, California, 75: Abs 689F

157. N. Longo, K. Siriwardena, A. Feigenbaum, D. Dimmock, B. Burton, S. Stockler, S. Waisbren, W. Lang, E. Jurecki, S. Prasad. Long-Term Developmental Progression in Young Children Taking Sapropterin for Phenylketonuria: A Two-Year Analysis of Safety and Efficacy. 2013 ACMG Annual Clinical Genetics Meeting, Phoenix, AZ, 19-23 March 2013, Abstract 169.

158. K Viau, SL Ernst, M Pasquali, LD Botto, GL Hedlund, **N Longo** (2013) Biochemical predictors of long-term outcome in patients with Guanidinoacetate Methyltransferase (GAMT) deficiency. J Inherit Metab Dis 36: (suppl 2): S104 (0-008) International Congress Inborn errors of Metabolism ICIEM 2013, Barcelona, Spain, 3-6 Sep 2013

159. B Lee, M Mokhtarani, GA Diaz, W Rhead, U Lichter-Konecki, A Feigenbaum, SA Berry, J Bartley, **N Longo**, W Berquist, W Smith, R Gallagher, CO Harding, S McCandless, A Schulze, S Nagamani, C Le Mons, K Dickinson, DF Coakley, TL Moors, D Millikien, M Marino, BF Scharschmidt. (2013) Optimizing ammonia (NH3) control in Urea Cycle Disorder (UCD) patients: short and long-term implications. J Inherit Metab Dis 36: (suppl 2): S138 (P-089) International Congress Inborn errors of Metabolism ICIEM 2013, Barcelona, Spain, 3-6 Sep 2013

160. M Pasquali, E Schwarz, T Yuzyuk, I De Biase, **N Longo** (2013) Feasibility of newborn screening for guanidinoacetate methyltransferase (GAMT) deficiency. J Inherit Metab Dis 36: (suppl 2): S315-316 (P-782) International Congress Inborn errors of Metabolism ICIEM 2013, Barcelona, Spain, 3-6 Sep 2013

161. Naleway JJ, Harlan FK, Longo N, Pasquali M, Batchelor RH, Luska JS. 2014. High-content screening using metabolic lysosomal enzyme probes. Mol Genet Metab 111(2): S80 (Abs 176)

162. Pano A, Goker-Alpanb O, **Longo N**, McDonald M, Shankar S, Shen Y, Chang P, Schiffmann R. 2014. Safety and effect of open-label agalsidase alfa in treatment-naïve children with Fabry disease. Mol Genet Metab 111(2): S84 (Abs 189)

163. N Longo, LB. Price, E Gappmaier, NL. Cantor, S L. Ernst, C Bailey, M Pasquali. 2014. ANAPLEROTIC THERAPY IN PROPIONIC ACIDEMIA. Mol Genet Metab 111(3): 269 (Abs 62)

164. SC Nagamani, GA Diaz, W Rhead, SA Berry, C Le Mons, U Lichter-Konecki, J Bartley, A Feigenbaum, N Longo, WBerquist, R Gallagher, D Bartholomew, CO Harding, MS Korson, SE McCandless, W Smith, J Vockley, D Kronn, R Zori, D Coakley, BF Scharschmidt, M Mokhtarani, B Lee. 2014. TREATMENT-RELATED SIGNS AND SYMPTOMS AMONG PATIENTS WITH UREA CYCLE DISORDERS (UCDS) DURING TREATMENT WITH SODIUM PHENYLBUTYRATE (NAPBA) AND GLYCEROL PHENYLBUTYRATE (GPB). Mol Genet Metab 111(3): 286 (Abs 98).

165. **Longo N**, Amat di San Filippo C, Yin X, Frigeni M, Mao R, Pasquali M (2014) Multiple mutations in the SLC22A5 gene in patients with OCTN2 carnitine transporter deficiency identified by newborn screening. SSIEM Meeting 2-5 September 2014, Innsbruck, Austria, J Inherit Metab Dis 37 (Suppl 1): S55 (Abs P-043).

166. Scharschmidt BF, Diaz GA, Lee B, Bartley J, **Longo N**, Berquist W, Le Mons C, Rudolh-Angelich I, Porter M, Mokhtarani M, Hook D. Protein and calorie intake among adult subjects with urea cycle disorders (UCDs) participating in the pivotal glycerol phenylbutyrate (GPB) clinical trial. SSIEM Meeting 2-5 September 2014, Innsbruck, Austria, J Inherit Metab Dis 37 (Suppl 1): S57 (Abs P-050).

167. Scharschmidt BF, Lee B, Diaz GA, Rhead W, Lichter-Konecki U, Feigenbaum A, Berry SA, Le Mons C, Bartley J, **Longo N**, Nagamani SC, Berquist W, Gallagher R, Bartholomew D, Harding CO, Korson MS, McCandless SE, Smith W, Cederbaum S, Merritt II JL, Schulze A, Vockley J, Kronn D, Zori R, Summar M, Milikien DA, Marino M, Coackley DF (2014) Blood ammonia and glutamine as predictors of hyperammonemic crises (HACs) in patients with urea cycle disorders (UCDs). SSIEM Meeting 2-5 September 2014, Innsbruck, Austria, J Inherit Metab Dis 37 (Suppl 1): S83 (Abs P-150).

168. Pastores GM, Rhead WJ, Charrow J, **Longo N**, Harmatz P, Hangartner TN, Wang N, Crombez E, Smith L (2014) LONG-TERM VELAGLUCERASE ALFA ENZYME REPLACEMENT THERAPY IN CHILDREN WITH TYPE 1 GAUCHER DISEASE. SSIEM Meeting2-5 September 2014, Innsbruck, Austria, J Inherit Metab Dis 37 (Suppl 1): S153 (Abs P-422).

169. Janet Thomas, **Nicola Longo**, Roberto Zori3, Barbara Burton4, Melissa Wasserstein5, Dorothy Grange6, Jerry Vockley7, Richard Hillman8, Cary Harding9, Gregory Rice10, Natasha Shur11, Williiam Rizzo12, Chester Whitley13, Kara Goodin14, Kim McBride15, Celeste Decker16, Markus Merilainen. Evaluation of Multiple Dosing Regimens in Phase 2 Studies of rAvPAL-PEG (BMN 165) for Control of Blood Phenylalanine Levels in Adults with Phenylketonuria. 2015 ACMG Annual Clinical Genetics Meeting, Salt Lake City, UT, 24-27 March 2015, Abs 49.

170. **Nicola Longo**, Kara Goodin2, Dorothy Grange3, Cary Harding4, Bolt Kendra5, Janet Thomas6, Melissa Wasserstein7, Barbara Burton8, Jerry Vockley9, Richard Hillman10, David Dimmock11, Natasha Shur12, Darius Adams13, William Rizzo14, Chester Whitley15, Celeste Decker16, Becky Schweighardt. Evaluation of Long-term Safety and Efficacy with rAvPAL-PEG (BMN 165) for Control of Blood Phenylalanine levels in Adults with Phenylketonuria (PKU). 2015 ACMG Annual Clinical Genetics Meeting, Salt Lake City, UT, 24-27 March 2015, Abs 53.

171. Cary Harding, **Nicola Longo**, Janet Thomas3, Barbara Burton4, Roberto Zori5, Deborah Bilder2, John Posner6, Phillip Lieberman7, Markus Merilainen8, Zhonghua Gu8, Becky Schweighardt8, Holly Weng8, Harvey Levy. Phase 2 Studies Contribute to rAvPAL-PEG Phase 3 Trial Design. 2015 ACMG Annual Clinical Genetics Meeting, Salt Lake City, UT, 24-27 March 2015, Abs 735.

172. Roberto Zori1, Janet Thomas2, Natasha Shur3, Williiam Rizzo4, Celeste Decker5, Markus Merilainen5, Mingjin Li5, Becky Schweighardt5, **Nicola Longo**. Evaluation of an Induction, Titration, and Maintenance Dosing Regimen in a Phase 2 Study of rAvPAL-PEG (BMN 165) for Control of Blood Phenylalanine Levels in Adults with Phenylketonuria (PKU). 2015 ACMG Annual Clinical Genetics Meeting, Salt Lake City, UT, 24-27 March 2015, Abs 737

173. Marta Frigeni1, F. Iacobazzi1,2, X. Yin1, **N. Longo**. Domain-swapping with OCTN2 shows tolerance to substitutions in the octn1 ergothioneine transporter. 38 SIMD Annual Meeting, Salt Lake City, UT, 28-31 March 2015, *Molecular Genetics and Metabolism* 114 (2015) 337-338: Abs 25

174. Claudia F. Reis1,2, A. Warnock1, M. Pasquali1, and **N. Longo**. Identification of citrin deficiency by newborn screening. 38 SIMD Annual Meeting, Salt Lake City, UT, 28-31 March 2015, Molecular Genetics and Metabolism 114 (2015) 338: Abs 26

175. Elisa Ballarini1,2, H.R. Underhill1, L. Botto1, M. Pasquali1, **N. Longo.** Multiple Acyl CoA dehydrogenase deficiency: Diagnosis and outcome after expanded newborn screening. 38 SIMD Annual Meeting, Salt Lake City, UT, 28-31 March 2015, Molecular Genetics and Metabolism 114 (2015) 351-52: Abs 55

176. Janet A. Thomas**, Nicola Longo**, Roberto Zori3, Barbara K. Burton4, Melissa Wasserstein5, Dorothy K. Grange6, Jerry Vockley7, Richard Hillman8, Cary Harding9, Natasha Shur10, Darius Adams11, Gregory M. Rice12, William B. Rizzo13, Chester Whitley14, Kara M. Goodin15, Kim L. McBride16, Celeste Decker17, Markus Merilainen17, Mingjin Li17, Becky Schweighardt17, David P. Dimmock. Evaluation of multiple dosing regimens in phase 2

studies of rAvPAL-PEG (BMN 165) for control of blood phenylalanine levels in adults with phenylketonuria. 38 SIMD Annual Meeting, Salt Lake City, UT, 28-31 March 2015, Molecular Genetics and Metabolism 114 (2015) 364-65: Abs 83

177. **Nicola Longo**, Janet A. Thomas2, Melissa P. Wasserstein3, Barbara K. Burton4, Jerry Vockley5, Dorothy K. Grange6, Richard Hillman7, Cary Harding8, David Dimmock9, Natasha Shur10, Darius Adams11, William B. Rizzo12, Chester Whitley13, Kara M. Goodin14, Celeste Decker15, Kendra Bolt15, Yinpu Chen15, Becky Schweighardt15, Roberto Zori. 84) Evaluation of long-term safety and efficacy with RAVPAL-PEG (BMN 165) for control of blood phenylalanine levels in adults with phenylketonuria (PKU). 38 SIMD Annual Meeting, Salt Lake City, UT, 28-31 March 2015, Molecular Genetics and Metabolism 114 (2015) 366: Abs 84.

178. Cary Harding, **Nicola Longo**, Janet A. Thomas3, Barbara K. Burton4, Roberto Zori5, Deborah A. Bilder6, John Posner7, Phil Lieberman8, Markus Merilainen9, Zhonghua Gu9, Becky Schweighardt9, Holly Weng9, Harvey Levy. Phase 2 studies contribute to RAVPAL-PEG phase 3 trial design. 38 SIMD Annual Meeting, Salt Lake City, UT, 28-31 March 2015, Molecular Genetics and Metabolism 114 (2015) 366: Abs 85.

179. Roberto Zori, Janet A. Thomas2, Natasha Shur3, William B Rizzo4, Celeste Decker5, Markus Merilainen5, Mingjin Li5, Becky Schweighart5, **Nicola Longo**. Evaluation of an induction, titration, and maintenance dosing regimen in a phase 2 study of RAVPAL-PEG (BMN 165) for control of blood phenylalanine levels in adults with phenylketonuria (PKU). 38 SIMD Annual Meeting, Salt Lake City, UT, 28-31 March 2015, Molecular Genetics and Metabolism 114 (2015) 368-9: Abs 88.

180**. N Longo,** J A Thomas, M Wasserstein, B K Burton, J Vockley, D K, Grange, R Hillman, C Harding, D Dimmock, N Shur, D Adams, W B Rizzo, C Whitley, K M Goodin, C Decker, K Bolt, Y Chen, B Schweighardt, R Zori. Evaluation of long-term safety and efficacy with rAvPAL-PEG for control of blood phenylalanine levels in adults with phenylketonuria (PKU). Annual Symposium of the Society for the Study of Inborn Errors of Metabolism LYON, FRANCE, J Inherit Metab Dis (2015) 38 (Suppl 1):S41, Abstract O-016

181. D Adams, J A Thomas, **N Longo**, R Zori, B K Burton, M Wasserstein, D K Grange, J Vockley, R Hillman, C Harding, N Shur, G M Rice, W B Rizzo, C Whitley, K M Goodin, K L McBride, C Decker, M Merilainen, M Li, B Schweighardt, D Dimmock. Evaluation of multiple dosing regimens in phase 2 studies of 'rAvPAL-PEG' for control of blood phenylalanine levels in adults with phenylketonuria. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism LYON, FRANCE, J Inherit Metab Dis (2015) 38 (Suppl 1):S116, Abstract P-149

182. R Zori, J A Thomas, N Shur, W B Rizzo, C Decker, M Merilainen, M Li, B Schweighardt, **N Longo.** Evaluation of an induction, titration, and maintenance dosing regimen in a phase 2 study of 'rAvPAL-PEG' for control of blood phenylalanine levels in adults with phenylketonuria (PKU). Annual Symposium of the Society for the Study of Inborn Errors of Metabolism LYON, FRANCE, J Inherit Metab Dis (2015) 38 (Suppl 1):S117, Abstract P-150

183. B K Burton, C Harding, **N Longo**, J A Thomas, R Zori, D A Bilder, J Posner, P Lieberman, M Merilainen, Z Gu, B Schweighardt, H Weng, H Levy. Phase 2 studies contribute to rAvPAL-PEG Phase 3 trial design. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism LYON, FRANCE, J Inherit Metab Dis (2015) 38 (Suppl 1):S117, Abstract P-151

184. B F Scharschmidt, B Lee, G A Diaz, W Rhead, U Lichter-Konecki, A Feigenbaum, S A Berry, C Le Mons, J Bartley, **N Longo**, S C Nagamani, W Berquist, R Gallagher, C Harding, S E McCandless, W Smith, A Schulze, M Marino, D F Coakley, MMokhtarani. Glutamine and hyperammonemic crises (HAC) in patients with urea cycle disorders (UCDS). Annual Symposium of the Society for the Study of Inborn Errors of Metabolism LYON, FRANCE, J Inherit Metab Dis (2015) 38 (Suppl 1):S147, Abstract P-225

185. **N Longo**, L B Price, E Gappmaier, N L Cantor, S L Ernst, C Bailey, M Pasquali. Anaplerotic therapy in propionic acidemia. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism LYON, FRANCE, J Inherit Metab Dis (2015) 38 (Suppl 1):S168, Abstract P-274.

186. M Frigeni, X Yin, M Pasquali, **N Longo.** Functional and molecular evaluation of patients with primary carnitine deficiency. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism LYON, FRANCE, J Inherit Metab Dis (2015) 38 (Suppl 1):S199, Abstract P-356.

187. Irene De Biase1,2,3, Krista S. Viau4, Aiping Liu3, Tatiana Yuzyuk1,2,3, Lorenzo D. Botto4, Marzia Pasquali1,2,3,4, **Nicola Longo1**,2,3,4 DIAGNOSIS, TREATMENT AND CLINICAL OUTCOME OF PATIENTS WITH LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE (LCHAD) DEFICIENCY. 39 SIMD Annual Meeting, 3-6 April 2016, Ponte Vedra Beach, FL. Molecular Genetics and Metabolism 117 (2016) Abs 36

188. Marta Frigeni1, Xue Yin1, Bijina Balakrishnan1, Marzia Pasquali1,2,3, **Nicola Longo1**,2,3 FUNCTIONAL STUDIES REMAIN THE BEST STRATEGY TO CONFIRM THE DIAGNOSIS OF PRIMARY CARNITINE DEFICIENCY. 39 SIMD Annual Meeting, 3-6 April 2016, Ponte Vedra Beach, FL. Molecular Genetics and Metabolism 117 (2016) 257: Abs 44

189. **Nicola Longo**, Stephen Amato, Jerry Vockley3, Klaas Wierenga4, Hong Li5, Deborah Bilder1, Barbara Burton6, David Dimmock7, CaryHarding8, John Posner9, Janet A. Thomas10, Roberto Zori11, Feriandas Greblikas12, Zhonghua Gu12, Markus Merilainen12, Haoling H.Weng12, Harvey Levy13 PRISM 301: AN OPEN-LABEL, RANDOMIZED, PHASE 3 CLINICAL TRIAL EVALUATING EFFICACY AND SAFETY OF PEGVALIASE FOR THE TREATMENT OF ADULTS WITH PHENYLKETONURIA. 39 SIMD Annual Meeting, 3-6 April 2016, Ponte Vedra Beach, FL. Molecular Genetics and Metabolism 117 (2016) 268: Abs 68

190. Jerry Vockley, BarbaraBurton, Gerard Berry, **Nicola Longo**, John A Phillips, Amarilis Sanchez-Valle, Pranoot Tanpaiboon, Stephanie Grunewald, Elaine Murphy, Alexandra Bowden, Jason Cataldo, Deborah Marsden, Emil Kakkis INTERIM RESULTS FROM AN OPEN-LABEL PHASE 2 STUDY TO ASSESS SAFETY AND CLINICAL EFFECTS OF INVESTIGATIONAL UX007 (TRIHEPTANOIN) IN SUBJECTS WITH LONG-CHAIN FATTY ACID OXIDATION DISORDERS (LC-FAOD). 39 SIMD Annual Meeting, 3-6 April 2016, Ponte Vedra Beach, FL. Molecular Genetics and Metabolism 117 (2016) 291: Abs 119

191. Tatiana Yuzyuk, Aiping Liu, Amanda Thomas, JoDell E Wilson, Irene De Biase, **Nicola Longo,** Marzia Pasquali SIMULTANEOUS QUANTIFICATION OF ALPHA-AMINOADIPIC SEMIALDEHYDE/PIPERIDEINE-6-CARBOXYLATE AND PIPECOLIC ACID IN PLASMA AND URINE. 39 SIMD Annual Meeting, 3-6 April 2016, Ponte Vedra Beach, FL. Molecular Genetics and Metabolism 117 (2016) 296: Abs 129

192. Tatiana Yuzyuk, Amanda Thomas, Aiping Liu, Irene De Biase, Krista S. Viau, Lorenzo D. Botto, Marzia Pasquali, **Nicola Longo** LYSINE RESTRICTION AND ARGININE SUPPLEMENTATION IN TREATMENT OF TWO PATIENTS WITH PYRIDOXINE DEPENDENT EPILEPSY. 39 SIMD Annual Meeting, 3-6 April 2016, Ponte Vedra Beach, FL. Molecular Genetics and Metabolism 117 (2016) 296: Abs 130

193. **N Longo**, J Thomas, M Wasserstein, B Burton, J Vockley, D Grange, C Decker, H H Weng, M Li, B Schweighardt, R Zori. Evaluation of long-term safety and efficacy of pegvaliase treatment for adults with phenylketonuria: updated year 4 results. SSIEM 2016 Annual Symposium, Rome, Italy, 6-9 September 2016. J Inherit Metab Dis (2016) 39 (Suppl 1):S39, O-013

194. M Frigeni, B Balakrishnan, F Ingoglia, X Yin, M Pasquali, **N Longo**. Decreased stability of the OCTN2 carnitine transporter in patients with primary carnitine deficiency. SSIEM 2016 Annual Symposium, Rome, Italy, 6-9 September 2016. J Inherit Metab Dis (2016) 39 (Suppl 1):S46, O-035

195. C Harding, S Amato, J Vockley, K Wierenga, H Li, D Bilder, B Burton, D Dimmock, **N Longo**, J Posner, J Thomas, R Zori, O Rosen, F Greblikas, K Gu, M Li, M Merilainen, H H Weng, H Levy. Phase 3 PRISM-1 and PRISM-2 clinical trial results: to evaluate the efficacy and safety of pegvaliase for the treatment of adults with phenylketonuria (PKU). SSIEM 2016 Annual Symposium, Rome, Italy, 6-9 September 2016. J Inherit Metab Dis (2016) 39 (Suppl 1):S107, P-172

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196. H Levy, C Harding, **N Longo**, D Bilder, B Burton, R Zori, J Posner, J Thomas, J Olbertz, O Rosen, Z Gu, M Merilainen, H H Weng, D Dimmock. Phase 3 PRISM-2 long-term extension evaluating efficacy and safety of pegvaliase for treatment of adults with phenylketonuria. SSIEM 2016 Annual Symposium, Rome, Italy, 6-9 September 2016. J Inherit Metab Dis (2016) 39 (Suppl 1):S108, P-173

197. J Vockley, B Burton, G T Berry, **N Longo**, J Phillips, A Sanchez-Valle, P Tanpaiboon, S Grunewald, E Murphy, A Bowden, L Zhang, J Cataldo, ,D Marsden, E Kakkis. Interim results from an open-label phase 2 study assessing the safety and clinical effects of investigational UX007 (triheptanoin) in subjects with long-chain fatty acid oxidation disorders (LC-FAOD). SSIEM 2016 Annual Symposium, Rome, Italy, 6-9 September 2016. J Inherit Metab Dis (2016) 39 (Suppl 1):S151, P-321

198. **N Longo**, D R Anderson, K Viau, M Pasquali. Clinical and biochemical outcome of patients with medium-chain acyl-CoA dehydrogenase deficiency. SSIEM 2016 Annual Symposium, Rome, Italy, 6-9 September 2016. J Inherit Metab Dis (2016) 39 (Suppl 1):S152, P-324.

199. Jerry Vockley1, Harvey Levy2, Stephen Amato3, Roberto Zori4, Janet Thomas5, Barbara Burton6, Cary O. Harding7, **Nicola Longo**, John Posner9, Deborah Bilder8, Joy Olbertz10, Zonghua Gu10, Kelly Lau10, Mimi Lin10, Kevin Larimore10, and David Dimmock11 Phase 3 PRISM-2 Long-term Extension Study Evaluating Efficacy and Safety of Pegvaliase for Treatment of Adults with Phenylketonuria. International Congress of Inborn Errors of Metabolism, September 5-8, 2017, Rio de Janeiro, Brazil, Journal of Inborn Errors of Metabolism & Screening, p. 128 Abs 285

200. Roberto Zori1, Harvey Levy2, **Nicola Longo**3, Kelly Lau4, Orli Rosen4, Mingjin Li4, Kevin Larimore4, Celeste Decker4, Haoling Weng4, and Jerry Vockley5. Phase 2 Long-term Pegvaliase Treatment for Adults With Phenylketonuria: Updated Year 5 Safety and Efficacy Data from the PAL-003 Extension. . International Congress of Inborn Errors of Metabolism, September 5-8, 2017, Rio de Janeiro, Brazil, Journal of Inborn Errors of Metabolism & Screening, p. 130 Abs 288

201. Joshua Lilienstein1, Barbara Burton2, Mitzie Grant3, Can Ficicioglu4, Jessica Kopesky5, Mina D. Nguyen-Driver6, Caroline Moore1, Elaina Jurecki1, and **Nicola Longo7** - Interim Analysis of the Phenylketonuria (PKU) Patients Enrolled in the PKUDOS Registry. International Congress of Inborn Errors of Metabolism, September 5-8, 2017, Rio de Janeiro, Brazil, Journal of Inborn Errors of Metabolism & Screening, p. 134 Abs 297

202. Francois Feillet1, Can Ficicioglu2, Florian B. Lagler3, **Nicola Longo**, Jan Alm5, Ania C. Muntau6, Alberto Burlina7, Amaya Belanger-Quintana8, Friedrich K. Trefz9, Raivo Kittus10, Elaina Jurecki11, Ignacio Alvarez10, Joshua Lilienstein11, and Barbara Burton12 An Interim Analysis of the Kuvan® Adult Maternal Pediatric European Registry (KAMPER) and Phenylketonuria Developmental Outcomes and Safety (PKUDOS) Registries: Pregnancies. International Congress of Inborn Errors of Metabolism, September 5-8, 2017, Rio de Janeiro, Brazil, Journal of Inborn Errors of Metabolism & Screening, p. 135 Abs 298

203. **Nicola Longo**. Effect of Liver Transplantation on Hyperammonemia and Metabolic Control in Propionic Acidemia. International Congress of Inborn Errors of Metabolism, September 5-8, 2017, Rio de Janeiro, Brazil, Journal of Inborn Errors of Metabolism & Screening, p. 185 Abs 410

204. Jerry Vockley1, Barbara Burton2, Gerard Berry3, **Nicola Longo**4, John Phillips5, Amarilis Sanchez-Valle6, Pranoot Tanpaiboon7, Stephanie Grunewald8, Elaine Murphy9, Alexandra Bowden10, Wencong Chen10, Chao-Yin Chen10, Jason Cataldo10, Deborah Marsden10, and Emil Kakkis. Results From a 78-Week Single-Arm, Open-Label Phase 2 Study to Evaluate UX007 in Pediatric and Adult Patients With Moderate to Severe Long-Chain Fatty Acid Oxidation Disorders (LC-FAOD). International Congress of Inborn Errors of Metabolism, September 5-8, 2017, Rio de Janeiro, Brazil, Journal of Inborn Errors of Metabolism & Screening, p. 218 Abs 482

205. Jill Mayhew1, Jerry Vockley2, Barbara Burton3, Gerard Berry4, **Nicola Longo**, John Phillips6, Amarilis Sanchez-Valle7, Pranoot Tanpaiboon8, Stephanie Grunewald9, Elaine Murphy10, Wencong Chen1, Chao-Yin Chen1, Jason Cataldo1, Deborah Marsden1, Alexandra Bowden1, Emil Kakkis1, Alison Skrinar1, and Reed Humphrey. Assessments of Exercise Tolerance and Muscle Function in Long Chain-Fatty Acid Oxidation Disorders (LC-FAOD): Results From a Phase 2 Open Label Study of UX007. International Congress of Inborn Errors of Metabolism, September 5-8, 2017, Rio de Janeiro, Brazil, Journal of Inborn Errors of Metabolism & Screening, p. 222 Abs 491.

206. Alexandre Nonino1, Joel Charrow2, Cristina Fraga3, Xuefan Gu4, Hiroyuki Ida5, **Nicola Longo**, Elena Lukina7, Sebastiaan Gaemers8, Marie-Helene Jouvin8, Jing Li8, Yaoshi Wu8, Yong Xue8, and M. Judith Peterschmitt. Once Versus Twice-Daily Dosing of Eliglustat in Patients With Gaucher disease Type 1: The Phase 3, Randomized, Double-Blind EDGE Trial. International Congress of Inborn Errors of Metabolism, September 5-8, 2017, Rio de Janeiro, Brazil, Journal of Inborn Errors of Metabolism & Screening, p. 313 Abs 697.

207. F. Feillet1, C. Ficicioglu, F.B. Lagler, **N. Longo**, J. Alm, A.C Muntau, A. Burlina, A. Belanger-Quintana, F.K. Trefz, R. Kittus, E.R. Jurecki, I. Alvarez, J. Lilienstein, and B. Burton on behalf of the KAMPER and PKUDOS investigators. AN INTERIM ANALYSIS OF THE KUVAN® ADULT MATERNAL PAEDIATRIC EUROPEAN REGISTRY (KAMPER) AND PHENYLKETONURIA DEVELOPMENTAL OUTCOMES AND SAFETY (PKUDOS) REGISTRIES: PREGNANCIES.). 40 SIMD Annual Meeting, 11-14 March 2018, San Diego, CA. Molecular Genetics and Metabolism 123 (2018) 230, (Abs 42).

208. J. Lilienstein, B. Burton, M. Grant, C. Ficicioglu, J. Kopesky, M.D. Nguyen-Driver, C. Moore, E.R. Jurecki, and **N. Longo** on behalf of the PKUDOS investigators. INTERIM ANALYSIS OF THE PHENYLKETONURIA (PKU) PATIENTS ENROLLED IN THE PKUDOS REGISTRY. 40 SIMD Annual Meeting, 11-14 March 2018, San Diego, CA. Molecular Genetics and Metabolism 123 (2018) 248, (Abs 76).

209. Jerry Vockley, MD, PhD; Barbara Burton, MD; Gerard T. Berry, MD; **Nicola Longo,** MD; John Phillips, MD; Amarilis Sanchez-Valle, MD; Pranoot Tanpaiboon, MD; Stephanie Grunewald, MD, PhD, Elaine Murphy, MD; Alexandra Bowden, PhD; Wencong Chen, PhD; Yunming Mu, PhD; Jason Cataldo, DO; Deborah Marsden, MD; Emil Kakkis, MD, PhD. RESULTS FROM A 78-WEEK SINGLE-ARM, OPEN-LABEL PHASE 2 STUDY TO EVALUATE UX007 IN PEDIATRIC AND ADULT PATIENTS WITH MODERATE TO SEVERE LONG-CHAIN FATTY ACID OXIDATION DISORDERS (LC-FAOD). 40 SIMD Annual Meeting, 11-14 March 2018, San Diego, CA. Molecular Genetics and Metabolism 123 (2018) 274, (Abs 124).

210. Chong J-L, Longo N, Pasquali M. Creatine Metabolism in Urea Cycle Disorders. Annual Clinical Genetics Meeting 2018. CHARLOTTE, NORTH CAROLINA | APRIL 10-14, 2018. Abs 87

211. Vockley J2, **Longo N**, Madden M1, Dwyer L1, Mu Y1 , Chen C Y1, Cataldo J1. Impact of UX007 and dietary management on major clinical events in a 78-week single-arm open-label phase 2 LC-FAOD study. SSIEM 2018 Annual Symposium, Athens, Greece, 4-7 September 2018. J Inherit Metab Dis (2018) 41 (Suppl 1):S54, 0-048

212. Burton B4, **Longo N**, Stuy M5, Vockley J6, Van Backle J2, Lane P3, Alvarez I3, Lilienstein J2, Jurecki E. The burden of illness in adults with phenylketonuria (PKU): Interim analysis of a cross-sectional study. SSIEM 2018 Annual Symposium, Athens, Greece, 4-7 September 2018. J Inherit Metab Dis (2018) 41 (Suppl 1):S95-96, P-099

213. Burton B K1, Harding C O2, Thomas J A3, **Longo N**, Posner J5, Dimmock D6, Zori R7, Weng H H8, Olbertz J8, Gershman A8, Rosen O8, Gupta S8, Jones S8, Gu Z8, Vockley J. Long-term safety of induction, titration, and maintenance dosing of pegvaliase treatment in adults with phenylketonuria. SSIEM 2018 Annual Symposium, Athens, Greece, 4-7 September 2018. J Inherit Metab Dis (2018) 41 (Suppl 1):S104, P-123

214. Harding C O2, Thomas J A3, Burton B K1, Zori R6 , Dimmock D, Vockley J8, Weng H H7, Olbertz J7, Gershman A7, Rosen O7, Jones S7, Li M7, **Longo N**. Characterization of hypophenylalaninemia in pegvaliase treated adults with PKU. SSIEM 2018 Annual Symposium, Athens, Greece, 4-7 September 2018. J Inherit Metab Dis (2018) 41 (Suppl 1):S104, P-124

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215. Feillet F1, Ficicioglu C2, Lagler F B3, **Longo N**, Alm J5, Muntau A C6, Burlina A7, Belanger-Quintana A8, Trefz F K9, Kittus R11, Jurecki E10, Alvarez I11, Lilienstein J10, Burton B. An interim analysis of the KAMPER and PKUDOS registries: Efficacy and safety of sapropterin before and during pregnancy. SSIEM 2018 Annual Symposium, Athens, Greece, 4-7 September 2018. J Inherit Metab Dis (2018) 41 (Suppl 1):S104-S105, P-125
216. Muntau A3, Feillet F1, Burton B K6, MacDonald A7, WesselA8, Alvarez I5, Lilienstein J4, Lane P5, Jurecki E4, **Longo N**. A meta-analysis of growth outcomes in phenylketonuria patients treated with phenylalanine-restricted diet + sapropterin. . SSIEM 2018 Annual Symposium, Athens, Greece, 4-7 September 2018. J Inherit Metab Dis (2018) 41 (Suppl 1):S105, P-126
217. Myrl D. Holida, John Bernat, **Nicola Longo**, Ozlem Goker-Alpan, William Wilcox (2019) Once every 4 weeks - 2 mg/kg of pegunigalsidase alfa for treating Fabry disease Preliminary results of a phase 3 study. Molecular Genetics and Metabolism 126(2): s73
218. David Warnock, Wallace Eric, Raphael Schiffmann, Myrl Holida, **Nicola Longo** (2019) Analysis of the baseline characteristics of Fabry disease patients screened for the pegunigalsidase alfa phase III BALANCE study. Molecular Genetics and Metabolism 126(2): s150-s151
219. Barbara Burton1, **Nicola Longo2**, Francois Maillot3, Yusof Rahman4, Rani Singh5, H. Serap Sivri6, Mary Stuy7, Jerry Vockley8, Joost Van Backle9, Ashok Jha10, Paul Lane10, Joshua Lilienstein9, Elaina Jurecki9 THE BURDEN OF ILLNESS IN ADULTS WITH PHENYLKETONURIA (PKU): INTERIM ANALYSIS OF A CROSS-SECTIONAL STUDY CONDUCTED IN NORTH AMERICA AND EUROPE. SIMD 2019 Annual Meeting, Seattle, WA, Molecular Genetics and Metabolism 126 (2019) 293 (poster 35).
220. Jason Cataldo1, **Nicola Longo2**, Megan Madden1, Lauren Dwyer1, Yunming Mu1, Chao-Yin Chen1, Jerry Vockley3. IMPACT OF UX007 AND DIETARY MANAGEMENT ON MAJOR CLINICAL EVENTS IN A 78-WEEK, SINGLE-ARM OPEN-LABEL PHASE 2 STUDY IN PATIENTS WITH LC-FAOD. SIMD 2019 Annual Meeting, Seattle, WA, Molecular Genetics and Metabolism 126 (2019) 293-294 (poster 37).
221. Francois Feillet1, Can Ficicioglu2, Florian Lagler3, **Nicola Longo4**, Jan Alm5, Ania Muntau6, Alberto Burlina7, Amaya Berlanger-Quintana8, Friedrich Trefz9, Elaina Jurecki10, Ignacio Alvarez11, Joshua Lilienstein10, Barbara Burton12 AN INTERIM ANALYSIS OF THE KAMPER AND PKUDOS REGISTRIES: EFFICACY AND SAFETY OF SAPROPTERIN BEFORE AND DURING PREGNANCY SIMD 2019 Annual Meeting, Seattle, WA, Molecular Genetics and Metabolism 126 (2019) 300 (poster 54).
222. Filippo Ingoglia1, Jean-Leon Chong1, Marzia Pasquali1**, Nicola Longo1** CREATINE METABOLISM IN UREA CYCLE DISORDERS SIMD 2019 Annual Meeting, Seattle, WA, Molecular Genetics and Metabolism 126 (2019) 307-308 (poster 73).
223. **Longo N**2, Diaz G2, Kok TMY3, Canavan C3, Vescio T3 and Berry SA4 GLYCEROL PHENYLBUTYRATE EFFICACY AND SAFETY FROM AN OPEN LABEL STUDY IN PEDIATRIC PATIENTS UNDER 2 MONTHS OF AGE WITH UREA CYCLE DISORDERS SIMD 2019 Annual Meeting, Seattle, WA, Molecular Genetics and Metabolism 126 (2019) 314-315 (poster 93).
224. Ania Muntau1, Francois Feillet2, Barbara Burton3, Anita MacDonald4, Ann Wessel5, Ignacio Alvarez6, Joshua Lilienstein7, Paul Lane6, Elaina Jurecki7, **Nicola Longo8** A META-ANALYSIS OF GROWTH OUTCOMES IN PHENYLKETONURIA PATIENTS TREATED WITH A PHENYLALANINE-RESTRICTED DIET + SAPROPTERIN SIMD 2019 Annual Meeting, Seattle, WA, Molecular Genetics and Metabolism 126 (2019) 317-318 (poster 101).
225. Fran Rohr1, Barbara K. Burton2, **Nicola Longo3**, Janet A. Thomas4, Cary O. Harding5, Orli Rosen6, Zhonghu Gu6, Joy Olbertz6, Haoling H. Weng PHASE 3 PRISM CLINICAL TRIALS: EVALUATING CHANGE IN DIET WITH PEGVALIASE TREATMENT IN ADULTS WITH PHENYLKETONURIA SIMD 2019 Annual Meeting, Seattle, WA, Molecular Genetics and Metabolism 126 (2019) 323 (poster 116).
226. Valentina Rovelli1, Krista Viau2, Marzia Pasquali3, **Nicola Longo3** CLINICAL AND BIOCHEMICAL OUTCOME OF PATIENTS WITH VERY LONG-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY SIMD 2019 Annual Meeting, Seattle, WA, Molecular Genetics and Metabolism 126 (2019) 323-324 (poster 118).
227. Neil Smith1, **Nicola Longo2**, Nenad Blau3 PHASE I CLINICAL EVALUATION OF CNSA-001 (SEPIAPTERIN), A NOVEL PHARMACOLOGICAL TREATMENT FOR PHENYLKETONURIA AND TETRAHYDROBIOPTERIN DEFICIENCIES, IN HEALTHY VOLUNTEERS SIMD 2019 Annual Meeting, Seattle, WA, Molecular Genetics and Metabolism 126 (2019) 328 (poster 131).
228. Jerry Vockley1, Roberto Zori2, Janet A. Thomas3, Mary Stuy4, Barbara K. Burton5, **Nicola Longo6**, Orli Rosen7, Zhonghu Gu7, Joy Olbertz7, Haoling H. Weng7 SUBGROUP ANALYSIS OF PEGVALIASE IN ADULTS WITH PHENYLKETONURIA IN PHASE 3 PRISM STUDIES: EVALUATING EFFICACY AND SAFETY BASED ON PREVIOUS SAPROPTERIN RESPONSE OR DIETARY PHENYLALANINE INTAKE SIMD 2019 Annual Meeting, Seattle, WA, Molecular Genetics and Metabolism 126 (2019) 332 (poster 140).
229. Vockley J, Zori R, Thomas J, Stuy M, Burton B K, **Longo N**, Rosen O, Gu Z, Olbertz J, Weng H H Efficacy & safety of pegvaliase by sapropterin response or dietary phenylalanine intake in adults with phenylketonuria. SSIEM 2019 Annual Symposium, Rotterdam, The Netherlands, 3-6 September 2019. J Inherit Metab Dis (2019) 42 (Suppl 1): Page 101, P-142
230. **Longo N**, Thomas J, Jurecki E, Lane P, Olbertz J, Gershman A, Wang B, Harding C O, Burton B K, Rohr F, Van Calcar S. Dietary intakes and adverse events in pegvaliase-treated phenylketonuria adults who had low blood phenylalanine Levels. SSIEM 2019 Annual Symposium, Rotterdam, The Netherlands, 3-6 September 2019. J Inherit Metab Dis (2019) 42 (Suppl 1): Page 103-104, P-147
231. **Longo N**, Rohr F, Burton B K, Thomas J, Harding C O, Rosen O, Gu Z, Olbertz J, Weng HH\. Phase 3 PRISM clinical trials: Evaluating change in diet with pegvaliase treatment in adults with phenylketonuria. SSIEM 2019 Annual Symposium, Rotterdam, The Netherlands, 3-6 September 2019. J Inherit Metab Dis (2019) 42 (Suppl 1): Page 104, P-148
232. Rovelli V, Viau K, Pasquali M, **Longo N** Clinical and Biochemical Outcome of Patients with very Long-Chain ACYL-COA Dehydrogenase Deficiency. SSIEM 2019 Annual Symposium, Rotterdam, The Netherlands, 3-6 September 2019. J Inherit Metab Dis (2019) 42 (Suppl 1): Page 184, P-328
233. Vockley J, Burton B, Berry G, **Longo N**, Phillips J, Sanchez-Valle A, Chapman K, Tanpaiboon P, Grunewald S, Murphy E, Bowden A, Mu Y, Cataldo J P-332 Interim results from an open-label, long-term extension study to evaluate the safety and efficacy of triheptanoin (UX007) in LC-FAOD. SSIEM 2019 Annual Symposium, Rotterdam, The Netherlands, 3-6 September 2019. J Inherit Metab Dis (2019) 42 (Suppl 1): Page 185, P-332
234. Jurecki E, Burton B, Ilan A, Delaney K, Madden D, **Longo N**, Harding C O, Thomas J Concept elicitation and outcomes assessment tool mapping with an international cohort of adult phenylketonuria patients. SSIEM 2019 Annual Symposium, Rotterdam, The Netherlands, 3-6 September 2019. J Inherit Metab Dis (2019) 42 (Suppl 1): Page 321, E-031
235. Tahseen Mozaffar, John W. Day, **Nicola Longo,** Jordi Diaz Manera, Volker Straub, Miko Maruoka, Cong Hane, Cheryl Wong Po Foo, Angela R. Smith, Nathan Bachtell. AT845 gene replacement therapy for late onset Pompe disease: Overview of clinical data from FORTIS, a phase 1/2 open-label clinical study. WORLDSymposium 2022, San Diego CA, 7-11 Feb, 2022. Molecular Genetics and Metabolism, Volume 135, Issue 2, February 2022, Pages S85-S86 (Abs 206)

PI ON CURENTLY APPROVED IRB PROTOCOLS

Nicola Longo MD PhD

IRB_00110412 PBD-001: A Phase 1/2, Open-Label, Randomized Parallel Arm, Intra-patient Dose Titration Study to Evaluate the Safety, Pharmacokinetics and Preliminary Efficacy of CNSA-001 (Sepiapterin) in Adult and Adolescent Primary Tetrahydrobiopterin Deficient Patients
Approved Longo 5/26/2021
IRB_00090396 018CTXX15001: An Observational, Multicenter Study of the Prevalence of Cerebrotendinous Xanthomatosis (CTX) in Patient Populations Diagnosed with Early-Onset Idiopathic Bilateral Cataracts
Approved Longo 6/22/2021
IRB_00121194 REN001-102: An Open Label Study to Determine the Safety and Tolerability of 12 Weeks Treatment with Oral REN001 in Subjects with Fatty Acid Oxidation Disorders (FAOD).
Approved Longo 10/8/2020
IRB_00111681 Modify ACT-434964: A Multicenter, Double-Blind, Randomized, Placebo-Controlled, Parallel-Group Study to Determine the Efficacy and Safety of Lucerastat oral monotherapy in Adult Subjects with Fabry Disease.
Approved Longo 10/7/2020
IRB_00031175 PKUDOS – PKU Demographics, Outcomes, and Safety Registry PKU MOMS- Subregistry for pregnant PKU moms
Approved Longo 5/21/2021
IRB_00082746 UX007-CL202: An Open-label Long-Term Safety and Efficacy Extension Study in Subjects with Long-Chain Fatty Acid Oxidation Disorders (LC-FAOD) Previously Enrolled in UX007 or Triheptanoin Studies
Approved Longo 11/20/2020
IRB_00126507 307-902: A Prospective Study of Phenylketonuria (PKU)
Approved Longo 12/3/2021
IRB_00117942 ATB200-03 (Propel): a phase 3 double-blind randomized study to assess the efficacy and safety of intravenous atb200 co administered with oral AT2221 in adult subjects with late onset pompe disease compared with alglucosidase alfa/placebo.
Approved Longo 2/18/2021
IRB_00106354 Trial of dichloroacetate (DCA) in pyruvate dehydrogenase complex (PDC) Deficiency
Approved Longo 2/26/2021
IRB_00093689 PB-102-F20: A Randomized, Double blind, Active Control Study of the Safety and Efficacy of PRX-102 compared to Agalsidase Beta on Renal Function in Patients with Fabry Disease Previously Treated With Agalsidase Beta (BALANCE)
Approved Longo 10/8/2020
IRB_00085855 Natural history of patients with metabolic disorders
Approved Longo
IRB_00075237 AGLU06909/LTS13930: A Prospective Safety Sub-Registry to Assess Anaphylaxis and Severe Allergic Reactions, and Severe Cutaneous and Systemic Immune-Mediated Reactions with Alglucosidase Alfa Treatment
Approved Longo 11/21/2020
IRB_00034057 Clinical Surveillance Program for Patients with Mucopolysaccharidosis Type VI
Approved Longo 3/27/2021
IRB_00128835 ATB200-07: A Phase 3 open-label extension study to assess the long-term safety and efficacy of intravenous ATB200 co-administered with oral AT2221 in adult subjects with late-onset Pompe disease.
Approved Longo 5/5/2021
IRB_00105142 HPN-100-021: A Randomised, Controlled, Open-Label Parallel Arm Study of the Safety, Pharmacokinetics and Ammonia Control of RAVICTI® (Glycerol Phenylbutyrate [GPB]) Oral Liquid and Sodium Phenylbutyrate (NaPBA) in Phenylbutyrate Treatment Naïve Patients
Approved Longo 11/19/2020
IRB_00062414 GOS: Gaucher Disease Outcome Survey
Approved Longo 4/18/2021
IRB_00116015 PB-102-F51: Open Label Extension Study to Evaluate the Long-Term Safety and Efficacy of Pegunigalsidase Alfa (PRX-102) 2 mg/kg Administered by Intravenous Infusion Every 4 Weeks in Patients with Fabry Disease
Approved Longo 2/25/2021
IRB_00124126 PEACE (Pegzilarginase Effect on Arginase 1 Deficiency Clinical Endpoints): A Randomized, Double-Blind, Placebo-Controlled Phase 3 Study of the Efficacy and Safety of Pegzilarginase in Children and Adults with Arginase 1 Deficiency

Approved Longo 10/22/2020
IRB_00109350 MSC12790/AGLUO7710: A Phase 3/4 Prospective Study to Characterize the Pharmacokinetics of Alglucosidase Alfa in Patients with Pompe Disease
Approved Longo 4/22/2021
IRB_00116016 PB-102-F60: Open Label Extension Study to Evaluate the Long-Term Safety and Efficacy of Pegunigalsidase Alfa (PRX-102) in Patients with Fabry Disease
Approved Longo 1/21/2021
IRB_00117942 ATB200-03 (Propel): a phase 3 double-blind randomized study to assess the efficacy and safety of intravenous atb200 co administered with oral AT2221 in adult subjects with late onset pompe disease compared with alglucosidase alfa/placebo.
Approved Longo 2/18/2021
IRB_00106354 Trial of dichloroacetate (DCA) in pyruvate dehydrogenase complex (PDC) Deficiency
Approved Longo 2/26/2021
IRB_00093689 PB-102-F20: A Randomized, Double blind, Active Control Study of the Safety and Efficacy of PRX-102 compared to Agalsidase Beta on Renal Function in Patients with Fabry Disease Previously Treated With Agalsidase Beta (BALANCE)
Approved Longo 10/8/2020
IRB_00120894 mRNA-3704-P101: A Global, Phase 1/2, Open Label, Dose Escalation Study to Evaluate the Safety, Pharmacodynamics, and Pharmacokinetics of mRNA-3704 in Patients with Isolated Methylmalonic Acidemia Due to Methylmalonyl-CoA Mutase Deficiency
Approved Longo 12/3/2020
IRB_00104191 PB-102-F50: A Phase 3, Open Label, Switch Over Study to Assess the Safety, Efficacy and Pharmacokinetics of pegunigalsidase alfa (PRX-102) 2 mg/kg Administered by Intravenous Infusion Every 4 Weeks for 52 weeks in Patients with Fabry Disease Currently Tr
Approved Longo 9/7/2020
IRB_00076357 B3031002: A Multicenter, Multicountry, Postmarketing, Active Surveillance Taliglucerase Alfa Registry in Patients with Gaucher Disease.
Approved Longo 10/7/2020
IRB_00097168 EFC14028: A phase 3 randomized, multicenter, multinational, double-blinded study comparing the efficacy and safety of repeated biweekly infusions of neoGAA (GZ402666) and alglucosidase alfa in treatment-naïve patients with late-onset Pompe disease (COMET
Approved Longo 5/11/2021
IRB_00121646 HMI-102-101: A Phase 1/2 Open-Label, Randomized, Concurrently-Controlled, Dose Escalation Study to Evaluate the Safety and Efficacy of HMI-102 in Adult PKU Subjects with PAH Deficiency
Approved Longo 9/18/2020
IRB_00042999 Rare Disease Registry Program
Approved Longo 2/28/2021
IRB_00131563 AT100-01: An AAV8 Neutralizing Antibody Seroprevalence Study in Subjects with Late Onset Pompe Disease
Approved Longo 6/24/2021
IRB_00107419 SPIMM-301: A Phase 3 Randomized, Double-Blind, Parallel-Group, Placebo-Controlled Trial to Evaluate the Efficacy and Safety of Daily Subcutaneous Injections of Elamipretide in Subjects with Primary Mitochondrial Myopathy Followed by an Open-Label Treatme
Approved Longo 7/21/2020

236. **ORAL PRESENTATIONS**

1.**Longo N**, Franchi-Gazzola R, Dall'Asta V, Bussolati O, Gazzola GC (1983) Trasporto di membrana di L-cistina in fibroblasti umani in coltura: aspetti cinetici e regolativi. Atti II Congresso Nazionale Associazione di Biologia Cellulare e del Differenziamento, Bressanone 2-4 maggio 1983, 88.

2.**Longo N**, Gazzola GC, Elsas LJ (1986) Insulin regulation of membrane transport by normal and insulin-receptor mutant human fibroblasts. **Clin Res** *34*(1): 242A.

3.**Longo N**, Elsas LJ (1987) Restriction fragment length polymorphism for the *alpha* subunit of the insulin receptor gene in inherited severe insulin resistance. **Clin Res** *35*(3): 624A.

4.**Longo N**, Visigalli R, Franchi-Gazzola R, Dall'Asta V, Bussolati O, Guidotti GG, Gazzola GC (1989) Inhibition of aspartate transport in *ras*-transformed or phorbol ester treated NIH 3T3 cells. **Eur J Cell Biol** *49* (Suppl 28) : 33 (abs 93)

5.**Longo N**, Bell GI, Shuster RC, Griffin LD, Langley SD, Elsas LJ (1990) Human fibroblasts express the insulin-responsive glucose transporter (GLUT4). **Clin Res** 38(2): 436A.

6.**Longo N**, Shuster RC, Griffin LD, Langley SD, Elsas LJ (1990) Activation of the insulin-receptor kinase modifies glucose transporter gene expression in fibroblasts cultured from a patient with leprechaunism. Proceedings of the 5□ International Congress of Inborn Errors of Metabolism, Pacific Grove, CA, 1-5 June 1990, W16.3.

7.**Longo N**, Shuster RC, Griffin LD, Langley SD, Elsas LJ (1992) Activation of insulin receptor signaling by a single amino acid substitution in the transmembrane domain. **Clin Res** 40(2): 321A.

8.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1992) Homozygosity for a natural mutation in the insulin receptor *alpha* subunit activates glucose transport. **Clin Res** 40(2): 329A.

9.**Longo N**, Langley SD, Griffin LD, Elsas LJ (1994) Prenatal diagnosis in leprechaunism: two novel mutations in the insulin receptor gene. Proceedings VI International Congress Inborn Errors of Metabolism, Milano, Italy, May 27-31, 1994, W10.5, p 95.

10.**Longo N**, Giannella-Neto D, Langley SD, Wang Y, Scaglia F, Elsas LJ (1997) Genotype-phenotype correlation in patients with inherited insulin-resistant syndromes. 7th International Congress of Inborn Errors of Metabolism. p. 80 (Abs O42) Vienna Austria May 1997.

11.**Longo N**, Scaglia F, Wang Y, Singh RH, Dembure PP, Pasquali M, Evinger JD, Bagnasco S, Fernhoff P (1997) Physiological characterization of the carnitine transporter responsible for primary carnitine deficiency. 7th International Congress of Inborn Errors of Metabolism. p. 219 (Abs O106), Vienna Austria May 1997.

12.**Longo N**, Wang Y, Smith S, Langley SD, Scaglia F (1998) Genotype-phenotype correlation in patients with inherited insulin resistant syndromes. **Am J Hum Genet** 63: A24 (Abs 124) Annual Meeting, American Society of Human Genetics, Denver, CO, Oct 1998.

13.**Longo N**, Meadows T, Wang Y (2000) Impaired sodium stimulation of carnitine transport in primary carnitine deficiency. **J Inherit Metab Dis** 23 (Suppl 1): 117 (Abs 233-O) 8th International Congress of Inborn Errors of Metabolism Cambridge UK 13-17 September 2000.

14**.Longo N**, Wang Y, Taroni F, Garavaglia B (2000) Functional analysis of mutations in the OCTN2 transporter causing primary carnitine deficiency: Lack of genotype-phenotype correlation**. J Inherit Metab Dis** 23 (Suppl 1): 118 (Abs 235-O) 8th International Congress of Inborn Errors of Metabolism Cambridge UK 13-17 September 2000.

15. **Longo N,** Amat di San Filippo C (2006) Glycosylation affects membrane maturation of the OCTN2 carnitine/organic cation transporter. **Acta BioMed** 77(suppl 3): 73-74.

16. **Longo N** (2007) The challenge for the long term surveillance of metabolic disorders detected by newborn screening. ACMG Annual Clinical Genetics Meeting, Nashville TN March 21-25,2007, 162 (Abs 282) (invited)

17. **Longo N** (2008) Endocrine evaluation of the obese adolescent child. XI International Meeting of Pediatric Laboratory Medicine, Fortaleza, Brazil, September 2008 (Invited).

18. **Longo N** (2009) Sugar, small molecules, and cardiomyopathy. Annual Meeting American College of Medical Genetics, Tampa, FL, 25-29 March 2009 (Invited)

19. K Viau, SL Ernst, M Pasquali, LD Botto, GL Hedlund, **N Longo** (2013) Biochemical predictors of long-term outcome in patients with Guanidinoacetate Methyltransferase (GAMT) deficiency. *J Inherit Metab Dis* 36: (suppl 2): S104 (0-008) International Congress Inborn errors of Metabolism ICIEM 2013, Barcelona, Spain, 3-6 Sep 2013.

20. **N Longo**, J A Thomas, M Wasserstein, B K Burton, J Vockley, D K, Grange, R Hillman, C Harding, D Dimmock, N Shur, D Adams, W B Rizzo, C Whitley, K M Goodin, C Decker, K Bolt, Y Chen, B Schweighardt, R Zori. Evaluation of long-term safety and efficacy with rAvPAL-PEG for control of blood phenylalanine levels in adults with phenylketonuria (PKU). Annual Symposium of the Society for the Study of Inborn Errors of Metabolism LYON, FRANCE, J Inherit Metab Dis (2015) 38 (Suppl 1):S41, Abstract O-016

INVITED SPEAKER AT NATIONAL AND INTERNATIONAL CONFERENCES

Meeting of the Italian Society for the Study of Inherited Metabolic Disorders, Genoa, Italy, November 1992 (Longo N (1992) Difetti del recettore dell'insulina. Pathologica 85: 28-30).

-European School of Medical Genetics, Sestri Levante , Italy, 28 March-3 April 1993.

"Transfection of mutant genes in eukaryotic cells"

"Identification of mutations in disease genes"

First International Symposium on Molecular Genetics in Endocrinology, Sao Paulo, Brazil, 4-7 December 1997

a.Longo N (1997) Molecular genetics of inherited severe insulin-resistance.

b.Longo N (1997) Molecular genetics of glucose transporters.

2001 FASEB Summer Research Conference “New perspectives in transporter biology”, Tucson AZ July 21 - 26, 2001. Transporters in fatty acid oxidation.

2002 “Transporters International Conference”, Munich, Germany, 1-5 September 2002. The OCTN2 carnitine transporter.

Longo N. Primary carnitine deficiency. 2003 “Ross Conference: Metabolic disorders”, Houston, TX, April 2003.

Longo N. Inherited metabolic disorders. 2004 “XLII Jornadas Medicas Nacionales”, Panama City, Republic of Panama, October 2004.

Longo N. Breakthrough research in tetrahydrobiopterin (BH4) therapy: Phenylketonuria and BH4 deficiency. 2006 Ancillary Meeting, American Society of Human Genetics, New Orleans 10 Oct 2006.

Longo N (2007) The challenge for the long term surveillance of metabolic disorders detected by newborn screening. ACMG Annual Clinical Genetics Meeting, Nashville TN March 21-25,2007, 162 (Abs 282)

Longo N (2008) Endocrine evaluation of the obese adolescent child. XI International Meeting of Pediatric Laboratory Medicine, Fortaleza, Brazil, September 2008

2008. Medical Management of Pediatric Neurotransmitter Disorders. Tetrahydrobiopterin deficiency. Washington DC 18 July 2008

2008. Medical Management of Pediatric Neurotransmitter Disorders. Sapropterin reductase deficiency and the BH4 deficiency database. Washington DC 19 July 2008

Longo N (2009) Sugar, small molecules, and cardiomyopathy. Annual Meeting American College of Medical Genetics, Tampa, FL, 25-29 March 2009

Longo N (2009) Phenylalanine ammonia lyase in phenylketonuria. Annual Meeting European Society for Phenylketonuria, Belek, Turkey, 30 Oct-1 November 2009.

Longo N (2009) Phenylalanine ammonia lyase and sapropterin therapy in phenylketonuria. Annual Meeting European Society for Phenylketonuria, Belek, Turkey, 30 Oct-1 November 2009

Longo N (2010) Phenylalanine ammonia lyase enzyme substitution therapy for phenylketonuria. 2nd European Phenylketonuria Group Meeting, "Advances and Challenges in PKU", Munich, Germany, Annual January 22-23, 2010

Longo N, Pasquali M (2010) Disorders of the carnitine cycle. 2010 ACMG Annual Clinical Genetics Meeting, Albuquerque, NM, March 24-28, 2010, Abstract 428 (Invited talk)

Longo N (2010) Mitochondrial function in Wolf-Hirschhorn syndrome. 4p- support group meeting, Salt Lake City, UT, 31 July 2010.

Longo N (2010) Current end emerging treatments for phenylketonuria, France Foundation, Denver CO, 24 September 2010.

Longo N, Pasquali M (2010) Newborn screening for metabolic disorders. Newborn screening in the Campania region of Italy. Naples, Italy, 30 October 2010.

Longo N (2010) New therapies for phenylketonuria. Milano Pediatria 2010, Italy, 19 Nov 2010.

Longo N (2010) Enzyme substitution therapy in PKU. Milano Pediatria 2010, Italy, 20 Nov 2010.

Longo N (2010) Carnitine and human diseases. Pharmacology Rounds, University of Utah, 29 Nov 2010.

Longo N, Pasquali M (2011) Carnitine and carnitine palmitoyl transferase II deficiency. SIMD Annual Meeting, Plenary lecture, February 28, 2011

Longo N (2011) New and emerging metabolic interventions for phenylketonuria. ACMG Annual Meeting, Satellite symposium, Vancouver BC, 17 March 2011

Longo N (2011) Enzyme Substitution Therapy and Other New Treatments for Phenylketonuria. Pediatric Academic Societies/Asian Society for Pediatric Research Annual Meeting, Denver CO, April 30, Invited Plenary Lecture, p. 42-34 (Abs)

Longo N (2012) ADAPT: A diversified approach to phenylketonuria treatment. Metabolic Academy, Denver CO, 24 February 2012.

Longo N (2012) Sapropterin Therapy in young children with Phenylketonuria. Metabolic Academy, Denver CO, 25 February 2012.

Longo N (2012) Carnitine and fatty acid oxidation. New perspectives in medicine, Florence, Italy, 3 May 2012.

Longo N (2012) Newborn screening for metabolic disorders. Nevada newborn screening program Las Vegas, NV, 20 June 2012 and Reno, NV, 28 June 2012.

Longo N (2013) 50 years of newborn screening for metabolic disorders. Nevada newborn screening program Las Vegas, NV, 21 Sep 2013 and Reno, NV, 14 September 2013.

Longo N (2013) Update on phenylketonuria: Invited Oral Presentation, 4 Sep 2013, International Congress Inborn errors of Metabolism ICIEM 2013, Barcelona, Spain, 3-6 Sep 2013.

Longo N (2014) Presidential address: Membrane transport and metabolic disorders. SIMD annual meeting, March 9–12, 2014, Asilomar Conference Center Pacific Grove, CA

Longo N (2014) Primary carnitine deficiency. Faroe Islands Health System, Faroe Islands, Kingdom of Denmark, 2 May 2014.

Longo N (2014) PEG-PAL: the latest evidence. 6 June 2014, Zagreb, Croatia. 2014 Annual Multidisciplinary Phenylketonuria Symposium.

Longo N (2014) PKU: USA and European guidelines workshop, 6 June 2014, Zagreb, Croatia. 2014 Annual Multidisciplinary Phenylketonuria Symposium.

Longo N (2014) Carnitine and fatty acid oxidation, China Lecture Tour, Beijing, Shenyang, Wuhan, Shangaii, 26 June-4 July 2014.

Longo N (2014) CLINICAL GUIDELINES FOR FATTY ACID OXIDATION (FAO) DISORDERS. Annual Symposium of the International Network for Fatty Acid Oxidation Research and Management (INFORM), Innsbruck, Austria, 6 Sep 2014.

Longo N (2015) New treatments for phenylketonuria. University of Bologna, Pediatric Grand Rounds, Bologna, Italy, 18 May 2015.

Longo N (2015) Next generation sequencing in medical genetics. Panama City, Republic of Panama, 8 Aug 2015

Longo N (2015) Diet (or no diet) and outcome in children with glutaric aciduria type I, aged more than 6 years. SSIEM Meeting, Lyon, France, 1 Sep 2015.

Longo N (2015) Primary carnitine deficiency and defects in carnitine biosynthesis: novel functions of carnitine? 2nd Annual Symposium of the International Network for Fatty Acid Oxidation Research and Management (INFORM), Lyon, France, 5 Sep 2015.

Longo N (2015) Magistral lecture: Phenylketonuria update. Fano Metabolic Meeting, Italy, 9 Sep 2015

Longo N (2015) Defects of fatty acid oxidation. Fano Metabolic Meeting, Italy, 9 Sep 2015

Longo N (2016) Clinical Experience & Treatment of Metabolic Diseases, Rare Diseases Days, Guatemala City, Guatemala, 29 Feb 2016.

Longo N (2016) Social and medical awareness about rare diseases in the USA. Rare Diseases Days, Guatemala City, Guatemala, 1 Mar 2016.

Longo N (2016) Carnitine deficiency. Fatty Acid Oxidation Organic Acidemia Organization Meeting, Denver CO, 8 July 2016

Longo N (2016) Liver transplantation for methylmalonic and propionic academia. Fatty Acid Oxidation Organic Acidemia Organization Meeting, Denver CO, 8 July 2016

Longo N (2016) Disorders of fatty acid oxidation. Southeastern Regional Genetics Group, Ponte Vedra, FL, 15 July 2016.

Longo N (2016) Primary carnitine deficiency. Society for the study of inborn error of metabolism, Annual Meeting, Rome, Italy, 4 Sep 2016.

Longo N (2016) CARNITINE DEFICIENCY AND FATTY ACID OXIDATION DISORDERS. European association of Pediatric Societies, Geneva, Switzerland, 23 Oct 2016.

Longo N (2016) Carnitine and Fatty Acid Oxidation. VUMC Medical Center, Amsterdam, The Netherlands, 12 Dec 2016.

Longo N (2016) Management of phenylketonuria. European Phenylketonuria Meeting, Amersfoort, The Netherlands, 14 Dec 2016.

Longo N (2016) Tetrahydrobiopterin in children age 0-4 with phenylketonuria. European Phenylketonuria Meeting, Amersfoort, The Netherlands, 14 Dec 2016.

Longo N (2017) Brain creatine deficiency syndromes. San Paolo Hospital, 19 Jan 2017, Milan, Italy

Longo N (2017) Neonatal seizures. Milano Pediatria 2017. 20 Jan 2017, Milan, Italy

Longo N (2017) New therapies for phenylketonuria. Milano Pediatria, 21 Jan 2017, Milan, Italy

Longo N (2017) Newborn Screening. Children's Hospital of Los Angeles, 6 Apr 2017

Longo N (2017) Carnitine and Fatty Acid Oxidation. Aaron Michael Graham Jr., Annual Grand Rounds, Children's Hospital of Los Angeles, April 7 2017

Longo N (2017) The impact of newborn screening on fatty acid oxidation disorders. Keynote address. INFORM Symposium, Rio de Janeiro, Brazil, 3 Sep 2017

Longo N (2017) Comparing short and long-term outcomes/complications of hyperammonaemia in patients treated with long term vs other management strategies. Symposium: STABILISE. RATIONALISE. OPTIMISE. Exploring long-term therapy options for hyperammonaemia in organic acidaemias, 13th International Congress of Inborn Errors of Metabolism, , Rio de Janeiro, Brazil, 7 Sep 2017

Longo N (2017) Efficacy and Safety of Pegvaliase for the Treatment of Adults with PKU: Phase 3 PRISM Clinical Trials. Japanese Society for Inborn Errors of Metabolism, Toyo, Japan, 13 Oct 2017.

Longo N (2019) Invited Professor. Advanced Course on Diagnosis and Treatment of Metabolic Diseases. Porto Alegre, Brazil, 4-8 Aug 2019. Lectures on: 1. Introduction 2. Clinical approach to the diagnosis and treatment of intermediate metabolism diseases. 3. Overview of the treatment of intermediate metabolism diseases. 4. Metabolic emergencies and approach to selected problems of intermediate metabolism diseases. 5. Perspective I screening for inborn errors of metabolism. 6. Review of clinical cases.

Longo N (2020) Management of Long-Chain Fatty Acid Oxidation Disorders. ASEMBIA Meeting (online), 7 May 2020

Longo N (2020) Breaking down long chain fatty acid oxidation disorders. GMDI meeting (on line), 7 May 2020

Longo N (2020) Liver transplantation in inborn errors of metabolism. American Liver Foundation (Online), 3 September 2020

Longo N (2017) Carnitine and Fatty Acid Oxidation. Grand Rounds, University Medical Center, Las Vegas NV, Children's Hospital of Los Angeles, 16 April 2021