INFORMATION NOTICE FOR BABYNEXT NEONATAL DNA SCREENING

Introduction

Neonatal screening for genetic diseases

Extended Neonatal Screening (SNE) is a secondary preventive healthcare program that for several years has been offered to newborns throughout Italy. SNE allows early identification of individuals affected with childhood-onset diseases to direct the child to the specific treatment for the identified disease and subsequent follow-up. In the event of a positive SNE result, it is generally necessary to take additional diagnostic tests (https://www.iss.it/screening-neonatali s.d.).

SNE, through biochemical investigations carried out on a simple blood draw from the heel of the newborn, early identifies 40 serious diseases in a neonatal time period for which there are treatment options.

The conditions included in SNE are all present in the Recommended Uniform Screening Panel (RUSP), a list of conditions that the U.S. Department of Public Health recommends to assess in newborns.

What is Babynext?

Babynext is a screening test aimed at the early diagnosis of more than 280 genetic conditions in the child in order to prevent the onset of clinical manifestations and/or cure them. **The results obtained from Babynext permit planning targeted clinical checks, implementing lifestyle changes, particularly dietary changes, or applying specific therapeutic protocols.**

Babynext not only analyses the conditions recommended by the RUSP, but also includes conditions with a potential for early intervention and treatment that cannot be identified with extended biochemical screening.

Genetic screening can also provide more accurate information about the actual presence of the conditions investigated by the SNE.

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The conditions investigated by Babynext may have different types of transmission:

- Autosomal recessive (AR) transmission: most of the conditions investigated by Babynext are transmitted in autosomal recessive mode (fig. 1). In this case, an individual, whether male or female, is affected when they have two variants of each of the two copies of the gene they carry, i.e. whether they are homozygous or compound heterozygous. The two variants are inherited from the parents who are generally simple heterozygous for one variant only and are therefore healthy carriers. Two healthy carrier parents have a 25% chance of having an affected child as they will both transmit the variant they carry; in the remaining cases the children will be carriers as one or the other parent (50%) or healthy without being carriers (25%). For all diseases transmitted in autosomal recessive mode. Babynext will be able to detect both the presence of a genotype at risk for the disease and the status of carrier. Identifying the status of a child as a healthy carrier can also provide useful information for parents' future reproductive choices and give instructions on performing targeted investigations on the parents. **Cystic fibrosis, congenital deafness related to gene GJB2, and beta-thalassemia are the most common autosomal recessive diseases and are included in Babynext.**

- Autosomal dominant (AD) transmission: many of the conditions investigated by Babynext are transmitted in autosomal dominant mode (fig.2). In this case, an individual, whether male or female, is affected when they have a single variant incurred by only one copy of the gene, i.e. when they are simple heterozygous for the variant. Although it is not possible to be a healthy carrier of an AD transmission disease, the clinical manifestations in this case may be variable and are not always present as the penetrance (ratio between clinically affected individuals and total individuals with the genetic variant) may be incomplete. Variants in genes related to AD diseases may be inherited from a parent or arise de novo in the conception of that individual. An individual affected with an AD disease has a 50% chance at each conception of transmitting the variant and thus the disease. **Neurofibromatosis type 1 and Alport syndrome are two examples AD conditions investigated by Babynext.**

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- X- linked (XLR/XLD) transmission: The X chromosome is constitutionally present in a single copy in males and in duplicate in females. X-linked recessive diseases (fig. 3 and 4) generally occur in male individuals who exhibit the disorder on their single X chromosome (i.e., they are hemizygous). Conversely, women who carry a single alteration on only one of their two X chromosomes (i.e., heterozygotes) will generally not show signs of the disease, except in rare cases. Women carrying a variant in a gene located on the X chromosome have, at each conception, a 50% risk of transmitting the altered X chromosome, but the disease will typically only manifest in males (25%) and not in females (25%) who will in their turn be carriers. If the X chromosome is not transmitted with the alteration, the children, whether male or female, will be neither affected nor carriers and will therefore not be at risk of developing or transmitting the disease. Males affected by the condition when transmitting their single X chromosome will have daughters who will therefore have to be carriers. On the other hand, in the case of transmission of the Y chromosome, there will be a male child and, since the chromosome carrying the alteration is excluded, none of the male children will be affected, but neither will they be a carrier or at risk of transmitting the condition. One of the most common conditions transmitted by X- linked mode investigated by Babynext is glucose 6-phosphate-dehydrogenase deficiency, commonly known as favism.

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Figure 1: Autosomal recessive (AR) inheritance

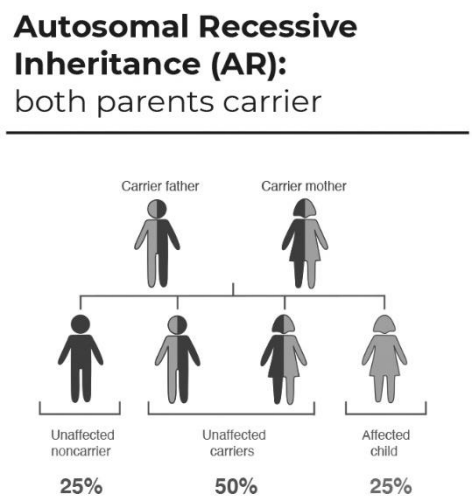


Figure 2: Autosomal dominant (AD) inheritance

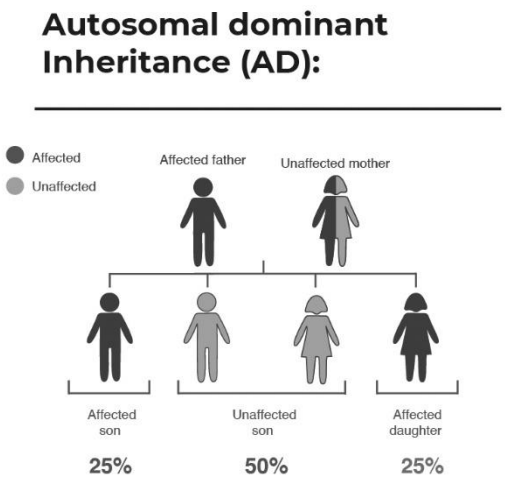
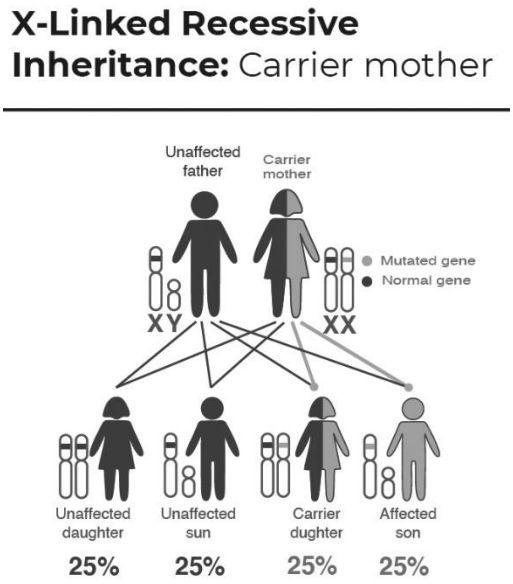


Figure 3 and 4: X-linked recessive inheritance (XLR/XLD)



Babynext also includes the sequencing of the entire coding region of the Human Genome (WES, Whole Exome Sequencing), defined as exome.

Whole Exome Sequencing (WES)

DNA is divided into thousands of segments that are called genes. Genes are in their turn formed by

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two types of regions with different functions: exons, which are the DNA regions that act as a template (coding) for proteins, and introns, which are non-coding DNA regions, but which indirectly intervene in the regulation of the processes that lead to the formation of the protein. Each gene consists of a long sequence of chemical letters, called nitrogen bases. The set of all the genes comprising an individual's genetic makeup is called the “genome” and the set of all the coding portions of the genome is called the “exome”. In 2000, the entire human genome was sequenced (read) for the first time, and it was defined that different individuals are characterized by the presence of many variants in a unique and individual-specific combination. The presence of different DNA sequences ensures variability between individuals, and in most cases the variants are “benign” meaning they cause no disease of any kind.

A minority of DNA variants cause disease (pathogenetic) and more than 80% of pathogenetic genetic variants are present in the exome (van Dijk EL et al. 2014). Whole Exome Sequencing is the sequencing of the entire coding region of the human genome corresponding to the analysis of approximately 20,000 genes.

WES is employed in a wide range of situations, including:

- ✓ differential diagnosis of a complex clinical picture due to multiple causes
- ✓ a well-defined clinical picture, but characterized by a wide genetic heterogeneity, i.e. many genes can give the same clinical picture
- ✓ a clinical picture for which the molecular bases are not yet known and therefore the gene causing it has not yet been identified
- ✓ genetic tests carried out previously without a conclusive

It is possible to request an advanced level of Babynext (Babynext Caring for life) which provides for the retention of exome data up to the child's 18th year of age in order to allow immediate consultation in case of need.

Babynext and Babynext Caring for Life is also available in a *Plus* version. This version includes two genetic tests for evaluating variants predisposed to the development of lactose intolerance and celiac disease.

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- *Celiac disease* is a chronic immune-mediated bowel condition triggered by gluten exposure in genetically predisposed individuals. The human leukocyte antigen (HLA) system, in particular the HLA-DQA1 and HLA-DQB1 genes that code for HLA-DQ2 and HLA-DQ8 molecules play a crucial role. In a person with celiac disease, gluten exposure causes a significant inflammatory response in the small intestine. Early diagnosis and intervention are therefore important in avoiding both symptoms of celiac disease and complications that could result from this condition. Classic symptoms can occur early after weaning, which involves the introduction of substances containing gluten, or later in growth. If alleles predisposing to celiac disease are present, it is important to report this predisposition to the attending pediatrician who will evaluate the possible presence of manifestations of the condition, whether to start confirmatory diagnostic tests (e.g. plasma antibody dosage and/or intestinal biopsy), and only as a last resort, whether to start a gluten-free diet.
- *Lactose intolerance* is an extremely common condition in the world. It is caused by a more or less marked reduction in the function of an intestinal enzyme called lactase, which is responsible for the digestion of lactose, a sugar resulting from the union of two simple sugars (galactose and glucose) contained in milk and its derivates. Some variants of the lactase gene (LTC) have been associated with lactase deficiency. When lactase does not function properly, lactose is not absorbed in the small intestine (lactose malabsorption) and reaches the colon where it ferments due to the action of the bacterial flora, causing a high production of fatty acids, water and gas. All this results in the onset of symptoms such as abdominal pain, swelling, diarrhea, bloating, and flatulence. Almost all children are born with the ability to digest lactose, but lactase production generally decreases with age. A genetic predisposition to lactose intolerance will therefore show any sign after weaning and may not manifest itself in any way in the first months of life. If alleles predisposing to lactose intolerance are present, it is important to report this predisposition to the attending pediatrician who will evaluate the possible presence of manifestations of the condition, whether to start confirmatory diagnostic tests (e.g. Lactose Breath Test) and, only as a last resort, whether to start a low/lactose-free diet or consider the use of exogenous enzymes to compensate lactase deficiency.

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Why and when to carry out Babynext?

Babynext performs an early screening of over 280 genetic diseases that can help parents and paediatricians identify such diseases in the early stages of life and allow early admittance to clinical tests, treatment and lifestyle changes. Babynext screening can be performed from birth **without any specific clinical indications**, i.e., symptoms and obvious signs of disease. The test will have to be preceded by an interview with a doctor or geneticist who will, in addition to the child's clinical information, collect details of the personal and family medical history of both parents. He or she will then inform the parents of the technical details of the test, explaining in particular its purpose and possible results.

Levels of investigation:

Babynext First: Gene analysis of the Babynext panel to screen for early-onset conditions.

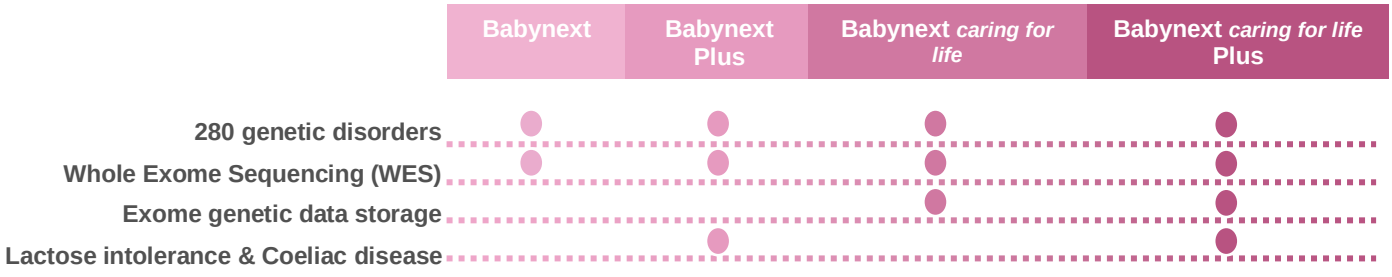
Babynext First Plus: combines Babynext First newborn screening with the investigation of primary food sensitivities, celiac disease and lactose.

Babynext Caring for Life: in addition to reporting the genes of the panels, Babynext allows for the storage of Whole Exome Sequencing data which can be consulted at a later stage. In the course of a child's life, in fact, genetic tests may be requested following clinical doubts.

WES is to date the ultimate frontier of molecular genetic diagnostics as it permits simultaneous sequencing of the coding region of approximately 20,000 genes, unlike most genetic tests that focus on a single gene or a pre-defined number of genes. Storing the exome data and biological sample allows quick access to this data, reducing waiting times for responses and the need for further testing.

In the event of a need to access exome data, genetic counselling will be possible with one of the geneticists of the Eurofins Genoma Group laboratory who will collect the clinical information and initiate the analysis of the exome. The results of this verification will then be discussed in a new genetic restitution consultation.

Babynext Caring for Life Plus: combines Babynext Caring for Life newborn screening with the investigation of primary food sensitivities, celiac disease and lactose.



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How is Babynext performed?

Sample

The child's DNA sample is obtained from cells collected with a buccal swab.

Genetic counselling

Babynext must be preceded by specialist counselling for a preliminary clinical assessment and to explain the purposes, technical details and limitations of the testing. If the request to perform the testing does not come from a specialist, pre-test genetic counselling may be carried out with a geneticist from the Eurofins Genoma Group laboratory.

Reassessment of the stored exome data may be carried out following dedicated genetic counselling. If the request does not come from a specialist in Medical Genetics, it will be possible to carry out pre-test genetic counselling with a geneticist from the Eurofins Genoma Group laboratory.

What results can be obtained from Babynext?

Babynext screening can give the following results:

- *Positive:* Babynext has shown one or more genetic variants of a clear or probable pathogenetic significance in the 288 genes investigated. This result may be indicative of:

  HEALTHY CARRIER: this result is obtained when there is a single variant in heterozygosis for AR diseases or a variant in heterozygosis in females for XLR diseases. Carrier status does not indicate that there is a risk of developing the condition but may give indications to perform further genetic investigations on the child or parents.

  AFFECTED: this result is obtained in the case of compound heterozygosis/homozygosis for variants in AR diseases, in the case of simple heterozygosis for AD diseases and in the case of hemizygosis in XLR diseases. The state of “genetically” affected might not coincide with the state of “clinically” affected, so it is important that each result be discussed and interpreted in the clinical context.

Sometimes a positive result may include variants of uncertain clinical significance, identified by the acronym VUS or VOUS (Variant of Uncertain Significance). These are variants for which current scientific knowledge does not allow defining a pathogenetic or benign nature, although the classification of these variants might change over time. These variants will only be reported under AR

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transmission conditions if in the same individual a variant of clear pathogenetic significance has been identified in the same gene.

- *Negative:* A negative report indicates that no mutation with known or probably pathogenic significance has been detected in the genes examined. A negative result reduces but does not eliminate the risk for the newborn to be affected by these genetic diseases or other genetic diseases not investigated by Babynext. The test is intended solely for identifying mutations in DNA that cause genetic diseases (pathogenetic or probable pathogenetic). Variants with uncertain clinical significance.

(VOUS) are not reported in the Babynext test barring the exceptions already specified.

Re-analysis of the exome data obtained from Babynext will be performed under specific clinical indications and may show positive, negative and uncertain results. The possible results will be explained in dedicated genetic counselling and after receiving dedicated information.

How soon can I get the results of Babynext?

The results of the screening test on over 280 genetic diseases will be available within 40 business days. This time could be extended in the event of repeating the test, further diagnostic investigations, or interpretation doubts.

If it is necessary to extend the screening test to full exome analysis, genetic counselling will be carried out with one of the geneticists of the Eurofins Genoma Group laboratory within 10 business days of the request and the reprocessing of the retained data will take place within 15 business days of the consultation.

Technical details of Babynext

Analysis techniques and target coverage

Babynext through a massive parallel sequencing (MPS) process, which employs Next Generation Sequencing (NGS) techniques, sequences approximately 95% of the entire exome, with a coverage of at least 50x and approximately 2.5% with a coverage of less than 20x. The gene sequences obtained are analysed using bioinformatics analysis to determine the presence of any variants in the genes listed in table 1.

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The SMN1 gene test is followed with the AmplideX® SMA Plus Kit in accordance with the manufacturer's instructions for quantitative determination of exon 7 of genes SMN1 and SMN2.

Parameters used for reporting genetic variants

Variants identified with Babynext will be classified, and therefore reported, according to different criteria. Consideration will be given to the presence of the variant in scientific literature (using several search engines and dedicated databases), in the ClinVar database (https://www.ncbi.nlm.nih.gov/clinvar/)or in other specific mutation databases of the genes being investigated.

An assessment of pathogenicity may also be given following the guidance of the American College of Medical Genetics (ACMG) and using dedicated bioinformatics instruments such as VarSome.

Accuracy

Current new-generation DNA sequencing (NGS) techniques produce results with an accuracy of more than 99% for sequence variants.

The AmplideX® SMA Plus kit has a specificity of >99% and a sensitivity of between 71 and 95% in relation to ethnicity.

Limits of the test

The Babynext test provides information on the presence of variants in the genes included in Table 1. Without a specific request, no variants of other genes will be reported, regardless of their clinical significance.

Variants classified as having uncertain significance at the time of reporting will not be reported, except as already specified, but may be reclassified as pathogenic or probably pathogenic variants after reporting. Re-filtering of the results based on additional clinical information can be done, when indicated, even after the final report has been issued.

The identified genetic variants do not provide information on the exact type of clinical manifestations, severity or age of onset of a disease. The genetic data obtained should be discussed in genetic counselling and interpreted in the clinical context.

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The Babynext test using NGS is unable to highlight:

- Variants located in intron regions in a position over +/- 5 nucleotides from the exon-intron junction,
- Large deletions, duplications or other gene rearrangements > 20 bp
- Variants of the mitochondrial genome
- Repeated expansions of triplets
- Genes with pseudogenes
- Epigenetic defects
- Low percentage of Mosaicisms
- Germline mosaicisms (i.e., mutations present only in gametes).

An inherent limitation of the NGS methodology used is the lack of coverage uniformity for each gene region analysed. Potential causes of non-uniformity include a suboptimal quantity and quality of the extracted DNA. This limit is reflected in the possibility, inherent in NGS methods, that specific mutations of the selected genes might not be detected by the test.

A variant of the SMN1 gene sequence can be identified in a minority of individuals affected with spinal muscular atrophy. The AmplideX® SMA Plus kit is designed to perform the quantitative determination of exon 7 of genes SMN1 and SMN2, while it is unable to detect nonsense, missense, or frameshift variants of the gene sequence.

Approximately 3-8% of healthy Spinal Muscular Atrophy carriers have two copies of the SMN1 gene on a single chromosome and zero copies on the other chromosome; these individuals are referred to as “silent carriers” or “2/0 carriers”. Quantitative analysis performed by the AmplideX® SMA Plus kit is not able to identify silent carrier status.

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Table 1: Genes included in the **Babynext**

Gene	*OMIM	Pathology	OMIM	Transmission mode	Pathology category
HBB	*141900	*Sickle Cell Anemia*	#603903	AR	Hematology
HBB	*141900	*Beta Thalassemia*	#613985	AR	Hematology
ANK1	*612641	*Hereditary spherocytosis, type 1*	#182900	AD/AR	Hematology
EPB42	*177070	*Hereditary spherocytosis, type 5*	#612690	AR	Hematology
F8	*300841	*Hemophilia A*	#306700	XLR	Hematology
F9	* 300746	*Hemophilia B*	#306900	XLR	Hematology
G6PD	*305900	*Glucose-6-phosphate dehydrogenase deficiency (Favism)*	#300908	XLD/XLR	Hematology
MPL	*159530	*Tombocytopenia*	#254450	AR	Hematology
SLC4A1	*109270	*Hereditary spherocytosis, type 4*	#612653	AD	Hematology
SPTA1	*182860	*Hereditary spherocytosis, type 3*	#270970	AR	Hematology
SPTB	*182870	*Hereditary spherocytosis, type 2*	#616649	AD	Hematology
DUOX2	*606759	*Familial thyroid dyshormonogenesis*	#607200	AR	Endocrinology
DUOXA2	*612772	*Familial thyroid dyshormonogenesis*	#274900	AR	Endocrinology
GLIS3	*610192	*Neonatal diabetes mellitus with congenital hypothyroidism*	# 610199	AR	Endocrinology
HSD3B2	*613890	*Congenital adrenal hyperplasia from 3-beta-hydroxysteroid dehydrogenase deficiency*	#201810	AR	Endocrinology
IYD	*612025	*Familial thyroid dyshormonogenesis 4*	#274800	AR	Endocrinology
NKX2-5	*600584	*Congenital hypothyroidism not associated with goiter*	#225250	AD	Endocrinology
PAX8	*167415	*Hypothyroidisimus conegnitus associated with thyroid hypoplasia and dysgenesis*	#218700	AD	Endocrinology
SLC5A5	*601843	*Thyroid Dysmorphogenesis*	#274400	AR	Endocrinology
TG	*612025	*Thyroid dyshormonogenesis type 3 (primitive congenital hypothyroidism)*	#274800	AR	Endocrinology
THRA	*190120	*Congenital Hypothyroidism Type 6*	#614450	AD	Endocrinology
TPO	*606765	*Thyroid dyshormonogenesis type 2*	#274500	AR	Endocrinology
TRHR	*188545	*Congenital hypothyroidism type 7*	#618573	AR	Endocrinology
TSHB	*188540	*Congenital hypothyroidism type 4*	#275100	AR	Endocrinology

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TSHR	*603372	*Congenital hypothyroidism type 1*	#275200	AR	Endocrinology
POU1F1	*173110	*Combined non-acquired pituitary hormone deficiency*	#613038	AR	Endocrinology
PROP1	*601538	*Combined pituitary hormone deficiency*	#262600	AR	Endocrinology
ABCD1	*300371	*X-linked adrenoleukodystrophy*	#300100	XLD	Endocrinology - Neurology
ABCD3	*170995	*Congenital defect of bile acid synthesis, type 5*	#616278	AR	Gastroenterology
ACOX2	*601641	*Congenital defect of bile acid synthesis, type 6*	#617308	AR	Gastroenterology
AKR1D1	*604741	*Congenital defect of bile acid synthesis, type 2*	#235555	AR	Gastroenterology
AMACR	*604489	*Congenital defect of bile acid synthesis, type 4*	#214950	AR	Gastroenterology
CYP7B1	*603711	*Congenital defect of bile acid synthesis, type 3*	#613812	AR	Gastroenterology
HSD3B7	*607764	*Congenital defect of bile acid synthesis, type 1*	#607765	AR	Gastroenterology
ADA	*608958	*Severe combined immunodeficiency from ADA deficiency*	#102700	AR	Immune
AK2	*103020	*Reticular dysgenesis*	#267500	AR	Immune
BTK	*300300	*X-linked agammaglobulinemia*	#300755	XLR	Immune
CD247	*186780	*Immunodeficiency 25 or Immunodeficiency due to defect in CD3-zeta*	# 610163	AR	Immune
CD3D	*186790	*Immunodeficiency 19*	#615617	AR	Immune
CD3E	*186830	*Immunodeficiency 18 - Immunodeficiency 18 variant SCID*	#615615	AR	Immune
CYBA	*608508	*Chronic, autosomal, CYBA-deficient or type 4 granulomatous disease*	#233690	AR	Immune
CYBB	*300481	*CYBB-related chronic granulomatous disease*	#306400	XLR	Immune
DCLRE1C	*605988	*Severe combined immunodeficiency, Athabascan*	#602450	AR	Immune
DCLRE1C	*605988	*Omenn syndrome/ / Severe combined immunodeficiency of Athabaskan type*	#603554	AR	Immune
HAX1	*605998	*Kostmann's syndrome*	# 610738	AR	Immune
IL2RG	*308380	*Severe X-linked combined immunodeficiency* *(IL2RG also causes moderate X-linked combined immunodeficiency OMIM 312863)*	#300400	XLR	Immune

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IL7R	*146661	*Severe combined immunodeficiency negative T lymphocyte B lymphocyte/positive natural killer cell type*	#608971	AR	Immune
JAK3	*600173	*Severe combined immunodeficiency*	#600802	AR	Immune
NHEJ1	*611290	*Cernunnos deficiency*	#611291	AR	Immune
PNP	*164050	*Purine nucleoside phosphorylase deficiency*	#613179	AR	Immune
PRF1	*170280	*Familial hemophagocytic lymphohistiocytosis*	#603553	AR	Immune
PTPRC	*151460	*PTPRC-related severe combined immunodeficiency*	#608971	AR	Immune
RAG1	*179615	*Combined cellular and humoral immunodeficiency with granulomas*	#233650	AR	Immune
RAG1	*179615	*Severe combined immunodeficiency, B-cell negative*	#601457	AR	Immune
RAG1	*179615	*Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity*	#609889	AR	Immune
RAG1	*179615	*Omenn syndrome to other RAG1-related disorders*	#603554	AR	Immune
RAG2	*179616	*Combined cellular and humoral immunodeficiency with granulomas*	#233650	AR	Immune
RAG2	*179616	*Severe combined immunodeficiency, B-cell negative*	#601457	AR	Immune
RAG2	*179616	*RAG2-related Omenn syndrome*	#603554	AR	Immune
STX11	*605014	*Familial haemophogocytic lymphohistocytosis*	#603552	AR	Immune
UNC13D	*608897	*Familial hemophagocytic lymphohistiocytosis type 3*	#608898	AR	Immune
ZAP70	*176947	*Immunodeficiency 48 (IMD48) or ZAP70 deficiency combined immunodeficiency*	#269840	AR	Immune
ACADM	*607008	*Deficiency of medium-chain acyl-CoA dehydrogenase*	#201450	AR	Metabolic
ACADVL	*609575	*Very long chain acyl-CoA dehydrogenase deficiency*	#201475	AR	Metabolic
ACAT1	*607809	*Beta-ketothiolase deficiency*	#203750	AR	Metabolic
ASL	*608310	*Argininosuccinic aciduria*	#207900	AR	Metabolic
ASS1	*603470	*Citrullinemia type 1*	#215700	AR	Metabolic

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BCKDHA	*608348	*Classic maple syrup urine disease, type 1a*	#248600	AR	Metabolic
BCKDHB	*248611	*Classic maple syrup urine disease, type 1a*	#248600	AR	Metabolic
BTD	*609019	*Biotinidase deficiency*	#253260	AR	Metabolic
CBS	*613381	*Homocystinuria, vitamin B6 responsive and nonresponsive type*	#236200	AR	Metabolic
CD320	*606475	*Methylmalonic aciduria, transient, due to transcobalamin receptor defect*	#613646	AR	Metabolic
DBT	*248610	*Maple syrup urine disease*	#248600	AR	Metabolic
FAH	*613871	*Tyrosinemia, type 1*	#276700	AR	Metabolic
GAA	*606800	*Glycogenosis type 2 - Pompe's disease*	#232300	AR	Metabolic
GALC	*606890	*Krabbe's disease*	#245200	AR	Metabolic
GALT	*606999	*Galactosemia*	#230400	AR	Metabolic
GCDH	*608801	*Glutaric aciduria type 1*	#231670	AR	Metabolic
GCH1	*600225	*Hyperphenylalaninemia (HPA) from tetrahydrobiopterin (BH4) deficiency*	#233910	AR	Metabolic
GLA	*300644	*Fabry disease*	#301500	XLR	Metabolic
GNMT	*606628	*N-methyltransferase deficiency*	#606664	AR	Metabolic
HADHA	*600890	*Deficiency of long-chain 3-hydroxyacyl-CoA dehydrogenase*	#609016	AR	Metabolic
HADHB	*143450	*Trifunctional protein deficiency*	# 609015	AR	Metabolic
HLCS	*609018	*Holocarboxylase synthetase deficiency*	#253270	AR	Metabolic
HMGCL	*613898	*Deficiency of 3-hydroxy-3-methylglutaryl-CoA lyase or Deficiency of HMG-CoA lyase or 3-hydroxy-3-methylglutaric aciduria or Hydroxy-methyl-glutaricaciduria*	#246450	AR	Metabolic
IDS	*607764	*Mucopolysaccharidosis II*	#607765	XLR	Metabolic
IDUA	*252800	*Mucopolysaccharidosis type Ih (I-h I-s I-h/s)*	#607014	AR	Metabolic
IDUA	*252800	*Mucopolysaccharidosis type Ih/s (I-h I-s I-h/s)*	#607015	AR	Metabolic
IDUA	*252800	*Type of mucopolysaccharidosis Is (I-h I-s I-h/s)*	#607016	AR	Metabolic
IVD	*607036	*Isovaleric acidemia*	#243500	AR	Metabolic

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LDLR	*606945	*Familial hypercholesterolemia*	#143890	AD	Metabolic
LPL	*609708	*Lipoprotein lipase deficiency*	#238600	AR	Metabolic
MAT1A	*610550	*Hypermethioninemia from methionine adenosyltransferase deficiency*	#250850	AR	Metabolic
MCCC1	*609010	*Isolated deficiency of 3-methylcrotonyl-CoA carboxylase type 1*	#210200	AR	Metabolic
MCCC2	*609014	*Isolated deficiency of 3-methylcrotonyl-CoA carboxylase type 2*	#210210	AR	Metabolic
MLYCD	*606761	*Malonic aciduria*	#248360	AR	Metabolic
MMAA	*607481	*Vitamin B12-responsive methylmalonic acidemia, cblA type*	#251100	AR	Metabolic
MMAB	*607568	*Vitamin B12-responsive methylmalonic acidemia, cblb type*	#251110	AR	Metabolic
MTHFR	*607093	*Homocostinuria from reduced activity of methylenetetrahydrofolate reductase*	#236250	AR	Metabolic
MTR	*156570	*megaloblastic anemia*	#250940	AR	Metabolic
MTRR	*602568	*Homocystinuria*	#236270	AR	Metabolic
MTTP	*157147	*Abetalipoproteinemia*	#200100	AR	Metabolic
MUT	*603058	*Methylmalonic acidemia refractory to vitB12*	#251000	AR	Metabolic
OPA3	*606580	*Costeff's syndrome or 3-methylglutaconic aciduria type III*	#258501	AR	Metabolic
PAH	*612349	*Phenylketonuria*	#261600	AR	Metabolic
PCBD1	*126090	*Hyperphenylalaninemia from tetrahydrobiopterin deficiency*	#264070	AR	Metabolic
PCCA	*232000	*Propionic acidemia alpha*	#606054	AR	Metabolic
PCCB	*232050	*Propionic beta acidemia*	#606054	AR	Metabolic
PTS	*612719	*BH4 deficiency hyperphenylalaninemia, A*	#261640	AR	Metabolic
QDPR	*612676	*BH4 deficiency hyperphenylalaninemia, C*	#261630	AR	Metabolic
SLC22A5	*603377	*Primary systemic carnitine deficiency*	#212140	AR	Metabolic
TAZ	*300394	*Barth's Syndrome*	#302060	XLR	Metabolic

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ABCD4	*603214	*Methylmalonic aciduria and homocystinuria, type cblJ*	#614857	AR	Metabolic
ACAD8	*604773	*Deficiency of isobutyryl-CoA dehydrogenase*	#611283	AR	Metabolic
ACADS	*606885	*Acyl-CoA dehydrogenase deficiency, short chain*	#201470	AR	Metabolic
ACADSB	*600301	*2-methylbutyryl glycinuria*	#610006	AR	Metabolic
ADK	*102750	*Hypermethioninemia from adenosine kinase deficiency*	#614300	AR	Metabolic
AHCY	*180960	*Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency*	#613752	AR	Metabolic
ARG1	*608313	*Argininemia*	#207800	AR	Metabolic
AUH	* 600529	*3-Methylglutaconic aciduria, type 1*	#250950	AR	Metabolic
CPT1A	* 600528	*Carnitine palmitoyltransferase type I , hepatic, type IA deficiency*	#255120	AR	Metabolic
CPT2	* 600650	*Carnitine palmitoyltransferase type II deficiency*	#255110	AD, AR	Metabolic
DNAJC19	*608977	*3-Methylglutaconic aciduria, type 5 / Dilated cardiomyopathy with ataxia*	#610198	AR	Metabolic
ETFA	*608053	*Glutaric acidemia, type 2a*	#231680	AR	Metabolic
ETFB	*130410	*Glutaric acidemia, type 2b*	#231680	AR	Metabolic
ETFDH	*231675	*Glutaric acidemia, type 2c*	#231680	AR	Metabolic
GALE	*606953	*Galactose epimerase deficiency*	#230350	AR	Metabolic
GALK1	*604313	*Galactokinase deficiency with cataracts*	#230200	AR	Metabolic
HADH	*601609	*Short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency hyperinsulinism*	#231530	AR	Metabolic
HPD	*609695	*Tyrosinemia, type 3 (also causes Hawkinsinuria MIM 140350 with AD transmission)*	#276710	AR	Metabolic
LMBRD1	*612625	*Methylmalonic acidemia with homocystinuria, cblF type*	#277380	AR	Metabolic
MCEE	*608419	*Methylmalonic acidemia from methylmalonyl-CoA epimerase deficiency*	#251120	AR	Metabolic
MMACHC	*609831	*Methylmalonic aciduria with homocystinuria, cblC type*	#277400	AR	Metabolic
MMADHC	*611935	*Metabolism OF COBALAMIN ASSOCIATED C*	#277410	AR	Metabolic

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NADK2	*615787	*Deficiency of 2,4-dienoyl-CoA reductase*	#616034	AR	Metabolic
SLC25A13	*603859	*Citrullinemia type II onset in adulthood*	#603471	AR	Metabolic
SLC25A13	*603859	*Citrullinemia type II neonatal onset*	#605814	AR	Metabolic
SLC25A20	*613698	*Carnitine-acylcarnitine translocase deficiency*	#212138	AR	Metabolic
TAT	*613018	*Tyrosinemia TYPE 2*	#276600	AR	Metabolic
NAGS	*608300	*N-acetylglutamate deficiency hyperammoniemia*	#237310	AR	Metabolic
OTC	*300461	*Ornithine carbamoyltransferase deficiency*	#311250	XL	Metabolic
AGL	*610860	*Glycogen storage disease type III*	#232400	AR	Metabolic
AGXT	*604285	*Primary hyperoxaluria, type 1*	#259900	AR	Metabolic
ALDH4A1	*606811	*Hyperprolinemia, type II*	#239510	AR	Metabolic
ALDOB	*612724	*Hereditary fructose intolerance*	#229600	AR	Metabolic
ARSA	*607574	*Metachromatic leukodystrophy*	#250100	AR	Metabolic
ARSB	*611542	*Mucopolysaccharidosis type VI*	#253200	AR	Metabolic
CTNS	*219800	*Nephropathic cystinosis*	#219800	AR	Metabolic
FBP1	*611570	*Fructose 1,6-diphosphatase deficiency*	#229700	AR	Metabolic
G6PC	*613742	*Glycogen storage disease, type Ia*	#232200	AR	Metabolic
GYS2	*138571	*Hepatic glycogen synthase deficiency glycogenosis*	#240600	AR	Metabolic
HMGCS2	*600234	*Deficiency of 3-hydroxy-3-methylglutaryl-CoA synthetase*	#605911	AR	Metabolic
HOGA1	*613597	*Primary hyperoxaluria, type 3*	#613616	AR	Metabolic
PRODH	*606810	*Hyperprolinemia, type 1*	#239500	AR	Metabolic
SLC37A4	*602671	*Glycogenosis TYPE 1B*	#232220	AR	Metabolic
SLC7A7	*603593	*Lysinuric protein intolerance*	#222700	AR	Metabolic
TRMU	*610230	*Transient childhood liver failure*	#613070	AR	Metabolica
ATP6V0A4	*605239	*Distal renal tubular acidosis 3 with or without sensorineural hearing loss*	#602722	AR	Metabolic
ATP6V1B1	*192132	*Distal renal tubular acidosis 2 with progressive sensorineural hearing loss*	#267300	AR	Nephrology
COL4A3	*120070	*Alport syndrome 2, linked to COL4A3*	#203780	AR	Nephrology

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COL4A3	*120070	*Alport syndrome 3, autosomal dominant*	#104200	AD	Nephrology
GRHPR	*604296	*Primary hyperoxaluria, type 2*	#260000	AR	Nephrology
SLC3A1	*104614	*Cystinuria A*	#220100	AR	Nephrology
SLC4A1	*109270	*Distal renal tubular acidosis 1 (DRTA1)*	#179800	AD	Nephrology
SLC7A9	*604144	*Cystinuria B*	#220100	AR	Nephrology
COL4A4	*120131	*Autosomal recessive Alport syndrome*	#203780	AR	Nephrology - Otolaryngology
COL4A5	*303630	*Alport syndrome*	#301050	XLD	Nephrology - Otolaryngology
SMN1	*600354	*Spinal muscular atrophy-1*	#253300	AR	Neurology
SMN2	*600354	*Spinal muscular atrophy-2*	#253550	AR	Neurology
SMN3	*600354	*Spinal muscular atrophy-3*	#253400	AR	Neurology
SMN4	*600354	*Spinal muscular atrophy-4*	#271150	AR	Neurology
RB1	*614041	*Retinoblastoma*	#180200	AD	Oncology
SMARCA4	*603254	*Predisposition to rhabdoid tumor type 2*	#613325	AD	Oncology
SMARCB1	*601607	*Predisposition to rhabdoid tumor type 1*	#609322	AD	Oncology
WT1	*607102	*Nephroblastoma or Wilms' tumor*	#194070	AD	Oncology
ACTG1	*102560	*Autosomal dominant deafness 20/26*	#604717	AD	Otolaryngology
ATP11A	*605868	*Autosomal dominant deafness 84*	#619810	AD	Otolaryngology
ATP2B2	*108733	*Autosomal dominant deafness 82*	#619804	AD	Otolaryngology
CABP2	*607314	*Autosomal dominant deafness 93*	#614899	AR	Otolaryngology
CDC14A	*603504	*Autosomal recessive deafness 32 with or without immotile sperm*	#608653	AR	Otolaryngology
CDH23	*605516	*Autosomal recessive deafness 12*	#601386	AR	Otolaryngology
CEACAM16	*614591	*Autosomal dominant deafness 4B*	#614614	AD	Otolaryngology
CEACAM16	*614591	*Autosomal recessive deafness 113*	#618410	AR	Otolaryngology
CIB2	*605564	*Autosomal recessive deafness 48*	#609439	AR	Otolaryngology
CLDN14	*605608	*Autosomal recessive deafness 29*	#614035	AR	Otolaryngology
COCH	*603196	*Autosomal dominant deafness 9*	#601369	AD	Otolaryngology
COL11A1	*120280	*Autosomal dominant deafness 37*	#618533	AD	Otolaryngology
COL11A2	*120290	*Autosomal dominant deafness 13*	#601868	AD	Otolaryngology
COL11A2	*120290	*Autosomal recessive deafness 53*	#609706	AR	Otolaryngology

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CRYM	*123740	*Autosomal dominant deafness 40*	#616357	AD	Otolaryngology
DIABLO	*605219	*Autosomal dominant deafness 64*	#614152	AD	Otolaryngology
DIAPH1	*602121	*Autosomal dominant 1 deafness with or without thrombocytopenia*	#124900	AD	Otolaryngology
EPS8L2	*614988	*Autosomal recessive deafness 106*	#617637	AR	Otolaryngology
ESPN	*606351	*Autosomal recessive deafness 36*	#609006	AR	Otolaryngology
ESPN	*606351	*Sensorineural deafness without autosomal dominant vestibular involvement*	#609006	AR	Otolaryngology
ESRRB	*602167	*Autosomal recessive deafness 35*	#608565	AR	Otolaryngology
EYA4	*603550	*Autosomal dominant deafness 10*	#601316	AD	Otolaryngology
FOXI1	*601093	*Deafness with enlarged vestibular aqueduct*	#600791	AR	Otolaryngology
GIPC3	*608792	*Autosomal recessive deafness 15*	#601869	AR	Otolaryngology
GJB2	*121011	*Autosomal dominant deafness type 3A*	#601544	AD	Otolaryngology
GJB2	*121011	*Autosomal recessive congenital deafness type 1A*	#220290	AR, DD	Otolaryngology
GJB3	*603324	*Autosomal dominant deafness 2B*	#612644	AD	Otolaryngology
GJB3	*603324	*GJB2/GJB3 digenic deafness*	#220290	AR, DD	Otolaryngology
GJB6	*604418	*Autosomal dominant deafness type 3B*	#612643	AD	Otolaryngology
GJB6	*604418	*Autosomal recessive congenital deafness type 1B*	# 612645	AR	Otolaryngology
GJB6	*604418	*GJB2/GJB6 congenital digenic deafness*	# 220290	AR, DD	Otolaryngology
GRAP	*604330	*Autosomal recessive deafness 114*	#618456	AR	Otolaryngology
GREB1L	*617782	*Autosomal dominant deafness 80*	#619274	AD	Otolaryngology
GRHL2	*608576	*Autosomal dominant deafness 28*	#608641	AD	Otolaryngology
GRXCR1	*613283	*Autosomal recessive deafness 25*	#613285	AR	Otolaryngology
GSDME	*608798	*Autosomal dominant deafness 5*	#600994	AD	Otolaryngology
HGF	*142409	*Autosomal recessive deafness 39*	#608265	AR	Otolaryngology
ILDR1	*609739	*Autosomal recessive deafness 42*	#609646	AR	Otolaryngology
KARS1	*601421	*Autosomal recessive deafness 89*	#613916	AR	Otolaryngology
KCNJ10	*602208	*Enlarged vestibular digenic aqueduct*	#600791	AR	Otolaryngology
KCNQ4	*603537	*Autosomal dominant deafness 2A*	#600101	AD	Otolaryngology
KITLG	*184745	*Autosomal dominant 69 unilateral or asymmetric deafness*	#616697	AD	Otolaryngology

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LHFPL5	*609427	*Autosomal recessive deafness 67*	#610265	AR	Otolaryngology
LMX1A	*600298	*Autosomal dominant deafness 7*	#601412	AD	Otolaryngology
LOXHD1	*613072	*Autosomal recessive deafness 77*	#613079	AR	Otolaryngology
LRTOMT	*612414	*Autosomal recessive deafness 63*	#611451	AR	Otolaryngology
MARVELD2	*610572	*Autosomal recessive deafness 49*	#610153	AR	Otolaryngology
MIR96	*611606	*Autosomal dominant deafness 50*	#613074	AD	Otolaryngology
MPZL2	*604873	*Autosomal recessive deafness 111*	#618145	AR	Otolaryngology
MSRB3	*613719	*Autosomal recessive deafness 74*	#613718	AR	Otolaryngology
MYH14	*608568	*Autosomal dominant deafness 4A*	#600652	AD	Otolaryngology
MYH9	*160775	*Autosomal dominant deafness 17*	#603622	AD	Otolaryngology
MYO15A	*602666	*Autosomal recessive deafness 3*	#600316	AR	Otolaryngology
MYO3A	*606808	*Autosomal recessive deafness 30*	#607101	AR	Otolaryngology
MYO6	*600970	*Autosomal dominant deafness 22*	#606346	AD	Otolaryngology
MYO6	*600970	*Autosomal dominant deafness 22 with hypertrophic cardiomyopathy*	#606346	AD	Otolaryngology
MYO6	*600970	*Autosomal recessive deafness 37*	#607821	AR	Otolaryngology
MYO7A	*276903	*Autosomal dominant deafness 11*	#601317	AD	Otolaryngology
MYO7A	*276903	*Autosomal recessive deafness 2*	#600060	AR	Otolaryngology
NLRP3	*606416	*Autosomal dominant deafness 34 with or without inflammation*	#617772	AD	Otolaryngology
OSBPL2	*606731	*Autosomal dominant deafness 67*	#616340	AD	Otolaryngology
OTOA	*607038	*Autosomal recessive deafness 22*	#607039	AR	Otolaryngology
OTOF	*603681	*Autosomal recessive auditory neuropathy 1*	#601071	AR	Otolaryngology
OTOF	*603681	*Autosomal recessive deafness 9*	#601071	AR	Otolaryngology
OTOG	*604487	*Autosomal recessive deafness 18B*	#614945	AR	Otolaryngology
OTOGL	*614925	*Autosomal recessive deafness 84B*	#614944	AR	Otolaryngology
P2RX2	*600844	*Autosomal dominant deafness 41*	#608224	AD	Otolaryngology
PCDH15	*605514	*Autosomal recessive deafness 23*	#609533	AR	Otolaryngology
PDZD7	*612971	*Autosomal recessive deafness 57*	#618003	AR	Otolaryngology
PJVK	*610219	*Autosomal recessive deafness 59*	#610220	AR	Otolaryngology
PLS1	*602734	*Autosomal dominant deafness 76*	#618787	AD	Otolaryngology
PNPT1	*610316	*Autosomal recessive deafness 70*	#614934	AR	Otolaryngology
POU4F3	*602460	*Autosomal dominant deafness 15*	#602459	AD	Otolaryngology
PPIP5K2	*611648	*Autosomal recessive deafness 100*	#618422	AR	Otolaryngology
PTPRQ	*603317	*Autosomal dominant deafness 73*	#617663	AD	Otolaryngology

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PTPRQ	*603317	*Autosomal recessive deafness 84A*	#613391	AR	Otolaryngology
RDX	*179410	*Autosomal recessive deafness 24*	#611022	AR	Otolaryngology
REST	*600571	*Autosomal dominant deafness 27*	#612431	AD	Otolaryngology
RIPOR2	*611410	*Autosomal dominant deafness 21*	#607017	AD	Otolaryngology
S1PR2	*605111	*Autosomal recessive deafness 68*	#610419	AR	Otolaryngology
SIX1	*601205	*Autosomal dominant deafness 23*	#605192	AD	Otolaryngology
SLC12A2	*600840	*Autosomal dominant deafness 78*	#619081	AD	Otolaryngology
SLC17A8	*607557	*Autosomal dominant deafness 25*	#605583	AD	Otolaryngology
SLC26A4	*605646	*Pendred's syndrome*	#274600	AR	Otolaryngology
SLC26A4	*605646	*Deafness with vestibular aqueduct enlargement*	#600791	AR	Otolaryngology
SPATA5L1	*619578	*Autosomal recessive deafness 119*	#619615	AR	Otolaryngology
STRC	*606440	*Autosomal recessive deafness 16*	#603720	AR	Otolaryngology
SYNE4	*615535	*Autosomal recessive deafness 76*	#615540	AR	Otolaryngology
TBC1D24	*613577	*Autosomal dominant deafness 65*	#616044	AD	Otolaryngology
TBC1D24	*613577	*Autosomal recessive deafness 86*	#614617	AR	Otolaryngology
TECTA	*602574	*Autosomal dominant deafness 8/12*	#601543	AD	Otolaryngology
TECTA	*602574	*Autosomal recessive deafness 21*	#603629	AR	Otolaryngology
TMC1	*606706	*Autosomal dominant deafness 36*	#606705	AD	Otolaryngology
TMC1	*606706	*Autosomal recessive deafness 7*	#600974	AR	Otolaryngology
TMEM132E	*616178	*Autosomal recessive deafness 99*	#618481	AR	Otolaryngology
TMIE	*607237	*Autosomal recessive deafness 6*	#600971	AR	Otolaryngology
TMPRSS3	*605511	*Autosomal recessive deafness 8/10*	#601072	AR	Otolaryngology
TNC	*187380	*Autosomal dominant deafness 56*	#615629	AD	Otolaryngology
TPRN	*613354	*Autosomal recessive deafness 79*	#613307	AR	Otolaryngology
TRIOBP	*609761	*Autosomal recessive deafness 28*	#609823	AR	Otolaryngology
USH1C	*605242	*Autosomal recessive deafness 18A*	#602092	AR	Otolaryngology
WBP2	*606962	*Autosomal recessive deafness 107*	#617639	AR	Otolaryngology
WFS1	*606201	*Autosomal dominant deafness 14/06/38*	#600965	AD	Otolaryngology
WHRN	*607928	*Autosomal recessive deafness 31*	#607084	AR	Otolaryngology
CFTR	*602421	*Cystic Fibrosis*	#219700	AR	Pulmonary - Gastrology
NF1	*613113	*Neurofibromatosis type 1*	#162200	AD	Syndromic

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