Referto Analisi : PrenatalScreen® - Analisi prenatale 1020 malattie genetiche mediante sequenziamento NGS

Data Referto:02/10/2015 ***Ora:***17:15

Anagrafica Laboratorio / Medico

Centro Inviante:

Città:

Anagrafica Paziente

Cognome: ***Nome:***

Data di Nascita: ***Luogo di Nascita:***

Origine Etnica: N.A. ***Sesso:***

Medico inviante: ***Vs. Codice di riferimento:***

Indicazione:

Storia Clinica:

Dati Campione

Tipo Campione: Liquido Amniotico ***Ns. Codice campione:*** B50299

Data Accettazione: 15/09/2015 ***Ora Accettazione:*** 19:05 ***Data prelievo:***

Dati Analisi

Analisi effettuata/e: PrenatalScreen® - Analisi prenatale 1020 malattie genetiche mediante sequenziamento NGS

Codice OMIM: ***Ereditarietà:***

Gene investigato: ***OMIM:*** ***Sequenza riferimento:***

Metodo di analisi: Next Generation Sequencing (NGS)

Strategia diagnostica:

Data inizio analisi: 16/09/2015 ***Data fine analisi:*** 02/10/2015

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Risultati e Conclusioni

Risultato:

- gene BTD (Biotinidase deficiency):
Presenza della mutazione G45R (c.133 G>A) in eterozigosi.[rs34885143]

- gene RAPSN (Congenital myasthenic syndrome):
Presenza della mutazione N88K (c.264 C>A) in eterozigosi.[rs104894299]

- gene CFTR (Cystic fibrosis):
Presenza della variante aminoacidica R75Q (c.224 G>A) in eterozigosi.[rs1800076]

Interpretazione:

Il campione in esame presenta le mutazioni:
G45R (c.133 G>A) in ETEROZIGOSI a livello del gene BTD.
Ref: Norrgard (1999) Pediatr Res 46, 20

N88K (c.264 C>A) in ETEROZIGOSI a livello del gene RAPSN .
Ref: Ohno (2002) Am J Hum Genet 70, 875

Il campione in esame presenta inoltre la variante aminoacidica R75Q (c.224 G>A) in eterozigosi a livello del gene CFTR.
Ref: Jarvi (1998) Fertil Steril 70, 724

Note tecniche: Relazione tecnica in allegato

Commenti:

Suggerimenti: L'esame effettuato ha prodotto un risultato per il quale è consigliabile un colloquio di approfondimento con uno specialista in genetica medica. Qualora fosse di suo interesse ricevere una consulenza genetica, il ns. Centro offre gratuitamente la possibilità di un colloquio con la genetista del Centro.

Risultati verificati da: Giuliano Cottone ***Data verifica :*** 02/10/2015

Risultati validati da: Francesco Fiorentino ***Data validazione :*** 02/10/2015

Il presente referto costituisce copia conforme all'originale, il quale è depositato negli archivi del laboratorio Genoma Group Srl.

Il Genetista
Dr.ssa Marina Baldi

Il Direttore del laboratorio
Dr. Francesco Fiorentino

Genoma Group Srl

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Roma, 02 ottobre 2015

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GENOMA®
Molecular Genetics Laboratories Group

Direttore: dott. Francesco Fiorentino

Laboratorio Certificato
UNI EN ISO 9001:2008

Relazione tecnica

PrenatalScreen®
Analisi prenatale di 1.024 malattie genetiche

CHIAMATA GRATUITA
NUMEROVERDE
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PROSPETTO DELL'ANALISI

Paziente	
Tipo Campione	Liquido Amniotico
Codice Campione	B50299
Metodo	Next Generation Sequencing (NGS)
Analisi	PrenatalScreen® - Analisi prenatale 1020 malattie genetiche mediante sequenziamento NGS

Conclusioni
- gene BTD (Biotinidase deficiency):
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Genomica di nuova generazione e diagnosi prenatale

Negli ultimi anni, gli straordinari progressi conseguiti nel settore della genomica e delle biotecnologie hanno posto le basi per leggere e comprendere le informazioni contenute nel genoma fetale. In particolare **le nuove tecnologie di sequenziamento, *Next Generation Sequencing* (NGS)**, ci permettono oggi di accedere alla sequenza del DNA in modo più facile ed efficace, fornendo una valutazione approfondita dell'informazione genetica del feto.

Il test PrenatalScreen®

PrenatalScreen® è un test diagnostico, sviluppato da GENOMA Group, che permette di eseguire nel feto un'analisi multipla di **oltre 1.000 malattie genetiche**, tra cui quelle più frequenti nella popolazione italiana, come la Fibrosi Cistica, l'Anemia Falciforme, la Talassemia, la Sordità Ereditaria.

PrenatalScreen® consente alla gestante di conoscere, attraverso l'analisi del DNA fetale, se il bambino è affetto da gravi malattie genetiche.

Finalità dell'esame

PrenatalScreen® è viene eseguito su DNA estratto da cellule fetali presenti nel liquido amniotico, e prelevate mediante amniocentesi, oppure nei villi coriali, prelevate mediante villocentesi. La sua finalità è lo studio nel feto di malattie genetiche gravi, tra cui quelle più frequenti nella popolazione italiana. I geni elencati in Tabella 1, sono stati selezionati in base all'incidenza nella popolazione delle malattie causate da mutazioni in tali geni, alla gravità del fenotipo clinico alla nascita ed all'importanza del quadro patogenetico associato, seguendo le indicazioni dell'American College of Medical Genetics (ACMG)(Grody et al., Genet Med 2013:15:482–483).

Indicazioni al test PrenatalScreen®

PrenatalScreen® è indicato nei seguenti casi:

•Anamnesi personale/familiare di malattie genetiche ereditarie;

•Per le gestanti che desiderano ridurre il rischio di una malattia genetica nel feto;

•Per gravidanze ottenute sia tramite concepimento naturale che mediante l'accesso a tecniche di procreazione medicalmente assistita (PMA);

•Per le coppie che hanno fatto ricorso a tecniche di fecondazione eterologa.

Come viene effettuato il test PrenatalScreen®?

Il test **PrenatalScreen®** su liquido amniotico viene effettuato mediante il prelievo di 15-20 ml di liquido amniotico per via trans-addominale, sotto controllo ecografico, tra la 15° e la 18° settimana di gestazione. Il liquido prelevato viene centrifugato per separare la parte liquida dalla frazione corpuscolata, costituita dalle cellule fetali che sono in sospensione nel liquido amniotico. Tali cellule, definite amniociti, sono sottoposte ad estrazione del DNA.

Il test **PrenatalScreen®** su Villi Coriali viene effettuato mediante il prelievo di 20 mg circa di villi coriali per via trans-addominale sotto controllo ecografico, tra la 11° e la 13° settimana di gestazione. Il materiale prelevato viene prima lavato ed osservato al microscopio per separare il tessuto materno dal tessuto fetale, e successivamente sottoposto ad estrazione del DNA.

Il DNA isolato dalle cellule fetali viene quindi **amplificato mediante tecnica PCR.**. Successivamente, attraverso un processo tecnologico avanzato di **sequenziamento massivo parallelo** (**MPS**), che impiega tecniche di ***Next Generation Sequencing*** (**NGS**) utilizzando sequenziatori **ILLUMINA**, si sequenziano completamente **744 geni** (esoni e regioni introniche adiacenti, ± 5 nucleotidi)(Tabella 1) ad elevata profondità di lettura. Le sequenze geniche ottenute vengono analizzate attraverso un'**avanzata analisi bioinformatica**, per determinare la

presenza di eventuali mutazioni nei geni in esame.

Risultati ottenibili con il test PrenatalScreen®

"POSITIVO" – Presenza di una o più mutazioni: indica che il test ha rilevato una o più mutazioni a livello di uno (o più) geni. Il nostro genetista, in sede di consulenza genetica, spiegherà in maniera dettagliata il significato del risultato del test ed, eventualmente , prospetterà la necessità di estendere l'esame ai genitori, al fine di verificare la trasmissione ereditaria della variante riscontrata.

Le mutazioni riscontrabili tramite il test **PrenatalScreen®** possono rientrare nelle seguenti categorie prognostiche:

- **con significato patologico noto**;
- **con significato benigno** in quanto sono riscontrabili in individui normali e sono prive di significato patologico;
- **con significato incerto** in quanto non ancora note o caratterizzate dalla comunità medico-scientifica.

"NEGATIVO" - Assenza di mutazioni: indica che il test non ha rilevato la presenza di mutazioni nei geni esaminati. Tale risultato riduce notevolmente le possibilità che il feto abbia le malattie genetiche esaminate, ma non può garantire che il feto sia sano.

Parametri utilizzati per la refertazione delle varianti genetiche

L'analisi è mirata esclusivamente ai geni elencati in Tabella 1. Verranno refertate solo le mutazioni classificate come a significato patogenetiche noto, sulla base dei dati della letteratura scientifica e la classificazione presente nel database di riferimento Human Gene Mutation Database (HGMD), aggiornato alla data del prelievo. Inoltre, seguendo le indicazioni dell'American College of Medical Genetics (ACMG), sono state considerate come patogenetiche o presunte patogenetiche solo le mutazioni con un valore di Minor Allele Frequency (MAF) <5% (1000 Genomes Project), riferibile come la frequenza di ricorrenza dell'allele meno comune all'interno della popolazione.

Target Coverage

Si intende per *Target Coverage*, il numero medio di letture (*reads*) ottenute dal sequenziamento per ciascuna base nucleotidica costituente il gene. Le varianti con una profondità di lettura (numero di reads) inferiore a 30X non sono vengono evidenziate dall'algoritmo di analisi bioinformatica.

Accuratezza del test PrenatalScreen®

Le tecniche attuali di sequenziamento del DNA producono risultati con un'accuratezza superiore al 99%. Benché questo test sia molto accurato bisogna sempre considerare i limiti dell'esame, di seguito descritti.

Limiti del test PrenatalScreen®

Questo esame valuta solo le malattie genetiche ed i geni elencati in Tabella 1. Il test non evidenzia altre malattie genetiche o geni non specificamente investigati.

L'esame inoltre non è in grado di evidenziare:

•mutazioni localizzate nelle regioni introniche oltre ± 5 nucleotidi dai breakpoints;

•delezioni, inversioni o duplicazioni maggiori di 20 bp;

•mosaicismi della linea germinale (cioè mutazioni presenti solo nei gameti).

Un risultato **"NEGATIVO" - Assenza di mutazioni** per i geni investigati non esclude la possibilità che nel feto siano presenti mutazioni localizzate in una regione del genoma non investigata dall'esame.

E' possibile che alcune zone del proprio DNA non possano essere sequenziate o che abbiano una copertura inferiore ai limiti fissati dagli esperti di GENOMA Group per garantire un'analisi accurata delle varianti. Queste regioni non saranno quindi comprese nell'analisi qualora non superino gli standard qualitativi richiesti.

In alcuni casi, il risultato di un'analisi genomica può rivelare una variante o mutazione del DNA con un significato clinico non certo o non determinabile in base alle attuali conoscenze medico-scientifiche. Inoltre identificare una variante genetica non permette sempre di predire con certezza se e quando una persona svilupperà una certa patologia o la severità dei sintomi correlati. Il valore di alcune delle varianti riscontrate con il test può quindi non essere determinabile in base alle conoscenze mediche attuali.

L'interpretazione delle varianti genetiche si basa sulle più recenti conoscenze disponibili al momento dell'analisi. Tale interpretazione potrebbe cambiare in futuro con l'acquisizione di nuove informazioni scientifiche e mediche sulla struttura del genoma ed influire sulla valutazione stessa delle varianti.

Alcune patologie possono essere causate o regolate da più di una variante nel suo DNA in uno o più geni. Alcune di queste varianti possono non essere ancora state identificate o validate dalla comunità scientifica e quindi non essere riportate come patogenetiche al momento dell'analisi.

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Per un'interpretazione corretta dei risultati è importante avere informazioni accurate circa lo stato di salute ed eventuali patologie nella storia clinica dell'individuo e dei suoi familiari. Ciò permette una migliore interpretazione dei risultati genetici da parte dei nostri genetisti.
Limite intrinseco della metodologia NGS utilizzata è la mancanza di uniformità di coverage per ciascuna regione genica analizzata. La qualità e la quantità del DNA ottenuto da campioni biologici prenatali è una delle possibili cause di tale mancanza di uniformità, potenzialmente responsabile della mancata identificazione di mutazioni geniche. Tale limite si traduce nella possibilità, insita nelle metodiche NGS, che specifiche mutazioni dei geni selezionati potrebbero non essere state rilevate dal test.

Tabella 1: PrenatalScreen® - Elenco dei geni analizzati e della malattie genetiche investigate

	MALATTIA	PhenoMIM	GENE
1	17-alpha-hydroxylase/17,20-lyase deficiency	202110	CYP17A1
2	17-beta-hydroxysteroid dehydrogenase X deficiency	300438	HSD17B10
3	3-beta-hydroxysteroid dehydrogenase, type II, deficiency	201810	HSD3B2
4	3-hydroxy-3-methylglutaric aciduria	246450	HMGCL
5	3-methylglutaconic aciduria type 1	250950	AUH
6	3-methylglutaconic aciduria type 3 (AR optic atrophy-3 or optic atrophy plus syndrome)	258501	OPA3
7	3-methylglutaconic aciduria, type V	610198	DNAJC19
8	46XY sex reversal 3	612965	NR5A1
9	4-hydroxybutyric aciduria	271980	ALDH5A1
10	ABCD syndrome	600501	EDNRB
11	Acampomelic campomelic dysplasia	114290	SOX9
12	Achalasia-addisonianism-alacrimia syndrome	231550	AAAS
13	Achondrogenesis type 1B	600972	SLC26A2

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14	Achondrogenesis, type IA	200600	TRIP11
15	Achondrogenesis, type II or hypochondrogenesis	200610	COL2A1
16	Achondroplasia	100800	FGFR3
17	Acyl-CoA dehydrogenase 9 deficiency	611126	ACAD9
18	Adenylosuccinase deficiency	103050	ADSL
19	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	202010	CYP11B1
20	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	613743	CYP11A1
21	Adrenoleukodystrophy	300100	ABCD1
22	Aicardi-Goutieres syndrome 2	610181	RNASEH2B
23	Aicardi-Goutieres syndrome 3	610329	RNASEH2C
24	Aicardi-Goutieres syndrome 4	610333	RNASEH2A
25	Aicardi-Goutieres syndrome 5	612952	SAMHD1
26	Aicardi-Goutieres syndrome 6	615010	ADAR
27	Alagille syndrome	118450	JAG1
28	Allan-Herndon-Dudley syndrome	300523	SLC16A2
29	Alpha-methylacyl-Coa Racemase deficiency	614307	AMACR
30	Alpha-thalassemia (HBA1)	604131	HBA1
31	Alpha-thalassemia (HBA2)	604131	HBA2
32	Alpha-thalassemia/mental retardation syndrome	301040	ATRX
33	Alport syndrome autosomal recessive (COL4A3)	203780	COL4A3
34	Alport syndrome autosomal recessive (COL4A4)	203780	COL4A4
35	Alport syndrome X-Linked	301050	COL4A5
36	Alström syndrome	203800	ALMS1
37	Aminoacylase 1 deficiency	609924	ACY1
38	Amish infantile epilepsy syndrome	609056	ST3GAL5

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39	Anauxetic dysplasia	607095	RMRP
40	Angelman syndrome	105830	UBE3A
41	Antenatal Bartter syndrome type 1	601678	SLC12A1
42	Antenatal Bartter syndrome type 2	241200	KCNJ1
43	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	201750	POR
44	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis	207410	FGFR2
45	Apert syndrome	101200	FGFR2
46	Aplasia/hypoplasia of limbs and pelvis	276820	WNT7A
47	Apparent mineralocorticoid excess	218030	HSD11B2
48	Argininemia	207800	ARG1
49	Argininosuccinic aciduria	207900	ASL
50	Aromatic L-amino acid decarboxylase deficiency	608643	DDC
51	Arthrogryposis - renal dysfunction - cholestasis	208085	VPS33B
52	Arthrogryposis, renal dysfunction, and cholestasis 2	613404	VIPAR
53	Ataxia - oculomotor apraxia type 1	208920	APTX
54	Ataxia with vitamin E deficiency	277460	TTPA
55	Ataxia-oculomotor apraxia 4	616267	PNKP
56	Ataxia-telangiectasia	208900	ATM
57	Atelosteogenesis type II	256050	SLC26A2
58	Atrial septal defect 2	607941	GATA4
59	Atrial septal defect 4	611363	TBX20
60	Atrial septal defect 6	613087	TLL1
61	Atrial septal defect 7, with or without AV conduction defects	108900	NKX2-5
62	Atrial septal defect 8	614433	CITED2
63	Atrial septal defect 9	614475	GATA6
64	Atrioventricular septal defect 3	600309	GJA1

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65	Atrioventricular septal defect 4	614430	GATA4
66	Atrioventricular septal defect 5	614474	GATA6
67	Atrioventricular septal defect, partial, with heterotaxy syndrome	606217	CRELD1
68	Autosomal dominant Charcot-Marie-Tooth disease type 2K	607831	GDAP1
69	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness	607706	GDAP1
70	Autosomal recessive dopa-responsive dystonia	605407	TH
71	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	608340	GDAP1
72	Autosomal recessive limb-girdle muscular dystrophy type 2I	607155	FKRP
73	Autosomal recessive malignant osteopetrosis 1	259700	TCIRG1
74	Autosomal recessive malignant osteopetrosis 4	611490	CLCN7
75	Autosomal recessive polycystic kidney disease	263200	PKHD1
76	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	270550	SACS
77	Baraitser-Winter syndrome 2	614583	ACTG1
78	Bardet-Biedl syndrome 1	209900	BBS1
79	Bardet-Biedl syndrome 10	615987	BBS10
80	Bardet-Biedl syndrome 11	615988	TRIM32
81	Bardet-Biedl syndrome 13	615990	MKS1
82	Bardet-Biedl syndrome 16	615993	SDCCAG8
83	Bardet-Biedl syndrome 2	615981	BBS2
84	Bardet-Biedl syndrome 3	600151	ARL6
85	Bardet-Biedl syndrome 6	605231	MKKS
86	Bardet-Biedl syndrome 9	615986	BBS9 (PTHB1)
87	Bartter syndrome, type 4a	602522	BSND

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88	Beare-Stevenson cutis gyrata syndrome	123790	FGFR2
89	Becker muscular dystrophy	300376	DMD
90	Beckwith-Wiedemann syndrome	130650	CDKN1C
91	Beckwith-Wiedemann syndrome	130650	NSD1
92	Bent bone dysplasia syndrome	614592	FGFR2
93	Bestrophinopathy, autosomal recessive	611809	BEST1
94	Beta-thalassemia	613985	HBB
95	Bifunctional enzyme deficiency	261515	HSD17B4
96	Biotinidase deficiency	253260	BTD
97	Björnstad syndrome	262000	BCS1L
98	Bloom syndrome	210900	BLM
99	Borjeson-Forssman-Lehmann syndrome	301900	PHF6
100	Brachiootic syndrome 3	608389	SIX1
101	Brachytelephalangic chondrodysplasia punctata	302950	ARSE
102	Brittle cornea syndrome	229200	ZNF469
103	Caffey disease	114000	COL1A1
104	Campomelic dysplasia	114290	SOX9
105	Campomelic dysplasia with autosomal sex reversal	114290	SOX9
106	Canavan disease	271900	ASPA
107	Carbamoylphosphate synthetase deficiency	237300	CPS1
108	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	604377	SCO2
109	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2	615119	COX15
110	Cardiofaciocutaneous syndrome	115150	BRAF
111	Cardiofaciocutaneous syndrome 2	615278	KRAS
112	Cardiofaciocutaneous syndrome 3	615279	MAP2K1

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113	Cardiofaciocutaneous syndrome 4	615280	MAP2K2
114	Carnitine deficiency, systemic primary	212140	SLC22A5
115	Carnitine palmitoyl transferase 1A deficiency	255120	CPT1A
116	Carnitine palmitoyl transferase II deficiency, infantile form	600649	CPT2
117	Carnitine palmitoyl transferase II deficiency, neonatal form	608836	CPT2
118	Carnitine-acylcarnitine translocase deficiency	212138	SLC25A20
119	Carpenter syndrome	201000	RAB23
120	Cartilage-hair hypoplasia	250250	RMRP
121	Cataract - intellectual deficit - hypogonadism_ Martsolf syndrome	212720	RAB3GAP2
122	Cataract 40, X-linked	302200	NHS
123	CATSHL syndrome	610474	FGFR3
124	Cerebellar ataxia - intellectual deficit - dysequilibrium syndrome	224050	VLDLR
125	Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome	609528	SNAP29
126	Cerebrotendinous xanthomatosis	213700	CYP27A1
127	Ceroid lipofuscinosis, neuronal, 1	256730	PPT1
128	Ceroid lipofuscinosis, neuronal, 10	610127	CTSD
129	Ceroid lipofuscinosis, neuronal, 2	204500	TPP1
130	Ceroid lipofuscinosis, neuronal, 7	610951	MFSD8
131	Charcot-Marie-Tooth disease, recessive intermediate C	615376	PLEKHG5
132	Charcot-Marie-Tooth disease, type 4A	214400	GDAP1
133	Charcot-Marie-Tooth disease, type 4B1	601382	MTMR2
134	Charcot-Marie-Tooth disease, type 4B2	604563	SBF2

135	Charcot-Marie-Tooth disease, type 4C	601596	SH3TC2
136	Charcot-Marie-Tooth disease, type 4D	601455	NDRG1
137	Charcot-Marie-Tooth disease, type 4F	614895	PRX
138	Charcot-Marie-Tooth disease, type 4H	609311	FGD4
139	Charcot-Marie-Tooth disease, X-linked recessive, 5	311070	PRPS1
140	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	302800	GJB1 (CX32)
141	CHARGE syndrome	214800	CHD7
142	CHARGE syndrome	214800	SEMA3E
143	Chediak-Higashi syndrome	214500	LYST
144	CHILD syndrome	308050	NSDHL
145	Chondrodysplasia punctata, rhizomelic, type 1	215100	PEX7
146	Chondrodysplasia, Blomstrand type	215045	PTH1R
147	Ciliary dyskinesia, primary, 1	244400	DNAI1
148	Ciliary dyskinesia, primary, 3	608644	DNAH5
149	Citrullinemia type I	215700	ASS1
150	CK syndrome	300831	NSDHL
151	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	201910	CYP21A2
152	Classic galactosemia	230400	GALT
153	Classical homocystinuria	236200	CBS
154	COACH syndrome	216360	CC2D2A
155	COACH syndrome	216360	RPGRIP1L
156	COACH syndrome	216360	TMEM67
157	Cockayne syndrome type A	216400	ERCC8
158	Cockayne syndrome type B	133540	ERCC6

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159	Coenzyme Q10 deficiency, primary, 1	607426	COQ2
160	Coenzyme Q10 deficiency, primary, 2- Deafness - encephaloneuropathy - obesity - valvulopathy	614651	PDSS1
161	Coenzyme Q10 deficiency, primary, 3- Leigh syndrome with nefrotic syndrome	614652	PDSS2
162	Coenzyme Q10 deficiency, primary, 4	612016	ADCK3
163	Coenzyme Q10 deficiency, primary, 5	614654	COQ9
164	Coffin-Lowry syndrome	303600	RPS6KA3
165	COFS syndrome 1	214150	ERCC6
166	Cohen Syndrome type 1	216550	VPS13B
167	Combined immunodeficiency with skin granulomas (gene RAG1)	233650	RAG1
168	Combined immunodeficiency with skin granulomas (gene RAG2)	233650	RAG2
169	Combined oxidative phosphorylation defect type 2	610498	MRPS16
170	Combined oxidative phosphorylation defect type 5	611719	MRPS22
171	Combined oxidative phosphorylation deficiency 4	610678	TUFM
172	Combined oxidative phosphorylation deficiency 6	300816	AIFM1
173	Combined pituitary hormone deficiencies, genetic forms	262600	PROP1
174	Combined pituitary hormone deficiency with spine abnormalities	221750	LHX3
175	Combined SAP deficiency	611721	PSAP
176	Complete androgen insensitivity syndrome	300068	AR

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177	Cone-rod dystrophy 13	608194	RPGRIP1
178	Cone-rod dystrophy 3	604116	ABCA4
179	Cone-rod dystrophy 6	601777	GUCY2D
180	Cone-rod retinal dystrophy-2	120970	CRX
181	Congenital bile acid synthesis defect type 4	214950	AMACR
182	Congenital disorder of glycosylation type Ia	212065	PMM2
183	Congenital disorder of glycosylation type Ib	602579	MPI
184	Congenital disorder of glycosylation type Ic	603147	ALG6
185	Congenital disorder of glycosylation type Ie	608799	DPM1
186	Congenital disorder of glycosylation type IIa	212066	MGAT2
187	Congenital disorder of glycosylation type IIc	266265	SLC35C1
188	Congenital disorder of glycosylation type IId	607091	B4GALT1
189	Congenital disorder of glycosylation type IIf	603585	SLC35A1
190	Congenital disorder of glycosylation type Ij	608093	DPAGT1
191	Congenital disorder of glycosylation type Ik	608540	ALG1
192	Congenital disorder of glycosylation, type Id	601110	ALG3
193	Congenital disorder of glycosylation, type If	609180	MPDU1
194	Congenital disorder of glycosylation, type Ig	607143	ALG12
195	Congenital disorder of glycosylation, type Ih	608104	ALG8
196	Congenital disorder of glycosylation, type Ii	607906	ALG2

197	Congenital disorder of glycosylation, type IIb	606056	MOGS
198	Congenital disorder of glycosylation, type IIe	608779	COG7
199	Congenital disorder of glycosylation, type IIg	611209	COG1
200	Congenital disorder of glycosylation, type IIh	611182	COG8
201	Congenital disorder of glycosylation, type IIi	613612	COG5
202	Congenital disorder of glycosylation, type IIj	613489	COG4
203	Congenital disorder of glycosylation, type IIj	613489	TMEM165
204	Congenital disorder of glycosylation, type III	614576	COG6
205	Congenital disorder of glycosylation, type Il	608776	ALG9
206	Congenital disorder of glycosylation, type Im	610768	DOLK (TMEM15)
207	Congenital disorder of glycosylation, type In	612015	RFT1
208	Congenital disorder of glycosylation, type Io	612937	DPM3
209	Congenital disorder of glycosylation, type Ip	613661	ALG11
210	Congenital disorder of glycosylation, type Iq	612379	SRD5A3
211	Congenital disorder of glycosylation, type Is	300884	ALG13
212	Congenital disorder of glycosylation, type It	614921	PGM1
213	Congenital heart defects, nonsyndromic, 1, X-linked	306955	ZIC3
214	Congenital lipoid adrenal hyperplasia	201710	STAR

215	Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells Diarrhea 4, malabsorptive, congenital	610370	NEUROG3
216	Congenital muscular dystrophy type 1A	607855	LAMA2
217	Congenital muscular dystrophy type 5B	606612	FKRP
218	Congenital myopathy with excess of muscle spindles	218040	HRAS
219	Conotruncal anomaly face syndrome	217095	TBX1
220	Conotruncal heart malformations, variable	217095	NKX2-5
221	Cornelia de Lange syndrome 1	122470	NIPBL
222	Cornelia de Lange syndrome 2	300590	SMC1A
223	Cornelia de Lange syndrome 3	610759	SMC3
224	Cornelia de Lange syndrome 4	614701	RAD21
225	Cornelia de Lange syndrome 5	300882	HDAC8
226	Corpus callosum agenesis - neuronopathy	218000	SLC12A6
227	Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity-hydrocephalus syndrome	307000	L1CAM
228	Corpus callosum, agenesis of, with mental retardation	300472	IGBP1
229	Costello syndrome	218040	HRAS
230	Craniofacial-deafness-hand syndrome	122880	PAX3
231	Craniofacial-skeletal-dermatologic dysplasia- Pfeiffer syndrome	101600	FGFR2
232	Craniofrontonasal dysplasia	304110	EFNB1
233	Craniosynostosis, type 1	123100	TWIST1
234	Crouzon syndrome	123500	FGFR2

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235	Crouzon syndrome with acanthosis nigricans	612247	FGFR3
236	Cutis laxa, autosomal recessive, type IIA	219200	ATP6V0A2
237	Cystic fibrosis; mucoviscidosis	219700	CFTR
238	Cystinosis	219800	CTNS
239	Danon disease	300257	LAMP2
240	Deafness, autosomal dominant 11	601317	MYO7A
241	Deafness, autosomal dominant 13	601868	COL11A2
242	Deafness, autosomal dominant 3B	612643	GJB6
243	Deafness, autosomal dominant type 1	124900	DIAPH1
244	Deafness, autosomal dominant type 12	601543	TECTA
245	Deafness, autosomal dominant type 15	602459	POU4F3
246	Deafness, autosomal dominant type 20	604717	ACTG1
247	Deafness, autosomal dominant type 22	606346	MYO6
248	Deafness, autosomal dominant type 23	605192	SIX1
249	Deafness, autosomal dominant type 25	605583	SLC17A8
250	Deafness, autosomal dominant type 28	608641	GRHL2
251	Deafness, autosomal dominant type 2A	600101	KCNQ4
252	Deafness, autosomal dominant type 2B	612644	GJB3
253	Deafness, autosomal dominant type 36	606705	TMC1
254	Deafness, autosomal dominant type 4	600652	MYH14
255	Deafness, autosomal dominant type 40	616357	CRYM

256	Deafness, autosomal dominant type 48	607841	MYO1A
257	Deafness, autosomal dominant type 5	600994	DFNA5
258	Deafness, autosomal dominant type 50	613074	MIR96
259	Deafness, autosomal dominant type 6	600965	WFS1
260	Deafness, autosomal dominant type 64	614152	DIABLO
261	Deafness, autosomal dominant type 9	601369	COCH
262	Deafness, autosomal recessive 12	601386	CDH23
263	Deafness, autosomal recessive 15	601869	GIPC3
264	Deafness, autosomal recessive 18A	602092	USH1C
265	Deafness, autosomal recessive 1A	220290	GJB2
266	Deafness, autosomal recessive 1B	612645	GJB6
267	Deafness, autosomal recessive 2	600060	MYO7A
268	Deafness, autosomal recessive 21	603629	TECTA
269	Deafness, autosomal recessive 22	607039	OTOA
270	Deafness, autosomal recessive 23	609533	PCDH15
271	Deafness, autosomal recessive 29	614035	CLDN14
272	Deafness, autosomal recessive 37	607821	MYO6
273	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	600791	SLC26A4
274	Deafness, autosomal recessive 53	609706	COL11A2
275	Deafness, autosomal recessive 7	600974	TMC1
276	Deafness, autosomal recessive 9	601071	OTOF
277	Dent disease 2	300555	OCRL
278	Desmosterolosis	602398	DHCR24
279	Diarrhea 1, secretory chloride, congenital	214700	SLC26A3
280	Diarrhea 3, secretory sodium, congenital, syndromic	270420	SPINT2

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281	Diarrhea 5, with tufting enteropathy, congenital	613217	EPCAM
282	Diastrophic dwarfism	222600	SLC26A2
283	DiGeorge syndrome	188400	TBX1
284	Dihydropyrimidine dehydrogenase deficiency	274270	DPYD
285	Donnai-Barrow syndrome	222448	LRP2
286	Double-outlet right ventricle	217095	CFC1
287	Double-outlet right ventricle	217095	GDF1
288	Duchenne muscular dystrophy	310200	DMD
289	Dyskeratosis congenita X-linked	305000	DKC1
290	Dystrophic epidermolysis bullosa pruriginosa	604129	COL7A1
291	Ectodermal dysplasia, hypohidrotic, with immune deficiency	300291	IKBKG
292	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency	300301	IKBKG
293	Ehlers-Danlos syndrome type 6 (kyphoscoliotic type)	225400	PLOD1
294	Ehlers-Danlos syndrome, cardiac valvular form	225320	COL1A2
295	Ehlers-Danlos syndrome, classic type	130000	COL1A1
296	Ehlers-Danlos syndrome, classic type (COL5A1)	130000	COL5A1
297	Ehlers-Danlos syndrome, classic type (COL5A2)	130000	COL5A2
298	Ehlers-Danlos syndrome, type III (hypermobile type)	130020	COL3A1
299	Ehlers-Danlos syndrome, type IV (vascular type)	130050	COL3A1
300	Ehlers-Danlos syndrome, type VIIA	130060	COL1A1
301	Ehlers-Danlos syndrome, type VIIB	130060	COL1A2
302	Eiken syndrome	600002	PTH1R

303	Ellis-van Creveld syndrome	225500	EVC2
304	elocardiofacial syndrome	192430	TBX1
305	Encephalopathy, neonatal severe	300673	MECP2
306	Epidermolysis bullosa simplex with muscular dystrophy	226670	PLEC
307	Epidermolysis bullosa simplex with pyloric atresia	612138	PLEC
308	Epilepsy, progressive myoclonic 2A (Lafora)	254780	EPM2A
309	Epilepsy, progressive myoclonic 2B (Lafora)	254780	NHLRC1
310	Epilepsy, pyridoxine-dependent	266100	ALDH7A1
311	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	300491	SYN1
312	Epileptic encephalopathy, early infantile, 1	308350	ARX
313	Epileptic encephalopathy, early infantile, 12	613722	PLCB1
314	Epileptic encephalopathy, early infantile, 15	615006	ST3GAL3
315	Epileptic encephalopathy, early infantile, 2	300672	CDKL5
316	Epileptic encephalopathy, early infantile, 3	609304	SLC25A22
317	Epileptic encephalopathy, early infantile, 8	300607	ARHGEF9
318	Epileptic encephalopathy, early infantile, 9	300088	PCDH19
319	Epiphyseal dysplasia, multiple 1	132400	COMP
320	Epiphyseal dysplasia, multiple, with myopia and deafness	132450	COL2A1
321	Escobar syndrome	265000	CHRNG
322	Ethylmalonic encephalopathy	602473	ETHE1
323	Exudative vitreoretinopathy 2, X-linked	305390	NDP

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324	Fabry disease	301500	GLA
325	Familial dysautonomia	223900	IKBKAP
326	Familial Mediterranean fever	249100	MEFV
327	Fanconi anemia complementation group C	227645	FANCC
328	Fanconi anemia, complementation group A	227650	FANCA
329	Fanconi anemia, complementation group B	300514	FANCB
330	Fanconi anemia, complementation group G	614082	FANCG
331	Fatal infantile lactic acidosis with methylmalonic aciduria	245400	SUCLG1
332	Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3	610505	TSFM
333	Fetal akinesia deformation sequence	208150	DOK7
334	Fetal akinesia deformation sequence	208150	RAPSN
335	Fetal Gaucher disease	608013	GBA
336	FG syndrome 4	300422	CASK
337	Fibrochondrogenesis 1	228520	COL11A1
338	Fibrochondrogenesis 2	614524	COL11A2
339	Fibular hypoplasia or aplasia - femoral bowing - oligodactyly	228930	WNT7A
340	Focal cortical dysplasia, Taylor balloon cell type	607341	TSC1
341	Fraser syndrome (gene FRAS1)	219000	FRAS1
342	Fraser syndrome (gene FREM2)	219000	FREM2
343	Free sialic acid storage disease, infantile form	269920	SLC17A5
344	French-Canadian type Leigh syndrome	220111	LRPPRC
345	Frontometaphyseal dysplasia	305620	FLNA

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346	Fucosidosis	230000	FUCA1
347	Fumaric aciduria	606812	FH
348	Galactokinase deficiency with cataracts	230200	GALK1
349	Galactose epimerase deficiency	230350	GALE
350	Gaucher disease type 2	230900	GBA
351	Gaucher disease type 3	231000	GBA
352	Gaucher disease type 3C	231005	GBA
353	Geleophysic dysplasia 1	231050	ADAMTSL2
354	Generalized junctional epidermolysis bullosa, non-Herlitz type	226650	COL17A1
355	Glutaric acidemia type 2	231680	ETFA
356	Glutaric acidemia type 2	231680	ETFB
357	Glutaric acidemia type 2	231680	ETFDH
358	Glutaryl-CoA dehydrogenase deficiency	231670	GCDH
359	Glutathione synthetase deficiency with 5-oxoprolinuria	266130	GSS
360	Glycine encephalopathy	605899	AMT
361	Glycine encephalopathy	605899	GCSH
362	Glycine encephalopathy	605899	GLDC
363	Glycogen storage disease due to acid maltase deficiency	232300	GAA
364	Glycogen storage disease due to glucose-6-phosphatase deficiency type 1a	232200	G6PC
365	Glycogen storage disease due to glucose-6-phosphatase deficiency type b	232220	SLC37A4
366	Glycogen storage disease due to glucose-6-phosphatase deficiency type c	232240	SLC37A4

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367	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form	232500	GBE1
368	Glycogen storage disease due to glycogen debranching enzyme deficiency	232400	AGL
369	GM1 gangliosidosis type 1	230500	GLB1
370	GM1 gangliosidosis type 2	230600	GLB1
371	GM1 gangliosidosis type 3	230650	GLB1
372	GRACILE syndrome	603358	BCS1L
373	Greenberg dysplasia	215140	LBR
374	Griscelli disease type 1	214450	MYO5A
375	Griscelli disease type 2	607624	RAB27A
376	Guanidinoacetate methyltransferase deficiency	612736	GAMT
377	HARP syndrome	607236	PANK2
378	Hemochromatosis, type 2A	602390	HFE2/HJV
379	Hemochromatosis, type 2B	613313	HAMP
380	Hemoglobin H disease, nondeletional	613978	HBA2
381	Hemophagocytic lymphohistiocytosis, familial, 2	603553	PRF1
382	Hemophagocytic lymphohistiocytosis, familial, 3	608898	UNC13D
383	Hemophagocytic lymphohistiocytosis, familial, 4	603552	STX11
384	Hemophagocytic lymphohistiocytosis, familial, 5	613101	STXBP2
385	Hemophilia A	306700	F8
386	Hemophilia B	306900	F9
387	Hepatic venoocclusive disease with immunodeficiency	235550	SP110
388	Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1	609060	GFM1

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389	Hereditary fructose intolerance	229600	ALDOB
390	Hermansky-Pudlak syndrome 1	203300	HPS1
391	Hermansky-Pudlak syndrome 2	608233	AP3B1
392	Hermansky-pudlak syndrome 9	614171	PLDN
393	Heterotaxy, visceral, 1, X-linked	306955	ZIC3
394	Heterotaxy, visceral, 5	270100	NODAL
395	Heterotopia, periventricular	300049	FLNA
396	Holocarboxylase synthetase deficiency	253270	HLCS
397	Holoprosencephaly-2	157170	SIX3
398	Holoprosencephaly-3	142945	SHH
399	Holoprosencephaly-5	609637	ZIC2
400	Holoprosencephaly-9	610829	GLI2
401	Holt-Oram syndrome	142900	TBX5
402	Homocystinuria due to MTHFR deficienc	236250	MTHFR
403	Hoyeraal-Hreidarsson syndrome	300240	DKC1
404	Hydrops fetalis, nonimmune (HBA1)	236750	HBA1
405	Hydrops fetalis, nonimmune (HBA2)	236750	HBA2
406	Hyperammonemia due to N-acetylglutamate synthetase deficiency	237310	NAGS
407	Hyper-IgE recurrent infection syndrome, autosomal recessive	243700	DOCK8
408	Hyperornithinemia-hyperammonemia-homocitrullinuria	238970	SLC25A15
409	Hyperphenylalaninemia, BH4-deficient, A	261640	PTS
410	Hyperphenylalaninemia, BH4-deficient, C	261630	QDPR
411	Hyperphenylalaninemia, BH4-deficient, D	264070	PCBD1

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412	Hypochondroplasia	146000	FGFR3
413	Hypogonadotropic hypogonadism 5	612370	CHD7
414	Hypomyelination - congenital cataract	610532	FAM126A
415	Hypoparathyroidism - intellectual deficit - dysmorphism syndrome	241410	TBCE
416	Hypophosphatemic rickets, AR	241520	DMP1
417	Hypophosphatemic rickets, autosomal recessive, 2	613312	ENPP1
418	Hypoplastic left heart syndrome 1	241550	GJA1
419	Hypoplastic left heart syndrome 2	614435	NKX2-5
420	Hypothryoidism, congenital, nongoitrous 4	275100	TSHB
421	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	218700	PAX8
422	Hypothyroidism, congenital, nongoitrous, 5	225250	NKX2-5
423	Hypothyroidism, congenital, nongoitrous, 6	614450	THRA
424	Ichthyosis follicularis - alopecia - photophobia	308205	MBTPS2
425	Ichthyosis, autosomal recessive 4B (harlequin)	242500	ABCA12
426	Ichthyosis, congenital, autosomal recessive 1	242300	TGM1
427	Ichthyosis, congenital, autosomal recessive 4A	601277	ABCA12
428	IMAGE syndrome	614732	CDKN1C
429	Immunodeficiency 17, CD3 gamma deficient	615607	CD3G
430	Immunodeficiency 18, SCID variant	615615	CD3E
431	Immunodeficiency 19	615617	CD3D
432	Immunodeficiency 27A, mycobacteriosis, AR	209950	IFNGR1

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433	Immunodeficiency 28, mycobacteriosis	614889	IFNGR2
434	Immunodeficiency 33	300636	IKBKG
435	Immunodeficiency 35	611521	TYK2
436	Immunodeficiency due to purine nucleoside phosphorylase deficiency	613179	PNP
437	Immunodeficiency with hyper IgM, type 5	608106	UNG
438	Immunodeficiency with hyper-IgM, type 2	605258	AICDA
439	Immunodeficiency with hyper-IgM, type 3	606843	CD40
440	Immunodeficiency, common variable, 1	607594	ICOS
441	Immunodeficiency, X-linked	300853	MAGT1
442	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	242860	DNMT3B
443	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	304790	FOXP3
444	Incontinentia pigmenti	308300	IKBKG
445	Infantile bilateral striatal necrosis Striatonigral degeneration, infantile	271930	NUP62
446	Interleukin 1 receptor antagonist deficiency	612852	IL1RN
447	Isolated CoQ-cytochrome C reductase deficiency Mitochondrial complex III deficiency, nuclear type 1	124000	BCS1L
448	Isolated growth hormone deficiency type III	307200	BTK
449	Isovaleric acidemia	243500	IVD
450	Jeune syndrome	611263	IFT80

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451	Johanson-Blizzard syndrome	243800	UBR1
452	Joubert syndrome 1	213300	INPP5E
453	Joubert syndrome 10	300804	OFD1
454	Joubert syndrome 15	614464	CEP41
455	Joubert syndrome 2	608091	TMEM216
456	Joubert syndrome 4	609583	NPHP1
457	Joubert syndrome 5	610188	CEP290
458	Joubert syndrome 6	610688	TMEM67
459	Joubert syndrome 7	611560	RPGRIP1L
460	Joubert syndrome 8	612291	ARL13B
461	Joubert syndrome 9	612285	CC2D2A
462	Joubert syndrome with ocular defect Joubert syndrome-3	608629	AHI1
463	Junctional epidermolysis bullosa - pyloric atresia	226730	ITGA6
464	Junctional epidermolysis bullosa - pyloric atresia	226730	ITGB4
465	Junctional epidermolysis bullosa, Herlitz type (gene LAMA3)	226700	LAMA3
466	Junctional epidermolysis bullosa, Herlitz type (gene LAMB3)	226700	LAMB3
467	Junctional epidermolysis bullosa, Herlitz type (gene LAMC2)	226700	LAMC2
468	Junctional epidermolysis bullosa, non Herlitz type (gene LAMA3)	226650	LAMA3
469	Junctional epidermolysis bullosa, non Herlitz type (gene LAMB3)	226650	LAMB3
470	Junctional epidermolysis bullosa, non Herlitz type (gene LAMC2)	226650	LAMC2
471	Junctional epidermolysis bullosa, non-Herlitz type	226650	ITGB4
472	Juvenile neuronal ceroid lipofuscinosis 3	204200	CLN3
473	Kabuki syndrome 1	147920	MLL2 (KMT2D)
474	Kabuki syndrome 2	300867	KDM6A

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475	Kahrizi syndrome	612713	SRD5A3
476	Kelley-Seegmiller syndrome	300323	HPRT1
477	Ketoacidosis due to beta-ketothiolase deficiency	203750	ACAT1
478	Kniest dysplasia	156550	COL2A1
479	Krabbe disease	245200	GALC
480	Krabbe disease, atypical	611722	PSAP
481	Lacticacidemia due to PDX1 deficiency	245349	PDHX
482	Late infantile neuronal ceroid lipofuscinosis 5	256731	CLN5
483	Late infantile neuronal ceroid lipofuscinosis 6	601780	CLN6
484	Late infantile neuronal ceroid lipofuscinosis 8	600143	CLN8
485	Lathosterolosis	607330	SC5DL
486	Leber congenital amaurosis 1	204000	GUCY2D
487	Leber congenital amaurosis 10	611755	CEP290
488	Leber congenital amaurosis 11	613837	IMPDH1
489	Leber congenital amaurosis 13	612712	RDH12
490	Leber congenital amaurosis 2	204100	RPE65
491	Leber congenital amaurosis 4	604393	AIPL1
492	Leber congenital amaurosis 6	613826	RPGRIP1
493	Leber congenital amaurosis 7	613829	CRX
494	Leber congenital amaurosis 8	613835	CRB1
495	Leber congenital amaurosis 9	608553	NMNAT1
496	Leigh syndrome	256000	BCS1L
497	Leigh syndrome	256000	NDUFA12
498	Leigh syndrome	256000	NDUFA2
499	Leigh syndrome	256000	NDUFA9
500	Leigh syndrome	256000	NDUFAF2
501	Leigh syndrome	256000	NDUFAF6
502	Leigh syndrome	256000	NDUFS4
503	Leigh syndrome	256000	NDUFS7

504	Leigh syndrome due to cytochrome c oxidase deficiency	256000	COX15
505	Leigh syndrome due to mitochondrial complex I deficiency	256000	FOXRED1
506	Leigh syndrome due to mitochondrial complex I deficiency	256000	NDUFS3
507	Leigh syndrome due to mitochondrial complex I deficiency	256000	NDUFS8
508	Leigh syndrome due to mitochondrial COX4 deficiency	256000	COX10
509	Leigh syndrome, due to COX deficiency	256000	SURF1
510	LEOPARD syndrome 1	151100	PTPN11
511	LEOPARD syndrome 2	611554	RAF1
512	LEOPARD syndrome 3	613707	BRAF
513	Lesch-Nyhan syndrome	300322	HPRT1
514	Lethal ataxia with deafness and optic atrophy	301835	PRPS1
515	Lethal congenital contractural syndrome 2	607598	ERBB3
516	Lethal congenital contracture syndrome type 1	253310	GLE1
517	Lethal osteosclerotic bone dysplasia	259775	FAM20C
518	Leukocyte adhesion deficiency, type III	612840	FERMT3
519	Leukodystrophy, hypomyelinating, 11	616494	POLR1C
520	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism	607694	POLR3A
521	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	611105	DARS2

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522	Limb girdle dystrophy with epidermolysis bullosa simplex	613723	PLEC
523	Lissencephaly 1	607432	PAFAH1B1
524	Lissencephaly 2, Norman-Roberts type	257320	RELN
525	Lissencephaly 3	611603	TUBA1A
526	Lissencephaly 5	615191	LAMB1
527	Lissencephaly, X-linked	300067	DCX
528	Lissencephaly, X-linked 2	300215	ARX
529	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	609016	HADH
530	Mandibuloacral dysplasia with type B lipodystrophy	608612	ZMPSTE24
531	Mannosidosis, alpha-, types I and II	248500	MAN2B1
532	Maple syrup urine disease type III Dihydrolipoamide dehydrogenase deficiency	246900	DLD
533	Maple syrup urine disease, type Ia	248600	BCKDHA
534	Maple syrup urine disease, type Ib	248600	BCKDHB
535	Maple syrup urine disease, type II	248600	DBT
536	Marinesco-Sjögren syndrome	248800	SIL1
537	Marshall syndrome	154780	COL11A1
538	Masa syndrome	303350	L1CAM
539	Meckel syndrome 2	603194	TMEM216
540	Meckel syndrome 3	607361	TMEM67
541	Meckel syndrome 4	611134	CEP290
542	Meckel syndrome 6	612284	CC2D2A
543	Meckel syndrome 7	267010	NPHP3
544	Meckel syndrome type 1	249000	MKS1
545	Meckel syndrome, type 5	611561	RPGRIP1L
546	Medium chain acyl-CoA dehydrogenase deficiency	201450	ACADM

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547	Megalencephalic leukoencephalopathy with subcortical cysts	604004	MLC1
548	Melnick-Needles syndrome	309350	FLNA
549	Menkes disease	309400	ATP7A
550	Mental retardation and microcephaly and cerebellar hypoplasia	300749	CASK
551	Mental retardation syndrome, X-linked, Siderius type	300263	PHF8
552	Mental retardation, autosomal dominant type 1	156200	MBD5
553	Mental retardation, autosomal dominant type 12	614562	ARID1B
554	Mental retardation, autosomal dominant type 14	614607	ARID1A
555	Mental retardation, autosomal dominant type 15	614608	SMARCB1
556	Mental retardation, autosomal dominant type 16	614609	SMARCA4
557	Mental retardation, autosomal dominant type 20	613443	MEF2C
558	Mental retardation, autosomal dominant type 5	612621	SYNGAP1
559	Mental retardation, autosomal dominant type 6	613970	GRIN2B
560	Mental retardation, autosomal dominant type 9	614255	KIF1A
561	Mental retardation, autosomal recessive 1	249500	PRSS12
562	Mental retardation, autosomal recessive 12	611090	ST3GAL3
563	Mental retardation, autosomal recessive 13	613192	TRAPPC9
564	Mental retardation, autosomal recessive 15	614202	MAN1B1

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565	Mental retardation, autosomal recessive 18	614249	MED23
566	Mental retardation, autosomal recessive 2	607417	CRBN
567	Mental retardation, autosomal recessive 3	608443	CC2D1A
568	Mental retardation, autosomal recessive 34	614499	CRADD
569	Mental retardation, autosomal recessive 39	615541	TTI2
570	Mental retardation, autosomal recessive 46	616116	NDST1
571	Mental retardation, autosomal recessive 47	616193	FMN2
572	Mental retardation, autosomal recessive 5	611091	NSUN2
573	Mental retardation, autosomal recessive 7	611093	TUSC3
574	Mental retardation, autosomal recessive, 6	611092	GRIK2
575	Mental retardation, truncal obesity, retinal dystrophy, and micropenis	610156	INPP5E
576	Mental retardation, with or without nystagmus-FG syndrome 4	300422	CASK
577	Mental retardation, X-linked 1	309530	IQSEC2
578	Mental retardation, X-linked 19	300844	RPS6KA3
579	Mental retardation, X-linked 21/34	300143	IL1RAPL1
580	Mental retardation, X-linked 29 and others	300419	ARX
581	Mental retardation, X-linked 30/47	300558	PAK3
582	Mental retardation, X-linked 41	300849	GDI1
583	Mental retardation, X-linked 45	300498	ZNF81
584	Mental retardation, X-linked 46	300436	ARHGEF6
585	Mental retardation, X-linked 58	300210	TSPAN7
586	Mental retardation, X-linked 63	300387	ACSL4

587	Mental retardation, X-linked 72	300271	RAB39B
588	Mental retardation, X-linked 9	309549	FTSJ1
589	Mental retardation, X-linked 91	300577	ZDHHC15
590	Mental retardation, X-linked 93	300659	BRWD3
591	Mental retardation, X-linked 94	300699	GRIA3
592	Mental retardation, X-linked 96	300802	SYP
593	Mental retardation, X-linked 97	300803	ZNF711
594	Mental retardation, X-linked 98	300912	KIAA2022
595	Mental retardation, X-linked 99	300919	USP9X
596	Mental retardation, X-linked syndromic 5	304340	AP1S2
597	Mental retardation, X-linked syndromic, Christianson type	300243	SLC9A6
598	Mental retardation, X-linked syndromic, Lubs type	300260	MECP2
599	Mental retardation, X-linked syndromic, Nascimento-type	300860	UBE2A
600	Mental retardation, X-linked syndromic, Raymond type	300799	ZDHHC9
601	Mental retardation, X-linked syndromic, Turner type	300706	HUWE1
602	Mental retardation, X-linked, Snyder-Robinson type	309583	SMS
603	Mental retardation, X-linked, syndromic 13	300055	MECP2
604	Mental retardation, X-linked, syndromic 14	300676	UPF3B
605	Mental retardation, X-linked, syndromic 15 (Cabezas type)	300354	CUL4B
606	Mental retardation, X-linked, syndromic, Claes-Jensen type	300534	KDM5C
607	Mental retardation, X-linked, syndromic, Hedera type	300423	ATP6AP2
608	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	300486	OPHN1

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609	Mental retardation-hypotonic facies syndrome, X-linked	309580	ATRX
610	Metachromatic leukodystrophy	250100	ARSA
611	Metachromatic leukodystrophy due to SAP-b deficiency	249900	PSAP
612	Metaphyseal chondrodysplasia, Murk Jansen type	156400	PTH1R
613	Metaphyseal dysplasia without hypotrichosis	250460	RMRP
614	Methylmalonic acidemia with homocystinuria, type cblC	277400	MMACHC
615	Methylmalonic acidemia with homocystinuria, type cblD	277410	MMADHC
616	Mevalonic aciduria	610377	MVK
617	Microcephaly 5, primary, autosomal recessive	608716	ASPM
618	Microcephaly, postnatal progressive, with seizures and brain atrophy	613668	MED17
619	Microcephaly, seizures, and developmental delay	613402	PNKP
620	Microphthalmia with coloboma 5	611638	SHH
621	Microphthalmia, syndromic 2 (OCULOFACIOCARDIODENTAL SYNDROME)	300166	BCOR
622	Microphthalmia, syndromic 7	309801	HCCS
623	Mitochondrial complex I deficiency	252010	NDUFA1
624	Mitochondrial complex I deficiency	252010	NDUFA11
625	Mitochondrial complex I deficiency	252010	NDUFAF1
626	Mitochondrial complex I deficiency	252010	NDUFAF2
627	Mitochondrial complex I deficiency	252010	NDUFAF3
628	Mitochondrial complex I deficiency	252010	NDUFAF4
629	Mitochondrial complex I deficiency	252010	NDUFAF5
630	Mitochondrial complex I deficiency	252010	NDUFB3
631	Mitochondrial complex I deficiency	252010	NDUFS1

632	Mitochondrial complex I deficiency	252010	NDUFS2
633	Mitochondrial complex I deficiency	252010	NDUFS3
634	Mitochondrial complex I deficiency	252010	NDUFS4
635	Mitochondrial complex I deficiency	252010	NDUFS6
636	Mitochondrial complex I deficiency	252010	NDUFV1
637	Mitochondrial complex II deficiency	252011	SDHAF1
638	Mitochondrial complex IV deficiency	220110	COX6B1
639	Mitochondrial complex IV deficiency	220110	FASTKD2
640	Mitochondrial complex IV deficiency	220110	TACO1
641	Mitochondrial complex IV deficiency	220110	COX10
642	Mitochondrial complex IV deficiency	220110	SCO1
643	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	604273	ATPAF2
644	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	614052	TMEM70
645	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	603041	TYMP
646	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	612073	SUCLA2
647	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	612075	RRM2B
648	Mitochondrial DNA depletion syndrome 8B (MNGIE type)	612075	RRM2B

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649	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency 3	251880	DGUOK
650	Mitochondrial DNA depletion syndrome, myopathic form	609560	TK2
651	Mitochondrial respiratory chain complex III deficiency	124000	UQCRB
652	Mitochondrial respiratory chain complex III deficiency	124000	UQCRQ
653	Mitochondrial trifunctional protein deficiency	609015	HADHA
654	Mitochondrial trifunctional protein deficiency	609015	HADHB
655	Miyoshi muscular dystrophy 1	254130	DYSF
656	Mohr-Tranebjaerg syndrome	304700	TIMM8A
657	Molybdenum cofactor deficiency A	252150	MOCS1
658	Molybdenum cofactor deficiency B	252160	MOCS2
659	Molybdenum cofactor deficiency C	615501	GPHN
660	Mowat-Wilson syndrome	235730	ZEB2
661	Mucolipidosis type 2	252500	GNPTAB
662	Mucolipidosis type 3	252600	GNPTAB
663	Mucolipidosis type 4	252650	MCOLN1
664	Mucopolysaccharidosis Ih	607014	IDUA
665	Mucopolysaccharidosis Ih/s	607015	IDUA
666	Mucopolysaccharidosis Is	607016	IDUA
667	Mucopolysaccharidosis type 2	309900	IDS
668	Mucopolysaccharidosis type 3A (Sanfilippo syndrome type A)	252900	SGSH
669	Mucopolysaccharidosis type 4B (Morquio)	253010	GLB1
670	Mucopolysaccharidosis type 6 (Maroteaux-Lamy)	253200	ARSB
671	Mucopolysaccharidosis type 7	253220	GUSB
672	Mucopolysaccharidosis type IIIB (Sanfilippo B)	252920	NAGLU

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673	Mucopolysaccharidosis type IIIC (Sanfilippo C)	252930	HGSNAT
674	Muenke syndrome	602849	FGFR3
675	MULIBREY nanism	253250	TRIM37
676	Multiple epiphyseal dysplasia type 4	226900	SLC26A2
677	Multiple pterygium syndrome, lethal type	253290	CHRNA1
678	Multiple pterygium syndrome, lethal type	253290	CHRND
679	Multiple pterygium syndrome, lethal type	253290	CHRNG
680	Multiple sulfatase deficiency	272200	SUMF1
681	Muscle-eye-brain disease	613153	FKRP
682	Muscular dystrophy, limb-girdle, type 2A	253600	CAPN3
683	Muscular dystrophy, limb-girdle, type 2B	253601	DYSF
684	Muscular dystrophy, limb-girdle, type 2C	253700	SGCG
685	Muscular dystrophy, limb-girdle, type 2D	608099	SGCA
686	Muscular dystrophy, limb-girdle, type 2E	604286	SGCB
687	Muscular dystrophy, limb-girdle, type 2H	254110	TRIM32
688	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	614643	ISPD
689	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1	613155	POMT1
690	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2	613156	POMT2

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691	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3	613151	POMGNT1
692	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6	608840	LARGE
693	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	609308	POMT1
694	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	613158	POMT2
695	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3	613157	POMGNT1
696	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7	616052	ISPD
697	Myasthenia, limb-girdle, familial	254300	DOK7
698	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	616326	RAPSN
699	Myasthenic syndrome, congenital, 1A, slow-channel	601462	CHRNA1
700	Myasthenic syndrome, congenital, 1B, fast-channel	608930	CHRNA1
701	Myasthenic syndrome, congenital, 2A, slow-channel	606313	CHRNB1
702	Myasthenic syndrome, congenital, 3A, slow-channel	616321	CHRND
703	Myasthenic syndrome, congenital, 3B, fast-channel	616322	CHRND
704	Myasthenic syndrome, congenital, 4A, slow-channel	605809	CHRNE
705	Myasthenic syndrome, congenital, 4B, fast-channel	616324	CHRNE

706	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	608931	CHRNE
707	Myopathy, distal, with anterior tibial onset	606768	DYSF
708	Nance-Horan syndrome	302350	NHS
709	Navajo neurohepatopathy	256810	MPV17
710	Nemaline myopathy 2, autosomal recessive	256030	NEB
711	Nemaline myopathy 5, Amish type	605355	TNNT1
712	Nephronophthisis 1, juvenile	256100	NPHP1
713	Nephronophthisis 11	613550	TMEM67
714	Nephronophthisis 13	614377	WDR19
715	Nephronophthisis 2, infantile	602088	INVS
716	Nephronophthisis 3	604387	NPHP3
717	Nephronophthisis 4	606966	NPHP4
718	Nephronophthisis 7	611498	GLIS2
719	Nephronophthisis 9	613824	NEK8
720	Nephrotic syndrome, type 1	256300	NPHS1
721	Nephrotic syndrome, type 2	600995	NPHS2
722	Nephrotic syndrome, type 3	610725	PLCE1
723	Nephrotic syndrome, type 5, with or without ocular abnormalities	614199	LAMB2
724	Neu-Laxova syndrome1	256520	PHGDH
725	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	250620	HIBCH
726	Neurodegeneration due to cerebral folate transport deficiency	613068	FOLR1
727	Neurodegeneration with brain iron accumulation 1	234200	PANK2
728	Neurofibromatosis, type 1	162200	NF1
729	Neurofibromatosis, type 2	101000	NF2
730	Neutropenia, severe congenital 3, autosomal recessive	610738	HAX1

731	Niemann-Pick disease type A	257200	SMPD1
732	Niemann-Pick disease type B	607616	SMPD1
733	Niemann-Pick disease type C1	257220	NPC1
734	Niemann-Pick disease type C2	607625	NPC2
735	Noonan syndrome 1	163950	PTPN11
736	Noonan syndrome 3	609942	KRAS
737	Noonan syndrome 4	610733	SOS1
738	Noonan syndrome 5	611553	RAF1
739	Noonan syndrome 6	613224	NRAS
740	Noonan syndrome 7	613706	BRAF
741	Noonan-like syndrome with loose anagen hair	607721	SHOC2
742	Norrie disease	310600	NDP
743	Occipital horn syndrome	304150	ATP7A
744	Oculocerebrorenal syndrome-Lowe oculocerebralrenal syndrome	309000	OCRL
745	Oculodentodigital dysplasia	164200	GJA1
746	Oculodentodigital dysplasia, autosomal recessive	257850	GJA1
747	Ohdo syndrome, X-linked	300895	MED12
748	Omenn syndrome	603554	DCLRE1C
749	Omenn syndrome (gene RAG1)	603554	RAG1
750	Omenn syndrome (gene RAG2)	603554	RAG2
751	Opitz GBBB syndrome, type I	300000	MID1
752	Opitz-Kaveggia syndrome o FG Syndrome 1	305450	MED12
753	Ornithine transcarbamylase deficiency	311250	OTC
754	Orofaciodigital syndrome I	311200	OFD1
755	Osteogenesis imperfecta type 8	610915	LEPRE1
756	Osteogenesis imperfecta type VII	610682	CRTAP
757	Osteogenesis imperfecta, type I	166200	COL1A1
758	Osteogenesis imperfecta, type II	166210	COL1A2
759	Osteogenesis imperfecta, type II	166210	COL1A1

760	Osteogenesis imperfecta, type III	259420	COL1A2
761	Osteogenesis imperfecta, type III	259420	COL1A1
762	Osteogenesis imperfecta, type IV	166220	COL1A2
763	Osteogenesis imperfecta, type IV	166220	COL1A1
764	Osteogenesis imperfecta, type IX	259440	PPIB
765	Osteopetrosis with renal tubular acidosis Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	259730	CA2
766	Osteopetrosis, autosomal recessive 5	259720	OSTM1
767	Otopalatodigital syndrome, type I	311300	FLNA
768	Otopalatodigital syndrome, type II	304120	FLNA
769	Otospondylomegaepiphyseal dysplasia	215150	COL11A2
770	Otospondylomegaepiphyseal dysplasia	215150	COL2A1
771	Paget disease, juvenile	239000	TNFRSF11B
772	Partial androgen insensitivity syndrome	312300	AR
773	Partington syndrome	309510	ARX
774	PCWH syndrome	609136	SOX10
775	Pelizaeus-Merzbacher disease	312080	PLP1
776	Pelizaeus-Merzbacher-like due to GJC2 mutation	608804	GJC2
777	Pendred syndrome	274600	SLC26A4
778	Peroxisomal acyl-CoA oxidase deficiency	264470	ACOX1
779	Peroxisome biogenesis disorder 10A (Zellweger)	614882	PEX3
780	Peroxisome biogenesis disorder 11A (Zellweger)	614883	PEX13
781	Peroxisome biogenesis disorder 11B	614885	PEX13

782	Peroxisome biogenesis disorder 12A (Zellweger)	614886	PEX19
783	Peroxisome biogenesis disorder 13A (Zellweger)	614887	PEX14
784	Peroxisome biogenesis disorder 14B	614920	PEX11B
785	Peroxisome biogenesis disorder 1A (Zellweger)	214100	PEX1
786	Peroxisome biogenesis disorder 1B (NALD/IRD)	601539	PEX1
787	Peroxisome biogenesis disorder 2A (Zellweger)	214110	PEX5
788	Peroxisome biogenesis disorder 2B	202370	PEX5
789	Peroxisome biogenesis disorder 3A (Zellweger)	614859	PEX12
790	Peroxisome biogenesis disorder 3B	266510	PEX12
791	Peroxisome biogenesis disorder 4A (Zellweger)	614862	PEX6
792	Peroxisome biogenesis disorder 4B	614863	PEX6
793	Peroxisome biogenesis disorder 5A (Zellweger)	614866	PEX2
794	Peroxisome biogenesis disorder 5B	614867	PEX2
795	Peroxisome biogenesis disorder 6A (Zellweger)	614870	PEX10
796	Peroxisome biogenesis disorder 6B	614871	PEX10
797	Peroxisome biogenesis disorder 7A (Zellweger)	614873	PEX26
798	Peroxisome biogenesis disorder 8A, (Zellweger)	614876	PEX16
799	Peroxisome biogenesis disorder 8B	614877	PEX16

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800	Peroxisome biogenesis disorder 9B	614879	PEX7
801	Perrault syndrome	233400	HSD17B4
802	Persistent truncus arteriosus	217095	GATA6
803	Persistent truncus arteriosus-Conotruncal heart malformations	217095	NKX2-6
804	Phenylketonuria	261600	PAH
805	Phosphoglycerate dehydrogenase deficiency	601815	PHGDH
806	Pierson syndrome	609049	LAMB2
807	Pitt-Hopkins syndrome	610954	TCF4
808	Platyspondylic skeletal dysplasia, Torrance type	151210	COL2A1
809	Polymicrogyria, bilateral frontoparietal	606854	GPR56
810	Polymicrogyria, symmetric or asymmetric	610031	TUBB2B
811	Pontocerebellar hypoplasia type 1A	607596	VRK1
812	Pontocerebellar hypoplasia type 2B	612389	TSEN2
813	Pontocerebellar hypoplasia type 2C	612390	TSEN34
814	Pontocerebellar hypoplasia, type 6	611523	RARS2
815	Porphyria, congenital erythropoietic	263700	UROS
816	Progressive epilepsy - intellectual deficit, Finnish type	610003	CLN8
817	Propionic acidemia (gene PCCA)	606054	PCCA
818	Propionic acidemia (gene PCCB)	606054	PCCB
819	Proud syndrome	300004	ARX
820	Proximal spinal muscular atrophy type 1	253300	SMN1
821	Proximal spinal muscular atrophy type 2	253550	SMN1

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822	Proximal spinal muscular atrophy type 3	253400	SMN1
823	Proximal spinal muscular atrophy type 4	271150	SMN1
824	Pseudoachondroplasia	177170	COMP
825	Pseudohermaphroditism, male, with gynecomastia	264300	HSD17B3
826	Pseudovaginal perineoscrotal hypospadias	264600	SRD5A2
827	Pyridoxal phosphate-responsive seizures	610090	PNPO
828	Pyruvate carboxylase deficiency	266150	PC
829	Pyruvate dehydrogenase E1-alpha deficiency Leigh syndrome, X-linked	312170	PDHA1
830	Pyruvate dehydrogenase E1-beta deficiency	614111	PDHB
831	Pyruvate dehydrogenase E2 deficiency	245348	DLAT
832	Pyruvate dehydrogenase phosphatase deficiency	608782	PDP1
833	Refsumdisease	266500	PHYH
834	Renal-hepatic-pancreatic dysplasia	208540	NPHP3
835	Renal-hepatic-pancreatic dysplasia 2	615415	NEK8
836	Renpenning syndrome	309500	PQBP1
837	Restrictive dermopathy, lethal	275210	ZMPSTE24
838	Retinitis pigmentosa 12, autosomal recessive	600105	CRB1
839	Retinitis pigmentosa 19	601718	ABCA4
840	Retinitis pigmentosa 2	312600	RP2
841	Retinitis pigmentosa 20	613794	RPE65
842	Retinitis pigmentosa 3	300029	RPGR
843	Retinitis pigmentosa 39	613809	USH2A

844	Retinitis pigmentosa 40	613801	PDE6B
845	Retinitis pigmentosa 43	613810	PDE6A
846	Retinitis pigmentosa 59	613861	DHDDS
847	Retinitis pigmentosa 61	614180	CLRN1
848	Retinitis pigmentosa type 1, autosomal dominant	180100	RP1
849	Retinitis pigmentosa type 10, autosomal dominant	180105	IMPDH1
850	Retinitis pigmentosa type 11, autosomal dominant	600138	PRPF31
851	Retinitis pigmentosa type 13, autosomal dominant	600059	PRPF8
852	Retinitis pigmentosa type 17, autosomal dominant	600852	CA4
853	Retinitis pigmentosa type 18, autosomal dominant	601414	PRPF3
854	Retinitis pigmentosa type 27, autosomal dominant	613750	NRL
855	Retinitis pigmentosa type 30, autosomal dominant	607921	FSCN2
856	Retinitis pigmentosa type 31, autosomal dominant	609923	TOPORS
857	Retinitis pigmentosa type 33, autosomal dominant	610359	SNRNP200
858	Retinitis pigmentosa type 35, autosomal dominant	610282	SEMA4A
859	Retinitis pigmentosa type 4, autosomal dominant	613731	RHO
860	Retinitis pigmentosa type 42, autosomal dominant	612943	KLHL7
861	Retinitis pigmentosa type 48, autosomal dominant	613827	GUCA1B
862	Retinitis pigmentosa type 50, autosomal dominant	613194	BEST1
863	Retinitis pigmentosa type 7, autosomal dominant	608133	PRPH2

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864	Retinitis pigmentosa type 9, autosomal dominant	180104	RP9
865	Rett syndrome	312750	MECP2
866	Rett syndrome, congenital variant	613454	FOXG1
867	Rhizomelic chondrodysplasia punctata type 3	600121	AGPS
868	Right atrial isomerism	208530	GDF1
869	Roberts syndrome	269000	ESCO2
870	Rolandic epilepsy, mental retardation, and speech dyspraxia	300643	SRPX2
871	Saethre-Chotzen syndrome (FGFR2)	101400	FGFR2
872	Saethre-Chotzen syndrome (TWIST1)	101400	TWIST1
873	Salla disease	604369	SLC17A5
874	Sandhoff disease	268800	HEXB
875	Scaphocephaly, maxillary retrusion, and mental retardation	609579	FGFR2
876	Schizencephaly	269160	EMX2
877	Schizencephaly	269160	SHH
878	Schizencephaly	269160	SIX3
879	Schneckenbecken dysplasia	269250	SLC35D1
880	Schwartz-Jampel syndrome	255800	HSPG2
881	Seckel syndrome	210600	ATR
882	SED congenita	183900	COL2A1
883	Senior-Loken syndrome 1	266900	NPHP1
884	Senior-Loken syndrome 4	606996	NPHP4
885	Senior-Loken syndrome 5	609254	IQCB1
886	Senior-Loken syndrome 6	610189	CEP290
887	Senior-Loken syndrome 7	613615	SDCCAG8
888	Senior-Loken syndrome 8	616307	WDR19
889	SESAME syndrome	612780	KCNJ10
890	Severe combined immunodeficiency due to complete RAG1 deficiency	601457	RAG1

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891	Severe combined immunodeficiency due to complete RAG2 deficiency	601457	RAG2
892	Severe combined immunodeficiency due to DCLRE1C deficiency	602450	DCLRE1C
893	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611291	NHEJ1
894	Severe combined immunodeficiency with sensitivity to ionizing radiation	602450	LIG4
895	Severe combined immunodeficiency. deaminase deficiency	102700	ADA
896	Severe generalized recessive dystrophic epidermolysis bullosa	226600	COL7A1
897	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy	601705	FOXN1
898	Shaheen syndrome	615328	COG6
899	Short-rib thoracic dysplasia 3 with or without polydactyly	613091	DYNC2H1
900	Short-rib thoracic dysplasia 4 with or without polydactyly	613819	TTC21B
901	Short-rib thoracic dysplasia 5 with or without polydactyly	614376	WDR19
902	Short-rib thoracic dysplasia 6 with or without polydactyly	263520	NEK1
903	Shwachman-Diamond syndrome	260400	SBDS
904	Sialidosis, type I	256550	NEU1
905	Sialidosis, type II	256550	NEU1
906	Sickle cell anemia	603903	HBB

907	Simpson-Golabi-Behmel syndrome type 2	300209	OFD1
908	Simpson-Golabi-Behmel syndrome, type 1	312870	GPC3
909	Síndrome de Dursun	612541	G6PC3
910	Sjogren-Larsson syndrome	270200	ALDH3A2
911	SMED Strudwick type	184250	COL2A1
912	Smith-Lemli-Opitz syndrome	270400	DHCR7
913	Sotos syndrome 1	117550	NSD1
914	Spastic paraplegia type 2, X-linked	312920	PLP1
915	Spinal muscular atrophy with respiratory distress	604320	IGHMBP2
916	Spinal muscular atrophy, distal, autosomal recessive, 4	611067	PLEKHG5
917	Spondylocostal dysostosis 1, autosomal recessive	277300	DLL3
918	Spondyloepimetaphyseal dysplasia, aggrecan type	612813	ACAN
919	Spondyloepiphyseal dysplasia, Kimberley type	608361	ACAN
920	Spondyloperipheral dysplasia	271700	COL2A1
921	Stargardt dusease 1	248200	ABCA4
922	Stickler sydrome, type I, nonsyndromic ocular	609508	COL2A1
923	Stickler syndrome, type I	108300	COL2A1
924	Stickler syndrome, type II	604841	COL11A1
925	Stickler syndrome, type III	184840	COL11A2
926	Stickler syndrome, type IV	614134	COL9A1
927	Stocco dos Santos X-linked mental retardation syndrome	300434	SHROOM4
928	Stüve-Wiedemann syndrome	601559	LIFR
929	Subcortical laminal heteropia, X-linked	300067	DCX
930	Succinyl CoA:3-oxoacid CoA transferase deficiency	245050	OXCT1

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931	Sudden infant death with dysgenesis of the testes syndrome	608800	TSPYL1
932	Sulfocysteinuria	272300	SUOX
933	Surfactant metabolism dysfunction, pulmonary, 1	265120	SFTPB
934	Surfactant metabolism dysfunction, pulmonary, 2	610913	SFTPC
935	Surfactant metabolism dysfunction, pulmonary, 3	610921	ABCA3
936	Syndromic microphthalmia type 9	601186	STRA6
937	Tay-Sachs disease	272800	HEXA
938	T-B+ severe combined immunodeficiency due to gamma chain deficiency	300400	IL2RG
939	T-B+ severe combined immunodeficiency due to JAK3 deficiency	600802	JAK3
940	T-B+ severe combined immunodeficiency, X-linked	312863	IL2RG
941	Tetra-amelia, autosomal recessive	273395	WNT3
942	Tetralogy of Fallot	187500	GATA4
943	Tetralogy of Fallot	187500	GATA6
944	Tetralogy of Fallot	187500	JAG1
945	Tetrology of Fallot	187500	GDF1
946	Tetrology of Fallot	187500	NKX2-5
947	Tetrology of Fallot	187500	TBX1
948	Thanatophoric dysplasia, type I	187600	FGFR3
949	Thanatophoric dysplasia, type II	187601	FGFR3
950	Thrombocytopenia-absent radius syndrome	274000	RBM8A
951	Thrombotic thrombocytopenic purpura, familial	274150	ADAMTS13
952	Thryoid dyshormonogenesis 6	607200	DUOX2
953	Thyroid dyshormonogenesis 1	274400	SLC5A5
954	Thyroid dyshormonogenesis 2A	274500	TPO

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955	Thyroid dyshormonogenesis 3	274700	TG
956	Thyroid dyshormonogenesis 4	274800	IYD
957	Thyroid dyshormonogenesis 5	274900	DUOXA2
958	Tietz albinism-deafness syndrome	103500	MITF
959	Transposition of great arteries, dextro-looped 3	613854	GDF1
960	Transposition of the great arteries, dextro-looped 1	608808	MED13L
961	Transposition of the great arteries, dextro-looped 2	613853	CFC1
962	Treacher Collins syndrome 1	154500	TCOF1
963	Treacher Collins syndrome 3	248390	POLR1C
964	Trichothiodystrophy 1, photosensitive	601675	ERCC2
965	Trichothiodystrophy 2, photosensitive	616390	ERCC3
966	Trichothiodystrophy 3, photosensitive	616395	GTF2H5
967	Tuberous sclerosis-1	191100	TSC1
968	Tuberous sclerosis-2	613254	TSC2
969	Tyrosinemia type 1	276700	FAH
970	Tyrosinemia type 2	276600	TAT
971	Tyrosinemia type 3	276710	HPD
972	Usher syndrome type 1	276900	MYO7A
973	Usher syndrome type 1C	276904	USH1C
974	Usher syndrome type 1D	601067	CDH23
975	Usher syndrome type 1D/F digenic	601067	CDH23
976	Usher syndrome type 1F	602083	PCDH15
977	Usher syndrome type 1G	606943	USH1G
978	Usher syndrome type 2A	276901	USH2A
979	Usher syndrome type 2C	605472	GPR98
980	Usher syndrome type 3A	276902	CLRN1
981	Usher syndrome type 3B	614504	HARS
982	VACTERL association, X-linked	314390	ZIC3
983	Ventricular septal defect 1	614429	GATA4

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984	Ventricular septal defect 2	614431	CITED2
985	Ventricular septal defect 3	614432	NKX2-5
986	Very long chain acyl-CoA dehydrogenase deficiency	201475	ACADVL
987	Vitamin B12-responsive methylmalonic acidemia type cblA	251100	MMAA
988	Vitamin B12-responsive methylmalonic acidemia type cblB	251110	MMAB
989	Vitamin B12-unresponsive methylmalonic acidemia type mut-	251000	MUT
990	Vitamin D-dependent rickets, type I	264700	CYP27B1
991	Vitamin D-dependent rickets, type II A	277440	VDR
992	Vitelliform macular dystrophy 2	153700	BEST1
993	Vitreoretinochoroidopathy	193220	BEST1
994	Waardenburg syndrome, type 1	193500	PAX3
995	Waardenburg syndrome, type 2A	193510	MITF
996	Waardenburg syndrome, type 2E, with or without neurologic involvement	611584	SOX10
997	Waardenburg syndrome, type 3	148820	PAX3
998	Waardenburg syndrome, type 4C	613266	SOX10
999	Waardenburg-Shah syndrome 4A (Sindrome di Waardenburg, tipo 4)	277580	EDNRB
1000	Waardenburg-Shah syndrome 4B	613265	EDN3
1001	Walker-Warburg syndrome (gene POMGNT1)	253280	POMGNT1
1002	Walker-Warburg syndrome (gene POMT1)	236670	POMT1
1003	Walker-Warburg syndrome (gene POMT2)	613150	POMT2
1004	Warburg micro syndrome 1	600118	RAB3GAP1
1005	Warburg micro syndrome 2	614225	RAB3GAP2
1006	Weissenbacher-Zweymuller syndrome	277610	COL11A2

1007	Wilson disease	277900	ATP7B
1008	Wilson-Turner syndrome	309585	HDAC8
1009	Wolcott-Rallison syndrome	226980	EIF2AK3
1010	Wolfram syndrome	222300	WFS1
1011	Xeroderma pigmentosum complementation group A	278700	XPA
1012	Xeroderma pigmentosum complementation group E	278740	DDB2
1013	Xeroderma pigmentosum, group C	278720	XPC
1014	Xeroderma pigmentosum/Cockayne syndrome complex complementation group B	610651	ERCC3
1015	Xeroderma pigmentosum/Cockayne syndrome complex complementation group D	278730	ERCC2
1016	Xeroderma pigmentosum/Cockayne syndrome complex complementation group F	278760	ERCC4
1017	Xeroderma pigmentosum/Cockayne syndrome complex complementation group G	278780	ERCC5
1018	X-linked agammaglobulinemia	300755	BTK
1019	X-linked centronuclear myopathy	310400	MTM1
1020	X-linked creatine transporter deficiency	300352	SLC6A8
1021	X-linked hyper-IgM syndrome Immunodeficiency, X-linked, with hyper-IgM	308230	CD40LG
1022	X-linked intellectual deficit with marfanoid habitus- Lujan-Fryns syndrome	309520	MED12
1023	X-linked spinal muscular atrophy type 2	301830	UBA1

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1024	Zellweger syndrome 7A	614872	PEX26

Roma, 02/10/2015

Il Genetista
Dr.ssa Marina Baldi

Il Direttore
Dr. F. Fiorentino

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