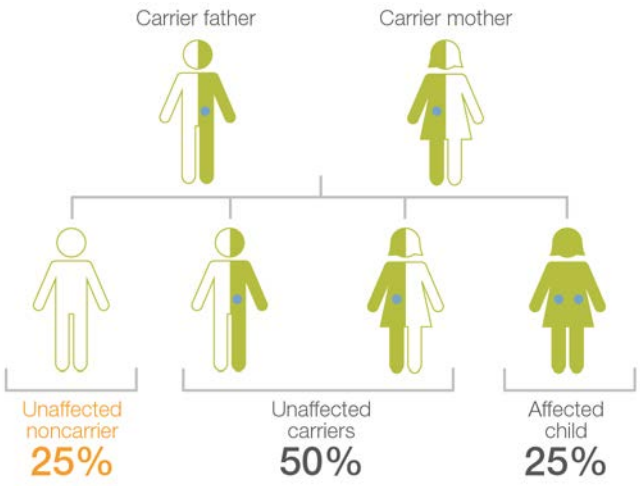
genescreen®

La genomica di nuova generazione

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 nostro DNA, il **genoma**. In particolare **le nuove tecnologie di sequenziamento, *Next Generation Sequencing* (NGS)**, ci permettono oggi di accedere alla sequenza del nostro DNA in modo più facile ed efficace, fornendo una valutazione approfondita dell'informazione genetica di ogni singolo individuo.

Ogni persona nasce, infatti, con caratteristiche genetiche che la differenziano dagli altri e che la rendono unica. Mentre la maggior parte delle differenze nella sequenza del DNA tra persone diverse è innocua, alcuni cambiamenti, definiti **mutazioni genetiche**, possono alterare la funzionalità genomica e rendere quella persona portatrice di una specifica malattia genetica trasmissibile ai propri figli.



I portatori di malattie genetiche sono tipicamente individui sani, completamente privi di sintomi ed inconsapevoli di essere a rischio di trasmettere tale "errore" del DNA ai figli.

Il test GeneScreen® EXPANDED

GeneScreen® Expanded è un test diagnostico, sviluppato da GENOMA Group, che permette di eseguire un'analisi multipla di **1467 malattie genetiche ereditarie**, tra cui quelle più frequenti nella popolazione italiana, come la Fibrosi Cistica, l'Anemia Falciforme, la Talassemia, la Sordità Ereditaria.

GeneScreen® consente alla coppia di conoscere, attraverso l'analisi del loro DNA, se si è portatori di gravi malattie genetiche. Il test, quindi, permette di identificare le coppie a rischio di trasmettere ai loro figli una specifica malattia genetica.

Indicazioni al test GeneScreen® EXPANDED

GeneScreen® EXPANDED è indicato:

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- Per le coppie che progettano di diventare genitori, sia tramite concepimento naturale che mediante l'accesso a tecniche di procreazione medicalmente assistita (PMA);
- Per le coppie che sono in attesa di un figlio, e che desiderano ridurre il rischio di trasmettere a quest'ultimo una malattia genetica ereditaria;
- Per le coppie che fanno ricorso a tecniche di fecondazione eterologa, al fine di individuare un donatore di gameti che non sia portatore di mutazioni nei medesimi geni riscontrate in uno dei partners della coppia.

L'esame può essere effettuato su un singolo individuo o, preferibilmente, su **entrambi i partners** della coppia.

Come viene effettuato il test GeneScreen® Expanded?

Il test viene eseguito mediante il prelievo di un campione ematico. Tramite un'analisi complessa di laboratorio, Il DNA viene isolato dalle cellule nucleate ed **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 **925 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.
L'analisi per individuare la delezione degli esoni 7 e 8 del gene SMN1 viene effettuata mediante tecnica MLPA e successiva elettroforesi capillare in sequenziatore automatico a tecnologia fluorescente.
La valutazione dell'espansione delle triplette nucleotidiche ripetute nel sito fragile FRAXA viene eseguita mediante PCR fluorescente e successiva elettroforesi capillare in sequenziatore automatico.
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).

Risultati ottenibili con il test GeneScreen® EXPANDED

"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 all'altro partner della coppia, al fine di verificare che quest'ultimo non sia portatore delle medesima malattia genetica, nel qual caso si ravviserebbe un rischio di trasmissione della patologia ai figli.
Le mutazioni riscontrabili tramite il test **GeneScreen® EXPANDED** possono rientrare nelle seguenti categorie prognostiche:

- **con significato patologico noto**;
- **con significato incerto** in quanto non ancora note o caratterizzate dalla comunità medico-scientifica. Tali varianti verranno refertate solo nei test eseguiti su entrambi i partners della coppia, nel caso in cui in uno dei partners dovesse essere stata riscontrata una mutazione a significato patologico noto.

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Se entrambi i partners della coppia dovessero risultare positivi per il test, portatori di una mutazione con significato patologico noto nel medesimo gene, il nostro genetista potrà fornire una panoramica sulle opzioni diagnostiche attualmente disponibili per verificare lo stato di salute del feto, in caso di futura gravidanza.

"NEGATIVO" - Assenza di mutazioni: indica che il test non ha rilevato la presenza di mutazioni nei geni esaminati.

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 o con significato incerto, sulla base dei dati della letteratura scientifica e la classificazione presente nel database di riferimento Human Gene Mutation Database (HGMD), NCBI Clinvar, aggiornati 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 GeneScreen® EXPANDED

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 GeneScreen® EXPANDED

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 di essere portatori di una mutazione localizzata 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 determinabile in base alle attuali conoscenze medico-scientifiche.

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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.
Limite intrinseco della metodologia NGS utilizzata è la mancanza di uniformità di coverage per ciascuna regione genica analizzata. Tale limite si traduce nella possibilità, insita nelle metodiche NGS, che specifiche mutazioni dei geni selezionati potrebbero non essere state rilevate dal test.
La valutazione dell'espansione delle triplette nucleotidiche ripetute nel sito fragile FRAXA mediante Polymerase Chain Reaction (PCR), per i limiti intrinseci della metodica, potrebbe non evidenziare la presenza di espansioni di triplette di grosse dimensioni. Quindi, in caso pazienti di sesso femminile nelle quali viene evidenziato un genotipo omozigote, la PCR potrebbe non aver amplificato un allele con un espansione di grosse dimensioni, e quindi trattarsi di un falso omozigote. In quest'ultimo caso, il risultato dovrebbe essere confermato mediante esame di secondo livello.

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Tabella 1: GeneScreen® Expanded - Elenco dei geni analizzati

Gene	Malattia Genetica	PhenoMIM
AAAS	Achalasia-addisonianism-alacrimia syndrome	231550
ABCA12	Ichthyosis, autosomal recessive 4B (harlequin)	242500
ABCA12	Ichthyosis, congenital, autosomal recessive 4A	601277
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3	610921
ABCA4	Cone-rod dystrophy 3	604116
ABCA4	Fundus flavimaculatus	248200
ABCA4	Macular degeneration, age-related, 2	153800
ABCA4	Retinal dystrophy, early-onset severe	248200
ABCA4	Retinitis pigmentosa 19	601718
ABCA4	Stargardt disease 1	248200
ABCB11	Cholestasis, benign recurrent intrahepatic, 2	605479
ABCB11	Cholestasis, progressive familial intrahepatic 2	601847
ABCB4	Cholestasis, intrahepatic, of pregnancy, 3	614972
ABCB4	Cholestasis, progressive familial intrahepatic 3	602347
ABCB4	Gallbladder disease 1	600803
ABCB7	Anemia, sideroblastic, with ataxia	301310
ABCC2	Dubin-Johnson syndrome	237500
ABCC6	Arterial calcification, generalized, of infancy, 2	614473
ABCC6	Pseudoxanthoma elasticum	264800
ABCC6	Pseudoxanthoma elasticum, forme fruste	177850
ABCC8	Diabetes mellitus, noninsulin-dependent	125853
ABCC8	Diabetes mellitus, permanent neonatal	606176
ABCC8	Diabetes mellitus, transient neonatal 2	610374
ABCC8	Hyperinsulinemic hypoglycemia, familial, 1	256450
ABCC8	Hypoglycemia of infancy, leucine-sensitive	240800
ABCD1	Adrenoleukodystrophy	300100
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	614857
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	611283
ACAD9	Acyl-CoA dehydrogenase 9 deficiency	611126

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ACADL	ACYL-CoA DEHYDROGENASE, LONG-CHAIN	609576
ACADM	Medium chain acyl-CoA dehydrogenase deficiency	201450
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of	201470
ACADSB	2-methylbutyrylglycinuria	610006
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency	201475
ACAT1	Ketoacidosis due to beta-ketothiolase deficiency	203750
ACE	Angiotensin I-converting enzyme, benign serum increase	
ACE	Microvascular complications of diabetes 3	612624
ACE	Myocardial infarction, susceptibility to	
ACE	Renal tubular dysgenesis	267430
ACE	SARS, progression of	
ACE	Stroke, hemorrhagic	614519
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	264470
ACSF3	Combined malonic and methylmalonic aciduria	614265
ACSL4	Mental retardation, X-linked 63	300387
ACTN4	Glomerulosclerosis, focal segmental, 1	603278
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency	102700
ADAMTS 13	Thrombotic thrombocytopenic purpura, familial	274150
ADAMTS 2	Ehlers-Danlos syndrome, dermatosparaxis type	225410
ADAMTS L2	Geleophysic dysplasia 1	231050
ADCK3	Autosomal recessive ataxia due to ubiquinone deficiency	612016
ADGRG1	Polymicrogyria, bilateral frontoparietal	606854
ADGRG1	Polymicrogyria, bilateral perisylvian	615752
ADK	Hypermethioninemia due to adenosine kinase deficiency	614300
AFF2	Mental retardation, X-linked, FRAXE type	309548
AGA	Aspartylglucosaminuria	208400
AGL	Glycogen storage disease due to glycogen debranching enzyme deficiency	232400
AGPS	Rhizomelic chondrodysplasia punctata type 3	600121
AGT	Hypertension, essential, susceptibility to	145500
AGT	Preeclampsia, susceptibility to	

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AGT	Renal tubular dysgenesis	267430
AGTR1	Hypertension, essential	145500
AGTR1	Renal tubular dysgenesis	267430
AGTR2	ANGIOTENSIN II RECEPTOR, TYPE 2	300034
AGXT	Hyperoxaluria, primary, type 1	259900
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	613752
AHI1	Joubert syndrome with ocular defect	608629
AIPL1	Cone-rod dystrophy	604393
AIPL1	Leber congenital amaurosis 4	604393
AIPL1	Retinitis pigmentosa, juvenile	604393
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	240300
ALAS2	Anemia, sideroblastic, 1	300751
ALAS2	Protoporphyria, erythropoietic, X-linked	300752
ALDH3A2	Sjogren-Larsson syndrome	270200
ALDH4A1	Hyperprolinemia, type II	239510
ALDH5A1	4-hydroxybutyric aciduria	271980
ALDH7A1	Epilepsy, pyridoxine-dependent	266100
ALDOA	Glycogen storage disease XII	611881
ALDOB	Hereditary fructose intolerance	229600
ALG1	Congenital disorder of glycosylation type Ik	608540
ALG12	Congenital disorder of glycosylation, type Ig	607143
ALG2	Congenital disorder of glycosylation, type Ii	607906
ALG3	Congenital disorder of glycosylation, type Id	601110
ALG6	Congenital disorder of glycosylation type Ic	603147
ALG8	Congenital disorder of glycosylation, type Ih	608104
ALG9	Congenital disorder of glycosylation, type Il	608776
ALMS1	Alström syndrome	203800
ALPL	Childhood-onset hypophosphatasia	241510
ALPL	Infantile hypophosphatasia	241500
ALS2	Amyotrophic lateral sclerosis 2, juvenile	205100
ALS2	Primary lateral sclerosis, juvenile	606353

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ALS2	Spastic paralysis, infantile onset ascending	607225
AMACR	Alpha-methylacyl-Coa Racemase deficiency	614307
AMACR	Congenital bile acid synthesis defect type 4	214950
AMPD1	Myopathy due to myoadenylate deaminase deficiency	615511
AMT	Glycine encephalopathy	605899
ANO5	Gnathodiaphyseal dysplasia	166260
ANO5	Miyoshi muscular dystrophy 3	613319
ANO5	Muscular dystrophy, limb-girdle, autosomal recessive 12	611307
ANTXR2	Hyaline fibromatosis syndrome	228600
AP1S1	MEDNIK syndrome	609313
AP1S2	Mental retardation, X-linked syndromic 5	304340
AP3B1	Hermansky-Pudlak syndrome 2	608233
APTX	Ataxia - oculomotor apraxia type 1	208920
AQP2	Diabetes insipidus, nephrogenic	125800
AR	Complete androgen insensitivity syndrome	300068
AR	Kennedy disease	313200
AR	Partial androgen insensitivity syndrome	312300
ARG1	Argininemia	207800
ARHGEF6	Mental retardation, X-linked 46	300436
ARHGEF9	Epileptic encephalopathy, early infantile, 8	300607
ARL13B	Joubert syndrome 8	612291
ARL6	Bardet-Biedl syndrome 1, modifier of	209900
ARL6	Bardet-Biedl syndrome 3	600151
ARL6	Retinitis pigmentosa 55	613575
ARSA	Metachromatic leukodystrophy	250100
ARSB	Mucopolysaccharidosis type 6	253200
ARSE	Brachytelephalangic chondrodysplasia punctata	302950
ARSF	ARYLSULFATASE F	300003
ARX	Early infantile epileptic encephalopathy	308350
ASL	Argininosuccinic aciduria	207900
ASNS	Asparagine synthetase deficiency	615574

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ASPA	Canavan disease	271900
ASPM	Microcephaly 5, primary, autosomal recessive	608716
ASS1	Citrullinemia type I	215700
ATIC	AICA-ribosiduria due to ATIC deficiency	608688
ATM	Ataxia-telangiectasia	208900
ATP6AP2	Mental retardation, X-linked, syndromic, Hedera type	300423
ATP6AP2	Parkinsonism with spasticity, X-linked	300911
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA	219200
ATP6V0A2	Wrinkly skin syndrome	278250
ATP6V1B1	Renal tubular acidosis with deafness	267300
ATP7A	Menkes disease	309400
ATP7A	Occipital horn syndrome	304150
ATP7A	X-linked distal spinal muscular atrophy	300489
ATP7B	Wilson disease	277900
ATP8B1	Cholestasis, benign recurrent intrahepatic	243300
ATP8B1	Cholestasis, intrahepatic, of pregnancy, 1	147480
ATP8B1	Cholestasis, progressive familial intrahepatic 1	211600
ATR	Seckel syndrome	210600
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic	300448
ATRX	Alpha-thalassemia/mental retardation syndrome	301040
ATRX	Mental retardation-hypotonic facies syndrome, X-linked	309580
AUH	3-methylglutaconic aciduria type 1	250950
B4GALT1	Congenital disorder of glycosylation type 2d	607091
B9D2	Joubert syndrome 34	614175
B9D2	Meckel syndrome 10	614175
BBS1	Bardet-Biedl syndrome 1	209900
BBS10	Bardet-Biedl syndrome 10	615987
BBS12	Bardet-Biedl syndrome 12	615989
BBS2	Bardet-Biedl syndrome 2	615981
BBS2	Retinitis pigmentosa 74	616562

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

BCHE	Apnea, postanesthetic, susceptibility to, due to BCHE deficiency	617936
BCHE	Butyrylcholinesterase deficiency	617936
BCKDHA	Maple syrup urine disease (gene BCKDHA)	248600
BCKDHB	Maple syrup urine disease (gene BCKDHB)	248600
BCOR	Microphthalmia, syndromic 2	300166
BCS1L	Björnstad syndrome	262000
BCS1L	GRACILE syndrome	603358
BCS1L	Isolated CoQ-cytochrome C reductase deficiency	124000
BCS1L	Leigh syndrome	256000
BEST1	Bestrophinopathy, autosomal recessive	611809
BEST1	Macular dystrophy, vitelliform, 2	153700
BEST1	Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma	193220
BEST1	Retinitis pigmentosa, concentric	613194
BEST1	Retinitis pigmentosa-50	613194
BEST1	Vitreoretinochoroidopathy	193220
BLM	Bloom syndrome	210900
BRCA2	Breast cancer, male, susceptibility to	114480
BRCA2	Breast-ovarian cancer, familial, 2	612555
BRCA2	Fanconi anemia, complementation group D1	605724
BRCA2	Glioblastoma 3	613029
BRCA2	Medulloblastoma	155255
BRCA2	Pancreatic cancer 2	613347
BRCA2	Prostate cancer	176807
BRCA2	Wilms tumor	194070
BRIP1	Breast cancer, early-onset, susceptibility to	114480
BRIP1	Fanconi anemia, complementation group J	609054
BRWD3	Mental retardation, X-linked 93	300659
BSCL2	Encephalopathy, progressive, with or without lipodystrophy	615924
BSCL2	Lipodystrophy, congenital generalized, type 2	269700
BSCL2	Neuropathy, distal hereditary motor, type VA	600794
BSCL2	Silver spastic paraplegia syndrome	270685

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BSND	Bartter syndrome, type 4a	602522
BSND	Sensorineural deafness with mild renal dysfunction	602522
BTD	Biotinidase deficiency	253260
BTK	Isolated growth hormone deficiency type III	307200
BTK	X-linked agammaglobulinemia	300755
C10orf2	Infantile onset spinocerebellar ataxia	271245
C3	C3 deficiency	613779
C3	Hemolytic uremic syndrome, atypical, susceptibility to, 5	612925
C3	Macular degeneration, age-related, 9	611378
CA2	Osteopetrosis with renal tubular acidosis	259730
CANT1	Desbuquois dysplasia 1	251450
CANT1	Epiphyseal dysplasia, multiple, 7	617719
CAPN3	Muscular dystrophy, limb-girdle, autosomal dominant 4	618129
CAPN3	Muscular dystrophy, limb-girdle, autosomal recessive 1	253600
CASK	FG syndrome 4	300422
CASK	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	300749
CASK	Mental retardation, with or without nystagmus	300422
CASP10	Autoimmune lymphoproliferative syndrome, type II	603909
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2	611938
CBS	Classical homocystinuria	236200
CC2D2A	COACH syndrome	216360
CC2D2A	Joubert syndrome 9	612285
CC2D2A	Meckel syndrome 6	612284
CCDC103	Ciliary dyskinesia, primary, 17	614679
CCDC39	Ciliary dyskinesia, primary, 14	613807
CD19	Immunodeficiency, common variable, 3	613493
CD247	Immunodeficiency 25	610163
CD2AP	Glomerulosclerosis, focal segmental, 3	607832
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect	613646
CD3D	Immunodeficiency 19	615617
CD3E	Immunodeficiency 18, SCID variant	615615

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CD3G	Immunodeficiency 17, CD3 gamma deficient	615607
CD40LG	X-linked hyper-IgM syndrome	308230
CDH23	Autosomal recessive nonsyndromic sensorineural deafness type DFNB12	601386
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy	225280
CDH3	Hypotrichosis, congenital, with juvenile macular dystrophy	601553
CDHR1	Cone-rod dystrophy 15	613660
CDHR1	Retinitis pigmentosa 65	613660
CDK5RAP2	Microcephaly 3, primary, autosomal recessive	604804
CDKL5	Epileptic encephalopathy, early infantile, 2	300672
CENPJ	Microcephaly 6, primary, autosomal recessive	608393
CENPJ	Seckel syndrome 4	613676
CEP152	Microcephaly 9, primary, autosomal recessive	614852
CEP152	Seckel syndrome 5	613823
CEP290	Joubert syndrome with oculorenal defect 5	610188
CEP290	Senior-Loken syndrome	610189
CERKL	Retinitis pigmentosa 26	608380
CFH	Basal laminar drusen	126700
CFH	Complement factor H deficiency	609814
CFH	Hemolytic uremic syndrome, atypical, susceptibility to, 1	235400
CFH	Macular degeneration, age-related, 4	610698
CFP	Properdin deficiency, X-linked	312060
CFTR	Cystic fibrosis; mucoviscidosis	219700
CHM	Choroideremia	303100
CHRNA1	Multiple pterygium syndrome, lethal type	253290
CHRNA1	Myasthenic syndrome, fast-channel congenital	608930
CHRNA1	Myasthenic syndrome, slow-channel congenital	601462
CHRND	Multiple pterygium syndrome, lethal type	253290
CHRND	Myasthenic syndrome, fast-channel congenital	608930
CHRND	Myasthenic syndrome, slow-channel congenital	601462
CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel	605809
CHRNE	Myasthenic syndrome, congenital, 4B, fast-channel	616324

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CHRNE	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	608931
CHRNG	Escobar syndrome	265000
CHRNG	Multiple pterygium syndrome, lethal type	253290
CHRNG	Myasthenia gravis, neonatal transient	100730
CHST6	Macular corneal dystrophy	217800
CIITA	Bare lymphocyte syndrome, type II, complementation group A	209920
CIITA	Rheumatoid arthritis, susceptibility to	180300
CLCN1	Myotonia congenita, dominant	160800
CLCN1	Myotonia congenita, recessive	255700
CLCN1	Myotonia levior, recessive	
CLCN5	Dent disease	300009
CLCN5	Hypophosphatemic rickets	300554
CLCN5	Nephrolithiasis, type I	310468
CLCN5	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis 308990	
CLCN7	Autosomal recessive malignant osteopetrosis 4	611490
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis 607626	
CLDN14	Deafness, autosomal recessive 29	614035
CLDN19	Familial hypomagnesemia - hypercalciuria - nephrocalcinosis - severe ocular involvement	248190
CLN3	Juvenile neuronal ceroid lipofuscinosis 3	204200
CLN5	Late infantile neuronal ceroid lipofuscinosis 5	256731
CLN6	Adult neuronal ceroid lipofuscinosis 4A	204300
CLN6	Late infantile neuronal ceroid lipofuscinosis 6	601780
CLN8	Late infantile neuronal ceroid lipofuscinosis 8	600143
CLN8	Progressive epilepsy - intellectual deficit, Finnish type	610003
CLRN1	Usher syndrome type 3A	276902
CNGA1	Retinitis pigmentosa 49	613756
CNGA3	Achromatopsia 2	216900
CNGB1	Retinitis pigmentosa 45	613767
CNGB3	Achromatopsia 3	262300
CNGB3	Macular degeneration, juvenile	248200
COG1	Congenital disorder of glycosylation, type IIg	611209

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COG7	Congenital disorder of glycosylation, type IIe	608779
COG8	Congenital disorder of glycosylation, type IIh	611182
COL11A1	Fibrochondrogenesis 1	228520
COL11A1	Lumbar disc herniation, susceptibility to	603932
COL11A1	Marshall syndrome	154780
COL11A1	Stickler syndrome, type II	604841
COL17A1	Generalized junctional epidermolysis bullosa, non-Herlitz type	226650
COL18A1	Knobloch syndrome, type 1	267750
COL1A1	Caffey disease	114000
COL1A1	Ehlers-Danlos syndrome, type I	130000
COL1A1	Ehlers-Danlos syndrome, type VIIA	130060
COL1A1	Osteogenesis imperfecta, type I	166200
COL1A1	Osteogenesis imperfecta, type II	166210
COL1A1	Osteogenesis imperfecta, type III	259420
COL1A1	Osteogenesis imperfecta, type IV	166220
COL1A2	Ehlers-Danlos syndrome, cardiac valvular type	225320
COL2A1	Achondrogenesis, type II or hypochondrogenesis	200610
COL2A1	Avascular necrosis of the femoral head	608805
COL2A1	Czech dysplasia	609162
COL2A1	Epiphyseal dysplasia, multiple, with myopia and deafness	132450
COL2A1	Kniest dysplasia	156550
COL2A1	Legg-Calve-Perthes disease	150600
COL2A1	Osteoarthritis with mild chondrodysplasia	604864
COL2A1	Platyspondylic skeletal dysplasia, Torrance type	151210
COL2A1	SED congenita	183900
COL2A1	SMED Strudwick type	184250
COL2A1	Spondyloepiphyseal dysplasia, Stanescu type	616583
COL2A1	Spondyloperipheral dysplasia	271700
COL2A1	Stickler sydrome, type I, nonsyndromic ocular	609508
COL2A1	Stickler syndrome, type I	108300
COL2A1	Vitreoretinopathy with phalangeal epiphyseal dysplasia	

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

COL4A3	Alport syndrome autosomal recessive (gene COL4A3)	203780
COL4A4	Alport syndrome autosomal recessive (gene COL4A4)	203780
COL4A5	Alport syndrome	301050
COL6A1	Bethlem myopathy	158810
COL6A1	Ullrich congenital muscular dystrophy	254090
COL6A2	Bethlem myopathy	158810
COL6A2	Ullrich congenital muscular dystrophy	254090
COL6A3	Bethlem myopathy	158810
COL6A3	Ullrich congenital muscular dystrophy	254090
COL7A1	Dystrophic epidermolysis bullosa pruriginosa	604129
COL7A1	Severe generalized recessive dystrophic epidermolysis bullosa	226600
COL9A1	Epiphyseal dysplasia, multiple, 6	614135
COL9A1	Stickler syndrome, type IV	614134
COL9A2	Epiphyseal dysplasia, multiple, 2	600204
COL9A2	Stickler syndrome, type V	614284
COQ2	Leigh syndrome with nephrotic syndrome	607426
COQ9	Coenzyme Q10 deficiency, primary, 5	614654
COX10	Leigh syndrome due to mitochondrial COX4 deficiency	256000
COX10	Mitochondrial complex IV deficiency	220110
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2	615119
COX15	Leigh syndrome due to cytochrome c oxidase deficiency	256000
COX6B1	Mitochondrial complex IV deficiency	220110
CPS1	Carbamoylphosphate synthetase deficiency	237300
CPT1A	Carnitine palmitoyl transferase 1A deficiency	255120
CPT2	Carnitine palmitoyl transferase II deficiency, infantile form	600649
CPT2	Carnitine palmitoyl transferase II deficiency, neonatal form	608836
CRB1	Leber congenital amaurosis 8	613835
CRB1	Pigmented paravenous chorioretinal atrophy	172870
CRB1	Retinitis pigmentosa-12	600105
CRLF1	Cold-induced sweating syndrome	272430
CRTAP	Osteogenesis imperfecta type VII	610682

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CRX	Cone-rod retinal dystrophy-2	120970
CRX	Leber congenital amaurosis 7	613829
CSTB	Unverricht-Lundborg disease	254800
CTH	Cystathioninuria	219500
CTH	Homocysteine, total plasma, elevated	
CTNS	Cystinosis	219800
CTSC	Haim-Munk syndrome	245010
CTSC	Papillon-Lefevre syndrome	245000
CTSC	Periodontitis 1, juvenile	170650
CTSD	Adult neuronal ceroid lipofuscinosis 10	610127
CTSK	Pycnodysostosis	265800
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type)	300354
CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA	233690
CYBB	Chronic granulomatous disease, X-linked	306400
CYBB	Immunodeficiency 34, mycobacteriosis, X-linked	300645
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	613743
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	202010
CYP11B1	Aldosteronism, glucocorticoid-remediable	103900
CYP11B2	Aldosterone to renin ratio raised	
CYP11B2	Hypoaldosteronism, congenital, due to CMO I deficiency	203400
CYP11B2	Hypoaldosteronism, congenital, due to CMO II deficiency	610600
CYP11B2	Low renin hypertension, susceptibility to	
CYP17A1	17-alpha-hydroxylase/17,20-lyase deficiency	202110
CYP19A1	Aromatase deficiency	613546
CYP19A1	Aromatase excess syndrome	139300
CYP1B1	Anterior segment dysgenesis 6, multiple subtypes	617315
CYP1B1	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset	231300
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	201910
CYP27A1	Cerebrotendinous xanthomatosis	213700
CYP27B1	Vitamin D-dependent rickets, type I	264700
CYP4V2	Bietti crystalline corneoretinal dystrophy	210370

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CYP7B1	Bile acid synthesis defect, congenital, 3	613812
CYP7B1	Spastic paraplegia 5A, autosomal recessive	270800
D2HGDH	D-2-hydroxyglutaric aciduria	600721
DBT	Classic maple syrup urine disease	248600
DCLRE1C	Omenn syndrome	603554
DCLRE1C	Severe combined immunodeficiency due to DCLRE1C deficiency	602450
DCX	Lissencephaly, X-linked	300067
DCX	Subcortical laminal heteropia, X-linked	300067
DDB2	Xeroderma pigmentosum complementation group E	278740
DDC	Aromatic L-amino acid decarboxylase deficiency	608643
DFNB59	Deafness, autosomal recessive 59	610220
DGUOK	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency 3	251880
DHCR24	Desmosterolosis	602398
DHCR7	Smith-Lemli-Opitz syndrome	270400
DHDDS	Congenital disorder of glycosylation, type 1bb	613861
DHDDS	Developmental delay and seizures with or without movement abnormalities	617836
DHDDS	Retinitis pigmentosa 59	613861
DKC1	Dyskeratosis congenita X-linked	305000
DKC1	Hoyeraal-Hreidarsson syndrome	300240
DLD	Leigh syndrome	256000
DLD	Maple syrup urine disease	248600
DLG3	Mental retardation, X-linked 90	300850
DLL3	Autosomal recessive spondylocostal dysostosis 1	277300
DMD	Becker muscular dystrophy	300376
DMD	Duchenne muscular dystrophy	310200
DMP1	Autosomal recessive hypophosphatemic rickets 1	241520
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus	608644
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus	244400
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus	612444
DNAJC19	Dilated cardiomyopathy with ataxia	610198
DNAL1	Ciliary dyskinesia, primary, 16	614017

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DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	242860
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive	243700
DOK7	Fetal akinesia deformation sequence	208150
DOK7	Myasthenia, limb-girdle, familial	254300
DOLK	Congenital disorder of glycosylation, type Im	610768
DPAGT1	Congenital disorder of glycosylation type 1j	608093
DPM1	Congenital disorder of glycosylation type 1e	608799
DPYD	Dihydropyrimidine dehydrogenase deficiency	274270
DSP	Lethal acantholytic epidermolysis bullosa	609638
DUOX2	Thyroid dyshormonogenesis 6	607200
DUOXA2	Thyroid dyshormonogenesis 5	274900
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	613091
DYSF	Miyoshi muscular dystrophy 1	254130
DYSF	Muscular dystrophy, limb-girdle, autosomal recessive 2	253601
DYSF	Myopathy, distal, with anterior tibial onset	606768
EDA	Muscular dystrophy, limb-girdle, autosomal recessive 2	253601
EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked	305100
EDA	Tooth agenesis, selective, X-linked 1	313500
EDAR	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant	129490
EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive	224900
EDAR	Hair morphology 1, hair thickness	612630
EDN3	Waardenburg-Shah syndrome 4B	613265
EDNRB	ABCD syndrome	600501
EDNRB	Waardenburg-Shah syndrome 4A	277580
EFEMP2	Cutis laxa, autosomal recessive, type IB	614437
EFNB1	Craniofrontonasal dysplasia	304110
EGR2	Charcot-Marie-Tooth disease type 4E	605253
EIF2AK3	Wolcott-Rallison syndrome	226980
EIF2B5	Leukoencephalopathy with vanishing white matter	603896
EIF2B5	Ovarioleukodystrophy	603896
ELK1	MEMBER OF ETS ONCOGENE FAMILY	311040

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EMD	Emery-Dreifuss muscular dystrophy 1, X-linked	310300
ENO3	Glycogen storage disease XIII	612932
ENPP1	Autosomal recessive hypophosphatemic rickets 2	613312
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora)	254780
ERBB3	Lethal congenital contractural syndrome 2	607598
ERCC2	Xeroderma pigmentosum/Cockayne syndrome complex complementation group D	278730
ERCC3	Xeroderma pigmentosum/Cockayne syndrome complex complementation group B	610651
ERCC4	Xeroderma pigmentosum/Cockayne syndrome complex complementation group F	278760
ERCC5	Xeroderma pigmentosum/Cockayne syndrome complex complementation group G	278780
ERCC6	Cockayne syndrome type B	133540
ERCC6	COFS syndrome 1	214150
ERCC8	Cockayne syndrome type A	216400
ESCO2	Roberts syndrome	269000
ESPN	Deafness, autosomal recessive 36	609006
ESPN	Deafness, neurosensory, without vestibular involvement, autosomal dominant	
ESRRB	Deafness, autosomal recessive 35	608565
ETFA	Glutaric acidemia type 2 (gene ETFA)	231680
ETFB	Glutaric acidemia type 2 (gene ETFB)	231680
ETFDH	Glutaric acidemia type 2 (gene ETFDH)	231680
ETHE1	Ethylmalonic encephalopathy	602473
EVC	Ellis-van Creveld syndrome	225500
EVC	Weyers acrodental dysostosis	193530
EVC2	Ellis-van Creveld syndrome	225500
EXOSC3	Pontocerebellar hypoplasia, type 1B	614678
EYS	Retinitis pigmentosa 25	602772
F11	Factor XI deficiency, autosomal dominant	612416
F11	Factor XI deficiency, autosomal recessive	612416
F2	Dysprothrombinemia	613679

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

F2	Hypoprothrombinemia	613679
F2	Pregnancy loss, recurrent, susceptibility to, 2	614390
F2	Stroke, ischemic, susceptibility to	601367
F2	Thrombophilia due to thrombin defect	188050
F5	Budd-Chiari syndrome	600880
F5	Factor V deficiency	227400
F5	Pregnancy loss, recurrent, susceptibility to, 1	614389
F5	Stroke, ischemic, susceptibility to	601367
F5	Thrombophilia due to activated protein C resistance	188055
F5	Thrombophilia, susceptibility to, due to factor V Leiden	188055
F8	Hemophilia A	306700
F9	Hemophilia B	306900
FAH	Tyrosinemia type 1	276700
FAM126A	Hypomyelination - congenital cataract	610532
FAM161A	Retinitis pigmentosa 28	606068
FAM20C	Lethal osteosclerotic bone dysplasia	259775
FANCA	Fanconi anemia, complementation group A	227650
FANCB	Fanconi anemia, complementation group B	300514
FANCC	Fanconi anemia complementation group C	227645
FANCD2	Fanconi anemia, complementation group D2	227646
FANCE	Fanconi anemia, complementation group E	600901
FANCG	Fanconi anemia, complementation group G	614082
FANCI	Fanconi anemia, complementation group I	609053
FANCL	Fanconi anemia, complementation group L	614083
FANCM	Premature ovarian failure 15	618096
FANCM	Spermatogenic failure 28	618086
FAS	Autoimmune lymphoproliferative syndrome, type IA	601859
FASLG	Autoimmune lymphoproliferative syndrome, type IB	601859
FASTKD2	Mitochondrial complex IV deficiency	220110
FBLN5	Cutis laxa, autosomal dominant 2	614434
FBLN5	Cutis laxa, autosomal recessive, type IA	219100

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FBLN5	Macular degeneration, age-related, 3	608895
FERMT3	Leukocyte adhesion deficiency, type III	612840
FGA	Congenital fibrinogen deficiency (gene FGA)	202400
FGB	Afibrinogenemia, congenital/Hypofibrinogenemia, congenital	202400
FGB	Dysfibrinogenemia, congenital	616004
FGD1	Aarskog-Scott syndrome	305400
FGD1	Mental retardation, X-linked syndromic 16	305400
FGD4	Charcot-Marie-Tooth disease type 4H	609311
FH	Fumaric aciduria	606812
FHL1	Uruguay faciocardiomusculoskeletal syndrome	300280
FHL1	Emery-Dreifuss muscular dystrophy 6, X-linked	300696
FHL1	Myopathy, X-linked, with postural muscle atrophy	300696
FHL1	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset	300717
FHL1	Reducing body myopathy, X-linked 1b, with late childhood or adult onset	300718
FHL1	Scapuloperoneal myopathy, X-linked dominant	300695
FIG4	Amyotrophic lateral sclerosis 11	612577
FIG4	Charcot-Marie-Tooth disease, type 4J	611228
FIG4	Polymicrogyria, bilateral temporooccipital	612691
FIG4	Yunis-Varon syndrome	216340
FKRP	Autosomal recessive limb-girdle muscular dystrophy type 2I	607155
FKRP	Congenital muscular dystrophy type 5B	606612
FKRP	Muscle-eye-brain disease	613153
FKTN	Autosomal recessive limb-girdle muscular dystrophy type 2M	611588
FKTN	Congenital muscular dystrophy type 4B	613152
FKTN	Fukuyama congenital muscular dystrophy	253800
FLNA	Cardiac valvular dysplasia, X-linked	314400
FLNA	Congenital short bowel syndrome	300048
FLNA	FG syndrome 2	300321
FLNA	Frontometaphyseal dysplasia 1	305620
FLNA	Heterotopia, periventricular, 1	300049
FLNA	Intestinal pseudoobstruction, neuronal	300048

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FLNA	Melnick-Needles syndrome	309350
FLNA	Otopalatodigital syndrome, type I	311300
FLNA	Otopalatodigital syndrome, type II	304120
FLNA	Terminal osseous dysplasia	300244
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa	609033
FMR1	Fragile X syndrome	300624
FMR1	Fragile X tremor/ataxia syndrome	300623
FMR1	Premature ovarian failure 1	311360
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	613068
FOXG1	Rett syndrome, congenital variant	613454
FOXN1	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy	601705
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	304790
FRAS1	Fraser syndrome (gene FRAS1)	219000
FREM2	Fraser syndrome (gene FRAS2)	219000
FTCD	Glutamate formiminotransferase deficiency	229100
FTSJ1	Mental retardation, X-linked 9	309549
FUCA1	Fucosidosis	230000
FXN	Friedreich ataxia	229300
FXN	Friedreich ataxia with retained reflexes	229300
G6PC	Glycogen storage disease due to glucose-6-phosphatase deficiency type 1a	232200
G6PC3	Síndrome de Dursun	612541
G6PD	Favism	134700
G6PD	Hemolytic anemia due to G6PD deficiency	300908
GAA	Glycogen storage disease due to acid maltase deficiency	232300
GALC	Krabbe disease	245200
GALE	Galactose epimerase deficiency	230350
GALK1	Galactokinase deficiency with cataracts	230200
GALNS	Mucopolysaccharidosis IVA	253000
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1	211900
GALT	Classic galactosemia	230400
GAMT	Guanidinoacetate methyltransferase deficiency	612736

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GAN	Giant axonal neuropathy-1	256850
GBA	Fetal Gaucher disease	608013
GBA	Gaucher disease type 2	230900
GBA	Gaucher disease type 3	231000
GBA	Gaucher disease type 3C	231005
GBE1	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form	232500
GCDH	Glutaryl-CoA dehydrogenase deficiency	231670
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia	128230
GCH1	Hyperphenylalaninemia, BH4-deficient, B	233910
GCSH	Glycine encephalopathy	605899
GDAP1	Autosomal dominant Charcot-Marie-Tooth disease type 2K	607831
GDAP1	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness	607706
GDAP1	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A	608340
GDAP1	Charcot-Marie-Tooth disease type 4A	214400
GDF5	Acromesomelic dysplasia, Hunter-Thompson type	201250
GDF5	Brachydactyly, type A1, C	615072
GDF5	Brachydactyly, type A2	112600
GDF5	Brachydactyly, type C	113100
GDF5	Chondrodysplasia, Grebe type	200700
GDF5	Du Pan syndrome	228900
GDF5	Multiple synostoses syndrome 2	610017
GDF5	Osteoarthritis-5	612400
GDF5	Symphalangism, proximal, 1B	615298
GDI1	Mental retardation, X-linked 41	300849
GFM1	Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1	609060
GHRHR	Growth hormone deficiency, isolated, type IV	618157
GJA1	Atrioventricular septal defect 3	600309
GJA1	Craniometaphyseal dysplasia, autosomal recessive	218400
GJA1	Erythrokeratodermia variabilis et progressiva 3	617525
GJA1	Hypoplastic left heart syndrome 1	241550

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GJA1	Oculodentodigital dysplasia	164200
GJA1	Oculodentodigital dysplasia, autosomal recessive	257850
GJA1	Palmoplantar keratoderma with congenital alopecia	104100
GJA1	Syndactyly, type III	186100
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	302800
GJB2	Autosomal recessive nonsyndromic sensorineural deafness type DFNB1A (gene GJB2)	220290
GJB3	Deafness, autosomal dominant 2B	612644
GJB3	Deafness, autosomal dominant, with peripheral neuropathy	
GJB3	Deafness, autosomal recessive	
GJB3	Deafness, digenic, GJB2/GJB3	220290
GJB3	Erythrokeratodermia variabilis et progressiva 1	133200
GJB6	Deafness, autosomal dominant 3B	612643
GJB6	Deafness, autosomal recessive 1B	612645
GJB6	Deafness, digenic GJB2/GJB6	220290
GJB6	Ectodermal dysplasia 2, Clouston type	129500
GJC2	Pelizaeus-Merzbacher-like due to GJC2 mutation	608804
GK	Glycerol kinase deficiency	307030
GLA	Fabry disease	301500
GLB1	GM1 gangliosidosis type 1	230500
GLB1	GM1 gangliosidosis type 2	230600
GLB1	GM1 gangliosidosis type 3	230650
GLB1	Mucopolysaccharidosis type 4B	253010
GLDC	Glycine encephalopathy	605899
GLE1	Lethal congenital contracture syndrome type 1	253310
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism	610199
GM2A	GM2-gangliosidosis, AB variant	272750
GNAS	ACTH-independent macronodular adrenal hyperplasia	219080
GNAS	McCune-Albright syndrome, somatic, mosaic	174800
GNAS	Osseous heteroplasia, progressive	166350
GNAS	Pituitary adenoma 3, multiple types, somatic	617686
GNAS	Pseudohypoparathyroidism Ia	103580

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

GNAS	Pseudohypoparathyroidism Ib	603233
GNAS	Pseudohypoparathyroidism Ic	612462
GNAS	Pseudopseudohypoparathyroidism	612463
GNE	Nonaka myopathy	605820
GNE	Sialuria	269921
GNMT	Glycine N-methyltransferase deficiency	606664
GNPTAB	Mucolipidosis type 2	252500
GNPTAB	Mucolipidosis type 3	252600
GNPTG	Mucolipidosis III gamma	252605
GNRHR	Fertile eunuch syndrome	228300
GNRHR	Hypogonadotropic hypogonadism 7 without anosmia	146110
GNS	Mucopolysaccharidosis type IIID	252940
GORAB	Geroderma osteodysplasticum	231070
GP1BA	Bernard-Soulier syndrome, type A1 (recessive)	231200
GP1BA	Bernard-Soulier syndrome, type A2 (dominant)	153670
GP1BA	Nonarteritic anterior ischemic optic neuropathy, susceptibility to	258660
GP1BA	von Willebrand disease, platelet-type	177820
GP1BB	Bernard-Soulier syndrome, type B	231200
GP1BB	Giant platelet disorder, isolated	231200
GP9	Bernard-Soulier syndrome, type C	231200
GPC3	Simpson-Golabi-Behmel syndrome, type 1	312870
GPR143	Nystagmus 6, congenital, X-linked	300814
GPR143	Ocular albinism, type I, Nettleship-Falls type	300500
GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive	614565
GPR98	Usher syndrome type 2C	605472
GRHPR	Hyperoxaluria, primary, type II	260000
GRIA3	Mental retardation, X-linked 94	300699
GRIK2	Mental retardation, autosomal recessive, 6	611092
GRM6	Night blindness, congenital stationary (complete), 1B, autosomal recessive	257270
GRXCR1	Deafness, autosomal recessive 25	613285
GSS	Glutathione synthetase deficiency with 5-oxoprolinuria	266130

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GTF2H5	Trichothiodystrophy, complementation group A	601675
GUCY2D	Choroidal dystrophy, central areolar 1	215500
GUCY2D	Cone-rod dystrophy 6	601777
GUCY2D	Leber congenital amaurosis 1	204000
GUSB	Mucopolysaccharidosis type 7	253220
HADH	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	609016
HADHA	Mitochondrial trifunctional protein deficiency	609015
HADHB	Mitochondrial trifunctional protein deficiency	609015
HAL	Histidinemia	235800
HAMP	Histidinemia	235800
HAX1	Neutropenia, severe congenital 3, autosomal recessive	610738
HBA1	Alpha-thalassemia	604131
HBA2	Erythrocytosis 7	617981
HBA2	Heinz body anemia	140700
HBA2	Hemoglobin H disease, deletional and nondeletional	613978
HBA2	Thalassemia, alpha-	604131
HBB	Beta-thalassemia	613985
HBB	Sickle cell anemia	603903
HCCS	Linear skin defects with multiple congenital anomalies 1	309801
HESX1	Combined pituitary hormone deficiencies, genetic forms	182230
HEXA	Tay-Sachs disease	272800
HEXB	Sandhoff disease	268800
HFE	Alzheimer disease, susceptibility to	104300
HFE	Hemochromatosis	235200
HFE	Microvascular complications of diabetes 7	612635
HFE	Porphyria cutanea tarda, susceptibility to	176100
HFE	Porphyria variegata, susceptibility to	176200
HFE	Transferrin serum level QTL2	614193
HFE2	Hemochromatosis, type 2A	602390
HGD	Alkaptonuria	203500
HGF	Deafness, autosomal recessive 39	608265

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HGSNAT	Sanfilippo syndrome type C	252930
HIBCH	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency	250620
HLCS	Holocarboxylase synthetase deficiency	253270
HMGCL	3-hydroxy-3-methylglutaric aciduria	246450
HMOX1	Heme oxygenase-1 deficiency	614034
HMOX1	Pulmonary disease, chronic obstructive, susceptibility to	606963
HOGA1	Hyperoxaluria, primary, type III	613616
HP	Anhaptoglobinemia	614081
HP	Hypohaptoglobinemia	614081
HPD	Tyrosinemia type 3	276710
HPRT1	Kelley-Seegmiller syndrome	300323
HPRT1	Lesch-Nyhan syndrome	300322
HPS1	Hermansky-Pudlak syndrome 1	203300
HPS3	Hermansky-Pudlak syndrome 3	614072
HSD11B2	Apparent mineralocorticoid excess	218030
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency	300438
HSD17B3	Pseudohermaphroditism, male, with gynecomastia	264300
HSD17B4	Bifunctional enzyme deficiency	261515
HSD17B4	Perrault syndrome	233400
HSD3B2	3-beta-hydroxysteroid dehydrogenase, type II, deficiency	201810
HSPD1	Leukodystrophy, hypomyelinating, 4	612233
HSPD1	Spastic paraplegia 13, autosomal dominant	605280
HSPG2	Schwartz-Jampel syndrome	255800
HTRA1	CARASIL syndrome	600142
HTRA1	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2	616779
HTRA1	Macular degeneration, age-related, 7	610149
HTRA1	Macular degeneration, age-related, neovascular type	610149
HUWE1	Mental retardation, X-linked syndromic, Turner type	300706
HYAL1	Mucopolysaccharidosis type IX	601492
HYLS1	Hydrolethalus syndrome	236680
ICOS	Immunodeficiency, common variable, 1	607594

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IDH3B	Retinitis pigmentosa 46	612572
IDS	Mucopolysaccharidosis type 2	309900
IDUA	Mucopolysaccharidosis Ih	607014
IDUA	Mucopolysaccharidosis Ih/s	607015
IDUA	Mucopolysaccharidosis Is	607016
IFNGR1	Immunodeficiency 27A, mycobacteriosis, AR	209950
IFNGR2	Immunodeficiency 28, mycobacteriosis	614889
IFT80	Jeune syndrome	611263
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia	300472
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency	608747
IGHMBP2	Spinal muscular atrophy with respiratory distress	604320
IKBKAP	Familial dysautonomia	223900
IKBKG	Ectodermal dysplasia, hypohidrotic, with immune deficiency	300291
IKBKG	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency	300301
IKBKG	Immunodeficiency 33	300636
IKBKG	Incontinentia pigmenti, type II	308300
IL12B	Immunodeficiency 29, mycobacteriosis	614890
IL12RB1	Immunodeficiency 30	614891
IL1RAPL1	Mental retardation, X-linked 21/34	300143
IL1RN	Interleukin 1 receptor antagonist deficiency	612852
IL2RA	Diabetes, mellitus, insulin-dependent, susceptibility to, 10	601942
IL2RA	Immunodeficiency 41 with lymphoproliferation and autoimmunity	606367
IL2RG	T-B+ severe combined immunodeficiency due to gamma chain deficiency	300400
IL2RG	T-B+ severe combined immunodeficiency, X-linked	312863
IMPDH1	Leber congenital amaurosis 11	613837
IMPDH1	Retinitis pigmentosa 10	180105
IMPG2	Macular dystrophy, vitelliform, 5	616152
IMPG2	Retinitis pigmentosa 56	613581
INPP5E	Joubert syndrome 1	213300
INPP5E	Mental retardation, truncal obesity, retinal dystrophy, and micropenis	610156
INSR	Leprechaunism	246200

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

INVS	Nephronophthisis 2, infantile	602088
IQCB1	Senior-Loken syndrome 5	609254
IQSEC2	Mental retardation, X-linked 1/78	309530
ISCU	Myopathy with lactic acidosis, hereditary	255125
ITGA6	Junctional epidermolysis bullosa - pyloric atresia	226730
ITGB4	Junctional epidermolysis bullosa with piloric atresia	226730
ITGB4	Junctional epidermolysis bullosa, non-Herlitz type	226650
IVD	Isovaleric acidemia	243500
IYD	Thyroid dyshormonogenesis 4	274800
JAK3	T-B+ severe combined immunodeficiency due to JAK3 deficiency	600802
KCNJ1	Antenatal Bartter syndrome	241200
KCNJ11	Diabetes mellitus, transient neonatal, 3	610582
KCNJ11	Diabetes mellitus, type 2, susceptibility to	125853
KCNJ11	Diabetes, permanent neonatal, with or without neurologic features	606176
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2	601820
KCNJ11	Maturity-onset diabetes of the young, type 13	616329
KCNJ13	Leber congenital amaurosis 16	614186
KCNJ13	Snowflake vitreoretinal degeneration	193230
KCNV2	Retinal cone dystrophy 3B	610356
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type	300534
KIAA2022	Mental retardation, X-linked 98	300912
KIF7	Acrocallosal syndrome	200990
KIF7	Al-Gazali-Bakalinova syndrome	607131
KIF7	Hydrolethalus syndrome 2	614120
KIF7	Joubert syndrome 12	200990
L1CAM	Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity-hydrocephalus syndrome	307000
L1CAM	Masa syndrome	303350
LAMA2	Congenital muscular dystrophy type 1A	607855
LAMA3	Junctional epidermolysis bullosa, Herlitz type (gene LAMA3)	226700
LAMA3	Junctional epidermolysis bullosa, Herlitz type (gene LAMB3)	226700
LAMA3	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMA3)	226650
LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities	614199

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LAMB2	Pierson syndrome	609049
LAMB3	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMB3)	226650
LAMC2	Junctional epidermolysis bullosa, Herlitz type (gene LAMC2)	226700
LAMC2	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMC2)	226650
LAMP2	Danon disease	300257
LARGE	Congenital muscular dystrophy type 1D	608840
LARGE	Muscle-eye-brain disease	613154
LBR	Greenberg dysplasia	215140
LCA5	Leber congenital amaurosis 5	604537
LDHA	Glycogen storage disease XI	612933
LDLR	Hypercholesterolemia, familial, 1	143890
LDLR	LDL cholesterol level QTL2	143890
LDLRAP1	Hypercholesterolemia, familial, 4	603813
LEPRE1	Osteogenesis imperfecta type 8	610915
LHCGR	Leydig cell adenoma, somatic, with precocious puberty	176410
LHCGR	Leydig cell hypoplasia with hypergonadotropic hypogonadism	238320
LHCGR	Leydig cell hypoplasia with pseudohermaphroditism	238320
LHCGR	Luteinizing hormone resistance, female	238320
LHCGR	Precocious puberty, male	176410
LHFPL5	Deafness, autosomal recessive 67	610265
LHX3	Combined pituitary hormone deficiency with spine abnormalities	221750
LIFR	Stüve-Wiedemann syndrome	601559
LIG4	Severe combined immunodeficiency with sensitivity to ionizing radiation	602450
LIPA	Cholesteryl ester storage disease	278000
LIPA	Wolman disease	278000
LIPH	Hypotrichosis 7	604379
LIPH	Woolly hair, autosomal recessive 2 with or without hypotrichosis	604379
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	277380
LMNA	Charcot-Marie-Tooth disease axonal type 2B1	605588
LMNA	Lethal restrictive dermopathy	275210
LMNA	Mandibuloacral dysplasia with type A lipodystrophy	248370
LOXHD1	Deafness, autosomal recessive 77	613079
LPL	Combined hyperlipidemia, familial	144250

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LPL	High density lipoprotein cholesterol level QTL 11	238600
LPL	Lipoprotein lipase deficiency	238600
LRAT	Leber congenital amaurosis 14	613341
LRP2	Retinal dystrophy, early-onset severe	613341
LRP2	Retinal dystrophy, early-onset severe	613341
LRP2	Retinitis pigmentosa, juvenile	613341
LRP2	Donnai-Barrow syndrome	222448
LRP5	Bone mineral density variability 1	601884
LRP5	Exudative vitreoretinopathy 4	601813
LRP5	Hyperostosis, endosteal	144750
LRP5	Osteopetrosis, autosomal dominant 1	607634
LRP5	Osteoporosis	166710
LRP5	Osteoporosis-pseudoglioma syndrome	259770
LRP5	Osteosclerosis	144750
LRP5	Polycystic liver disease 4 with or without kidney cysts	617875
LRP5	van Buchem disease, type 2	607636
LRPPRC	French-Canadian type Leigh syndrome	220111
LRTOMT	Deafness, autosomal recessive 63	611451
LYST	Chediak-Higashi syndrome	214500
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia	300853
MAK	Retinitis pigmentosa 62	614181
MAN2B1	Mannosidosis, alpha-, types I and II	248500
MARVELD2	Deafness, autosomal recessive 49	610153
MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency	250850
MAT1A	Methionine adenosyltransferase deficiency, autosomal recessive	250850
MATN3	Epiphyseal dysplasia, multiple, 5	607078
MATN3	Osteoarthritis susceptibility 2	140600
MATN3	Spondyloepimetaphyseal dysplasia	608728
MBTPS2	Ichthyosis follicularis - alopecia - photophobia	308205
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	210200
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	210210

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MCEE	Methylmalonyl-CoA epimerase deficiency	251120
MCOLN1	Mucolipidosis type 4	252650
MCPH1	Microcephaly 1, primary, autosomal recessive	251200
MECP2	Severe neonatal-onset encephalopathy with microcephaly	300673
MED12	X-linked intellectual deficit with marfanoid habitus	309520
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	613668
MED25	Basel-Vanagait-Smirin-Yosef syndrome	616449
MED25	Charcot-Marie-Tooth disease, type 2B2	605589
MEFV	Familial Mediterranean fever	249100
MERTK	Retinitis pigmentosa 38	613862
MESP2	Spondylocostal dysostosis 2, autosomal recessive	608681
MFRP	Microphthalmia, isolated 5	611040
MFSD8	Nanophthalmos 2	609549
MFSD8	Nanophthalmos 2	609549
MFSD8	Late infantile neuronal ceroid lipofuscinosis	610951
MGAT2	Congenital disorder of glycosylation type 2a	212066
MID1	Opitz GBBB syndrome, type I	300000
MKKS	Bardet-Biedl syndrome 6	605231
MKS1	McKusick-Kaufman syndrome	236700
MKS1	McKusick-Kaufman syndrome	236700
MKS1	Meckel syndrome type 1	249000
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	604004
MLYCD	Malonyl-CoA decarboxylase deficiency	248360
MMAA	Vitamin B12-responsive methylmalonic acidemia type cblA	251100
MMAB	Vitamin B12-responsive methylmalonic acidemia type cblB	251110
MMACHC	Methylmalonic acidemia with homocystinuria, type cblC	277400
MMACHC	Methylmalonic acidemia with homocystinuria, type cblD	277410
MMADHC	Homocystinuria, cblD type, variant 1	277410
MOCS1	Methylmalonic aciduria and homocystinuria, cblD type	277410
MOCS1	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A (gene MOCS1)	252150
MOCS2	Methylmalonic aciduria and homocystinuria, cblD type	277410
MOCS2	Methylmalonic aciduria, cblD type, variant 2	277410

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MOCS2	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A (gene MOCS2)	252150
MOGS	Congenital disorder of glycosylation, type IIb	606056
MPDU1	Congenital disorder of glycosylation, type If	609180
MPI	Congenital disorder of glycosylation type 1b	602579
MPL	Thrombocythemia 2	601977
MPL	Thrombocytopenia, congenital amegakaryocytic	604498
MPV17	Methylmalonic aciduria, cblD type, variant 2	277410
MPV17	Methylmalonic aciduria, cblD type, variant 2	277410
MPV17	Navajo neurohepatopathy	256810
MPZ	Charcot-Marie-Tooth disease, type 1B	118200
MPZ	Charcot-Marie-Tooth disease, type 2I	607677
MPZ	Charcot-Marie-Tooth disease, type 2J	607736
MPZ	Dejerine-Sottas disease	145900
MPZ	Neuropathy, congenital hypomyelinating	605253
MPZ	Roussy-Levy syndrome	180800
MRE11	Ataxia-telangiectasia-like disorder 1	604391
MRPS16	Combined oxidative phosphorylation defect type 2	610498
MRPS22	Combined oxidative phosphorylation defect type 5	611719
MTHFR	Homocystinuria due to MTHFR deficiency	236250
MTM1	X-linked centronuclear myopathy	310400
MTMR2	Charcot-Marie-Tooth disease, type 4B1	601382
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type	250940
MTR	Neural tube defects, folate-sensitive, susceptibility to	601634
MTRR	Homocystinuria-megaloblastic anemia, cbl E type	236270
MTRR	Neural tube defects, folate-sensitive, susceptibility to	601634
MTTP	Abetalipoproteinemia	200100
MUT	Metabolic syndrome, protection against	605552
MUT	Metabolic syndrome, protection against	605552
MUT	Vitamin B12-unresponsive methylmalonic acidemia type mut-	251000
MVK	Mevalonic aciduria	610377
MYD88	Macroglobulinemia, Waldenstrom	153600
MYD88	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	612260

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MYO15A	Deafness, autosomal recessive 3	600316
MYO3A	Deafness, autosomal recessive 30	607101
MYO5A	Griscelli disease type 1	214450
MYO6	Deafness, autosomal dominant 22	606346
MYO7A	Autosomal recessive nonsyndromic sensorineural deafness type DFNB2	600060
MYO7A	Usher syndrome type 1	276900
NAGA	Kanzaki disease	609242
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)	252920
NAGS	Hyperammonemia due to N-acetylglutamate synthetase deficiency	237310
NBN	Aplastic anemia	609135
NBN	Nijmegen breakage syndrome	251260
NDP	Exudative vitreoretinopathy 2, X-linked	305390
NDP	Norrie disease	310600
NDRG1	Charcot-Marie-Tooth disease, type 4D	601455
NDUFA1	Mitochondrial complex I deficiency, nuclear type 12	301020
NDUFA1	Mitochondrial complex I deficiency	252010
NDUFA7	Mitochondrial complex I deficiency, nuclear type 12	301020
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 12	301020
NDUFAF2	Leigh syndrome	256000
NDUFAF2	Mitochondrial complex I deficiency	252010
NDUFAF4	Mitochondrial complex I deficiency	252010
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16	618238
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency	256000
NDUFS3	Mitochondrial complex I deficiency	252010
NDUFS4	Leigh syndrome	256000
NDUFS4	Mitochondrial complex I deficiency	252010
NDUFS5	NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 5	603847
NDUFS6	Complex I, mitochondrial respiratory chain, deficiency of	252010
NDUFS7	Leigh syndrome	256000
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency	256000
NDUFV1	Mitochondrial complex I deficiency	252010
NEB	Nemaline myopathy 2	256030
NEFL	Charcot-Marie-Tooth disease, dominant intermediate G	617882

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NEU1	Sialidosis, type I	256550
NEU1	Sialidosis, type II	256550
NEUROG 3	Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells	610370
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611291
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora)	254780
NHP2	Dyskeratosis congenita, autosomal recessive 2	613987
NHS	Cataract 40, X-linked	302200
NHS	Nance-Horan syndrome	302350
NKX2-1	Chorea, hereditary benign	118700
NKX2-1	Choreoathetosis, hypothyroidism, and neonatal respiratory distress	610978
NKX2-1	Thyroid cancer, nonmedullary, 1	188550
NKX2-5	Atrial septal defect 7, with or without AV conduction defects	108900
NKX2-5	Conotruncal heart malformations, variable	217095
NKX2-5	Hypoplastic left heart syndrome 2	614435
NKX2-5	Hypothyroidism, congenital nongoitrous, 5	225250
NKX2-5	Tetralogy of Fallot	187500
NKX2-5	Ventricular septal defect 3	614432
NLGN3	Asperger syndrome susceptibility, X-linked 1	300494
NLGN3	Autism susceptibility, X-linked 1	300425
NLGN4X	Mental retardation, X-linked	300495
NLRP7	Hydatidiform mole, recurrent, 1	231090
NMNAT1	Leber congenital amaurosis 9	608553
NOP10	Dyskeratosis congenita, autosomal recessive 1	224230
NPC1	Niemann-Pick disease type C1	257220
NPC2	Niemann-Pick disease type C2	607625
NPHP1	Joubert syndrome 4	609583
NPHP3	Renal-hepatic-pancreatic dysplasia	208540
NPHP3	Senior-Loken syndrome 1	266900
NPHP4	Senior-Loken syndrome	606996
NPHS1	Nephrotic syndrome, type 1	256300
NPHS2	Nephrotic syndrome, type 2	600995

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NR0B1	46XY sex reversal 2, dosage-sensitive	300018
NR2E3	Enhanced S-cone syndrome	268100
NR2E3	Retinitis pigmentosa 37	611131
NR5A1	46XY sex reversal 3	612965
NR5A1	Adrenocortical insufficiency	612965
NSD1	Beckwith-Wiedemann syndrome	130650
NSD1	Sotos syndrome 1	117550
NSDHL	CHILD syndrome	308050
NSDHL	CK syndrome	300831
NSUN2	Mental retardation, autosomal recessive 5	611091
NTRK1	Hereditary sensory and autonomic neuropathy type 4	256800
NUP62	Infantile bilateral striatal necrosis	271930
NXF5	NUCLEAR RNA EXPORT FACTOR 5	300319
NYX	Night blindness, congenital stationary (complete), 1A, X-linked	310500
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia	258870
OCA2	Albinism, brown oculocutaneous	203200
OCA2	Albinism, oculocutaneous, type II	203200
OCA2	Skin/hair/eye pigmentation 1, blond/brown hair	227220
OCA2	Skin/hair/eye pigmentation 1, blue/nonblue eyes	227220
OCRL	Dent disease 2	300555
OCRL	Oculocerebrorenal syndrome	309000
OFD1	Simpson-Golabi-Behmel syndrome type 2	300209
OPA3	3-methylglutaconic aciduria type 3	258501
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	300486
ORAI1	Immunodeficiency 9	612782
ORAI1	Myopathy, tubular aggregate, 2	615883
OSTM1	Osteopetrosis, autosomal recessive 5	259720
OTC	Ornithine transcarbamylase deficiency	311250
OTOA	Deafness, autosomal recessive 22	607039
OTOF	Auditory neuropathy, autosomal recessive, 1	601071
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency	245050
PAH	Phenylketonuria	261600

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PAK3	Mental retardation, X-linked 30/47	300558
PALB2	Fanconi anemia, complementation group N	610832
PANK2	Pantothenate kinase-associated neurodegeneration	234200
PAX3	Craniofacial-deafness-hand syndrome	122880
PAX3	Rhabdomyosarcoma 2, alveolar	268220
PAX3	Waardenburg syndrome, type 1	193500
PAX3	Waardenburg syndrome, type 3	148820
PAX6	Aniridia	106210
PAX6	Anterior segment dysgenesis 5, multiple subtypes	604229
PAX6	Cataract with late-onset corneal dystrophy	106210
PAX6	Coloboma of optic nerve	120430
PAX6	Coloboma, ocular	120200
PAX6	Foveal hypoplasia 1	136520
PAX6	Keratitis	148190
PAX6	Morning glory disc anomaly	120430
PAX6	Optic nerve hypoplasia	165550
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	218700
PC	Pyruvate carboxylase deficiency	266150
PCBD1	Hyperphenylalaninemia, BH4-deficient, D	264070
PCCA	Propionic acidemia (gene PCCA)	606054
PCCB	Propionic acidemia (gene PCCB)	606054
PCDH15	Deafness, autosomal recessive 23	609533
PCDH15	Usher syndrome, type 1D/F digenic	601067
PCDH15	Usher syndrome, type 1F	602083
PCDH19	Epileptic encephalopathy, early infantile, 9	300088
PDE6A	Retinitis pigmentosa 43	613810
PDE6B	Night blindness, congenital stationary, autosomal dominant 2	163500
PDE6B	Retinitis pigmentosa-40	613801
PDE6C	Cone dystrophy 4	613093
PDE6G	Retinitis pigmentosa 57	613582
PDHA1	Leigh syndrome, X-linked	308930
PDHB	Pyruvate dehydrogenase E1-beta deficiency	614111
PDHX	Lacticacidemia due to PDX1 deficiency	245349

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PDP1	Pyruvate dehydrogenase phosphatase deficiency	608782
PDSS1	Deafness - encephaloneuropathy - obesity - valvulopathy	614651
PDSS2	Leigh syndrome with nephrotic syndrome	614652
PDX1	Diabetes mellitus, type II, susceptibility to	125853
PDX1	MODY, type IV	606392
PDX1	Pancreatic agenesis 1	260370
PDZD7	Deafness, autosomal recessive 57	618003
PDZD7	Retinal disease in Usher syndrome type IIA, modifier of	276901
PDZD7	Usher syndrome, type IIC, GPR98/PDZD7 digenic	605472
PEPD	Prolidase deficiency	170100
PEX1	Zellweger syndrome 1A	214100
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)	614870
PEX10	Peroxisome biogenesis disorder 6B	614871
PEX12	Neonatal adrenoleukodystrophy (gene PEX12)	266510
PEX13	Peroxisome biogenesis disorder 11A (Zellweger)	614883
PEX13	Peroxisome biogenesis disorder 11B	614885
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)	614866
PEX26	Peroxisome biogenesis disorder 5B	614867
PEX26	Peroxisome biogenesis disorder 5B	614867
PEX26	Neonatal adrenoleukodystrophy (gene PEX26)	614873
PEX26	Zellweger syndrome 7A	614872
PEX5	Neonatal adrenoleukodystrophy (gene PEX5)	202370
PEX6	Heimler syndrome 2	616617
PEX7	Peroxisome biogenesis disorder 4A (Zellweger)	614862
PEX7	Peroxisome biogenesis disorder 4B	614863
PEX7	Rhizomelic chondrodysplasia punctata type 1	215100
PFKM	Glycogen storage disease VII	232800
PGK1	Phosphoglycerate kinase 1 deficiency	300653
PGM1	Congenital disorder of glycosylation, type It	614921
PHF8	Mental retardation syndrome, X-linked, Siderius type	300263
PHGDH	Neu-Laxova syndrome 1	256520
PHGDH	Phosphoglycerate dehydrogenase deficiency	601815
PHKG2	Cirrhosis due to liver phosphorylase kinase deficiency	

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

PHKG2	Glycogen storage disease IXc	613027
PHYH	Refsum disease	266500
PKHD1	Autosomal recessive polycystic kidney disease	263200
PKLR	Hemolytic anemia due to red cell pyruvate kinase deficiency	266200
PLA2G6	Infantile neuroaxonal dystrophy 2A	256600
PLA2G6	Infantile neuroaxonal dystrophy 2B	610217
PLCE1	Nephrotic syndrome, tupe 3	610725
PLDN	Hermansky-pudlak syndrome 9	614171
PLEC	Epidermolysis bullosa simplex with muscular dystrophy	226670
PLEC	Epidermolysis bullosa simplex with pyloric atresia	612138
PLEC	Limb girdle dystrophy with epidermolysis bullosa simplex	613723
PLEKHG5	Autosomal recessive distal spinal muscular atrophy type 4	611067
PLG	Plasminogen deficiency type 1	217090
PLOD1	Ehlers-Danlos syndrome type 6	225400
PLP1	Spastic paraplegia type 2, X-linked	312920
PMM2	Congenital disorder of glycosylation type 1a	212065
PMP22	Charcot-Marie-Tooth disease, type 1A	118220
PMP22	Charcot-Marie-Tooth disease, type 1E	118300
PMP22	Dejerine-Sottas disease	145900
PMP22	Roussy-Levy syndrome	180800
PNPO	Pyridoxal phosphate-responsive seizures	610090
POLG	Alpers syndrome	203700
POLG	Autosomal recessive progressive external ophthalmoplegia	258450
POLG	Mitochondrial neurogastrointestinal encephalomyopathy	613662
POLG	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis	607459
POLR1C	Leukodystrophy, hypomyelinating, 11	616494
POMGNT1	Treacher Collins syndrome 3	248390
POMGNT1	Treacher Collins syndrome 3	248390
POMGNT1	Autosomal recessive limb-girdle muscular dystrophy type C	613157
POMGNT1	Congenital muscular dystrophy with cerebellar involvement	613151
POMGNT1	Walker-Warburg syndrome (gene POMGNT1)	253280

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POMT1	Autosomal recessive limb-girdle muscular dystrophy type C	609308
POMT1	Congenital muscular dystrophy with cerebellar involvement	613155
POMT1	Walker-Warburg syndrome (gene POMT1)	236670
POMT2	Autosomal recessive limb-girdle muscular dystrophy type C	613158
POMT2	Congenital muscular dystrophy with cerebellar involvement	613156
POMT2	Walker-Warburg syndrome (gene POMT2)	613150
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	201750
POU1F1	Combined pituitary hormone deficiencies, genetic forms	613038
POU3F4	Deafness, X-linked 2	304400
PPT1	Adult neuronal ceroid lipofuscinosis	256730
PQBP1	Renpenning syndrome	309500
PRCD	Retinitis pigmentosa 36	610599
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	603553
PRKRA	Dystonia 16	612067
PRODH	Hyperprolinemia, type I	239500
PRODH	Schizophrenia, susceptibility to, 4	600850
PROM1	Cone-rod dystrophy 12	612657
PROP1	Macular dystrophy, retinal, 2	608051
PROP1	Combined pituitary hormone deficiencies, genetic forms	262600
PRPS1	Retinitis pigmentosa 41	612095
PRPS1	Lethal ataxia with deafness and optic atrophy	301835
PRPS1	X-linked Charcot-Marie-Tooth disease type 5	311070
PRSS12	Macular dystrophy, retinal, 2	608051
PRSS12	Retinitis pigmentosa 41	612095
PRSS12	Stargardt disease 4	603786
PRSS12	Stargardt disease 4	603786
PRSS12	Mental retardation, autosomal recessive 1	249500
PRX	Charcot-Marie-Tooth disease type 4F	614895
PSAP	Encephalopathy due to prosaposin deficiency	611721
PSAP	Krabbe disease	611722
PSAP	Metachromatic leukodystrophy	249900
PSAT1	Phosphoserine aminotransferase deficiency	610992

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PTEN	Neu-Laxova syndrome 2	616038
PTEN	Neu-Laxova syndrome 2	616038
PTEN	Bannayan-Riley-Ruvalcaba syndrome	153480
PTEN	Cowden syndrome 1	158350
PTEN	Lhermitte-Duclos syndrome	158350
PTEN	Macrocephaly/autism syndrome	605309
PTH1R	Chondrodysplasia, Blomstrand type	215045
PTH1R	Eiken syndrome	600002
PTH1R	Failure of tooth eruption, primary	125350
PTH1R	Metaphyseal chondrodysplasia, Murk Jansen type	156400
PTS	Hyperphenylalaninemia, BH4-deficient, A	261640
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1	600462
PYGM	Glycogen storage disease due to muscle glycogen phosphorylase deficiency	232600
QDPR	Hyperphenylalaninemia, BH4-deficient, C	261630
RAB23	Carpenter syndrome	201000
RAB27A	Griscelli disease type 2	607624
RAB39B	Mental retardation, X-linked 72	300271
RAB3GAP 1	Micro syndrome	600118
RAB3GAP 2	Cataract - intellectual deficit - hypogonadism	212720
RAD51C	Fanconi anemia, complementation group O	613390
RAG1	Breast-ovarian cancer, familial, susceptibility to, 3	613399
RAG1	Breast-ovarian cancer, familial, susceptibility to, 3	613399
RAG1	Combined immunodeficiency with skin granulomas	233650
RAG1	Omenn syndrome (gene RAG1)	603554
RAG1	Severe combined immunodeficiency due to complete RAG1/2 deficiency	601457
RAG2	Combined immunodeficiency with skin granulomas	233650
RAG2	Omenn syndrome (gene RAG2)	603554
RAG2	Severe combined immunodeficiency due to complete RAG1/2 deficiency	601457
RAPSN	Fetal akinesia deformation sequence	208150
RARS2	Pontocerebellar hypoplasia, type 6	611523
RAX	Microphthalmia, isolated 3	611038
RDH12	Leber congenital amaurosis 13	612712

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RDX	Deafness, autosomal recessive 24	611022
RELN	Lissencephaly syndrome, Norman-Roberts type	257320
REN	Hyperproreninemia	
REN	Hyperuricemic nephropathy, familial juvenile 2	613092
REN	Renal tubular dysgenesis	267430
RFT1	Congenital disorder of glycosylation, type In	612015
RGR	Retinitis pigmentosa	613769
RHO	Night blindness, congenital stationary, autosomal dominant 1	610445
RHO	Retinitis pigmentosa 4, autosomal dominant or recessive	613731
RHO	Retinitis punctata albescens	136880
RLBP1	Bothnia retinal dystrophy	607475
RLBP1	Fundus albipunctatus	136880
RLBP1	Newfoundland rod-cone dystrophy	607476
RLBP1	Retinitis punctata albescens	136880
RMRP	Anauxetic dysplasia	607095
RMRP	Cartilage-hair hypoplasia	250250
RMRP	Metaphyseal dysplasia without hypotrichosis	250460
RNASEH2A	Aicardi-Goutieres syndrome 4	610333
RNASEH2B	Aicardi-Goutieres syndrome 2	610181
RNASEH2C	Aicardi-Goutieres syndrome 3	610329
RP2	Retinitis pigmentosa 2	312600
RPE65	Leber congenital amaurosis 2	204100
RPE65	Retinitis pigmentosa 20	613794
RPGR	Cone-rod dystrophy, X-linked, 1	304020
RPGR	Macular degeneration, X-linked atrophic	300834
RPGR	Retinitis pigmentosa 3	300029
RPGR	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness	300455
RPGRIP1L	Joubert syndrome with hepatic defect	216360
RPGRIP1L	Meckel syndrome, type 5	611561
RPL10	Autism, susceptibility to, X-linked 5	300847
RPS6KA3	Coffin-Lowry syndrome	303600

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RPS6KA3	Mental retardation, X-linked 19	300844
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	612075
RRM2B	Mitochondrial DNA depletion syndrome 8B (MNGIE type)	612075
RS1	Retinoschisis	312700
RYR1	Central core disease	117000
RYR1	King-Denborough syndrome	145600
RYR1	Malignant hyperthermia susceptibility 1	145600
RYR1	Minicore myopathy with external ophthalmoplegia	255320
RYR1	Neuromuscular disease, congenital, with uniform type 1 fiber	117000
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	270550
SAG	Oguchi disease-1	258100
SAG	Retinitis pigmentosa 47	613758
SAMD9	MIRAGE syndrome	617053
SAMD9	Tumoral calcinosis, familial, normophosphatemic	610455
SAMHD1	Aicardi-Goutieres syndrome 5	612952
SAMHD1	Chilblain lupus 2	614415
SBDS	Shwachman-Diamond syndrome	260400
SBF2	Charcot-Marie-Tooth disease, type 4B2	604563
SC5DL	Lathosterolosis	607330
SCN2A	Epileptic encephalopathy, early infantile, 11	613721
SCN2A	Seizures, benign familial infantile, 3	607745
SCNN1A	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1A)	264350
SCNN1B	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1B)	264350
SCNN1G	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1G)	264350
SCO1	Mitochondrial complex IV deficiency	
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	604377
SEMA4A	Cone-rod dystrophy 10	610283
SEMA4A	Retinitis pigmentosa 35	610282
SEPN1	Rigid spine syndrome	602771
SEPSECS	Pontocerebellar hypoplasia type 2D	613811
SERPINA1	Emphysema due to AAT deficiency	613490

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SERPINA 1	Emphysema-cirrhosis, due to AAT deficiency	613490
SERPINA 1	Hemorrhagic diathesis due to antithrombin Pittsburgh	613490
SERPINA 1	Pulmonary disease, chronic obstructive, susceptibility to	606963
SETX	Amyotrophic lateral sclerosis 4, juvenile	602433
SETX	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2	606002
SFTPB	Surfactant metabolism dysfunction, pulmonary, 1	265120
SFTPC	Surfactant metabolism dysfunction, pulmonary, 2	610913
SGCA	Muscular dystrophy, limb-girdle, autosomal recessive 3	608099
SGCB	Muscular dystrophy, limb-girdle, autosomal recessive 4	604286
SGCD	Cardiomyopathy, dilated, 1L	606685
SGCD	Muscular dystrophy, limb-girdle, autosomal recessive 6	601287
SGCG	Muscular dystrophy, limb-girdle, autosomal recessive 5	253700
SGSH	Mucopolysaccharidosis type 3A (Sanfilippo syndrome type A)	252900
SH2D1A	X-linked lymphoproliferative disease	308240
SH3TC2	Charcot-Marie-Tooth disease, type 4C	601596
SH3TC2	Mononeuropathy of the median nerve, mild	613353
SHROOM 4	Stocco dos Santos X-linked mental retardation syndrome	300434
SIL1	Marinesco-Sjögren syndrome	248800
SIX6	Optic disc anomalies with retinal and/or macular dystrophy	212550
SLC12A1	Antenatal Bartter syndrome type 1	601678
SLC12A3	Gitelman syndrome	263800
SLC12A6	Corpus callosum agenesis - neuronopathy	218000
SLC16A2	Allan-Herndon-Dudley syndrome	300523
SLC17A5	Free sialic acid storage disease, infantile form	269920
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	249270
SLC22A5	Carnitine deficiency, systemic primary	212140
SLC24A1	Night blindness, congenital stationary (complete), 1D, autosomal recessive	613830
SLC25A13	Citrullinemia, adult-onset type II	603471
SLC25A13	Citrullinemia, type II, neonatal-onset	605814
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria	238970
SLC25A20	Carnitine-acylcarnitine translocase deficiency	212138

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SLC25A22	Early infantile epileptic encephalopathy	609304
SLC26A2	Achondrogenesis type 1B	600972
SLC26A2	Atelosteogenesis type II	256050
SLC26A2	Diastrophic dwarfism	222600
SLC26A2	Multiple epiphyseal dysplasia type 4	226900
SLC26A3	Diarrhea 1, secretory chloride, congenital	214700
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	600791
SLC26A4	Pendred syndrome	274600
SLC26A5	Deafness, autosomal recessive 61	613865
SLC35A1	Congenital disorder of glycosylation type 2f	603585
SLC35C1	Congenital disorder of glycosylation type 2c	266265
SLC35D1	Schneckenbecken dysplasia	269250
SLC37A4	Glycogen storage disease due to glucose-6-phosphatase deficiency type b	232220
SLC37A4	Glycogen storage disease due to glucose-6-phosphatase deficiency type c	232240
SLC39A4	Acrodermatitis enteropathica	201100
SLC3A1	Cystinuria	220100
SLC45A2	Albinism, oculocutaneous, type IV	606574
SLC45A2	Skin/hair/eye pigmentation 5, black/nonblack hair	227240
SLC45A2	Skin/hair/eye pigmentation 5, dark/fair skin	227240
SLC45A2	Skin/hair/eye pigmentation 5, dark/light eyes	227240
SLC46A1	Folate malabsorption, hereditary	229050
SLC4A11	Congenital hereditary endothelial dystrophy type II	217700
SLC4A11	Corneal dystrophy - perceptive deafness	217400
SLC5A5	Folate malabsorption, hereditary	229050
SLC6A19	Hartnup disorder	234500
SLC6A19	Hyperglycinuria	138500
SLC6A19	Iminoglycinuria, digenic	242600
SLC6A8	X-linked creatine transporter deficiency	300352
SLC7A7	Lysinuric protein intolerance	222700
SLC7A9	Cystinuria	220100
SLC9A6	Mental retardation, X-linked syndromic, Christianson type	300243
SLX4	Fanconi anemia, complementation group P	613951
SMARCA	Fanconi anemia, complementation group P	613951

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L1		
SMN1	Proximal spinal muscular atrophy type 1	253300
SMN1	Proximal spinal muscular atrophy type 2	253550
SMN1	Proximal spinal muscular atrophy type 3	253400
SMN1	Proximal spinal muscular atrophy type 4	271150
SMN2	Spinal muscular atrophy, type III, modifier of	253400
SMPD1	Niemann-Pick disease type A	257200
SMPD1	Niemann-Pick disease type B	607616
SMS	Mental retardation, X-linked, Snyder-Robinson type	309583
SNAI2	Piebaldism	172800
SNAI2	Waardenburg syndrome, type 2D	608890
SNAP29	Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome	609528
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency	300123
SOX3	Panhypopituitarism, X-linked	312000
SP110	Hepatic venoocclusive disease with immunodeficiency	235550
SPG11	Amyotrophic lateral sclerosis 5, juvenile	602099
SPG11	Charcot-Marie-Tooth disease, axonal, type 2X	616668
SPG11	Spastic paraplegia 11, autosomal recessive	604360
SPG20	Troyer syndrome	275900
SPG7	Spastic paraplegia 7, autosomal recessive	607259
SRD5A2	Pseudovaginal perineoscrotal hypospadias	264600
SRD5A3	Congenital disorder of glycosylation, type Iq	612379
SRD5A3	Kahrizi syndrome	612713
SRPX2	Rolandic epilepsy, mental retardation, and speech dyspraxia	300643
ST3GAL3	Epileptic encephalopathy, early infantile, 15	615006
ST3GAL3	Mental retardation, autosomal recessive 12	611090
ST3GAL5	Amish infantile epilepsy syndrome	609056
STAR	Congenital lipoid adrenal hyperplasia	201710
STAT1	Immunodeficiency 31A, mycobacteriosis, autosomal dominant	614892
STAT1	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive	613796
STAT1	Immunodeficiency 31C, autosomal dominant	614162

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STIL	Microcephaly 7, primary, autosomal recessive	612703
STIM1	Immunodeficiency 10	612783
STIM1	Myopathy, tubular aggregate, 1	160565
STIM1	Stormorken syndrome	185070
STRA6	Syndromic microphthalmia type 9	601186
STRC	Deafness, autosomal recessive 16	603720
STX11	Hemophagocytic lymphohistiocytosis, familial, 4	603552
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5	613101
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	612073
SUCLG1	Fatal infantile lactic acidosis with methylmalonic aciduria	245400
SUMF1	Multiple sulfatase deficiency	272200
SUOX	Sulfocysteinuria	272300
SURF1	Leigh syndrome, due to COX deficiency	256000
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	300491
SYP	Mental retardation, X-linked 96	300802
TAF1	Dystonia-Parkinsonism, X-linked	314250
TAF1	Mental retardation, X-linked, syndromic 33	300966
TAT	Tyrosinemia type 2	276600
TAZ	Barth syndrome	302060
TBCE	Hypoparathyroidism - intellectual deficit - dysmorphism syndrome	241410
TCAP	Cardiomyopathy, hypertrophic, 25	607487
TCAP	Muscular dystrophy, limb-girdle, autosomal recessive 7	601954
TCF4	Pitt-Hopkins syndrome	610954
TCIRG1	Autosomal recessive malignant osteopetrosis 1	259700
TCN2	Transcobalamin II deficiency	275350
TECTA	Deafness, autosomal dominant 8/12	601543
TECTA	Deafness, autosomal recessive 21	603629
TERT	Dyskeratosis congenita, autosomal dominant 2	613989
TERT	Dyskeratosis congenita, autosomal recessive 4	613989
TERT	Leukemia, acute myeloid	601626
TERT	Melanoma, cutaneous malignant, 9	615134
TERT	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1	614742

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

TFR2	Hemochromatosis, type 3	604250
TG	Autoimmune thyroid disease, susceptibility to, 3	608175
TG	Thyroid dyshormonogenesis 3	274700
TGM1	Ichthyosis, congenital, autosomal recessive 1	242300
TH	Autosomal recessive dopa-responsive dystonia	605407
THRA	Hypothyroidism, congenital, nongoitrous, 6	614450
THRB	Thyroid hormone resistance	188570
THRB	Thyroid hormone resistance, autosomal recessive	274300
THRB	Thyroid hormone resistance, selective pituitary	145650
TIMM8A	Mohr-Tranebjaerg syndrome	304700
TK2	Mitochondrial DNA depletion syndrome, myopathic form	609560
TLR3	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2	613002
TLR3	HIV1 infection, resistance to	609423
TMC1	Deafness, autosomal dominant 36	606705
TMC1	Deafness, autosomal recessive 7	600974
TMEM216	Joubert syndrome 2	608091
TMEM216	Meckel syndrome 2	603194
TMEM67	COACH syndrome	216360
TMEM67	Joubert syndrome 6	610688
TMIE	Deafness, autosomal recessive 6	600971
TMPRSS3	Deafness, autosomal recessive 8/10	601072
TNFRSF11B	Paget disease, juvenile	239000
TNNT1	Nemaline myopathy 5, Amish type	605355
TPO	Thyroid dyshormonogenesis 2A	274500
TPP1	Neuronal ceroid lipofuscinosis 2	204500
TPRN	Deafness, autosomal recessive 79	613307
TRAPPC9	Mental retardation, autosomal recessive 13	613192
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness	615441
TREX1	Aicardi-Goutières syndrome	225750
TRIM32	Bardet-Biedl syndrome 11	615988
TRIM32	Muscular dystrophy, limb-girdle, autosomal recessive 8	254110
TRIM37	MULIBREY nanism	253250

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TRIOBP	Deafness, autosomal recessive 28	609823
TRMU	Deafness, mitochondrial, modifier of	580000
TRMU	Liver failure, transient infantile	613070
TSEN54	Pontocerebellar hypoplasia type 2A	277470
TSEN54	Pontocerebellar hypoplasia type 4	225753
TSFM	Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3	610505
TSHB	Isolated thyroid-stimulating hormone deficiency	275100
TSHR	Hyperthyroidism, familial gestational	603373
TSHR	Hyperthyroidism, nonautoimmune	609152
TSHR	Hypothyroidism, congenital, nongoitrous, 1	275200
TSHR	Thyroid adenoma, hyperfunctioning, somatic	
TSHR	Thyroid carcinoma with thyrotoxicosis	
TSPAN7	Mental retardation, X-linked 58	300210
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome	608800
TTC37	Trichohepatoenteric syndrome 1	222470
TTN	Cardiomyopathy, dilated, 1G	604145
TTN	Cardiomyopathy, familial hypertrophic, 9	613765
TTN	Muscular dystrophy, limb-girdle, autosomal recessive 10	608807
TTN	Myopathy, myofibrillar, 9, with early respiratory failure	603689
TTN	Salih myopathy	611705
TTN	Tibial muscular dystrophy, tardive	600334
TTPA	Ataxia with vitamin E deficiency	277460
TUBA1A	Lissencephaly 3	611603
TUFM	Combined oxidative phosphorylation deficiency 4	610678
TULP1	Leber congenital amaurosis 15	613843
TULP1	Retinitis pigmentosa 14	600132
TUSC3	ntal retardation, autosomal recessive 7	611093
TYK2	Immunodeficiency 35	611521
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	603041
TYR	Albinism, oculocutaneous, type IA	203100
TYR	Albinism, oculocutaneous, type IB	606952
TYR	Melanoma, cutaneous malignant, susceptibility to, 8	601800

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TYR	Skin/hair/eye pigmentation 3, blue/green eyes	601800
TYR	Skin/hair/eye pigmentation 3, light/dark/freckling skin	601800
TYR	Waardenburg syndrome/albinism, digenic	103470
TYRP1	Albinism, oculocutaneous, type III	203290
TYRP1	Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)	612271
UBA1	X-linked spinal muscular atrophy type 2	301830
UBE2A	Mental retardation, X-linked syndromic, Nascimento-type	300860
UBE3A	Angelman syndrome	105830
UBR1	Johanson-Blizzard syndrome	243800
UGT1A1	Bilirubin, serum level of, QTL1	601816
UGT1A1	Crigler-Najjar syndrome, type I	218800
UGT1A1	Crigler-Najjar syndrome, type II	606785
UGT1A1	Gilbert syndrome	143500
UGT1A1	Hyperbilirubinemia, familial transient neonatal	237900
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3	608898
UNC93B1	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1	610551
UPF3B	Mental retardation, X-linked, syndromic 14	300676
UQCRB	Mitochondrial respiratory chain complex III deficiency	124000
UQCRQ	Mitochondrial respiratory chain complex III deficiency	124000
UROS	Porphyria, congenital erythropoietic	263700
USH1C	Autosomal recessive nonsyndromic sensorineural deafness type DFNB18	602092
USH1C	Usher syndrome type 1C	276904
USH1G	Usher syndrome type 1G	606943
USH2A	Usher syndrome type 2A	276901
USP9X	Mental retardation, X-linked 99	300919
USP9X	Mental retardation, X-linked 99, syndromic, female-restricted	300968
VDR	Vitamin D-dependent rickets type 2A	277440
VIPAR	Arthrogryposis, renal dysfunction, and cholestasis 2	613404
VLDLR	Cerebellar ataxia - intellectual deficit - dysequilibrium syndrome	224050
VPS13A	Choreoacanthocytosis	200150
VPS13B	Cohen Syndrome type 1	216550
VPS33B	Arthrogryposis - renal dysfunction - cholestasis	208085
VRK1	Pontocerebellar hypoplasia type 1A	607596

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VSX2	Microphthalmia with coloboma 3	610092
VSX2	Microphthalmia, isolated 2	610093
VWF	von Willebrand disease, type 1	193400
VWF	von Willebrand disease, types 2A, 2B, 2M, and 2N	613554
VWF	von Willibrand disease, type 3	277480
WAS	Wiskott-Aldrich syndrome	301000
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations	604317
WFS1	Wolfram syndrome 1	222300
WHRN	Deafness, autosomal recessive 31	607084
WHRN	Usher syndrome, type 2D	611383
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood	208230
WISP3	Spondyloepiphyseal dysplasia tarda with progressive arthropathy	208230
WNT10A	X-linked severe congenital neutropenia	300299
WNT3	Tetra-amelia, autosomal recessive	273395
WNT7A	Aplasia/hypoplasia of limbs and pelvis	276820
WNT7A	Fibular hypoplasia or aplasia - femoral bowing - oligodactyly	228930
WRN	Werner syndrome	277700
XIAP	Lymphoproliferative syndrome, X-linked, 2	300635
XPA	Xeroderma pigmentosum complementation group A	278700
XPC	Xeroderma pigmentosum, group C	278720
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type	300799
ZEB2	Mowat-Wilson syndrome	235730
ZFYVE26	Spastic paraplegia 15, autosomal recessive	270700
ZIC3	Congenital heart defects, nonsyndromic, 1, X-linked	306955
ZIC3	Heterotaxy, visceral, 1, X-linked	306955
ZMPSTE24	Lethal restrictive dermopathy	275210
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy	608612
ZNF469	Brittle cornea syndrome	229200
ZNF711	Mental retardation, X-linked 97	300803

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