Azienda Ospedaliero - Universitaria "A. Meyer"
Clinica di Neurologia Pediatrica - Università degli Studi di Firenze
Direttore Prof. Renzo Guerrini

Laboratorio di Biologia Molecolare delle Malattie Neurometaboliche
Responsabile: Prof.ssa Amelia Morrone
Viale Pieraccini, 24 - 50139 Firenze
Tel. +39 055 5662543 - Fax +39 055 5662849; email: a.morrone@meyer.it

PRESTAZIONI DI NEXT GENERATION SEQUENCING (NGS) DEL LABORATORIO DI BIOLOGIA MOLECOLARE DELLE MALATTIE NEUROMETABOLICHE

Si può richiedere il sequenziamento dei seguenti pannelli di geni (elencati in blu), ma anche selezionare (barrare) un solo gene o più geni dagli elenchi dettagliati a seguire.

- ☐ **DISORDINI CONGENITI DELLA GLICOSILAZIONE (CDG) (141 GENI)**
- ☐ **ALTERATA GLICOSILAZIONE DELLA TRANSFERRINA SIERICA: 48 GENI CDG E 5 GENI PER DISORDINI CORRELATI (53 GENI)**
- ☐ **SINDROME DI WALKER-WARBURG (15 GENI)**
- ☐ **ALFA-DISTROGLICANOPATIA (19 GENI)**
- ☐ **SINDROME MIASTENICA CONGENITA CON DIFETTO DI GLICOSILAZIONE (5 GENI)**
- ☐ **RABDOMIOLISI (75 GENI)**
- ☐ **MALATTIA DA DEPOSITO DI GLICOGENO-FORME MUSCOLARI (21 GENI)**
- ☐ **DIFETTI DELLA BETA-OSSIDAZIONE DEGLI ACIDI GRASSI (28 GENI)**
- ☐ **DEFICIT DI SERINA E SINDROME DI NEU-LAXOVA (4 GENI)**
- ☐ **POLIMERASI GAMMA (2 GENI)**
- ☐ **RIFIUTO DELLA FRUTTA (4 GENI)**
- ☐ **GALATTOSEMIA (3 GENI)**
- ☐ **DISORDINI DI CHETOLISI (3 GENI)**
- ☐ **DISORDINI DI CHETOGENESI (2 GENI)**
- ☐ **MALATTIE DA ACCUMULO LISOSOMIALE-PANNELLO COMPLETO (64 GENI)**
- ☐ **CEROIDOLIPOFUSCINOSI NEURONALI (NCL) (13 GENI)**
- ☐ **MALATTIE PEROSSISOMIALI (28 GENI)**
- ☐ **IPERAMMONIEMIE (20 GENI)**
- ☐ **EPATOPATIA E/O COLESTASI (48 GENI)**
- ☐ **METABOLISMO DEI METALLI (13 GENI)**
- ☐ **METABOLISMO DEGLI STEROLI (11 GENI)**
- ☐ **IPEROSSALURIA (3 GENI)**
- ☐ **ACIDEMIA ISOVALERICA (1 GENE)**
- ☐ **ACIDEMIA METILMALONICA, METABOLISMO COBALAMINA, IPEROMOCISTEINEMIA, METABOLISMO FOLATI E DISORDINI CORRELATI (41 GENI)**

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- ☐ **ACIDEMIA PROPIONICA (2 GENI)**
- ☐ **ACIDURIA 2 METILBURRICA E ISOBUTIRRICA (2 GENI)**
- ☐ **ACIDURIA 2-IDROSSIGLUTARICA (4 GENI)**
- ☐ **ACIDURIA 2-METIL-3-IDROSSIBUTIRRICA (1 GENE)**
- ☐ **ACIDURIA 3 IDROSSI 3 METILGLUTARICA (1 GENE)**
- ☐ **ACIDURIA 3-IDROSSIISOBUTIRRICA (3 GENI)**
- ☐ **ACIDURIA 3-METILGLUTACONICA (16 GENI)**
- ☐ **ACIDURIA 4-IDROSSIBUTIRRICA o DEFICIT DI SUCCINICO SEMIALDEIDE DEIDROGENASI (1 GENE)**
- ☐ **ACIDURIA ALFA-METILACETOACETICA o DEFICIT DI BETA-CHETOTIOLASI (1 GENE)**
- ☐ **ACIDURIA FORMIMINOGLUTAMMICA (1 GENE)**
- ☐ **ACIDURIA GLICERICA (2 GENI)**
- ☐ **ACIDURIA GLUTARICA (5 GENI)**
- ☐ **ACIDURIA ISOBUTIRRICA (1 GENE)**
- ☐ **ACIDURIA MEVALONICA (1 GENE)**
- ☐ **ACIDURIA OXOGLUTARICA E DEFICIT DI ALFA-CHETOGLUTARATO DEIDROGENASI (3 GENI)**
- ☐ **ACIDURIA PIROGLUTAMMICA (2 GENI)**
- ☐ **ALCAPTONURIA (1 GENE)**
- ☐ **CISTINURIA (2 GENI)**
- ☐ **DEFICIT DI AMINOACILASI 1 (1 GENE)**
- ☐ **DEFICIT DI CARBOSSILASI CON C5OH ELEVATA ALLO SCREENING NEONATALE (4 GENI)**
- ☐ **DEFICIT DI LIPINA (1 GENE)**
- ☐ **DIFETTI DEL CICLO DI KREBS (12 GENI)**
- ☐ **IPERECPLESSIA TIPO 3 (1 GENE)**
- ☐ **IPERFENILALANINEMIA (12 GENI)**
- ☐ **IPERGLICEROLEMIA (1 GENE)**
- ☐ **IPERGLICINEMIA NON CHETOTICA (7 GENI)**
- ☐ **IPERGLICINURIA (3 GENI)**
- ☐ **IPERPROLINEMIA e IPOPROLINEMIA (5 GENI)**
- ☐ **IPOFOSFATASIA (1 GENE)**
- ☐ **MALATTIA DELLE URINE A SCIROPPO D'ACERO (MSUD) (4 GENI)**

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- ☐ **MALATTIA DI CANAVAN o DEFICIT DI AMINOACILASI 2 (1 GENE)**
- ☐ **MALONICO ACIDURIA (1 GENE)**
- ☐ **POLIMERASI GAMMA (2 GENI)**
- ☐ **PURINE E PIRIMIDINE (27 GENI)**
- ☐ **SINDROME DA IPOTIROIDISMO-RITARDO-DISMORFISMO (1 GENE)**
- ☐ **SULFOCISTINURIA (4 GENI)**
- ☐ **TIROSINEMIA (3 GENI)**
- ☐ **XANTOMATOSI CEREBROTENDINEA (1 GENE)**

ELENCO DETTAGLIATO DEI PANNELLI E DEI GENI INCLUSI

☐ **DISORDINI CONGENITI DELLA GLICOSILAZIONE (CDG) (141 GENI)**

Gene	Proteina	Malattia
ALG1	ALG1, Chitobiosyldiphosphodolichol Beta-Mannosyltransferase	ALG1-CDG (CDG Ik)
ALG2	ALG2, Alpha-1,3/1,6-Mannosyltransferase	ALG2-CDG (CDG Ii)
ALG3	ALG3, Alpha-1,3- Mannosyltransferase	ALG3-CDG (CDG Id)
ALG6	ALG6, Alpha-1,3-Glucosyltransferase	ALG6-CDG (CDG Ic)
ALG8	ALG8, Alpha-1,3-Glucosyltransferase	ALG8-CDG (CDG Ih)
ALG9	ALG9, Alpha-1,2-Mannosyltransferase	ALG9-CDG (CDG Il)
ALG10	ALG10, Alpha-1,2-Glucosyltransferase	ALG10-CDG (Long QT syndrome, acquired, reduced susceptibility to)
ALG11	ALG11, Alpha-1,2-Mannosyltransferase	ALG11-CDG (CDG Ip)
ALG12	ALG12, Alpha-1,6-Mannosyltransferase	ALG12-CDG (CDG Ig)
ALG13	ALG13, UDP-N-Acetylglucosaminyltransferase Subunit	ALG13-CDG (Epileptic encephalopathy, early infantile, 36)
ALG14	ALG14, UDP-N-Acetylglucosaminyltransferase Subunit	ALG14-CDG (Myasthenic syndrome, congenital, 15, without tubular aggregates)
ATP6AP1	ATPase H+ Transporting Accessory Protein 1	Immunodeficiency 47
ATP6AP2	ATPase H+ Transporting Accessory Protein 2	Mental retardation, X-linked, syndromic, Hedera type
ATP6V0A2	ATPase H+ Transporting V0 Subunit A2	Cutis laxa, autosomal recessive, type IIA; Wrinkly skin syndrome
ATP6V1A	ATPase H+ Transporting V1 Subunit A	Cutis laxa, autosomal recessive, type IID; Epileptic encephalopathy, infantile or early childhood, 3, autosomal dominant
ATP6V1E1	ATPase H+ Transporting V1 Subunit E1	Cutis laxa, autosomal recessive, type IIC
B3GALNT2	Beta-1,3-N-Acetylgalactosaminyltransferase 2	B3GALNT2-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11))
B3GALT6	Beta-1,3-Galactosyltransferase 6	B3GALT6-CDG (Ehlers-Danlos syndrome, progeroid type, 2)
B3GAT3	Beta-1,3-Glucuronyltransferase 3	B3GAT-CDG (Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects)
B3GLCT	Beta 3-Glucosyltransferase	B3GLCT-CDG (Peters-plus syndrome)
B4GALNT1	Beta-1,4-N-Acetyl-Galactosaminyltransferase 1	B4GALNT1-CDG (Spastic paraplegia 26, autosomal recessive)
B4GALT1	Beta-1,4-Galactosyltransferase 1	B4GALT1-CDG (CDG IId)

B4GALT7	Beta-1,4-Galactosyltransferase 7	B4GALT7-CDG (Ehlers-Danlos syndrome with short stature and limb anomalies)
B4GAT1	Beta-1,4-Glucuronyltransferase 1	B4GAT1-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
CAD	Carbamoyl-Phosphate Synthetase 2, Aspartate Transcarbamylase, And Dihydroorotase	CAD-CDG (CDG Iz)
CCDC115	Coiled-Coil Domain Containing 115	CCDC115-CDG (CDG IIo)
CHST3	Carbohydrate Sulfotransferase 3	Spondyloepiphyseal dysplasia with congenital joint dislocations
CHST6	Carbohydrate Sulfotransferase 6	Macular corneal dystrophy
CHST14	Carbohydrate Sulfotransferase 14	CHST14-CDG (Ehlers-Danlos syndrome, musculocontractural type 1)
CHSY1	Chondroitin Sulfate Synthase 1	Temtamy preaxial brachydactyly syndrome
COG1	Component Of Oligomeric Golgi Complex 1	COG1-CDG (CDG IIg)
COG2	Component Of Oligomeric Golgi Complex 2	COG2-CDG
COG4	Component Of Oligomeric Golgi Complex 4	COG4-CDG (CDG IIj)
COG5	Component Of Oligomeric Golgi Complex 5	COG5-CDG (CDG IIi)
COG6	Component Of Oligomeric Golgi Complex 6	COG6-CDG (CDG III)
COG7	Component Of Oligomeric Golgi Complex 7	COG7-CDG (CDG IIe)
COG8	Component Of Oligomeric Golgi Complex 8	COG8-CDG (CDG IIh)
CRPPA (ex. ISPD)	Isoprenoid Synthase Domain Containing	CRPPA-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
CSGALNACT1	Chondroitin Sulfate N-Acetylgalactosaminyltransferase 1	Mizumoto et al 2019, Hum Mutat (in press) doi:10.1002/humu.23952
DDOST	Dolichyl-Diphosphooligosaccharide--Protein Glycosyltransferase Non-Catalytic Subunit	DDOST-CDG (CDG Ir)
DHDDS	Dehydrodolichyl Diphosphate Synthase Subunit	DHDDS-CDG (Retinitis pigmentosa 59)
DLL3	Delta Like Canonical Notch Ligand 3	Spondylocostal dysostosis 1, autosomal recessive
DOLK	Dolichol Kinase	DOLK-CDG (CDG Im)
DPAGT1	Dolichyl-Phosphate N-Acetylglucosaminephosphotransferase 1	DPAGT1-CDG (CDG Ij)
DPM1	Dolichyl-Phosphate Mannosyltransferase Subunit 1, Catalytic	DPM1-CDG (CDG Ie)
DPM2	Dolichyl-Phosphate Mannosyltransferase Subunit 2, Regulatory	DPM2-CDG (CDG Iu)
DPM3	Dolichyl-Phosphate Mannosyltransferase Subunit 3	DPM3-CDG (CDG Io)
DSE	Dermatan Sulfate Epimerase	DSE-CDG (Ehlers-Danlos syndrome, musculocontractural type 2)
EOGT	EGF Domain Specific O-Linked N-Acetylglucosamine Transferase	EOGT-CDG (Adams-Oliver syndrome 4)
EXT1	Exostosin Glycosyltransferase 1	EXT1-CDG (Exostoses, multiple)
EXT2	Exostosin Glycosyltransferase 2	EXT2-CDG (Exostoses, multiple)
FKRP	Fukutin Related Protein	FKRP-CDG (Muscular dystrophy-dystroglycanopathy)
FKTN	Fukutin	FKTN-CDG (Fukuyama congenital muscular dystrophy)
FUT8	Fucosyltransferase 8	Congenital disorder of glycosylation with defective fucosylation
GALNT3	Polypeptide N-Acetylgalactosaminyltransferase 3	Tumoral calcinosis, hyperphosphatemic, familial, 1

GANAB	Glucosidase II Alpha Subunit	Polycystic kidney disease 3
GFPT1	Glutamine--Fructose-6-Phosphate Transaminase 1	GFPT1-CDG (Myasthenia, congenital, 12, with tubular aggregates)
GMPPA	GDP-Mannose Pyrophosphorylase A	GMPPA-CDG (Alacrima, achalasia, and mental retardation syndrome)
GMPPB	GDP-Mannose Pyrophosphorylase B	GMPPB-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
GNE	Glucosamine (UDP-N-Acetyl)-2-Epimerase/N-Acetylmannosamine Kinase	GNE-CDG (Autosomal dominant Sialuria; Autosomal recessive (Nonaka myopathy)
GPAA1	Glycosylphosphatidylinositol Anchor Attachment 1	Glycosylphosphatidylinositol biosynthesis defect 15
HES7	Hes Family BHLH Transcription Factor 7	Spondylocostal dysostosis 4, autosomal recessive
KRT5	Keratin	Dowling-Degos disease 1; Epidermolysis bullosa simplex
LARGE1	LARGE Xylosyl- And Glucuronyltransferase 1	LARGE1-CDG (Muscular dystrophy-dystroglycanopathy)
LFNG	LFNG O-Fucosylpeptide 3-Beta-N-Acetylglucosaminyltransferase	Spondylocostal dysostosis 3, autosomal recessive
MAGT1	Magnesium Transporter 1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia
MAN1A2	Mannosidase Alpha Class 1A Member 2	Periventricular heterotopia and polymicrogyria
MAN1B1	Mannosidase Alpha Class 1B Member 1	Mental retardation, autosomal recessive 15
MESP2	Mesoderm Posterior BHLH Transcription Factor 2	Spondylocostal dysostosis 2, autosomal recessive
MGAT2	Mannosyl (Alpha-1,6-)-Glycoprotein Beta-1,2-N-Acetylglucosaminyltransferase	MGAT2-CDG (CDG IIa)
MOGS	Mannosyl-Oligosaccharide Glucosidase	MOGS-CDG (CDG IIb)
MPDU1	Mannose-P-Dolichol Utilization Defect 1	MPDU1-CDG (CDG If)
MPI	Mannose Phosphate Isomerase	MPI-CDG (CDG Ib)
NANS	N-Acetylneuraminate Synthase	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type
NGLY1	N-Glycanase 1	Congenital disorder of deglycosylation
NUS1	NUS1 Dehydrodolichyl Diphosphate Synthase Subunit	NUS1-CDG (CDG 1aa)
OGT	O-Linked N-Acetylglucosamine (GlcNAc) Transferase	Mental retardation, X-linked 106
P4HA1	Prolyl 4-Hydroxylase Subunit Alpha 1	Early-onset joint hypermobility, joint contractures, muscle weakness, bone dysplasia and high myopia
PAPSS2	3'-Phosphoadenosine 5'-Phosphosulfate Synthase 2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes
PGAP1	Post-GPI Attachment To Proteins 1	PGAP1-CDG (Mental retardation, autosomal recessive 42)
PGAP2	Post-GPI Attachment To Proteins 2	PGAP2-CDG (Hyperphosphatasia with mental retardation syndrome 3)
PGAP3	Post-GPI Attachment To Proteins 3	PGAP3-CDG (Hyperphosphatasia with mental retardation syndrome 4)
PGM1	Phosphoglucomutase 1	PGM1-CDG (CDG It)
PGM3	Phosphoglucomutase 3	PGM3-CDG (Immunodeficiency 23)
PIGA	Phosphatidylinositol Glycan Anchor Biosynthesis Class A	PIGA-CDG (Multiple congenital anomalies-hypotonia-seizures syndrome)
PIGB	Phosphatidylinositol Glycan Anchor Biosynthesis Class B	PIGB-CDG (Hyperphosphatasia with mental retardation syndrome)

PIGC	Phosphatidylinositol Glycan Anchor Biosynthesis Class C	PIGC-CDG
PIGF	Phosphatidylinositol Glycan Anchor Biosynthesis Class F	PIGF-CDG
PIGG	Mental retardation, autosomal recessive 53	Phosphatidylinositol Glycan Anchor Biosynthesis Class G
PIGL	Phosphatidylinositol Glycan Anchor Biosynthesis Class L	PIGL-CDG (CHIME syndrome)
PIGM	Phosphatidylinositol Glycan Anchor Biosynthesis Class M	PIGM-CDG (Glycosylphosphatidylinositol deficiency)
PIGN	Phosphatidylinositol Glycan Anchor Biosynthesis Class N	PIGN-CDG (Multiple congenital anomalies-hypotonia-seizures syndrome)
PIGO	Phosphatidylinositol Glycan Anchor Biosynthesis Class O	PIGO-CDG (Hyperphosphatasia with mental retardation syndrome)
PIGP	Phosphatidylinositol Glycan Anchor Biosynthesis Class P	PIGP-CDG
PIGQ	Phosphatidylinositol Glycan Anchor Biosynthesis Class Q	PIGQ-CDG
PIGS	Phosphatidylinositol Glycan Anchor Biosynthesis Class S	Glycosylphosphatidylinositol biosynthesis defect 18
PIGT	Phosphatidylinositol Glycan Anchor Biosynthesis Class T	PIGT-CDG (Multiple congenital anomalies-hypotonia-seizures syndrome)
PIGV	Phosphatidylinositol Glycan Anchor Biosynthesis Class V	PIGV-CDG (Hyperphosphatasia with mental retardation syndrome)
PIGW	Phosphatidylinositol Glycan Anchor Biosynthesis Class W	PIGW-CDG (Hyperphosphatasia with mental retardation syndrome)
PIGY	Phosphatidylinositol Glycan Anchor Biosynthesis Class Y	PIGY-CDG (Hyperphosphatasia with mental retardation syndrome)
PMM2	Phosphomannomutase 2	PMM2-CDG (CDG Ia)
POFUT1	Protein O-Fucosyltransferase 1	POFUT1-CDG (Dowling-Degos disease 2)
POGLUT1	Protein O-Glucosyltransferase 1	POGLUT1-CDG (Dowling-Degos disease 4)
POMGNT1	Protein O-Linked Mannose N-Acetylglucosaminyltransferase 1 (Beta 1,2-)	POMGNT1-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
POMGNT2	Protein O-Linked Mannose N-Acetylglucosaminyltransferase 2 (Beta 1,4-)	POMGNT2-CDG ((Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
POMK	Protein-O-Mannose Kinase	POMK-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
POMT1	Protein O-Mannosyltransferase 1	POMT1-CDG (Muscular dystrophy-dystroglycanopathy)
POMT2	Protein O-Mannosyltransferase 2	POMT2-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
PRKCSH	Protein Kinase C Substrate 80K-H	Polycystic liver disease 1
RFT1	RFT1 Homolog	RFT1-CDG (CDG In)
RIPPLY2	Ripply Transcriptional Repressor 2	(Spondylocostal dysostosis 6)
RPN2	Ribophorin II	RPN2-CDG
RXYLT1	Ribitol Xylosyltransferase 1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10
SEC23B	Sec23 Homolog B, Coat Complex II Component	SEC23B-CDG (Autosomal dominant Cowden syndrome 7; Autosomal recessive Dyserythropoietic anemia, congenital type II)
SLC35A1	Solute Carrier Family 35 Member A1	SLC35A1-CDG (CDG IIf)

SLC35A2	Solute Carrier Family 35 Member A2	SLC35A2-CDG (CDG IIm)
SLC35A3	Solute Carrier Family 35 Member A3	SLC35A3-CDG (Arthrogryposis, mental retardation, and seizures)
SLC35C1	Solute Carrier Family 35 Member C1	SLC35C1-CDG (CDG IIc)
SLC35D1	Solute Carrier Family 35 Member D1	Schneckenbecken dysplasia
SLC39A8	Solute Carrier Family 39 Member 8	SLC39A8-CDG (CDG IIn)
SLC9A7	Solute Carrier Family 9 Member A7	Khayat et al 2019, Hum Mol Genet 28(4):598-614. X-linked, decrivono un apziente con mutazione de novo che ha TIEF alterato.
SRD5A3	Steroid 5 Alpha-Reductase 3	SRD5A3-CDG (CDG Iq; Kahrizi syndrome)
SSR3	Signal Sequence Receptor Subunit 3	Ng et al 2019, J Inherit Metab Dis, Apr 3 doi: 10.1002/jimd.12091. Descrivono 1 paziente con profile CDG tipo I.
SSR4	Signal Sequence Receptor Subunit 4	SSR4-CDG (CDG-Iy)
ST3GAL3	ST3 Beta-Galactoside Alpha-2,3-Sialyltransferase 3	Epileptic encephalopathy, early infantile, 15
ST3GAL5	ST3 Beta-Galactoside Alpha-2,3-Sialyltransferase 5	Salt and pepper developmental regression syndrome
ST6GAL2	ST6 Beta-Galactoside Alpha-2,6-Sialyltransferase 2	Autism spectrum disorder
STT3A	STT3A, Catalytic Subunit Of The Oligosaccharyltransferase Complex	STT3A-CDG (CDG Iw)
STT3B	STT3B, Catalytic Subunit Of The Oligosaccharyltransferase Complex	STT3B-CDG (CDG Ix)
TF	Transferrin	Atransferrinemia
TMEM165	Transmembrane Protein 165	TMEM165-CDG (CDG IIk)
TMEM199	Transmembrane Protein 199	TMEM199-CDG (CDG IIp)
TMEM5	Transmembrane Protein 5	TMEM5-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
TRAPPC11	Trafficking Protein Particle Complex 11	Muscular dystrophy, limb-girdle, autosomal recessive 18
TRAPPC12	Trafficking Protein Particle Complex 12	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity
TRAPPC6B	Trafficking Protein Particle Complex 6B	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy
TUSC3	Magnesium Uptake/Transporter TUSC3	Mental retardation, autosomal recessive 7
VMA21	VMA21 Vacuolar H+-ATPase Homolog (S. Cerevisiae)	Myopathy, X-linked, with excessive autophagy
VPS13B	Vacuolar Protein Sorting 13 Homolog B	Cohen syndrome
XYLT1	Xylosyltransferase 1	XYLT1-CDG (Desbuquois dysplasia 2)
XYLT2	Xylosyltransferase 2	XYLT2-CDG (Spondyloocular syndrome)

☐ ALTERATA GLICOSILAZIONE DELLA TRANSFERRINA SIERICA: 48 GENI CDG E 5 GENI PER DISORDINI CORRELATI (53 GENI).

Gene	Proteina	Malattia
☐ ALDOB	Aldolase, Fructose-Bisphosphate B	Hereditary fructose intolerance
☐ ALG1	ALG1, Chitobiosyldiphosphodolichol Beta-Mannosyltransferase	ALG1-CDG (CDG Ik)
☐ ALG2	ALG2, Alpha-1,3/1,6-Mannosyltransferase	ALG2-CDG (CDG Ii)
☐ ALG3	ALG3, Alpha-1,3- Mannosyltransferase	ALG3-CDG (CDG Id)
☐ ALG6	ALG6, Alpha-1,3-Glucosyltransferase	ALG6-CDG (CDG Ic)
☐ ALG8	ALG8, Alpha-1,3-Glucosyltransferase	ALG8-CDG (CDG Ih)
☐ ALG9	ALG9, Alpha-1,2-Mannosyltransferase	ALG9-CDG (CDG Il)
☐ ALG11	ALG11, Alpha-1,2-Mannosyltransferase	ALG11-CDG (CDG Ip)

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☐ ALG12	ALG12, Alpha-1,6-Mannosyltransferase	ALG12-CDG (CDG Ig)
☐ ALG13	ALG13, UDP-N-Acetylglucosaminyltransferase Subunit	ALG13-CDG (Epileptic encephalopathy, early infantile, 36)
☐ ATP6AP1	ATPase H+ Transporting Accessory Protein 1	Immunodeficiency 47
☐ ATP6V0A2	ATPase H+ Transporting V0 Subunit A2	Cutis laxa, autosomal recessive, type IIA; Wrinkly skin syndrome
☐ B4GALT1	Beta-1,4-Galactosyltransferase 1	B4GALT1-CDG (CDG IId)
☐ CCDC115	Coiled-Coil Domain Containing 115	CCDC115-CDG (CDG IIo)
☐ COG1	Component Of Oligomeric Golgi Complex 1	COG1-CDG (CDG IIg)
☐ COG2	Component Of Oligomeric Golgi Complex 2	COG2-CDG
☐ COG4	Component Of Oligomeric Golgi Complex 4	COG4-CDG (CDG IIj)
☐ COG5	Component Of Oligomeric Golgi Complex 5	COG5-CDG (CDG IIi)
☐ COG6	Component Of Oligomeric Golgi Complex 6	COG6-CDG (CDG III)
☐ COG7	Component Of Oligomeric Golgi Complex 7	COG7-CDG (CDG IIe)
☐ COG8	Component Of Oligomeric Golgi Complex 8	COG8-CDG (CDG IIh)
☐ DDOST	Dolichyl-Diphosphooligosaccharide--Protein Glycosyltransferase Non-Catalytic Subunit	DDOST-CDG (CDG Ir)
☐ DHDDS	Dehydrodolichyl Diphosphate Synthase Subunit	DHDDS-CDG (Retinitis pigmentosa 59)
☐ DOLK	Dolichol Kinase	DOLK-CDG (CDG Im)
☐ DPM1	Dolichyl-Phosphate Mannosyltransferase Subunit 1, Catalytic	DPM1-CDG (CDG Ie)
☐ DPM2	Dolichyl-Phosphate Mannosyltransferase Subunit 2, Regulatory	DPM2-CDG (CDG Iu)
☐ DPM3	Dolichyl-Phosphate Mannosyltransferase Subunit 3	DPM3-CDG (CDG Io)
☐ GALE	UDP-Galactose-4-Epimerase	Galactose epimerase deficiency
☐ GALK1	Galactokinase 1	Galactokinase deficiency with cataracts
☐ GALT	Galactose-1-Phosphate Uridylyltransferase	Galactosemia
☐ GMPPA	GDP-Mannose Pyrophosphorylase A	GMPPA-CDG (Alacrima, achalasia, and mental retardation syndrome)
☐ MAN1B1	Mannosidase Alpha Class 1B Member 1	Mental retardation, autosomal recessive 15
☐ MAGT1	Magnesium Transporter 1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia
☐ MGAT2	Mannosyl (Alpha-1,6-)-Glycoprotein Beta-1,2-N-Acetylglucosaminyltransferase	MGAT2-CDG (CDG IIa)
☐ MOGS	Mannosyl-Oligosaccharide Glucosidase	MOGS-CDG (CDG IIb)
☐ MPDU1	Mannose-P-Dolichol Utilization Defect 1	MPDU1-CDG (CDG If)
☐ MPI	Mannose Phosphate Isomerase	MPI-CDG (CDG Ib)
☐ NUS1	NUS1 Dehydrodolichyl Diphosphate Synthase Subunit	NUS1-CDG (CDG 1aa)
☐ PGM1	Phosphoglucomutase 1	PGM1-CDG (CDG It)
☐ PMM2	Phosphomannomutase 2	PMM2-CDG (CDG Ia)
☐ POMT2	Protein O-Mannosyltransferase 2	POMT2-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ RFT1	RFT1 Homolog	RFT1-CDG (CDG In)
☐ SLC35A1	Solute Carrier Family 35 Member A1	SLC35A1-CDG (CDG IIf)
☐ SLC35A2	Solute Carrier Family 35 Member A2	SLC35A2-CDG (CDG IIm)
☐ SLC35C1	Solute Carrier Family 35 Member C1	SLC35C1-CDG (CDG IIc)
☐ SLC39A8	Solute Carrier Family 39 Member 8	SLC39A8-CDG (CDG IIn)
☐ SRD5A3	Steroid 5 Alpha-Reductase 3	SRD5A3-CDG (CDG Iq; Kahrizi syndrome)
☐ SSR4	Signal Sequence Receptor Subunit 4	SSR4-CDG (CDG-Iy)

☐ STT3A	STT3A, Catalytic Subunit Of The Oligosaccharyltransferase Complex	STT3A-CDG (CDG Iw)
☐ STT3B	STT3B, Catalytic Subunit Of The Oligosaccharyltransferase Complex	STT3B-CDG (CDG Ix)
☐ TF	Transferrin	Atransferrinemia
☐ TMEM165	Transmembrane Protein 165	TMEM165-CDG (CDG IIk)
☐ TMEM199	Transmembrane Protein 199	TMEM199-CDG (CDG IIp)

☐ SINDROME DI WALKER-WARBURG (15 GENI)

Gene	Proteina	Malattia
☐ B3GALNT2	Beta-1,3-N-Acetylgalactosaminyltransferase 2	B3GALNT2-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11))
☐ B4GAT1	Beta-1,4-Glucuronyltransferase 1	B4GAT1-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ COL4A1	Collagen Type IV Alpha 1 Chain	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps
☐ CRPPA (ex. ISPD)	Isoprenoid Synthase Domain Containing	ISPD-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ DAG1	Dystroglycan 1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies)
☐ FKRP	Fukutin Related Protein	FKRP-CDG (Muscular dystrophy-dystroglycanopathy)
☐ FKTN	Fukutin	FKTN-CDG (Fukuyama congenital muscular dystrophy)
☐ LARGE1	LARGE Xylosyl- And Glucuronyltransferase 1	LARGE1-CDG (Muscular dystrophy-dystroglycanopathy)
☐ POMGNT1	Protein O-Linked Mannose N-Acetylglucosaminyltransferase 1 (Beta 1,2-)	POMGNT1-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ POMGNT2	Protein O-Linked Mannose N-Acetylglucosaminyltransferase 2 (Beta 1,4-)	POMGNT2-CDG ((Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ POMK	Protein-O-Mannose Kinase	POMK-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ POMT1	Protein O-Mannosyltransferase 1	POMT1-CDG (Muscular dystrophy-dystroglycanopathy)
☐ POMT2	Protein O-Mannosyltransferase 2	POMT2-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ RXYLT1	Ribitol Xylosyltransferase 1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10
☐ TMEM5	Transmembrane Protein 5	TMEM5-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))

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Viale Pieraccini, 24 - 50139 Firenze
Tel. +39 055 5662543 - Fax +39 055 5662849; email: a.morrone@meyer.it

☐ ALFA-DISTROGLICANOPATIA (19 GENI)

Gene	Proteina	Malattia
☐ ANO5	Anoctamin 5	Autosomal dominant Gnathodiaphyseal dysplasia; Autosomal recessive Muscular dystrophy, limb-girdle, type 2L
☐ B3GALNT2	Beta-1,3-N-Acetylgalactosaminyltransferase 2	B3GALNT2-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11))
☐ B4GAT1	Beta-1,4-Glucuronyltransferase 1	B4GAT1-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ CRPPA (ex. ISPD)	Isoprenoid Synthase Domain Containing	ISPD-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ DAG1	Dystroglycan 1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies)
☐ DPM1	Dolichyl-Phosphate Mannosyltransferase Subunit 1, Catalytic	DPM1-CDG (CDG Ie)
☐ DPM2	Dolichyl-Phosphate Mannosyltransferase Subunit 2, Regulatory	DPM2-CDG (CDG Iu)
☐ DPM3	Dolichyl-Phosphate Mannosyltransferase Subunit 3	DPM3-CDG (CDG Io)
☐ FKRP	Fukutin Related Protein	FKRP-CDG (Muscular dystrophy-dystroglycanopathy)
☐ FKTN	Fukutin	FKTN-CDG (Fukuyama congenital muscular dystrophy)
☐ GMPPB	GDP-Mannose Pyrophosphorylase B	GMPPB-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ LARGE1	LARGE Xylosyl- And Glucuronyltransferase 1	LARGE1-CDG (Muscular dystrophy-dystroglycanopathy)
☐ POMGNT1	Protein O-Linked Mannose N-Acetylglucosaminyltransferase 1 (Beta 1,2-)	POMGNT1-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ POMGNT2	Protein O-Linked Mannose N-Acetylglucosaminyltransferase 2 (Beta 1,4-)	POMGNT2-CDG ((Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ POMK	Protein-O-Mannose Kinase	POMK-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ POMT1	Protein O-Mannosyltransferase 1	POMT1-CDG (Muscular dystrophy-dystroglycanopathy)
☐ POMT2	Protein O-Mannosyltransferase 2	POMT2-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ RXYLT1	Ribitol Xylosyltransferase 1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10
☐ TMEM5	Transmembrane Protein 5	TMEM5-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))

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SINDROME MIASTENICA CONGENITA CON DIFETTO DI GLICOSILAZIONE (5 GENI)

Gene	Proteina	Malattia
ALG2	ALG2, Alpha-1,3/1,6-Mannosyltransferase	ALG2-CDG (CDG Ii)
ALG14	ALG14, UDP-N-Acetylglucosaminyltransferase Subunit	ALG14-CDG (Myasthenic syndrome, congenital, 15, without tubular aggregates)
DPAGT1	Dolichyl-Phosphate N-Acetylglucosaminephosphotransferase 1	DPAGT1-CDG (CDG Ij)
GFPT1	Glutamine--Fructose-6-Phosphate Transaminase 1	GFPT1-CDG (Myasthenia, congenital, 12, with tubular aggregates)
GMPPB	GDP-Mannose Pyrophosphorylase B	GMPPB-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))

RIFIUTO DELLA FRUTTA (4 GENI)

Gene	Proteina	Malattia
ALDOB	Aldolase, Fructose-Bisphosphate B	Hereditary fructose intolerance
FBP1	Fructose-1,6-Bisphosphatase 1	Fructose-1,6-bisphosphatase deficiency
GK	Glycerol Kinase	Glycerol kinase deficiency
KHK	Ketohexokinase	Fructosuria

GALATTOSEMIA (3 GENI)

Gene	Proteina	Malattia
GALE	UDP-Galactose-4-Epimerase	Galactose epimerase deficiency
GALK1	Galactokinase 1	Galactokinase deficiency with cataracts
GALT	Galactose-1-Phosphate Uridylyltransferase	Galactosemia

RABDOMIOLISI (75 GENI)

Gene	Proteina	Malattia
ACAD9	Very-Long-Chain Acyl-CoA Dehydrogenase VLCAD	Mitochondrial complex I deficiency due to ACAD9 deficiency
ACADVL	Acyl-CoA Dehydrogenase, Very Long Chain	VLCAD deficiency
ADSL	Adenylosuccinate Lyase	Adenylosuccinase deficiency
AGL	AGL Amylo-Alpha-1, 6-Glucosidase, 4-Alpha-Glucanotransferase	Glycogen storage disease IIIa; Glycogen storage disease IIIb
AHCY	Adenosylhomocysteinase	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
ALDOA	Aldolase, Fructose-Bisphosphate A	Glycogen storage disease XII
AMACR	Alpha-Methylacyl-CoA Racemase	Bile acid synthesis defect, congenital, 4
AMPD1	Adenosine Monophosphate Deaminase 1	Myopathy due to myoadenylate deaminase deficiency
ANO5	Anoctamin 5	Muscular dystrophy, limb-girdle, type 2L; Miyoshi muscular dystrophy 3; Gnathodiaphyseal dysplasia
ATP2A1	ATPase Sarcoplasmic/Endoplasmic Reticulum Ca2+ Transporting 1	Brody myopathy
ATP5F1D	ATP Synthase F1 Subunit Delta	Mitochondrial complex V (ATP synthase) deficiency
BCS1L	BCS1 Homolog, Ubiquinol-Cytochrome C Reductase Complex Chaperone	Bjornstad syndrome; GRACILE syndrome; Leigh syndrome ; Mitochondrial complex III deficiency, nuclear type 1
CACNA1S	Calcium Voltage-Gated Channel Subunit Alpha1 S	Hypokalemic periodic paralysis, type 1; Malignant hyperthermia susceptibility 5;
CASQ1	Calsequestrin 1	Myopathy, vacuolar, with CASQ1 aggregates

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☐ CAV3	Caveolin 3	Cardiomyopathy, familial hypertrophic; Creatine phosphokinase, elevated serum; Long QT syndrome 9; Myopathy, distal, Tateyama type; Rippling muscle disease
☐ COL4A1	Collagen Type IV Alpha 1 Chain	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps
☐ CPT2	Carnitine Palmitoyltransferase 2	CPT deficiency, hepatic, type II; CPT II deficiency, lethal neonatal; Myopathy due to CPT II deficiency; Encephalopathy, acute, infection-induced, 4, susceptibility to
☐ CRPPA (ex ISPD)	Isoprenoid Synthase Domain Containing	ISPD-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ CTDP1	CTD Phosphatase Subunit 1	Congenital cataracts, facial dysmorphism, and neuropathy
☐ CYP2C8	Cytochrome P450 Family 2 Subfamily C Member 8	{Drug metabolism, altered, CYP2C8-related}
☐ DGUOK	Deoxyguanosine Kinase	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type); Portal hypertension, noncirrhotic; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4
☐ DLD	Dihydrolipoamide Dehydrogenase	Dihydrolipoamide dehydrogenase deficiency; Pyruvate dehydrogenase component E3
☐ DMD	Dystrophin	Duchenne muscular dystrophy; Becker muscular dystrophy
☐ DYSF	Dysferlin	Miyoshi muscular dystrophy 1; Muscular dystrophy, limb-girdle, type 2B; Myopathy, distal, with anterior tibial onset
☐ ENO3	Enolase 3	Glycogen storage disease XIII
☐ ETFA	Electron Transfer Flavoprotein Alpha Subunit	Glutaric acidemia IIA; MADD
☐ ETFB	Electron Transfer Flavoprotein Beta Subunit	Glutaric acidemia IIB; MADD
☐ ETFDH	Electron Transfer Flavoprotein Dehydrogenase	Glutaric acidemia IIC; MADD
☐ FDX2	Ferredoxin 2	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy
☐ FKRP	Fukutin Related Protein	Muscular dystrophy-dystroglycanopathy
☐ FKTN	Fukutin	Fukuyama congenital muscular dystrophy-dystroglycanopathy
☐ GAA	Glucosidase Alpha	Glycogenosis type II/Pompe
☐ GBE1	1,4-Alpha-Glucan Branching Enzyme 1	Glycogen storage disease IV; Polyglucosan body disease, adult form
☐ GMPPB	GDP-Mannose Pyrophosphorylase B	GMPPB-CDG (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies))
☐ GYG1	Glycogenin 1	Glycogen storage disease XV; Polyglucosan body myopathy 2
☐ GYS1	Glycogen Synthase 1	Glycogen storage disease 0, muscle
☐ HADHA	Hydroxyacyl-CoA Dehydrogenase/3-Ketoacyl-CoA Thiolase/Enoyl-CoA Hydratase (Trifunctional Protein), Alpha Subunit	LCHAD deficiency; Fatty liver, acute, of pregnancy; HELLP syndrome, maternal, of pregnancy; Trifunctional protein deficiency
☐ HADHB	Hydroxyacyl-CoA Dehydrogenase/3-Ketoacyl-CoA Thiolase/Enoyl-CoA Hydratase (Trifunctional Protein), Beta Subunit	LCHAD deficiency; Trifunctional protein deficienc

☐ HMBS	Hydroxymethylbilane Synthase	Porphyria, acute intermittent; Porphyria, acute intermittent, nonerythroid variant
☐ HRAS	HRas Proto-Oncogene, GTPase	Congenital myopathy with excess of muscle spindles; Costello syndrome; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;
☐ ISCU	Iron-Sulfur Cluster Assembly Enzyme	Myopathy with lactic acidosis, hereditary
☐ KCNE3	Potassium Voltage-Gated Channel Subfamily E Regulatory Subunit 3	Brugada syndrome 6
☐ KCNJ11	Potassium Inwardly Rectifying Channel Subfamily J Member 11	Diabetes mellitus, transient neonatal, 3; Diabetes, permanent neonatal, with or without neurologic features; Hyperinsulinemic hypoglycemia, familial, 2; Maturity-onset diabetes of the young, type 13
☐ KCNJ2	Potassium Inwardly Rectifying Channel Subfamily J Member 2	Andersen syndrome; Atrial fibrillation, familial, 9
☐ LAMP2	Lysosomal Associated Membrane Protein 2	Danon
☐ LDHA	Lactate Dehydrogenase A	Glycogen storage disease XI
☐ LDHB	Lactate Dehydrogenase B	[Lactate dehydrogenase-B deficiency]
☐ LIPA	Lipase A, Lysosomal Acid Type	Wolman disease
☐ LPIN1	Lipin 1	Deficit of lipin 1
☐ MYH3	Myosin Heavy Chain 3	Arthrogryposis, distal, type 2A (Freeman-Sheldon); Arthrogryposis, distal, type 2B3 (Sheldon-Hall); Contractures, pterygia, and variable skeletal fusions syndrome 1A; Contractures, pterygia, and variable skeletal fusions syndrome 1B
☐ PFKM	Phosphofructokinase, Muscle	Glycogen storage disease VII
☐ PGAM2	Phosphoglycerate Mutase 2	Glycogen storage disease X
☐ PGK1	Phosphoglycerate Kinase 1	Phosphoglycerate kinase 1 deficiency
☐ PGM1	Phosphoglucomutase 1	Congenital disorder of glycosylation, type It
☐ PHKA1	Phosphorylase Kinase, Alpha 1 (Muscle)	Glycogen storage disease IX
☐ PHKB	Phosphorylase Kinase Beta Subunit	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive
☐ PHKG2	Phosphorylase Kinase Catalytic Subunit Gamma 2	Cirrhosis due to liver phosphorylase kinase deficiency; Glycogen storage disease IXc
☐ PRKAG2	Protein Kinase AMP-Activated Non-Catalytic Subunit Gamma 2	Glycogen storage disease of heart, lethal congenital
☐ PYGL	Glycogen Phosphorylase L	Glycogen storage disease VI
☐ PYGM	Phosphorylase, Glycogen, Muscle	McArdle disease; Glycogen storage disease V
☐ QARS1	Glutaminyl-TRNA Synthetase	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy
☐ RBCK1	RANBP2-Type And C3HC4-Type Zinc Finger Containing 1	Polyglucosan body myopathy 1 with or without immunodeficiency
☐ RYR1	Ryanodine Receptor 1	Central core disease; King-Denborough syndrome; Minicore myopathy with external ophthalmoplegia; Neuromuscular disease, congenital, with uniform type 1 fiber; Malignant hyperthermia susceptibility 1
☐ SCN4A	Sodium Voltage-Gated Channel Alpha Subunit 4	Hyperkalemic periodic paralysis, type 2; Hypokalemic periodic paralysis, type 2; Myasthenic syndrome, congenital, 16; Myotonia congenita, atypical, acetazolamide-responsive; Paramyotonia congenita
☐ SGCA	Sarcoglycan Alpha	Muscular dystrophy, limb-girdle, autosomal recessive 3
☐ SIL1	SIL1 Nucleotide Exchange Factor	Marinesco-Sjogren syndrome
☐ SLC16A1	Solute Carrier Family 16 Member 1	Erythrocyte lactate transporter defect; Hyperinsulinemic hypoglycemia, familial, 7;

		Monocarboxylate transporter 1 deficiency
☐ SLC22A5	Solute Carrier Family 22 (Organic Cation/Carnitine Transporter), Member 5	Carnitine deficiency, systemic primary
☐ SLC25A20	Carnitine-acylcarnitine translocase deficiency	Solute Carrier Family 25 Member 20
☐ SUCLA2	Succinate-CoA Ligase ADP-Forming Beta Subunit	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
☐ TANGO2	Transport And Golgi Organization 2 Homolog	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration
☐ TK2	Thymidine Kinase 2, Mitochondrial	Mitochondrial DNA depletion syndrome 2 (myopathic type)
☐ TSEN54	TRNA Splicing Endonuclease Subunit 54	Pontocerebellar hypoplasia
☐ TSFM	Ts Translation Elongation Factor, Mitochondrial	Combined oxidative phosphorylation deficiency 3
☐ XK	X-Linked Kx Blood Group	McLeod syndrome with or without chronic granulomatous disease

☐ MALATTIE DA DEPOSITO DI GLICOGENO-FORME MUSCOLARI (21 GENI)

Gene	Proteina	Malattia
☐ AGL	AGL Amylo-Alpha-1, 6-Glucosidase, 4-Alpha-Glucanotransferase	Glycogen storage disease IIIa; Glycogen storage disease IIIb
☐ ALDOA	Aldolase, Fructose-Bisphosphate A	Glycogen storage disease XII
☐ ENO3	Enolase 3	Glycogen storage disease XIII
☐ GAA	Glucosidase Alpha	Glycogenosis type II/Pompe
☐ GBE1	1,4-Alpha-Glucan Branching Enzyme 1	Glycogen storage disease IV; Polyglucosan body disease, adult form
☐ GYG1	Glycogenin 1	Glycogen storage disease XV; Polyglucosan body myopathy 2
☐ GYS1	Glycogen Synthase 1	Glycogen storage disease 0, muscle
☐ LAMP2	Lysosomal Associated Membrane Protein 2	Danon
☐ LDHA	Lactate Dehydrogenase A	Glycogen storage disease XI
☐ LDHB	Lactate Dehydrogenase B	[Lactate dehydrogenase-B deficiency]
☐ PFKM	Phosphofructokinase, Muscle	Glycogen storage disease VII
☐ PGAM2	Phosphoglycerate Mutase 2	Glycogen storage disease X
☐ PGK1	Phosphoglycerate Kinase 1	Phosphoglycerate kinase 1 deficiency
☐ PGM1	Phosphoglucomutase 1	Congenital disorder of glycosylation, type It
☐ PHKA1	Phosphorylase Kinase, Alpha 1 (Muscle)	Glycogen storage disease IX
☐ PHKB	Phosphorylase Kinase Beta Subunit	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive
☐ PHKG2	Phosphorylase Kinase Catalytic Subunit Gamma 2	Cirrhosis due to liver phosphorylase kinase deficiency; Glycogen storage disease IXc
☐ PRKAG2	Protein Kinase AMP-Activated Non-Catalytic Subunit Gamma 2	Glycogen storage disease of heart, lethal congenital
☐ PYGL	Glycogen Phosphorylase L	Glycogen storage disease VI
☐ PYGM	Phosphorylase, Glycogen, Muscle	McArdle disease; Glycogen storage disease V
☐ RBCK1	RANBP2-Type And C3HC4-Type Zinc Finger Containing 1	Polyglucosan body myopathy 1 with or without immunodeficiency

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PARALISI PERIODICA IPOKALIEMICA (4 GENI)

Gene	Proteina	Malattia
CACNA1S	Calcium Voltage-Gated Channel Subunit Alpha1 S	Hypokalemic periodic paralysis, type 1; Malignant hyperthermia susceptibility 5;
KCNE3	Potassium Voltage-Gated Channel Subfamily E Regulatory Subunit 3	Brugada syndrome 6
KCNJ2	Potassium Inwardly Rectifying Channel Subfamily J Member 2	Andersen syndrome; Atrial fibrillation, familial, 9
SCN4A	Sodium Voltage-Gated Channel Alpha Subunit 4	Hyperkalemic periodic paralysis, type 2; Hypokalemic periodic paralysis, type 2; Myasthenic syndrome, congenital, 16; Myotonia congenita, atypical, acetazolamide-responsive; Paramyotonia congenita

DIFETTI DELLA BETA-OSSIDAZIONE DEGLI ACIDI GRASSI (28 GENI)

Gene	Proteina	Malattia
ACACA	Acetyl-CoA Carboxylase Alpha	Acetyl-CoA carboxylase deficiency
ACAD9	Very-Long-Chain Acyl-CoA Dehydrogenase VLCAD	Mitochondrial complex I deficiency due to ACAD9 deficiency
ACADL	Acyl-CoA Dehydrogenase Long Chain	Bell et al 2011, Sci Transl Med 3(65):65ra4
ACADM	Acyl-CoA Dehydrogenase, C-4 To C-12 Straight Chain	MEDIUM-CHAIN ACYL-CoA DEHYDROGENASE; MCAD; MCADH
ACADS	Acyl-CoA Dehydrogenase, C-2 To C-3 Short Chain	SHORT-CHAIN ACYL-CoA DEHYDROGENASE; SCAD
ACADVL	Acyl-CoA Dehydrogenase, Very Long Chain	VLCAD deficiency
ACAT1	Acetoacetyl Coenzyme A Thiolase	MITOCHONDRIAL ACETOACETYL-CoA THIOLASE; MAT
CPT1A	Carnitine Palmitoyltransferase 1A	CPT deficiency, hepatic, type IA
CPT2	Carnitine Palmitoyltransferase 2	CPT deficiency, hepatic, type II; CPT II deficiency, lethal neonatal; Myopathy due to CPT II deficiency; Encephalopathy, acute, infection-induced, 4, susceptibility to
ECHS1	Enoyl-CoA Hydratase, Short Chain 1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency
ETFA	Electron Transfer Flavoprotein Alpha Subunit	Glutaric acidemia IIA; MADD
ETFB	Electron Transfer Flavoprotein Beta Subunit	Glutaric acidemia IIB; MADD
ETFDH	Electron Transfer Flavoprotein Dehydrogenase	Glutaric acidemia IIC; MADD
ETHE1	ETHE1, Persulfide Dioxygenase	Ethylmalonic encephalopathy
FLAD1	Flavin Adenine Dinucleotide Synthetase 1	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency
GLUD1	Glutamate Dehydrogenase 1	Hyperinsulinism-hyperammonemia syndrome
HADH	Hydroxyacyl-CoA Dehydrogenase	SCHAD; 3-hydroxyacyl-CoA dehydrogenase deficiency (SCHAD); Hyperinsulinemic hypoglycemia, familial, 4
HADHA	Hydroxyacyl-CoA Dehydrogenase/3-Ketoacyl-CoA Thiolase/Enoyl-CoA Hydratase (Trifunctional Protein), Alpha Subunit	LCHAD deficiency; Fatty liver, acute, of pregnancy; HELLP syndrome, maternal, of pregnancy; Trifunctional protein deficiency
HADHB	Hydroxyacyl-CoA Dehydrogenase/3-Ketoacyl-CoA Thiolase/Enoyl-CoA Hydratase (Trifunctional Protein), Beta Subunit	LCHAD deficiency; Trifunctional protein deficienc
HMGCL	3-Hydroxymethyl-3-Methylglutaryl-	HMG-CoA lyase deficiency

	CoA Lyase	
HMGCS2	3-Hydroxy-3-Methylglutaryl-CoA Synthase 2	HMG-CoA synthase-2 deficiency
OXCT1	3-Oxoacid CoA-Transferase 1	Succinyl CoA:3-oxoacid CoA transferase deficienc
SLC16A1	Solute Carrier Family 16 Member 1	Erythrocyte lactate transporter defect; Hyperinsulinemic hypoglycemia, familial, 7; Monocarboxylate transporter 1 deficiency
SLC22A5	Solute Carrier Family 22 (Organic Cation/Carnitine Transporter), Member 5	Carnitine deficiency, systemic primary
SLC25A20	Solute Carrier Family 25 (Carnitine/Acylcarnitine Translocase), Member 20	Carnitine-acylcarnitine translocase deficiency
SLC52A1	Solute Carrier Family 52 (Riboflavin Transporter), Member 1	Riboflavin deficiency
SLC52A2	Solute Carrier Family 52 (Riboflavin Transporter), Member 2	Brown-Vialetto-Van Laere syndrome 2
SLC52A3	Solute Carrier Family 52 (Riboflavin Transporter), Member 3	Brown-Vialetto-Van Laere syndrome 1; Fazio-Londe disease
TANGO2	Transport And Golgi Organization 2 Homolog	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration

DISORDINI DI CHETOLISI (3 GENI)

Gene	Proteina	Malattia
ACAT1	Acetoacetyl Coenzyme A Thiolase	MITOCHONDRIAL ACETOACETYL-CoA THIOLASE; MAT
OXCT1	3-Oxoacid CoA-Transferase 1	Succinyl CoA:3-oxoacid CoA transferase deficienc
SLC16A1	Solute Carrier Family 16 Member 1	Erythrocyte lactate transporter defect; Hyperinsulinemic hypoglycemia, familial, 7; Monocarboxylate transporter 1 deficiency

DISORDINI DI CHETOGENESI (2 GENI)

Gene	Proteina	Malattia
HMGCL	3-Hydroxymethyl-3-Methylglutaryl-CoA Lyase	HMG-CoA lyase deficiency
HMGCS2	3-Hydroxy-3-Methylglutaryl-CoA Synthase 2	HMG-CoA synthase-2 deficiency

DEFICIT DI SERINA E SINDROME DI NEU-LAXOVA (4 GENI)

Gene	Proteina	Malattia
PHGDH	Phosphoglycerate Dehydrogenas	Neu-Laxova syndrome 1; Phosphoglycerate dehydrogenase deficiency
PSAT1	Phosphoserine Aminotransferase 1	Phosphoserine aminotransferase deficiency; Neu-Laxova syndrome 2
PSPH	Phosphoserine Phosphatase	Phosphoserine phosphatase deficiency
SLC1A4	Solute Carrier Family 1 Member 4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly

Laboratorio di Biologia Molecolare delle Malattie Neurometaboliche
Responsabile: Prof.ssa Amelia Morrone
Viale Pieraccini, 24 - 50139 Firenze
Tel. +39 055 5662543 - Fax +39 055 5662849; email: a.morrone@meyer.it

DEFICIT POLIMERASI GAMMA (2 GENI)

Gene	Proteina	Malattia
POLG	Polymerase (DNA) Gamma, Catalytic Subunit	Mitochondrial DNA depletion syndrome (Alpers type or MNGIE type); Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE); Progressive external ophthalmoplegia
POLG2	DNA Polymerase Gamma 2, Accessory Subunit	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4

CEROIDOLIPOFUSCINOSI NEURONALI (NCL) (13 GENI)

Gene	Proteina	Malattia
ATP13A2	ATPase Type 13A2	NCL 12; Kufor-Rakeb syndrome
CLN3	Battenin	NCL 3; Batten disease; Spielmeyer-Vogt disease
CLN5	Protein CLN5	NCL 5
CLN6	Protein CLN6	NCL 6; Kufs type, adult onset
CLN8	Protein CLN8	NCL 8; Northern epilepsy variant
CTSD	Cathepsin D	NCL 10
CTSF	Cathepsin F	NCL 13; Kufs type
DNAJC5	DnaJ homolog subfamily C member 5	NCL 4; Parry type
GRN	Granulin	NCL 11; Aphasia, primary progressive; Frontotemporal lobar degeneration with ubiquitin-positive inclusions
KCTD7	BTB/POZ domain-containing protein KCTD7	NCL 14; Epilepsy, progressive myoclonic 3, with or without intracellular inclusions
MFSD8	Major Facilitator Superfamily Domain Containing 8	NCL 7; Macular dystrophy with central cone involvement
PPT1	Palmitoyl-Protein Thioesterase 1	NCL 1
TPP1	Tripeptidyl Peptidase 1	NCL 2; Spinocerebellar ataxia, autosomal recessive 7

MALATTIE DA ACCUMULO LISOSOMIALE-PANNELLO COMPLETO (64 GENI)

Gene	Proteina	Malattia
AGA	Aspartylglucosaminidase	Aspartylglucosaminuria
AP3B1	Clathrin Assembly Protein Complex 3 Beta-1 Large Chain	Hermansky-Pudlak syndrome 2
AP5Z1	Adaptor Related Protein Complex 5 Zeta 1 Subunit	Spastic paraplegia 48, autosomal recessive
ARSA	Arylsulfatase A	Metachromatic leucodistrophy
ARSB	Arylsulfatase B	MPS VI
ASAH1	N-Acylsphingosine Amidohydrolase (Acid Ceramidase)	Farber
ATP13A2	ATPase Type 13A2	NCL 12; Kufor-Rakeb syndrome
BLOC1S3	Biogenesis Of Lysosomal Organelles Complex 1 Subunit 3	Hermansky-Pudlak syndrome 8
CLN3	Battenin	NCL 3; Batten disease; Spielmeyer-Vogt disease
CLN5	Protein CLN5	NCL 5
CLN6	Protein CLN6	NCL 6; Kufs type, adult onset
CLN8	Protein CLN8	NCL 8; Northern epilepsy variant
CTNS	Cystinosin, Lysosomal Cystine Transporter	Cystinosis
CTSA	Cathepsin A	Galactosialidosis
CTSD	Cathepsin D	NCL 10
CTSF	Cathepsin F	NCL 13; Kufs type
CTSK	Cathepsin K	Pycnodysostosis
DNAJC5	DnaJ homolog subfamily C member 5	NCL 4; Parry type

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☐ DTNBP1	Dystrobrevin Binding Protein 1	Hermansky-Pudlak syndrome 7
☐ FUCA1	Alpha-L-Fucosidase 1	Fucosidosis
☐ GAA	Glucosidase Alpha	Glycogenosis type II/Pompe
☐ GALC	Galactosylceramidase	Krabbe
☐ GALNS	Galactosamine (N-Acetyl)-6-Sulfatase	MPS IVA
☐ GBA	Glucosylceramidase Beta	Gaucher
☐ GLA	Galactosidase Alpha	Fabry
☐ GLB1	Galactosidase Beta 1	MPS IVB/Gangliosidosis GM1
☐ GM2A	GM2 Ganglioside Activator	Gangliosidosis GM2, activator defect
☐ GNE	Acetylmannosamine Kinase	Sialuria
☐ GNPTAB	N-Acetylglucosamine-1-Phosphate Transferase Alpha And Beta Subunits	Mucolipidosis II alfa/beta, III alfa/beta
☐ GNPTG	N-Acetylglucosamine-1-Phosphate Transferase Gamma Subunit	Mucolipidosis III gamma
☐ GNS	Glucosamine (N-Acetyl)-6-Sulfatase	MPS III D (San Filippo D)
☐ GRN	Granulin	NCL 11; Aphasia, primary progressive; rontotemporal lobar degeneration with ubiquitin-positive inclusions
☐ GUSB	Glucuronidase Beta	MPS VII
☐ HEXA	Hexosaminidase Subunit Alpha	Gangliosidosis GM2, Tay Sachs
☐ HEXB	Hexosaminidase Subunit Beta	Gangliosidosis GM2, Sandhoff
☐ HGSNAT	Heparan-Alpha-Glucosaminide N-Acetyltransferase	MPS III C (San Filippo C)
☐ HPS1	Biogenesis Of Lysosomal Organelles Complex 3 Subunit 1	Hermansky-Pudlak syndrome 1
☐ HPS3	HPS3, Biogenesis Of Lysosomal Organelles Complex 2 Subunit 1	Hermansky-Pudlak syndrome 3
☐ HPS4	Biogenesis Of Lysosomal Organelles Complex 3 Subunit 2	Hermansky-Pudlak syndrome 4
☐ HPS5	Biogenesis Of Lysosomal Organelles Complex 2 Subunit 2	Hermansky-Pudlak syndrome 5
☐ HPS6	HPS6, Biogenesis Of Lysosomal Organelles Complex 2 Subunit 3	Hermansky-Pudlak syndrome 6
☐ HYAL1	Hyaluronoglucosaminidase 1	MPS IX
☐ IDS	Iduronate 2-Sulfatase	MPS II
☐ IDUA	Iduronidase, Alpha-L-	MPS I
☐ KCTD7	BTB/POZ domain-containing protein KCTD7	NCL 14; Epilepsy, progressive myoclonic 3, with or without intracellular inclusions
☐ LAMP2	Lysosomal Associated Membrane Protein 2	Danon
☐ LIPA	Lipase A, Lysosomal Acid Type	Wolman
☐ MAN2B1	Mannosidase Alpha Class 2B Member 1	alfa-mannosidosis
☐ MANBA	Mannosidase Beta	beta-mannosidosis
☐ MCOLN1	Mucolipin 1	Mucolipidosis IV
☐ MFSD8	Major Facilitator Superfamily Domain Containing 8	NCL 7; Macular dystrophy with central cone involvement
☐ NAGA	Alpha-N-Acetylgalactosaminidase	Schindler
☐ NAGLU	N-Acetyl-Alpha-Glucosaminidase	MPS III B (San Filippo B)
☐ NEU1	Neuraminidase 1	Sialidosis
☐ NPC1	Niemann-Pick C1 protein	Niemann-Pick type I
☐ NPC2	Epididymal secretory protein E1	Niemann-Pick type II
☐ PPT1	Palmitoyl-Protein Thioesterase 1	NCL 1
☐ PSAP	Prosaposin	Metachromatic leucodistrophy, Krabbe, Gaucher
☐ SCARB2	Scavenger Receptor Class B Member	epilepsy, progressive myoclonic 4, with or without

	2; LIMP-2	renal failure and unverricht-lundborg syndrome
☐ SGSH	N-Sulfoglucosamine Sulfohydrolase	MPS III A (San Filippo A)
☐ SLC17A5	Vesicular H(+)/Aspartate-Glutamate Cotransporter	Sialic acid storage disease
☐ SMPD1	Sphingomyelin Phosphodiesterase 1	Niemann-Pick
☐ SUMF1	Sulfatase Modifying Factor 1	Multiple sulfatase deficiency
☐ TPP1	Tripeptidyl Peptidase 1	NCL 2; Spinocerebellar ataxia, autosomal recessive 7

☐ MALATTIE PEROSSISOMIALI (28 GENI)

Gene	Proteina	Malattia
☐ ABCD1	ATP Binding Cassette Subfamily D Member 1	Adrenoleukodystrophy
☐ ACOX1	Palmitoyl Acyl-CoA Oxidase 1	Peroxisomal acyl-CoA oxidase deficiency
☐ AGPS	Alkylglycerone Phosphate Synthase	Rhizomelic chondrodysplasia punctata, type 3
☐ AGXT	Alanine-Glyoxylate Aminotransferase	Hyperoxaluria, primary, type 1
☐ AMACR	Alpha-Methylacyl-CoA Racemase	Alpha-methylacyl-CoA racemase deficiency; Bile acid synthesis defect, congenital, 4
☐ BAAT	Bile Acid-CoA:Amino Acid N-Acyltransferase	Hypercholanemia, familial
☐ BCAP31	B-Cell Receptor-Associated Protein 3	Deafness, dystonia, and cerebral hypomyelination
☐ DNM1L	Dynamin 1-Like	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission 1
☐ FAR1	Fatty Acyl-CoA Reductase 1	Peroxisomal fatty acyl-CoA reductase 1 disorder
☐ GNPAT	Glyceronephosphate O-Acyltransferase	Rhizomelic chondrodysplasia punctata, type 2
☐ HSD17B4	Hydroxysteroid (17-Beta) Dehydrogenase 4	D-bifunctional protein deficiency; Perrault syndrome 1
☐ PEX1	Peroxisomal Biogenesis Factor 1	Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD); Heimler syndrome 1
☐ PEX10	Peroxisomal Biogenesis Factor 10	Peroxisome biogenesis disorder 6A (Zellweger); Peroxisome biogenesis disorder 6B
☐ PEX11B	Peroxisomal Biogenesis Factor 11 Beta	Peroxisome biogenesis disorder 14B
☐ PEX12	Peroxisomal Biogenesis Factor 12	Peroxisome biogenesis disorder 3A (Zellweger); Peroxisome biogenesis disorder 3B
☐ PEX13	Peroxisomal Biogenesis Factor 13	Peroxisome biogenesis disorder 11A (Zellweger); Peroxisome biogenesis disorder 11B
☐ PEX14	Peroxisomal Biogenesis Factor 14	Peroxisome biogenesis disorder 13A (Zellweger);
☐ PEX16	Peroxisomal Biogenesis Factor 16	Peroxisome biogenesis disorder 8A (Zellweger); Peroxisome biogenesis disorder 8B
☐ PEX19	Peroxisomal Biogenesis Factor 19	Peroxisome biogenesis disorder 12A (Zellweger)
☐ PEX2	Peroxisomal Biogenesis Factor 2	Peroxisome biogenesis disorder 5A (Zellweger); Peroxisome biogenesis disorder 5B
☐ PEX26	Peroxisomal Biogenesis Factor 26	Peroxisome biogenesis disorder 7A (Zellweger); Peroxisome biogenesis disorder 7B
☐ PEX3	Peroxisomal Biogenesis Factor 3	Peroxisome biogenesis disorder 10A (Zellweger)
☐ PEX5	Peroxisomal Biogenesis Factor 5	Peroxisome biogenesis disorder 2A (Zellweger); Peroxisome biogenesis disorder 2B; Rhizomelic chondrodysplasia punctata, type 5
☐ PEX6	Peroxisomal Biogenesis Factor 6	Peroxisome biogenesis disorder 4A (Zellweger); Peroxisome biogenesis disorder 4B (NALD/IRD); Heimler syndrome 2
☐ PEX7	Peroxisomal Biogenesis Factor 7	Peroxisome biogenesis disorder 9B; Rhizomelic chondrodysplasia punctata, type 1

PHYH	Phytanoyl-CoA 2-Hydroxylase	Refsum disease
SCP2	Sterol Carrier Protein 2	Leukoencephalopathy with dystonia and motor neuropathy
SUGCT	Succinyl-CoA:Glutarate-CoA Transferase	Glutaric aciduria III

IPERAMMONIEMIE (20 GENI)

Gene	Proteina	Malattia
ALDH18A1	Pyrroline-5-Carboxylate Synthetase	Cutis laxa, autosomal recessive, type IIIA; Spastic paraplegia 9A, autosomal dominant; Spastic paraplegia 9B, autosomal recessiv
ARG1	Arginase 1	Argininemia
ASL	Argininosuccinate Lyase	Argininosuccinic aciduria
ASS1	Argininosuccinate Synthase 1	Citrullinemia
CA5A	Carbonic Anhydrase 5A, Mitochondrial	Hyperammonemia due to carbonic anhydrase 5A deficiency
CPS1	Carbamoyl-Phosphate Synthase 1	Carbamoylphosphate synthetase I deficiency; Neonatal susceptibility to pulmonary hypertension; Venoocclusive disease after bone marrow transplantation
GLUD1	Glutamate Dehydrogenase 1	Hyperinsulinism-hyperammonemia syndrome
GLUL	Glutamate-Ammonia Ligase	Glutamine deficiency, congenital
IVD	Isovaleryl-CoA Dehydrogenase	Isovaleric acidemia
MUT	Methylmalonyl-CoA Mutase	Methylmalonic aciduria, mut(0) type
NAGS	N-Acetylglutamate Synthase	N-acetylglutamate synthase deficiency
OAT	Ornithine Aminotransferase	Gyrate atrophy of choroid and retina with or without ornithinemia
OTC	Ornithine Carbamoyltransferase	Ornithine transcarbamylase deficiency
PC	Pyruvate Carboxylase	Pyruvate carboxylase deficiency
PCCA	Propionyl-CoA Carboxylase Alpha Subunit	Propionicacidemia
PCCB	Propionyl-CoA Carboxylase Beta Subunit	Propionicacidemia
SLC25A13	Mitochondrial Aspartate Glutamate Carrier 2	Citrullinemia type II;
SLC25A15	Ornithine Transporter 1	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
SLC7A7	Sol ute Carrier Family 7 (Amino Acid Transporter Light Chain, Y+L System), Member 7	Lysinuric protein intolerance
TMEM70	Transmembrane Protein 7	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2

EPATOPATIA E/O COLESTASI (69 GENI)

Gene	Proteina	Malattia
ABCB11	ATP Binding Cassette Subfamily B Member 11	Cholestasis
ABCB4	ATP Binding Cassette Subfamily B Member 4	Cholestasis
ACADM	Acyl-CoA Dehydrogenase, C-4 To C-12 Straight Chain	MEDIUM-CHAIN ACYL-CoA DEHYDROGENASE; MCAD; MCADH
ACOX1	Palmitoyl Acyl-CoA Oxidase 1	Peroxisomal acyl-CoA oxidase deficiency
ADK	Adenosine Kinase	Hypermethioninemia due to adenosine kinase

		deficiency
☐ AGK	Acylglycerol Kinase	Sengers syndrome ; Mitochondrial DNA depletion syndrome 10
☐ AKR1D1	Aldo-Keto Reductase Family 1 Member D1	Bile acid synthesis defect, congenital, 2
☐ ALDOB	Aldolase, Fructose-Bisphosphate B	Hereditary fructose intolerance
☐ AMACR	Alpha-Methylacyl-CoA Racemase	Alpha-methylacyl-CoA racemase deficiency; Bile acid synthesis defect, congenital, 4
☐ ATAD3A	ATPase Family, AAA Domain Containing 3A	global developmental delay, hypotonia, optic atrophy, axonal neuropathy, and hypertrophic cardiomyopathy
☐ ATP7B	ATPase Copper Transporting Beta	Wilson disease
☐ ATP8B1	ATPase Phospholipid Transporting 8B1	Cholestasis
☐ BAAT	Bile Acid-CoA:Amino Acid N-Acyltransferase	Hypercholanemia, familial
☐ BCS1L	BCS1 Homolog, Ubiquinol-Cytochrome C Reductase Complex Chaperone	Bjornstad syndrome; GRACILE syndrome; Leigh syndrome ; Mitochondrial complex III deficiency, nuclear type 1
☐ CLDN1	Claudin 1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis
☐ CYP27A1	Cytochrome P450 Family 27 Subfamily A Member 1	Cerebrotendinous xanthomatosis
☐ CYP7A1	Cytochrome P450 Family 7 Subfamily A Member 1	
☐ CYP7B1	Cytochrome P450 Family 7 Subfamily B Member 1	Bile acid synthesis defect, congenital, 3
☐ DCDC2	Doublecortin Domain Containing 2	Neonatal Sclerosing Cholangitis
☐ DGUOK	Deoxyguanosine Kinase	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)
☐ DLD	Dihydrolipoamide Dehydrogenase	Dihydrolipoamide dehydrogenase deficiency; Pyruvate dehydrogenase component E3
☐ DNM1L	Dynamin 1-Like	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission 1
☐ ETHE1	ETHE1, Persulfide Dioxygenase	Ethylmalonic encephalopathy
☐ FAR1	Fatty Acyl-CoA Reductase 1	Peroxisomal fatty acyl-CoA reductase 1 disorder
☐ FBP1	Fructose-1,6-Bisphosphatase 1	Fructose-1,6-bisphosphatase deficiency
☐ FGFR2	Fibroblast Growth Factor Receptor 2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis
☐ GALE	UDP-Galactose-4-Epimerase	Galactose epimerase deficiency
☐ GALK1	Galactokinase 1	Galactokinase deficiency with cataracts
☐ GALT	Galactose-1-Phosphate Uridylyltransferase	Galactosemia
☐ HSD17B4	Hydroxysteroid (17-Beta) Dehydrogenase 4	D-bifunctional protein deficiency; Perrault syndrome 1
☐ HSD3B7	Hydroxy-Delta-5-Steroid Dehydrogenase, 3 Beta- And Steroid Delta-Isomerase 7	Bile acid synthesis defect, congenital, 1
☐ IARS	Isoleucyl-TRNA Synthetase	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy
☐ ISCA2	Iron-Sulfur Cluster Assembly 2	Multiple mitochondrial dysfunctions syndrome 4
☐ JAG1	Jagged 1	Alagille syndrome 1; Tetralogy of Fallot
☐ LIPA	Lipase A, Lysosomal Acid Type	Cholesteryl ester storage disease; Wolman disease
☐ LYRM4	LYR Motif Containing 4	Combined oxidative phosphorylation deficiency 19
☐ MPV17	MPV17, Mitochondrial Inner Membrane Protein	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)

☐ MTTP	Microsomal Triglyceride Transfer Protein	Abetalipoproteinemia
☐ NPC1	Niemann-Pick C1 protein	Niemann-Pick type I
☐ NPC2	Epididymal secretory protein E1	Niemann-Pick type II
☐ NR1H4	Nuclear Receptor Subfamily 1 Group H Member 4	Cholestasis
☐ PEX1	Peroxisomal Biogenesis Factor 1	Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD); Heimler syndrome 1
☐ PEX10	Peroxisomal Biogenesis Factor 10	Peroxisome biogenesis disorder 6A (Zellweger); Peroxisome biogenesis disorder 6B
☐ PEX11B	Peroxisomal Biogenesis Factor 11 Beta	Peroxisome biogenesis disorder 14B
☐ PEX12	Peroxisomal Biogenesis Factor 12	Peroxisome biogenesis disorder 3A (Zellweger); Peroxisome biogenesis disorder 3B
☐ PEX13	Peroxisomal Biogenesis Factor 13	Peroxisome biogenesis disorder 11A (Zellweger); Peroxisome biogenesis disorder 11B
☐ PEX14	Peroxisomal Biogenesis Factor 14	Peroxisome biogenesis disorder 13A (Zellweger);
☐ PEX16	Peroxisomal Biogenesis Factor 16	Peroxisome biogenesis disorder 8A (Zellweger); Peroxisome biogenesis disorder 8B
☐ PEX19	Peroxisomal Biogenesis Factor 19	Peroxisome biogenesis disorder 12A (Zellweger)
☐ PEX2	Peroxisomal Biogenesis Factor 2	Peroxisome biogenesis disorder 5A (Zellweger); Peroxisome biogenesis disorder 5B
☐ PEX26	Peroxisomal Biogenesis Factor 26	Peroxisome biogenesis disorder 7A (Zellweger); Peroxisome biogenesis disorder 7B
☐ PEX3	Peroxisomal Biogenesis Factor 3	Peroxisome biogenesis disorder 10A (Zellweger)
☐ PEX5	Peroxisomal Biogenesis Factor 5	Peroxisome biogenesis disorder 2A (Zellweger); Peroxisome biogenesis disorder 2B; Rhizomelic chondrodysplasia punctata, type 5
☐ PEX6	Peroxisomal Biogenesis Factor 6	Peroxisome biogenesis disorder 4A (Zellweger); Peroxisome biogenesis disorder 4B (NALD/IRD); Heimler syndrome 2
☐ PEX7	Peroxisomal Biogenesis Factor 7	Peroxisome biogenesis disorder 9B; Rhizomelic chondrodysplasia punctata, type 1
☐ POLG	Polymerase (DNA) Gamma, Catalytic Subunit	Mitochondrial DNA depletion syndrome 4A (Alpers type); Mitochondrial DNA depletion syndrome 4B (MNGIE type); Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE); Progressive external ophthalmoplegia
☐ RRM2B	Ribonucleotide Reductase Regulatory TP53 Inducible Subunit M2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy); Mitochondrial DNA depletion syndrome 8B (MNGIE type)
☐ SERPINA 1	Serpin Family A Member 1	Emphysema-cirrhosis, due to AAT deficiency
☐ SLC25A13	Solute Carrier Family 25 Member 13	Citrullinemia, type II
☐ SLC27A5	Solute Carrier Family 27 Member 5	
☐ SLC30A10	Solute Carrier Family 30 Member 10	Hypermanganesemia with dystonia 1
☐ TALDO1	Transaldolase 1	Transaldolase deficiency
☐ TJP2	Tight Junction Protein 2	Cholestasis
☐ TK2	Thymidine Kinase 2, Mitochondrial	Mitochondrial DNA depletion syndrome 2 (myopathic type)
☐ TMEM70	Transmembrane Protein 7	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2
☐ TWNK (C10orf2)	Mitochondrial Twinkle Protein	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type); Perrault syndrome 5;

		Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3
☐ TYMP	Thymidine Phosphorylase	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
☐ VIPAS39	VPS33B Interacting Protein, Apical-Basolateral Polarity Regulator, Spe-39 Homolog	Arthrogryposis, renal dysfunction, and cholestasis 2
☐ VPS33B	VPS33B, Late Endosome And Lysosome Associated	Arthrogryposis, renal dysfunction, and cholestasis 1

☐ METABOLISMO DEI METALLI (13 GENI)

Gene	Proteina	Malattia
☐ AP1S1	Adaptor Related Protein Complex 1 Sigma 1 Subunit	MEDNIK syndrome
☐ ATP7A	ATPase Copper Transporting AlphA	Menkes disease
☐ ATP7B	ATPase Copper Transporting Beta	Wilson disease
☐ BCS1L	BCS1 Homolog, Ubiquinol-Cytochrome C Reductase Complex Chaperone	GRACILE syndrome
☐ HAMP	Hepcidin Antimicrobial Peptide	Hemochromatosis, type 2B
☐ HFE	Hemochromatosis	Hemochromatosis
☐ HFE2	Hemochromatosis Type 2	Hemochromatosis, type 2A
☐ PANK2	Pantothenate Kinase 2	HARP syndrome; Neurodegeneration with brain iron accumulation 1
☐ SLC30A2	Solute Carrier Family 30 Member 2	Zinc deficiency, transient neonatal
☐ SLC33A1	Solute Carrier Family 33 Member 1	Congenital cataracts, hearing loss, and neurodegeneration; Spastic paraplegia 42, autosomal dominant
☐ SLC39A4	Solute Carrier Family 39 Member 4	Acrodermatitis enteropathica
☐ TF	Transferrin	Atransferrinemia
☐ TFR2	Transferrin Receptor 2	Hemochromatosis, type 3

☐ METABOLISMO DEGLI STEROLI (11 GENI)

Gene	Proteina	Malattia
☐ DHCR24	24-Dehydrocholesterol Reductase	Desmosterolosis
☐ DHCR7	7-Dehydrocholesterol Reductase	Smith-Lemli-Opitz syndrome
☐ EBP	Emopamil Binding Protein (Sterol Isomerase)	Conradi-Hunermann chondrodysplasia punctata; MEND syndrome
☐ FGFR2	Fibroblast Growth Factor Receptor 2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis
☐ LBR	Lamin B Receptor	Greenberg skeletal dysplasia; Pelger-Huet anomaly
☐ MSMO1	Methylsterol Monooxygenase 1	Microcephaly, congenital cataract, and psoriasiform dermatitis
☐ MVK	Mevalonate Kinase	Mevalonic aciduria
☐ NSDHL	NAD(P) Dependent Steroid Dehydrogenase-Like	CHILD syndrome
☐ PEX7	Peroxisomal Biogenesis Factor 7	Rhizomelic chondrodysplasia punctata, type 1
☐ POR	Cytochrome P450 Oxidoreductase	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
☐ SC5D	Sterol-C5-Desaturase	Lathosterolosis

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Responsabile: Prof.ssa Amelia Morrone
Viale Pieraccini, 24 - 50139 Firenze
Tel. +39 055 5662543 - Fax +39 055 5662849; email: a.morrone@meyer.it

IPEROSSALURIA (3 GENI)

Gene	Proteina	Malattia
AGXT	Alanine-Glyoxylate Aminotransferas	Hyperoxaluria, primary, type I
GRHPR	Glyoxylate Reductase/Hydroxypyruvate Reductase	Hyperoxaluria, primary, type II
HOGA1	4-Hydroxy-2-Oxoglutarate Aldolase 1	Hyperoxaluria, primary, type III

MALATTIA DELLE URINE A SCIROPPO D'ACERO (MSUD) (4 GENI)

Gene	Proteina	Malattia
BCKDHA	Branched Chain Keto Acid Dehydrogenase E1, Alpha Polypeptide	MSUD tipo Ia
BCKDHB	Branched Chain Keto Acid Dehydrogenase E1, Beta Polypeptide	MSUD tipo Ib
DBT	Dihydrolipoamide Branched Chain Transacylase E2	MSUD tipo II
DLD	Dihydrolipoamide Dehydrogenase	MSUD tipo III

ACIDURIA 3-METILGLUTACONICA (16 GENI)

Gene	Proteina	Malattia
AGK	Acylglycerol Kinase	Sengers syndrome; Cataract 38, autosomal recessive
ATAD3A	ATPase Family, AAA Domain Containing 3A	global developmental delay, hypotonia, optic atrophy, axonal neuropathy, and hypertrophic cardiomyopathy
ATP5E	Mitochondrial ATP Synthase Epsilon Chain	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3
ATPAF2	ATP Synthase Mitochondrial F1 Complex Assembly Factor 2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1
AUH	AU RNA Binding Protein/Enoyl-CoA Hydratase	3-methylglutaconic aciduria, type I
CLPB	ClpB Homolog, Mitochondrial AAA ATPase Chaperonin	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia
DNAJC19	DnaJ Heat Shock Protein Family (Hsp40) Member C19	3-methylglutaconic aciduria, type V
HMGCL	3-Hydroxymethyl-3-Methylglutaryl-CoA Lyase	HMG-CoA lyase deficiency
OPA3	Optic Atrophy 3 protein	Optic atrophy 3 with cataract; 3-methylglutaconic aciduria, type III
POLG	Polymerase (DNA) Gamma, Catalytic Subunit	Mitochondrial DNA depletion syndrome (Alpers type or MNGIE type); Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE); Progressive external ophthalmoplegia
RANBP2	RAN Binding Protein 2	Encephalopathy, acute, infection-induced, 3, susceptibility to
RYR1	Ryanodine Receptor 1	Central core disease; King-Denborough syndrome; Minicore myopathy with external ophthalmoplegia; Neuromuscular disease, congenital, with uniform type 1 fiber; Malignant hyperthermia susceptibility 1
SERAC1	Serine Active Site Containing 1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome
SUCLA2	Succinate-CoA Ligase ADP-Forming Beta Subunit	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)

☐ TAZ	Tafazzin	Barth syndrome; 3-methylglutaconic aciduria type II
☐ TMEM70	Transmembrane Protein 7	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2

☐ DEFICIT DI CARBOSSILASI CON C5OH ELEVATA ALLO SCREENING NEONATALE (4 GENI)

Gene	Proteina	Malattia
☐ BTD	Biotinidase	Biotinidase deficiency
☐ HLCS	Holocarboxylase Synthetase	Holocarboxylase synthetase deficiency
☐ MCCC1	Methylcrotonoyl-CoA Carboxylase 1	3-Methylcrotonyl-CoA carboxylase 1 deficiency
☐ MCCC2	Methylcrotonoyl-CoA Carboxylase 2	3-Methylcrotonyl-CoA carboxylase2 deficiency

☐ ACIDEMIA PROPIONICA (2 GENI)

Gene	Proteina	Malattia
☐ PCCA	Propionyl-CoA Carboxylase Alpha Subunit	Propionicacidemia
☐ PCCB	Propionyl-CoA Carboxylase Beta Subunit	Propionicacidemia

☐ ACIDEMIA METILMALONICA, METABOLISMO COBALAMINA, IPEROMOCISTEINEMIA, METABOLISMO FOLATI E DISORDINI CORRELATI (41 GENI)

Gene	Proteina	Malattia
☐ ABCD4	ATP Binding Cassette Subfamily D Member 4	Methylmalonic aciduria and homocystinuria, **cbl J** type
☐ ACSF3	Acyl-CoA Synthetase Family Member 3	Combined malonic and methylmalonic aciduria
☐ ADK	Adenosine Kinase	Hypermethioninemia due to adenosine kinase deficiency
☐ AHCY	Adenosylhomocysteinase	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
☐ AMN	Amnion Associated Transmembrane Protein	Megaloblastic anemia-1, Norwegian type
☐ BHMT	Betaine--Homocysteine S-Methyltransferase	-
☐ CBS	Cystathionine-Beta-Synthase	Homocystinuria, B6-responsive and nonresponsive types; Thrombosis, hyperhomocysteinemic
☐ CD320	Transcobalamin Receptor	Methylmalonic aciduria due to transcobalamin receptor defect
☐ CTH	Cystathionine Gamma-Lyase	Cystathioninuria; Homocysteine, total plasma, elevated
☐ CUBN	Cubilin	Megaloblastic anemia-1, Finnish type
☐ DHFR	Dihydrofolate Reductase	Megaloblastic anemia due to dihydrofolate reductase deficiency
☐ FOLR1	Folate Receptor 1	Folate transport deficiency
☐ GIF	Gastric Intrinsic Factor (Vitamin B Synthesis)	Intrinsic factor deficiency
☐ GNMT	Glycine N-Methyltransferase	Glycine N-methyltransferase deficiency
☐ HCFC1	Host Cell Factor C1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, **cbl X** type)
☐ LMBRD1	LMBR1 Domain Containing 1	Methylmalonic aciduria and homocystinuria, **cbl F** type
☐ LRP2	LDL Receptor Related Protein 2	Donnai-Barrow syndrome (deficiency of folate transport into the brain)
☐ MAT1A	Methionine Adenosyltransferase 1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency;

		Methionine adenosyltransferase deficiency, autosomal recessive
☐ MAT2A	Methionine Adenosyltransferase 2A	-
☐ MAT2B	Methionine Adenosyltransferase 2B	-
☐ MCEE	Methylmalonyl-CoA Epimerase	Methylmalonyl-CoA epimerase deficiency
☐ MLYCD	Malonyl-CoA Decarboxylase	Malonyl-CoA decarboxylase deficiency
☐ MMAA	Methylmalonic Aciduria (Cobalamin Deficiency) CblA Type	Methylmalonic aciduria, vitamin B12-responsive; **cbl A** type
☐ MMAB	Methylmalonic Aciduria (Cobalamin Deficiency) CblB Type	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, **cbl B** complementation type
☐ MMACHC	Methylmalonic Aciduria (Cobalamin Deficiency) CblC Type, With Homocystinuria	Methylmalonic aciduria and homocystinuria, **cbl C** type
☐ MMADHC	Methylmalonic Aciduria And Homocystinuria, CblD Type	Methylmalonic aciduria and homocystinuria, **cbl D** type
☐ MTHFD1	Methenyltetrahydrofolate Cyclohydrolase, Formyltetrahydrofolate Synthetase	Susceptibility to abruptio placentae; Susceptibility to spina bifida folate-sensitive
☐ MTHFR	Methylenetetrahydrofolate Reductase	Homocystinuria due to MTHFR deficiency
☐ MTHFS	5,10-Methenyltetrahydrofolate Synthetase	Seizures, speech delay, mental retardation, recurrent infections
☐ MTR	5-Methyltetrahydrofolate-Homocysteine Methyltransferase	Homocystinuria-megaloblastic anemia, **cbl G** complementation type; Susceptibility to neural tube defects folate-sensitive
☐ MTRR	5-Methyltetrahydrofolate-Homocysteine Methyltransferase Reductase	Homocystinuria-megaloblastic anemia, **cbl E** type; Susceptibility to neural tube defects folate-sensitive
☐ MUT	Methylmalonyl-CoA Mutase	Methylmalonic aciduria, mut(0) type
☐ SHMT1	Serine Hydroxymethyltransferase 1	Pyridoxine Deficiency
☐ SHMT2	Serine Hydroxymethyltransferase 2	Pyridoxine Deficiency
☐ SLC46A1	Solute Carrier Family 46 Member 1	Folate malabsorption, hereditary
☐ SUCLA2	Succinate-CoA Ligase ADP-Forming Beta Subunit	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
☐ SUCLG1	Succinate-CoA Ligase Alpha Subunit	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)
☐ TCN1	Transcobalamin 1	-
☐ TCN2	Transcobalamin 2	Transcobalamin II deficiency
☐ TPMT	Thiopurine S-Methyltransferase	Thiopurines, poor metabolism of, 1
☐ TYMS	Thymidylate Synthetase	-

☐ ACIDURIA 2 METILBURRICA E ISOBUTIRRICA (2 GENI)

Gene	Proteina	Malattia
☐ ACADSB	Acyl-CoA Dehydrogenase, Short/Branched Chain	2-methylbutyrylglycinuria
☐ ACAD8	Acyl-CoA Dehydrogenase Family Member 8	Isobutyryl-CoA dehydrogenase deficiency

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ACIDURIA GLUTARICA (5 GENI)

Gene	Proteina	Malattia
GCDH	Glutaryl-CoA Dehydrogenase	Glutaricaciduria, type I
ETFDH	Electron Transfer Flavoprotein Dehydrogenase	Glutaricaciduria, type IIC
ETFA	Electron Transfer Flavoprotein Alpha Subunit	Glutaricaciduria, type IIA
ETFB	Electron Transfer Flavoprotein Beta Subunit	Glutaricaciduria, type IIB
SUGCT	Succinyl-CoA:Glutarate-CoA Transferase	Glutaricaciduria, type III

ACIDURIA PIROGLUTAMMICA (2 GENI)

Gene	Proteina	Malattia
GSS	Glutathione Synthetase	Glutathione synthetase deficiency; Hemolytic anemia due to glutathione synthetase deficiency
OPLAH	5-Oxoprolinase (ATP-Hydrolysing)	5-oxoprolinase deficiency

ACIDURIA GLICERICA (2 GENI)

Gene	Proteina	Malattia
GLYCTK	Glycerate Kinase	D-glyceric aciduria
GRHPR	Glyoxylate Reductase/Hydroxypyruvate Reductase	Hyperoxaluria, primary, type II

ACIDURIA 2-IDROSSIGLUTARICA (4 GENI)

Gene	Proteina	Malattia
D2HGDH	D-2-Hydroxyglutarate Dehydrogenase	D-2-hydroxyglutaric aciduria
IDH2	Isocitrate Dehydrogenase 2, Cytosolic	D-2-hydroxyglutaric aciduria 2
L2HGDH	L-2-Hydroxyglutarate Dehydrogenase	L-2-hydroxyglutaric aciduria
SLC25A1	Solute Carrier Family 25 Member 1	Combined D-2- and L-2-hydroxyglutaric aciduria

ACIDURIA OXOGLUTARICA E DEFICIT DI ALFA-CHETOGLUTARATO DEIDROGENASI (3 GENI)

Gene	Proteina	Malattia
DLST	Dihydrolipoamide S-Succinyltransferase	Alpha-Ketoglutarate Dehydrogenase Deficiency
OGDH	Oxoglutarate Dehydrogenase	Alpha-ketoglutarate dehydrogenase deficiency
SLC25A19	Solute Carrier Family 25 Member 19	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)

TIROSINEMIA (3 GENI)

Gene	Proteina	Malattia
FAH	Fumarylacetoacetate Hydrolase (Fumarylacetoacetase)	Tyrosinemia, type I
HPD	4-Hydroxyphenylpyruvate Dioxygenase	Tyrosinemia, type III; Hawkinsinuria
TAT	Tyrosine Aminotransferase	Tyrosinemia, type II

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☐ DIFETTI DEL CICLO DI KREBS (12 GENI)

Gene	Proteina	Malattia
☐ DLST	Dihydrolipoamide S-Succinyltransferase	Alpha-Ketoglutarate Dehydrogenase Deficiency
☐ FH	Fumarate Hydratase	Fumarase deficiency
☐ IDH2	Isocitrate Dehydrogenase 2, Cytosolic	D-2-hydroxyglutaric aciduria 2
☐ IDH3B	Isocitrate Dehydrogenase 3 Beta	Retinitis pigmentosa 46
☐ ISCU	Iron-Sulfur Cluster Assembly Enzyme	Myopathy with lactic acidosis, hereditary
☐ OGDH	Oxoglutarate Dehydrogenase	Alpha-ketoglutarate dehydrogenase deficiency
☐ SDHA	Succinate Dehydrogenase Complex Flavoprotein Subunit A	Cardiomyopathy, dilated, 1GG; Leigh syndrome; Mitochondrial respiratory chain complex II deficiency;
☐ SDHAF1	Succinate Dehydrogenase Complex Assembly Factor 1	Mitochondrial complex II deficiency
☐ SDHD	Succinate Dehydrogenase Complex Subunit D	Mitochondrial complex II deficiency;
☐ SLC25A19	Solute Carrier Family 25 Member 19	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)
☐ SUCLA2	Succinate-CoA Ligase ADP-Forming Beta Subunit	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
☐ SUCLG1	SUCLG1	Succinate-CoA Ligase Alpha Subunit

☐ IPERPROLINEMIA e IPOPROLINEMIA (5 GENI)

Gene	Proteina	Malattia
☐ ALDH4A1	Aldehyde Dehydrogenase 4 Family Member A1	Hyperprolinemia, type II
☐ PRODH	Proline Dehydrogenase 1	Hyperprolinemia, type I; Schizophrenia, susceptibility to, 4
☐ ALDH18A1	Pyrroline-5-Carboxylate Synthetase	Hypoprolinemia type I; Cutis laxa, autosomal recessive, type IIIA; Spastic paraplegia 9A, autosomal dominant; Spastic paraplegia 9B, autosomal recessiv
☐ PYCR1	Pyrroline-5-Carboxylate Reductase 1	Hypoprolinemia type II; Cutis laxa, autosomal recessive, type IIB; Cutis laxa, autosomal recessive, type IIIB
☐ PEPD	Peptidase D	Prolidase deficiency

☐ IPERGLICINURIA (3 GENI)

Gene	Proteina	Malattia
☐ SLC6A19	Solute Carrier Family 6 Member 19	Hartnup disorder; Hyperglycinuria; Iminoglycinuria, digenic
☐ SLC6A20	X TRANSPORTER PROTEIN 3;	Hyperglycinuria
☐ SLC36A2	PROTON/AMINO ACID TRANSPORTER 2	Hyperglycinuria

☐ CISTINURIA (2 GENI)

Gene	Proteina	Malattia
☐ SLC3A1	Solute Carrier Family 3 Member 1	Cystinuria
☐ SLC7A9	Solute Carrier Family 7 Member 9	Cystinuria

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IPERFENILALANINEMIA (12 GENI)

Gene	Proteina	Malattia
DBH	Dopamine Beta-Hydroxylase	Dopamine beta-hydroxylase deficiency; Dopamine-beta-hydroxylase activity levels, plasma
DDC	Aromatic L-Amino Acid Decarboxylase	Aromatic L-amino acid decarboxylase deficiency
GCH1	GTP Cyclohydrolase 1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia; Hyperphenylalaninemia, BH4-deficient, B
MAOA	Monoamine Oxidase A	Brunner syndrome; Antisocial behavior
PAH	Phenylalanine Hydroxylase	Phenylketonuria; Hyperphenylalaninemia, non-PKU mild
PCBD1	Pterin-4 Alpha-Carbinolamine Dehydratase 1	Hyperphenylalaninemia, BH4-deficient, D
PTS	6-Pyruvoyltetrahydropterin Synthase	Hyperphenylalaninemia, BH4-deficient, A
QDPR	Quinoid Dihydropteridine Reductase	Hyperphenylalaninemia, BH4-deficient, C
SLC18A2	Solute Carrier Family 18 Member A2	Brain Dopamine-Serotonin Vesicular Transport Disease
SLC6A3	Solute Carrier Family 6 Member 3; Dopamine trasporter (DAT)	Parkinsonism-dystonia, infantile; Nicotine dependence, protection against
SPR	Sepiapterin Reductase (7,8-Dihydrobiopterin:NADP+ Oxidoreductase)	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency
TH	Tyrosine Hydroxylase	Segawa syndrome, recessive

IPERGLICINEMIA NON CHETOTICA (7 GENI)

Gene	Proteina	Malattia
AMT	Aminomethyltransferase	Glycine encephalopathy
BOLA3	BolA Family Member 3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia
GCSH	Glycine Cleavage System Protein H	Glycine encephalopathy
GLDC	Glycine Decarboxylase	Glycine encephalopathy
GLRX5	Glutaredoxin 5	Spasticity, childhood-onset, with hyperglycinemia
LIAS	Lipoic Acid Synthetase	Hyperglycinemia, lactic acidosis, and seizures
SLC25A22	Solute Carrier Family 25 Member 22	Epileptic encephalopathy, early infantile, 3

ACIDURIA 3-IDROSSIISOBUTIRRICA (3 GENI)

Gene	Proteina	Malattia
ALDH6A1	Aldehyde Dehydrogenase 6 Family Member A1	Methylmalonate semialdehyde dehydrogenase deficiency
HIBADH	3-Hydroxyisobutyrate Dehydrogenase	3-Hydroxyisobutyric Aciduria
HIBCH	3-Hydroxyisobutyryl-CoA Hydrolase	3-hydroxyisobutryl-CoA hydrolase deficiency

SULFOCISTINURIA (4 GENI)

Gene	Proteina	Malattia
GPHN	Gephyrin	Molybdenum cofactor deficiency C
MOCS1	Molybdenum Cofactor Synthesis 1	Molybdenum cofactor deficiency A
MOCS2	Molybdenum Cofactor Synthesis 2	Molybdenum cofactor deficiency B
SUOX	Sulfite Oxidase	Sulfite oxidase deficiency

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☐ PURINE E PIRIMIDINE (27 GENI)

Gene	Proteina	Malattia
☐ ADA	Adenosine Deaminase	Adenosine deaminase deficiency, partial; Severe combined immunodeficiency due to ADA deficiency
☐ ADSL	Adenylosuccinate Lyase	Adenylosuccinase deficiency
☐ AMPD1	Adenosine Monophosphate Deaminase 1	Myopathy due to myoadenylate deaminase deficiency
☐ APRT	Adenine Phosphoribosyltransferase	Adenine phosphoribosyltransferase deficiency
☐ ATIC	5-Aminoimidazole-4-Carboxamide Ribonucleotide Formyltransferase/IMP Cyclohydrolase	AICA-ribosiduria due to ATIC deficiency
☐ CAD	Carbamoyl-Phosphate Synthetase 2, Aspartate Transcarbamylase, And Dihydroorotase	Congenital disorder of glycosylation, type Iz
☐ DGUOK	Deoxyguanosine Kinase	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)
☐ DHODH	Dihydroorotate Dehydrogenase	Miller syndrome
☐ DPYD	Dihydropyrimidine Dehydrogenase	Dihydropyrimidine dehydrogenase deficiency
☐ DPYS	Dihydropyrimidinase	Dihydropyrimidinuria
☐ GPHN	Gephyrin	Molybdenum cofactor deficiency C
☐ HPRT1	Hypoxanthine Phosphoribosyltransferase 1	Lesch-Nyhan syndrome
☐ IMPDH1	MP (Inosine 5'-Monophosphate) Dehydrogenase 1	Leber congenital amaurosis 11; Retinitis pigmentosa 10
☐ ITPA	Inosine Triphosphatase	Epileptic encephalopathy, early infantile, 35; Inosine triphosphatase deficiency
☐ MOCS1	Molybdenum Cofactor Synthesis 1	Molybdenum cofactor deficiency A
☐ MOCS2	Molybdenum Cofactor Synthesis 2	Molybdenum cofactor deficiency B
☐ NT5C3A	Pyrimidine 5-Nucleotidase 1	Anemia, hemolytic, due to UMPH1 deficiency
☐ PNP	Purine Nucleoside Phosphorylase	Immunodeficiency due to purine nucleoside phosphorylase deficiency
☐ PRPS1	Phosphoribosyl Pyrophosphate Synthetase 1	Charcot-Marie-Tooth disease, X-linked; Arts syndrome; Deafness, X-linked 1; Gout, PRPS-related; Phosphoribosylpyrophosphate synthetase superactivity
☐ RRM2B	Ribonucleotide Reductase Regulatory TP53 Inducible Subunit M2B	Mitochondrial DNA depletion syndrome 8A and 8B; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5
☐ SUOX	Sulfite Oxidase	Sulfite oxidase deficiency
☐ TK2	Thymidine Kinase 2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3; Mitochondrial DNA depletion syndrome 2 (myopathic type)
☐ TPMT	Thiopurine S-Methyltransferase	Thiopurines, poor metabolism of, 1
☐ TYMP	Thymidine Phosphorylase	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
☐ UMPS	Uridine Monophosphate Synthetase	Orotic aciduria
☐ UPB1	Ureidopropionase, Beta	Beta-ureidopropionase deficiency
☐ XDH	Xanthine Dehydrogenase	Xanthinuria, type I

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☐ **ACIDEMIA ISOVALERICA (1 GENE)**

IVD

☐ **ACIDURIA 3 IDROSSI 3 METILGLUTARICA (1 GENE)**

HMGCL

☐ **ACIDURIA MEVALONICA (1 GENE)**

MVK

☐ **ACIDURIA 2-METIL-3-IDROSSIBUTIRRICA (1 GENE)**

HSD17B10

☐ **ACIDURIA ISOBUTIRRICA (1 GENE)**

ACAD8

☐ **ACIDURIA 4-IDROSSIBUTIRRICA o DEFICIT DI SUCCINICO SEMIALDEIDE DEIDROGENASI (1 GENE)**

ALDH5A1

☐ **MALONICO ACIDURIA (1 GENE)**

☐ MLYCD

☐ **MALATTIA DI CANAVAN o DEFICIT DI AMINOACILASI 2 (1 GENE)**

ASPA

☐ **ALCAPTONURIA (1 GENE)**

HGD

☐ **ACIDURIA FORMIMINOGLUTAMMICA (1 GENE)**

FTCD

☐ **DEFICIT DI AMINOACILASI 1 (1 GENE)**

ACY1

☐ **IPERGLICEROLEMIA (1 GENE)**

GK

☐ **XANTOMATOSI CEREBROTENDINEA (1 GENE)**

CYP27A1

☐ **IPOFOSFATASIA (1 GENE)**

ALPL

☐ **DEFICIT DI LIPINA (1 GENE)**

LPIN1

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☐ ACIDURIA ALFA-METILACETOACETICA o DEFICIT DI BETA-CHETOTIOLASI (1 GENE)

ACAT1

☐ IPERECPLESSIA TIPO 3 (1 GENE)

Gene	Proteina	Malattia
SLC6A5	Solute Carrier Family 6 Member 5	Hyperekplexia 3

☐ SINDROME DA IPOTIROIDISMO-RITARDO-DISMORFISMO (1 GENE)

Gene	Proteina	Malattia
TBCE	Tubulin Folding Cofactor E	Hypoparathyroidism-retardation-dysmorphism syndrome; Kenny-Caffey syndrome, type 1