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ReadyGen

Patient	Sample	Referring Doctor
Patient Name: Test Patient Date of Birth: 11/01/2019 Reference #: SNSS00000000 Indication: Screening for Genetic Conditions and Pharmacogenetic Variants Test Type: ReadyGen Panel	Specimen Type: Buccal Swab Lab #: 192345678PN Date Collected: 11/14/2019 Date Received: 11/15/2019 Final Report: 11/27/2019	Doctor Doctor, MD Practice Phone: (123) 456-7890 Fax:

RESULT SUMMARY

THIS PATIENT WAS TESTED FOR 213 DISORDERS (185 GENES)

NEGATIVE for all diseases tested. Please note that carrier status for autosomal recessive diseases and variants of uncertain significance are not reported in this test.

Recommendations

It is recommended that state-mandated newborn screening is performed on all newborn infants. Newborn screening is the standard of care and should not be replaced by this postnatal screening panel.

These negative results reduce but do not eliminate the possibility that this individual is affected with one or more of the diseases tested. If the patient is clinically suspected to be affected with a disease on this panel, parents should seek medical attention for their child and further testing should be performed.

Interpretation

This patient was tested for a panel of diseases using a combination of sequencing, targeted genotyping and copy number analysis. These negative results reduce but do not eliminate the possibility that this individual is a carrier for one or more of the disorders tested. Please see Table of Residual Risks Based on Ethnicity for specific detection rates and residual risk estimates after a negative screening result. With individuals of mixed ethnicity, it is recommended to use the highest residual risk estimate. Only variants determined to have a high likelihood of pathogenicity (pathogenic or likely pathogenic) are reported in this postnatal screening test. Carrier status for autosomal recessive diseases is not reported. Next generation sequencing of the parental DNA was not performed. If indicated, only targeted testing (genotyping, Sanger sequencing, and/or copy number analysis) was performed on the parental DNA to confirm the inheritance pattern or phase of variants identified in the proband.

.REPORT_NOT_SIGNED

Laboratory Medical Consultant: George A. Diaz, M.D., Ph.D.

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Test Methods and Comments

Genomic DNA isolated from this patient was analyzed by one or more of the following methodologies, as applicable:

Genotyping
Multiplex PCR amplification and allele specific primer extension analyses using the MassARRAY® System were used to identify variants that are complex in nature or are present in low copy repeat regions and are, therefore, not amenable to next generation sequencing technologies. Rare sequence variants may interfere with assay performance.

Multiplex Ligation-Dependent Probe Amplification (MLPA)
MLPA® probe sets and reagents, from MRC-Holland, were used for the analysis of copy number of specific targets versus known control samples. Each target region was assayed with two adjacent oligonucleotide probes which following hybridization were ligated and used as template for subsequent rounds of amplification. Each complete probe within the assay has a unique length and amplicons are separated and identified by capillary electrophoresis. False positive or negative results may also occur due to rare sequence variants in target regions detected by MLPA probes. Analytical sensitivity and specificity of the MLPA method are both 99%.

For Alpha Thalassemia, the copy numbers of the *HBA1* and *HBA2* genes were analyzed. Alpha-globin gene deletions, triplications, and the Constant Spring (CS) mutation are assessed. However, this test does not detect all known alpha-thalassemia mutations such as point mutations. It is expected to detect approximately 90% of all alpha-thalassemia mutations, varying by ethnicity. Therefore, this result reduces, but does not eliminate, the chance that this patient is affected with alpha-thalassemia, defined as a loss of three or four copies of HBA.

For Spinal Muscular Atrophy (SMA), the copy number of the *SMN1* and *SMN2* genes were analyzed. The individual dosage of exons 7 and 8 as well as the combined dosage of exons 1, 4, 6 and 8 of *SMN1* and *SMN2* were assessed. Copy number gains and losses can be detected with this assay. Depending on ethnicity, approximately 2-5% of individuals affected with SMA will not be identified by dosage sensitive methods as this testing cannot detect individuals with that carry an intragenic mutation in *SMN1*.

Next Generation Sequencing (NGS)
NGS was performed on a panel of genes for the purpose of identifying pathogenic or likely pathogenic variants.

Agilent SureSelect™QXT technology was used with custom capture library to target the exonic regions and other clinically relevant regions of the below genes. These targeted regions were sequenced using the Illumina HiSeq2500 system with 100 bp paired-end reads. The DNA sequences were mapped to and analyzed in comparison with the published human genome build UCSC hg19 reference sequence. The targeted coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for the average depth of coverage and data quality threshold values. This technology may not detect all small insertion/deletions and is not diagnostic for large duplications/deletions, repeat expansions, and structural genomic variation. This testing will detect variants within the exons and the intron-exon boundaries of the target regions. Variants outside these regions will either not be detected or are not guaranteed to be detected. These regions include, but are not limited to, UTRs, promoters, and deep intronic areas or regions that fall within low copy repeat segments. In addition, a mutation(s) in a gene not included on the panel could be present in this patient. All potentially pathogenic variants may be confirmed by either a specific genotyping assay or Sanger sequencing, if indicated. Any benign variants, likely benign variants or variants of uncertain significance identified during this analysis were not reported. Carrier status for autosomal recessive diseases was not reported.

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Please note that any inconsistencies in the reported biological familial relationships could significantly change the interpretation of these results.

Variant interpretation and classification is performed based on the American College of Medical Genetics standards and guidelines for the interpretation of sequence variants (Richards et al, 2015). Frequency in control populations is evaluated based on the Genome Aggregation Database (gnomAD; http://gnomad.broadinstitute.org/). Variant interpretations, based on current knowledge, may change over time as more information arises.

Sanger Sequencing

Sanger sequencing, as indicated, was performed in both directions using BigDye Terminator chemistry with the ABI 3730 DNA analyzer with target specific amplicons. It also may be used to supplement specific guaranteed target regions that fail NGS sequencing due to poor quality or low depth of coverage <20 reads or as a confirmatory method for NGS positive results. False negative results may occur if rare variants interfere with amplification or annealing.

Disclaimer

Please note these tests were developed and their performance characteristics were determined by Mount Sinai Genomics, Inc. They have not been cleared or approved by the FDA. These analyses generally provide highly accurate information regarding the patient's carrier or affected status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

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Table of Residual Risks by Ethnicity

Please note: This table displays the residual risk of being affected with the disease after a negative result. **If a patient is reported to be affected with a disease, this table does not apply for that disease.**

Disease (Inheritance)	Gene	Ethnicity	Disease Frequency	Genotype Detection Rate	Residual Risk of Being Affected	Analytical Genotype Detection Rate
Abetalipoproteinemia (AR)	*MTTP*	African	<1 in 1,000,000	94%	1 in 16,900,000	94%
NM_000253.3		Ashkenazi Jewish	1 in 120,000	94%	1 in 2,100,000	
		East Asian	<1 in 1,000,000	65%	1 in 2,900,000	
		Caucasian	<1 in 1,000,000	63%	1 in 2,700,000	
		Latino	<1 in 1,000,000	94%	1 in 16,900,000	
		South Asian	<1 in 1,000,000	94%	1 in 16,900,000	
		Worldwide	<1 in 1,000,000	73%	1 in 3,700,000	
Acrodermatitis Enteropathica (AR)	*SLC39A4*	African	1 in 710,000	96%	1 in 17,900,000	96%
NM_130849.3		East Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Finnish	1 in 190,000	96%	1 in 4,700,000	
		Caucasian	1 in 400,000	96%	1 in 7,800,000	
		Latino	1 in 1,000,000	82%	1 in 5,500,000	
		South Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Worldwide	1 in 600,000	93%	1 in 9,000,000	
Acute Infantile Liver Failure (AR)	*TRMU*	African	<1 in 1,000,000	79%	1 in 4,700,000	98%
NM_018006.4		Ashkenazi Jewish	1 in 840,000	98%	1 in 42,400,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	84%	1 in 6,200,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	1 in 410,000	61%	1 in 1,100,000	
		Worldwide	<1 in 1,000,000	79%	1 in 4,800,000	
		Sephardic Jewish, Yemenite	1 in 4,600	66%	1 in 13,500	
Adenosine Deaminase Deficiency (AR)	*ADA*	African	1 in 33,000	85%	1 in 230,000	98%
NM_000022.2		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 610,000	85%	1 in 4,200,000	
		Latino	1 in 250,000	91%	1 in 2,900,000	
		South Asian	1 in 320,000	75%	1 in 1,300,000	
		Worldwide	1 in 370,000	83%	1 in 2,100,000	
Adrenoleukodystrophy, X-Linked (XL)	*ABCD1*	Worldwide	1 in 20,000	47%	1 in 38,000	89%
NM_000033.3						
Exception: Exons 8 and 9						
Alagille Syndrome 1 / Tetralogy of Fallot (AD)	*JAG1*	Worldwide	1 in 30,000	80%	1 in 150,000	99%
NM_000214.2						
Alpha-Mannosidosis (AR)	*MAN2B1*	African	1 in 340,000	98%	1 in 17,000,000	98%
NM_000528.3		East Asian	<1 in 1,000,000	77%	1 in 4,300,000	
		Finnish	1 in 190,000	98%	1 in 9,600,000	
		Caucasian	1 in 770,000	86%	1 in 5,700,000	
		Latino	<1 in 1,000,000	76%	1 in 4,200,000	
		South Asian	<1 in 1,000,000	47%	1 in 1,900,000	
		Worldwide	1 in 720,000	86%	1 in 5,300,000	
Alpha-Thalassemia (AR)	*HBA1 / HBA2*	Caucasian	<1 in 1,000,000	98%	1 in 20,000,000	98%
NM_000558.4 / NM_000517.4		Southeast Asian	1 in 50	98%	1 in 1,000	
		East Asian	1 in 650	98%	1 in 13,000	
		Worldwide	1 in 8,800	98%	1 in 180,000	

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Alport Syndrome (*COL4A3*-Related) (AR)	*COL4A3*	African	1 in 430,000	72%	1 in 1,600,000	98%
NM_000091.4		Ashkenazi Jewish	1 in 210,000	98%	1 in 10,300,000	
		East Asian	1 in 230,000	74%	1 in 900,000	
		Finnish	<1 in 1,000,000	65%	1 in 2,900,000	
		Caucasian	1 in 190,000	77%	1 in 830,000	
		Latino	1 in 150,000	77%	1 in 660,000	
		South Asian	1 in 520,000	80%	1 in 2,700,000	
		Worldwide	1 in 220,000	79%	1 in 1,000,000	
Alport Syndrome (*COL4A4*-Related) (AR)	*COL4A4*	African	1 in 540,000	57%	1 in 1,300,000	96%
NM_000092.4		Ashkenazi Jewish	<1 in 1,000,000	96%	1 in 25,300,000	
		East Asian	1 in 100,000	48%	1 in 190,000	
		Finnish	<1 in 1,000,000	96%	1 in 25,300,000	
		Caucasian	1 in 490,000	66%	1 in 1,400,000	
		Latino	1 in 520,000	88%	1 in 4,300,000	
		South Asian	1 in 690,000	86%	1 in 4,900,000	
		Worldwide	1 in 510,000	65%	1 in 1,500,000	
Alport Syndrome (*COL4A5*-Related) (XL)	*COL4A5*	Worldwide	1 in 60,000	80%	1 in 300,000	94%
NM_000495.3						
Argininemia (AR)	*ARG1*	African	<1 in 1,000,000	72%	1 in 3,600,000	90%
NM_000045.3		Ashkenazi Jewish	<1 in 1,000,000	9[illegible]	1 in 10,300,000	
		East Asian	<1 in 1,000,000	[illegible]%	1 in 1,200,000	
		Caucasian	<1 in 1,000,000	41%	1 in 1,700,000	
		Latino	1 in [illegible],000	43%	1 in 960,000	
		South Asian	1 in 1,[illegible]000	40%	1 in 1,700,000	
		Worldwide	<1 in 1,000,000	45%	1 in 1,800,000	
Argininosuccinic Aciduria (AR)	*ASL*	African	1 in 560,000	49%	1 in 1,100,000	98%
NM_000048.3		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	1 in 790,000	79%	1 in 3,700,000	
		Finnish	1 in 33,000	98%	1 in 1,700,000	
		Caucasian	1 in 55,000	81%	1 in 290,000	
		[illegible]	1 in 760,000	51%	1 in 1,600,000	
		South Asian	<1 in 1,000,000	67%	1 in 3,100,000	
		Worldwide	1 in 100,000	77%	1 in 450,000	
Ataxia With Isolated Vitamin E Deficiency (AR)	*TTPA*	African	1 in 410,000	98%	1 in 20,500,000	98%
NM_000370.3		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 150,000	98%	1 in 7,700,000	
Barth Syndrome (XL)	[illegible]	Worldwide	1 in 150,000	59%	1 in 370,000	97%
NM_000495.3						
Beta-Globin Related Hemoglobinopathies (AD/AR)	*HBB*	African	1 in 40,000	85%	1 in 267,000	98%
NM_000518.4		Ashkenazi Jewish	1 in 570,000	98%	1 in 28,500,000	
		East Asian	1 in 30,000	87%	1 in 231,000	
		Finnish	<1 in 1,000,000	23%	1 in 1,300,000	
		Caucasian	1 in 210,000	80%	1 in 1,050,000	
		Latino	<1 in 1,000,000	79%	1 in 4,760,000	
		South Asian	1 in 3,000	95%	1 in 60,000	
		Worldwide	1 in 30,000	91%	1 in 333,000	
		Mediterranean	1 in 3,100	>90%	1 in 31,000	
Beta-Globin Related Hemoglobinopathies: Sickling Disease (HbS and HbC) (AR)	*HBB*	African	1 in 300	>99%	1 in 30,000	>99%
NM_000518.4		Caucasian	<1 in 1,000,000	>99%	1 in 100,000,000	
		Latino	1 in 180,000	>99%	1 in 18,000,000	
		South Asian	<1 in 1,000,000	>99%	1 in 100,000,000	
Variants Tested: c.19G>A, p.E7K; c.20A>T, p.E7V		Worldwide	1 in 30,000	>99%	1 in 3,300,000	

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3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency (AR) NM_000198.3	*HSD3B2*	African	<1 in 1,000,000	79%	1 in 4,700,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	55%	1 in 2,200,000	
		Latino	<1 in 1,000,000	48%	1 in 1,900,000	
		South Asian	<1 in 1,000,000	74%	1 in 3,800,000	
		Worldwide	<1 in 1,000,000	63%	1 in 2,700,000	
Beta-Ketothiolase Deficiency (AR) NM_000019.3	*ACAT1*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	1 in 340,000	85%	1 in 2,300,000	
		Caucasian	<1 in 1,000,000	78%	1 in 4,600,000	
		Latino	1 in 120,000	92%	1 in 1,500,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	86%	1 in 7,000,000	
Beta-Mannosidosis (AR) NM_005908.3	*MANBA*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 960,000	90%	1 in 9,100,000	
		Latino	<1 in 1,000,000	90%	1 in 10,200,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	9[illegible]%	1 in 13,200,000	
BH4-Deficient Hyperphenylalaninemia C (AR) NM_000320.2	*QDPR*	Caucasian	<1 in 1,000,000	[illegible]%	1 in 1,400,000	98%
		Latino	<1 in 1,000,000	11%	1 in 1,100,000	
		South Asian	<1 in 1,000,000	44%	1 in 1,800,000	
		Worldwide	<1 in 1,000,000	30%	1 in 1,400,000	
BH4-Deficient Hyperphenylalaninemia D (AR) NM_000281.3	*PCBD1*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	1 in 710,000	98%	1 in 35,900,000	
		Caucasian	<1 in 1,000,000	66%	1 in 3,000,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	76%	1 in 4,200,000	
Biotinidase Deficiency (AR) NM_000060.3	*BTD*	African	1 in 11,000	87%	1 in 86,000	98%
		Ashkenazi Jewish	1 in 950	98%	1 in 47,000	
		East Asian	1 in 420,000	84%	1 in 2,600,000	
		Finnish	1 in 330	98%	1 in 16,000	
		Caucasian	1 in 540	96%	1 in 13,000	
		Latino	1 in 2,400	94%	1 in 38,000	
		South Asian	1 in 210	97%	1 in 6,200	
		Worldwide	1 in 690	96%	1 in 16,000	
Canavan Disease (AR) NM_000049.2	*ASPA*	African	<1 in 1,000,000	96%	1 in 25,300,000	96%
		Ashkenazi Jewish	1 in 9,900	96%	1 in 250,000	
		Finnish	1 in 230,000	96%	1 in 5,900,000	
		Caucasian	1 in 950,000	77%	1 in 4,100,000	
		Latino	<1 in 1,000,000	76%	1 in 4,200,000	
		South Asian	<1 in 1,000,000	38%	1 in 1,600,000	
		Worldwide	1 in 620,000	85%	1 in 4,200,000	
Carbamoylphosphate Synthetase I Deficiency (AR) NM_001875.4	*CPS1*	African	1 in 640,000	39%	1 in 1,100,000	96%
		Ashkenazi Jewish	<1 in 1,000,000	96%	1 in 25,300,000	
		East Asian	1 in 200,000	46%	1 in 370,000	
		Finnish	<1 in 1,000,000	53%	1 in 2,100,000	
		Caucasian	1 in 470,000	47%	1 in 880,000	
		Latino	<1 in 1,000,000	41%	1 in 1,700,000	
		South Asian	<1 in 1,000,000	21%	1 in 1,300,000	
		Worldwide	1 in 690,000	45%	1 in 1,300,000	
Carnitine Acylcarnitine Translocase Deficiency (AR) NM_001875.4	*SLC25A20*	African	<1 in 1,000,000	63%	1 in 2,700,000	85%
		East Asian	<1 in 1,000,000	85%	1 in 6,500,000	
		Caucasian	<1 in 1,000,000	49%	1 in 1,900,000	
		Latino	<1 in 1,000,000	12%	1 in 1,100,000	
		South Asian	<1 in 1,000,000	7%	1 in 1,100,000	
		Worldwide	<1 in 1,000,000	37%	1 in 1,600,000	

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Carnitine Palmitoyltransferase IA Deficiency (AR) NM_001876.3	*CPT1A*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	1 in 970,000	98%	1 in 48,500,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	1 in 280,000	93%	1 in 4,300,000	
		Caucasian	<1 in 1,000,000	88%	1 in 8,100,000	
		Latino	<1 in 1,000,000	24%	1 in 1,300,000	
		South Asian	<1 in 1,000,000	55%	1 in 2,200,000	
		Worldwide	<1 in 1,000,000	75%	1 in 4,000,000	
		Hutterite	1 in 1,000	>90%	1 in 10,000	
Carnitine Palmitoyltransferase II Deficiency (AR) NM_000098.2	*CPT2*	African	1 in 160,000	72%	1 in 560,000	98%
		Ashkenazi Jewish	1 in 6,700	98%	1 in 340,000	
		East Asian	1 in 280,000	51%	1 in 580,000	
		Finnish	1 in 250,000	98%	1 in 12,400,000	
		Caucasian	1 in 87,000	61%	1 in 220,000	
		Latino	1 in 250,000	87%	1 in 1,900,000	
		South Asian	<1 in 1,000,000	91%	1 in 11,700,000	
		Worldwide	1 in 110,000	73%	1 in 390,000	
Central Hypothyroidism and Testicular Enlargement (XL) NM_001170961.1	*IGSF1*	Worldwide	1 in 500,0	68%	1 in 1,600,000	99%
Cerebral Creatine Deficiency Syndrome 2 (AR) NM_000156.5	*GAMT*	African	<1 in 1,000,000	94%	1 in 2,200,000	96%
		Ashkenazi Jewish	<1 in 1,000,000	96%	1 in 25,300,000	
		East Asian	<1 in 1,000,000	25%	1 in 1,300,000	
		Caucasian	in 760,00	63%	1 in 2,100,000	
		Latino	in 1,000,0	54%	1 in 2,200,000	
		South Asian	<1 in 1,000,000	45%	1 in 1,800,000	
		World	<1 in 1,000,000	50%	1 in 2,000,000	
		Portugu e	1 in 63,000	>90%	1 in 630,000	
Cerebral Creatine Deficiency Syndrome 3 (AR) NM_001482.2	*GATM*	African	1 in 1,000,000	44%	1 in 1,800,000	98%
		st Asian	<1 in 1,000,000	76%	1 in 4,100,000	
		Cauc an	<1 in 1,000,000	16%	1 in 1,200,000	
		W dwide	<1 in 1,000,000	42%	1 in 1,700,000	
Cerebrotendinous Xanthomatosis (AR) NM_000784.3	*CYP27A1*	Afric	1 in 330,000	91%	1 in 3,500,000	98%
		zi Jewish	1 in 440,000	98%	1 in 22,000,000	
		East Asian	1 in 60,000	70%	1 in 200,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 300,000	86%	1 in 2,200,000	
		Latino	1 in 370,000	85%	1 in 2,400,000	
		South Asian	1 in 82,000	73%	1 in 300,000	
		Worldwide	1 in 210,000	83%	1 in 1,200,000	
		Sephardic Jewish - Moroccan	1 in 23,000	>90%	1 in 230,000	
Chronic Granulomatous Disease (*CYBA*-Related) (AR) NM_000101.2	*CYBA*	African	<1 in 1,000,000	60%	1 in 2,500,000	92%
		Finnish	<1 in 1,000,000	92%	1 in 12,800,000	
		Caucasian	<1 in 1,000,000	43%	1 in 1,800,000	
		Latino	<1 in 1,000,000	92%	1 in 12,800,000	
		South Asian	<1 in 1,000,000	37%	1 in 1,600,000	
		Worldwide	<1 in 1,000,000	49%	1 in 2,000,000	
		Sephardic Jewish - Moroccan	1 in 700	69%	1 in 2,200	
Chronic Granulomatous Disease (*CYBB*-Related) (XL) NM_000397.3	*CYBB*	Worldwide	1 in 100,000	83%	1 in 590,000	98%
Citrin Deficiency (AR) NM_014251.2	*SLC25A13*	African	1 in 760,000	56%	1 in 1,700,000	98%
		Ashkenazi Jewish	1 in 300,000	98%	1 in 15,000,000	
		East Asian	1 in 9,400	96%	1 in 220,000	
		Caucasian	<1 in 1,000,000	90%	1 in 9,800,000	
		Latino	<1 in 1,000,000	87%	1 in 7,600,000	
		South Asian	1 in 990,000	74%	1 in 3,800,000	
		Worldwide	1 in 430,000	87%	1 in 3,200,000	

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Citrullinemia, Type I (AR)	*ASS1*	African	1 in 460,000	75%	1 in 1,900,000	98%
NM_000050.4		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 420,000	76%	1 in 1,700,000	
		Latino	1 in 370,000	91%	1 in 4,100,000	
		South Asian	1 in 150,000	73%	1 in 540,000	
		Worldwide	1 in 460,000	76%	1 in 1,900,000	
Combined Pituitary Hormone Deficiency 1 (AD/AR)	*POU1F1*	East Asian	<1 in 1,000,000	3%	1 in 1,000,000	98%
NM_000306.3		Caucasian	<1 in 1,000,000	21%	1 in 1,300,000	
		Latino	<1 in 1,000,000	76%	1 in 4,100,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	76%	1 in 4,100,000	
Combined Pituitary Hormone Deficiency 2 (AR)	*PROP1*	Finnish	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_006261.4		Caucasian	1 in 930,000	69%	1 in 3,000,000	
		Latino	<1 in 1,000,000	85%	1 in 6,600,000	
		Worldwide	<1 in 1,000,000	74%	1 in 3,800,000	
Combined Pituitary Hormone Deficiency 3 (AR)	*LHX3*	East Asian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_014564.3		Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Congenital Adrenal Hyperplasia due to 11-Beta-Hydroxylase Deficiency (AR)	*CYP11B1*	African	1 in 240,000	29%	1 in 240,000 - 340,000	83%
NM_000497.3		East Asian	<1 in 1,000,000	3%	1 in 1,000,000	
		Caucasian	1 in 720,000	4%	1 in 720,000	
		Latino	1 in 1,300,000	20%	1 in 1,300,000	
		South Asian	1 in 190,000	0-47%	1 in 190,000 - 360,000	
Exception: Exons 3-7		Worldwide	1 in 770,000	2%	1 in 790,000	
Congenital Amegakaryocytic Thrombocytopenia (AR)	*MPL*	African	1 in 990,000	82%	1 in 5,600,000	98%
NM_005373.2		Ashkenazi Jewish	1 in 14,000	98%	1 in 720,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 230,000	85%	1 in 1,600,000	
		Latino	<1 in 1,000,000	72%	1 in 3,600,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 360,000	88%	1 in 2,900,000	
Congenital Bile Acid Synthesis Defect (*AKR1D1*-Related) (AR)	*AKR1D1*	East Asian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_005989.3		Caucasian	<1 in 1,000,000	77%	1 in 4,400,000	
		South Asian	<1 in 1,000,000	79%	1 in 4,700,000	
		Worldwide	<1 in 1,000,000	80%	1 in 5,100,000	
Congenital Bile Acid Synthesis Defect (*HSD3B7*-Related) (AR)	*HSD3B7*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
(*HSD3B7*-Related) (AR)		Ashkenazi Jewish	<1 in 1,000,000	4%	1 in 1,000,000	
NM_025193.3		East Asian	<1 in 1,000,000	12%	1 in 1,100,000	
		Caucasian	<1 in 1,000,000	71%	1 in 3,400,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	73%	1 in 3,600,000	
		Worldwide	<1 in 1,000,000	72%	1 in 3,500,000	
Congenital Disorder of Glycosylation Type Ib (AR)	*MPI*	African	<1 in 1,000,000	42%	1 in 1,700,000	98%
NM_002435.2		East Asian	1 in 780,000	63%	1 in 2,100,000	
		Finnish	<1 in 1,000,000	66%	1 in 2,900,000	
		Caucasian	1 in 890,000	84%	1 in 5,500,000	
		Latino	<1 in 1,000,000	85%	1 in 6,900,000	
		South Asian	<1 in 1,000,000	55%	1 in 2,200,000	
		Worldwide	<1 in 1,000,000	76%	1 in 4,200,000	
Congenital Hypothyroidism due to Thyroid Dysgenesis or Hypoplasia (AD)	*PAX8*	Worldwide	1 in 30,000	47%	1 in 57,000	99%
NM_003466.3						

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Congenital Neutropenia (*HAX1*-Related) (AR) NM_006118.3	*HAX1*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Congenital Nongoitrous Hypothryoidism 1 / Nonautoimmune Hyperthyroidism (AD/AR) NM_000369.2	*TSHR*	African	1 in 670,000	63%	1 in 1,800,000	92%
		Ashkenazi Jewish	1 in 260,000	74%	1 in 990,000	
		East Asian	1 in 34,000	37%	1 in 54,000	
		Finnish	<1 in 1,000,000	92%	1 in 12,800,000	
		Caucasian	1 in 360,000	51%	1 in 720,000	
		Latino	1 in 450,000	57%	1 in 1,000,000	
		South Asian	1 in 300,000	54%	1 in 660,000	
		Worldwide	1 in 280,000	53%	1 in 590,000	
Congenital Nongoitrous Hypothryoidism 4 (AR) NM_000549.4	*TSHB*	Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Congenital Nongoitrous Hypothryoidism 6 (AR) NM_199334.3	*THRA*	Worldwide	1 in 270,000	62%	1 in 710,000	99%
Corticosterone Methyloxidase Deficiency (AR) NM_000498.3	*CYP11B2*	African	<1 in 1,000,000	22%	1 in 1,300,000	67%
		East Asian	<1 in 1,000,000	2%	1 in 1,000,000	
		Finnish	<1 in 1,000,000	3%	1 in 1,000,000	
		Caucasian	<1 in 1,000,000	19%	1 in 1,200,000	
		Latino	<1 in 1,000,000	21%	1 in 1,300,000	
		South Asian	<1 in 1,000,000	17%	1 in 1,200,000	
		Worldwide	<1 in 1,000,000	17%	1 in 1,200,000	
Exception: Exons 3 - 7		Sephardic Jewish, Iranian	1 in 3600	>90%	1 in 36,000	
Crigler-Najjar Syndrome, Types 1 & 2 / Gilbert Syndrome (AR) NM_000463.2	*UGT1A1*	African	<1 in 1,000,000	51%	1 in 2,040,000	94%
		East Asian	1 in 90,000	38%	1 in 145,000	
		Finnish	<1 in 1,000,000	14%	1 in 1,200,000	
		Caucasian	1 in 500,000	33%	1 in 746,000	
		Latino	<1 in 1,000,000	28%	1 in 1,400,000	
		South Asian	1 in 40,000	63%	1 in 108,000	
		Worldwide	1 in 330,000	45%	1 in 600,000	
Cystic Fibrosis (AR) NM_000492.3	*CFTR*	African	1 in 13,000	83%	1 in 78,000	98%
		Ashkenazi Jewish	1 in 2,300	96%	1 in 60,000	
		East Asian	1 in 310,000	65%	1 in 870,000	
		Finnish	1 in 22,000	87%	1 in 180,000	
		Caucasian	1 in 2,100	90%	1 in 22,000	
		Latino	1 in 6,500	92%	1 in 86,000	
		South Asian	1 in 21,000	83%	1 in 120,000	
		Worldwide	1 in 4,500	88%	1 in 37,000	
Cystinosis (AR) NM_004937.2	*CTNS*	African	<1 in 1,000,000	46%	1 in 1,900,000	98%
		East Asian	1 in 620,000	89%	1 in 5,800,000	
		Caucasian	1 in 250,000	94%	1 in 3,900,000	
		Latino	<1 in 1,000,000	79%	1 in 4,800,000	
		South Asian	<1 in 1,000,000	63%	1 in 2,700,000	
		Worldwide	<1 in 1,000,000	82%	1 in 5,600,000	
		French Canadian - Saguenay-Lac St. Jean	1 in 6,000	81%	1 in 32,000	
		Sephardic Jewish - Moroccan	1 in 40,000	85%	1 in 270,000	
Distal Renal Tubular Acidosis and other *SLC4A1*-Related Disorders (AR) NM_000342.3	*SLC4A1*	African	<1 in 1,000,000	62%	1 in 2,600,000	88%
		East Asian	1 in 290,000	49%	1 in 580,000	
		Caucasian	<1 in 1,000,000	27%	1 in 1,400,000	
		Latino	<1 in 1,000,000	88%	1 in 8,600,000	
		South Asian	<1 in 1,000,000	67%	1 in 3,000,000	
		Worldwide	<1 in 1,000,000	41%	1 in 1,700,000	

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Dopa-Responsive Dystonia / BH4-Deficient Hyperphenylalaninemia B (AD/AR) NM_000161.2	*GCH1*	Worldwide	<1 in 1,000,000	64%	1 in 2,800,000	97%
Dyskeratosis Congenita (*DKC1*-Related) (XL) NM_001363.4	*DKC1*	Worldwide	1 in 5,000,000	73%	1 in 3,700,000	99%
Dyskeratosis Congenita (*RTEL1*-Related) (AR) NM_001283009.1	*RTEL1*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	1 in 49,000	98%	1 in 2,500,000	
		East Asian	1 in 590,000	81%	1 in 3,200,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	84%	1 in 6,400,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	91%	1 in 10,600,000	
Early Infantile Epileptic Encephalopathy 11/ Benign Familial Infantile Seizures 3 (AD) NM_021007.2	*SCN2A*	Worldwide	1 in 50,000	37%	1 in 79,000	98%
Early Infantile Epileptic Encephalopathy 13 / Benign Familial Infantile Seizures 5 (AD) NM_014191.3	*SCN8A*	Worldwide	1 in 7,000	42%	1 in 12,000	99%
Early Infantile Epileptic Encephalopathy 7 / Benign Neonatal Seizures 1 (AD) NM_172107.2	*KCNQ2*	Worldwide	1 in 100,000	61%	1 in 260,000	99%
Ethylmalonic Encephalopathy (AR) NM_014297.3	*ETHE1*	African	<1 in 1,000,000	96%	1 in 25,300,000	96%
		Caucasian	<1 in 1,000,000	39%	1 in 1,600,000	
		Latino	<1 in 1,000,000	86%	1 in 7,000,000	
		South Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Worldwide	<1 in 1,000,000	59%	1 in 2,400,000	
Fabry Disease (XL) NM_000169.2	*GLA*	Worldwide	1 in 4,000	74%	1 in 15,000	99%
Factor IX Deficiency (XL) NM_000133.3	*F9*	Worldwide	1 in 4,000	61%	1 in 10,000	98%
Familial Hypercholesterolemia (AR) NM_000527.4	*LDLR*	African	1 in 98,000	43%	1 in 170,000	92%
		Ashkenazi Jewish	<1 in 1,000,000	68%	1 in 3,100,000	
		East Asian	1 in 17,000	56%	1 in 39,000	
		Finnish	1 in 340,000	40%	1 in 570,000	
		Caucasian	1 in 56,000	34%	1 in 85,000	
		Latino	1 in 130,000	25%	1 in 180,000	
		South Asian	1 in 69,000	26%	1 in 93,000	
		Worldwide	1 in 64,000	35%	1 in 98,000	
		South African Afrikaner	1 in 20,000	88%	1 in 166,700	
Familial Hyperinsulinemic Hypoglycemia 4 / 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (AR) NM_005327.4	*HADH*	African	<1 in 1,000,000	43%	1 in 1,800,000	92%
		Caucasian	<1 in 1,000,000	65%	1 in 2,900,000	
		Latino	<1 in 1,000,000	23%	1 in 1,300,000	
		Worldwide	<1 in 1,000,000	41%	1 in 1,700,000	
Familial Hyperinsulinism (*ABCC8*-Related) (AR) NM_000352.4	*ABCC8*	African	1 in 260,000	22%	1 in 340,000	98%
		Ashkenazi Jewish	1 in 16,000	78%	1 in 69,000	
		East Asian	1 in 57,000	26%	1 in 76,000	
		Finnish	1 in 180,000	84%	1 in 1,100,000	
		Caucasian	1 in 150,000	33%	1 in 220,000	
		Latino	1 in 320,000	72%	1 in 1,200,000	
		South Asian	1 in 530,000	50%	1 in 1,100,000	
		Worldwide	1 in 140,000	40%	1 in 230,000	

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Familial Hyperinsulinism (*KCNJ11*-Related) (AR) NM_000525.3	*KCNJ11*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	<1 in 1,000,000	6%	1 in 1,100,000	
		Caucasian	<1 in 1,000,000	63%	1 in 2,700,000	
		Latino	<1 in 1,000,000	81%	1 in 5,200,000	
		South Asian	<1 in 1,000,000	75%	1 in 4,000,000	
		Worldwide	<1 in 1,000,000	51%	1 in 2,000,000	
Familial Infantile Convulsions with Paroxysmal Choreoathetosis (AD) NM_145239.2	*PRRT2*	Worldwide	1 in 200,000	77%	1 in 870,000	99%
Fanconi Anemia, Group A (AR) NM_000135.2	*FANCA*	African	1 in 98,000	74%	1 in 370,000	90%
		Ashkenazi Jewish	1 in 250,000	81%	1 in 1,300,000	
		East Asian	1 in 130,000	79%	1 in 650,000	
		Finnish	1 in 290,000	90%	1 in 2,900,000	
		Caucasian	1 in 87,000	76%	1 in 360,000	
		Latino	1 in 310,000	76%	1 in 1,300,000	
		South Asian	1 in 260,000	60%	1 in 660,000	
		Worldwide	1 in 110,000	77%	1 in 470,000	
Fanconi Anemia, Group C (AR) NM_000136.2	*FANCC*	African	1 in 940,0	76%	1 in 3,900,000	98%
		Ashkenazi Jewish	1 in 27,000	98%	1 in 1,300,000	
		East Asian	1 in 470,000	9	1 in 23,700,000	
		Finnish	<1 in 1,000,000	8%	1 in 50,300,000	
		Caucasian	1 740,000	93%	1 in 10,200,000	
		Latino	<1 in 000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,0 000	98%	1 in 50,300,000	
		Worldwide	in 790,0	94%	1 in 12,400,000	
Fanconi Anemia, Group G (AR) NM_001629.1	*FANCG*	African	1 in 980,000	98%	1 in 49,000,000	98%
		East A	1 in 450,000	52%	1 in 940,000	
		Finnis	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasia	in 1,000,000	96%	1 in 25,300,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		ou sian	<1 in 1,000,000	98%	1 in 50,300,000	
		W ldwide	<1 in 1,000,000	91%	1 in 10,500,000	
Fructose-1,6-Bisphosphatase Deficiency (AR) NM_000507.3	*FBP1*	Af n	<1 in 1,000,000	79%	1 in 4,800,000	79%
		East As n	<1 in 1,000,000	79%	1 in 4,800,000	
		Ca sian	<1 in 1,000,000	31%	1 in 1,400,000	
		Latino	<1 in 1,000,000	9%	1 in 1,100,000	
		South Asian	1 in 990,000	52%	1 in 2,000,000	
		Worldwide	<1 in 1,000,000	38%	1 in 1,600,000	
Galactokinase Deficiency (AR) NM_000154.1	*GALK1*	African	1 in 600,000	33%	1 in 900,000	96%
		East Asian	<1 in 1,000,000	30%	1 in 1,400,000	
		Finnish	<1 in 1,000,000	96%	1 in 25,300,000	
		Caucasian	<1 in 1,000,000	52%	1 in 2,100,000	
		Latino	<1 in 1,000,000	61%	1 in 2,600,000	
		South Asian	1 in 640,000	72%	1 in 2,300,000	
		Worldwide	<1 in 1,000,000	55%	1 in 2,200,000	
		Roma	1 in 9,000	>90%	1 in 90,000	
Galactose Epimerase Deficiency (AR) NM_000403.3	*GALE*	African	<1 in 1,000,000	9%	1 in 1,100,000	98%
		East Asian	1 in 43,000	37%	1 in 690,000	
		Caucasian	<1 in 1,000,000	43%	1 in 1,800,000	
		Latino	<1 in 1,000,000	74%	1 in 3,800,000	
		South Asian	<1 in 1,000,000	44%	1 in 1,800,000	
		Worldwide	<1 in 1,000,000	41%	1 in 1,700,000	

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Galactosemia (AR) NM_000155.3	*GALT*	African	1 in 30,000	87%	1 in 230,000	98%
		Ashkenazi Jewish	1 in 130,000	98%	1 in 8,300,000	
		East Asian	1 in 170,000	22%	1 in 220,000	
		Finnish	<1 in 1,000,000	46%	1 in 1,800,000	
		Caucasian	1 in 60,000	92%	1 in 800,000	
		Latino	1 in 190,000	91%	1 in 2,000,000	
		South Asian	1 in 470,000	81%	1 in 2,500,000	
		Worldwide	1 in 97,000	81%	1 in 500,000	
Gaucher Disease (AR) NM_000157.3	*GBA*	Ashkenazi Jewish	1 in 900	90%	1 in 9,200	90%
		Caucasian	1 in 110,000	76%	1 in 460,000	
		Worldwide	1 in 100,000	75%	1 in 400,000	
Generalized Thyrotropin-Releasing Hormone Resistance (AR) NM_003301.5	*TRHR*	East Asian	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Glucose Transporter 1 Deficiency Syndrome and Other *SLC2A1*-Related Disorders (AD/AR) NM_006516.2	*SLC2A1*	Worldwide	<1 in 1,000,000	64%	1 in 2,800,000	98%
Glutaric Acidemia, Type I (AR) NM_000159.3	*GCDH*	African	1 in 35,000	70%	1 in 120,000	98%
		East Asian	1 in 40,000	98%	1 in 8,400,000	
		Finnish	1 in 500,000	82%	1 in 2,700,000	
		Caucasian	1 in 160,000	86%	1 in 1,100,000	
		Latino	1 in 290,000	89%	1 in 2,600,000	
		South Asian	1 in 270,000	85%	1 in 1,800,000	
		Worldwide	1 in 160,000	79%	1 in 790,000	
		Oji-Cree First Nations (N. Manitoba)	1 in 300	>90%	1 in 3,000	
		Old Order Amish of Pennsylvania	1 in 500	>90%	1 in 5,000	
		Lumbee Native American	1 in 1,000	>90%	1 in 10,000	
Glutaric Acidemia, Type IIa (AR) NM_000126.3	*ETFA*	African	<1 in 1,000,000	72%	1 in 3,600,000	94%
		East Asian	<1 in 1,000,000	17%	1 in 1,200,000	
		Caucasian	<1 in 1,000,000	67%	1 in 3,000,000	
		Latino	<1 in 1,000,000	60%	1 in 2,500,000	
		South Asian	<1 in 1,000,000	94%	1 in 16,900,000	
		Worldwide	<1 in 1,000,000	69%	1 in 3,300,000	
Glutaric Acidemia, Type IIb (AR) NM_001985.2	*ETFB*	African	<1 in 1,000,000	81%	1 in 5,200,000	98%
		Ashkenazi Jewish	1 in 960,000	98%	1 in 48,500,000	
		Caucasian	<1 in 1,000,000	32%	1 in 1,500,000	
		Latino	1 in 420,000	98%	1 in 20,900,000	
		South Asian	<1 in 1,000,000	42%	1 in 1,700,000	
		Worldwide	<1 in 1,000,000	65%	1 in 2,900,000	
Glutaric Acidemia, Type IIc (AR) NM_004453.3	*ETFDH*	African	1 in 470,000	44%	1 in 840,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	1 in 32,000	43%	1 in 56,000	
		Finnish	<1 in 1,000,000	70%	1 in 3,300,000	
		Caucasian	1 in 450,000	64%	1 in 1,300,000	
		Latino	<1 in 1,000,000	34%	1 in 1,500,000	
		South Asian	<1 in 1,000,000	22%	1 in 1,300,000	
		Worldwide	1 in 460,000	51%	1 in 930,000	
Glutathione Synthetase Deficiency (AR) NM_000178.2	*GSS*	African	1 in 490,000	62%	1 in 1,300,000	94%
		Ashkenazi Jewish	1 in 47,000	94%	1 in 790,000	
		East Asian	<1 in 1,000,000	94%	1 in 16,900,000	
		Finnish	<1 in 1,000,000	94%	1 in 16,900,000	
		Caucasian	<1 in 1,000,000	47%	1 in 1,900,000	
		Latino	<1 in 1,000,000	94%	1 in 16,900,000	
		South Asian	<1 in 1,000,000	94%	1 in 16,900,000	
		Worldwide	<1 in 1,000,000	71%	1 in 3,400,000	

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Glycogen Storage Disease, Type 0 (AR)	*GYS2*	African	1 in 600,000	90%	1 in 6,200,000	90%
NM_021957.3		Ashkenazi Jewish	<1 in 1,000,000	>90%	1 in 10,000,000	
		East Asian	<1 in 1,000,000	90%	1 in 10,300,000	
		Finnish	<1 in 1,000,000	40%	1 in 1,700,000	
		Caucasian	1 in 220,000	64%	1 in 610,000	
		Latino	1 in 130,000	86%	1 in 960,000	
		South Asian	1 in 95,000	90%	1 in 970,000	
		Worldwide	1 in 340,000	71%	1 in 1,200,000	
Glycogen Storage Disease, Type Ia (AR)	*G6PC*	African	<1 in 1,000,000	78%	1 in 4,500,000	98%
NM_000151.3		Ashkenazi Jewish	1 in 22,000	98%	1 in 1,100,000	
		East Asian	1 in 54,000	52%	1 in 110,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 400,000	88%	1 in 3,500,000	
		Latino	1 in 480,000	79%	1 in 2,300,000	
		South Asian	<1 in 1,000,000	44%	1 in 1,800,000	
		Worldwide	1 in 380,000	82%	1 in 2,100,000	
Glycogen Storage Disease, Type II (AR)	*GAA*	African	1 in 20,000	67%	1 in 60,000	98%
NM_000152.3		Ashkenazi Jewish	1 in 23,00	95%	1 in 460,000	
		East Asian	1 in 16,00	61%	1 in 41,000	
		Finnish	1 in 540,000	35	1 in 820,000	
		Caucasian	1 in 9,600	%	1 in 54,000	
		Latino	in 36,000	74%	1 in 140,000	
		South Asian	1 1,000	83%	1 in 430,000	
		Worldwide	1 in 2 ,000	75%	1 in 81,000	
Glycogen Storage Disease, Type III (AR)	*AGL*	African	in 150,0	73%	1 in 550,000	98%
NM_000028.2		East Asian	1 in 1,000,000	98%	1 in 50,300,000	
		Finni	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucas	1 in 270,000	91%	1 in 3,000,000	
		Latino	1 in 880,000	93%	1 in 12,100,000	
		South Asia	<1 in 1,000,000	53%	1 in 2,100,000	
		W ide	1 in 400,000	84%	1 in 2,500,000	
		Sephardi wish ccan	1 in 5,000	>90%	1 in 50,000	
		Fa se	1 in 3,000	>90%	1 in 30,000	
Glycogen Storage Disease, Type IXb (AR)	*PHKB*	Afric	1 in 790,000	62%	1 in 2,100,000	88%
NM_000293.2		sian	1 in 630,000	88%	1 in 5,400,000	
		Caucasian	1 in 580,000	73%	1 in 2,200,000	
		Latino	1 in 680,000	17%	1 in 820,000	
		South Asian	1 in 580,000	88%	1 in 5,000,000	
		Worldwide	1 in 680,000	66%	1 in 2,000,000	
Glycogen Storage Disease, Type VI (AR)	*PYGL*	African	1 in 510,000	74%	1 in 2,000,000	90%
NM_002863.4		East Asian	1 in 420,000	71%	1 in 1,400,000	
		Finnish	<1 in 1,000,000	51%	1 in 2,000,000	
		Caucasian	1 in 830,000	52%	1 in 1,700,000	
		Latino	<1 in 1,000,000	78%	1 in 4,600,000	
		South Asian	1 in 870,000	19%	1 in 1,100,000	
		Worldwide	1 in 910,000	55%	1 in 2,000,000	
Hemolytic Anemia (*G6PD*-Related) (XL)	*G6PD*	African	1 in 4	91%	1 in 34	99%
NM_001042351.2		Ashkenazi Jewish	1 in 85	95%	1 in 1680	
		East Asian	1 in 18	61%	1 in 45	
		Caucasian	1 in 161	69%	1 in 520	
		Latino	1 in 65	65%	1 in 180	
		South Asian	1 in 11	84%	1 in 64	
		Worldwide	1 in 23	79%	1 in 106	

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Hereditary Fructose Intolerance (AR)	*ALDOB*	African	1 in 410,000	96%	1 in 10,300,000	96%
NM_000035.3		Ashkenazi Jewish	1 in 79,000	96%	1 in 2,000,000	
		East Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Finnish	1 in 40,000	96%	1 in 1,000,000	
		Caucasian	1 in 26,000	92%	1 in 320,000	
		Latino	1 in 220,000	88%	1 in 1,900,000	
		South Asian	1 in 620,000	91%	1 in 7,000,000	
		Worldwide	1 in 58,000	92%	1 in 750,000	
HMG-CoA Lyase Deficiency (AR)	*HMGCL*	African	<1 in 1,000,000	96%	1 in 25,300,000	96%
NM_000191.2		East Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Finnish	<1 in 1,000,000	96%	1 in 25,300,000	
		Caucasian	<1 in 1,000,000	46%	1 in 1,800,000	
		Latino	<1 in 1,000,000	96%	1 in 25,300,000	
		South Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Worldwide	<1 in 1,000,000	66%	1 in 3,000,000	
HMG-CoA Synthase 2 Deficiency (AR)	*HMGCS2*	African	1 in 520,000	87%	1 in 3,900,000	92%
NM_005518.3		East Asian	1 in 130,000	88%	1 in 1,100,000	
		Caucasian	1 in 910,00	58%	1 in 2,200,000	
		Latino	<1 in 1,000,0	52%	1 in 2,100,000	
		South Asian	1 in 770,000	87	1 in 5,900,000	
		Worldwide	1 in 940,000	%	1 in 2,900,000	
Holocarboxylase Synthetase Deficiency (AR)	*HLCS*	African	<1 in 1,000,000	84%	1 in 6,200,000	98%
NM_000411.6		East Asian	1 in 70,000	90%	1 in 4,900,000	
		Finnish	in 1,0 ,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,	76%	1 in 4,200,000	
		Latino	1 in 1,000,000	75%	1 in 4,000,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	83%	1 in 5,700,000	
Homocystinuria (*CBS*-Related) (AR)	*CBS*	African	1 in 140,000	90%	1 in 1,300,000	94%
NM_000071.2		kenazi Jewish	1 in 440,000	82%	1 in 2,400,000	
		sian	<1 in 1,000,000	53%	1 in 2,100,000	
		innish	1 in 450,000	88%	1 in 3,900,000	
		sian	1 in 80,000	80%	1 in 410,000	
		Latin	1 in 160,000	87%	1 in 1,300,000	
		Asian	<1 in 1,000,000	80%	1 in 5,000,000	
		Worldwide	1 in 130,000	82%	1 in 730,000	
Homocystinuria, Cobalamin E Type (AR)	*MTRR*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_002454.2		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	87%	1 in 7,700,000	
		Latino	1 in 960,000	92%	1 in 12,500,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	91%	1 in 11,200,000	
Homocystinuria-Megaloblastic Anemia, Cobalamin G Type (AR)	*MTR*	African	<1 in 1,000,000	43%	1 in 1,700,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	59%	1 in 2,400,000	
NM_000254.2		East Asian	1 in 490,000	98%	1 in 24,800,000	
		Finnish	<1 in 1,000,000	46%	1 in 1,800,000	
		Caucasian	<1 in 1,000,000	54%	1 in 2,200,000	
		Latino	<1 in 1,000,000	72%	1 in 3,600,000	
		Worldwide	<1 in 1,000,000	57%	1 in 2,300,000	
Hyperinsulinism-Hyperammonemia Syndrome (AD)	*GLUD1*	Worldwide	1 in 6,000	84%	1 in 37,000	99%
NM_005271.3						
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome (AR)	*SLC25A15*	East Asian	1 in 370,000	98%	1 in 18,400,000	98%
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
NM_014252.3		Caucasian	<1 in 1,000,000	60%	1 in 2,500,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	55%	1 in 2,200,000	
		Worldwide	<1 in 1,000,000	75%	1 in 4,000,000	
		Metis - Saskatchewan	1 in 1,400	>90%	1 in 14,000	

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Hypophosphatasia (AD/AR)	*ALPL*	African	<1 in 1,000,000	75%	1 in 4,000,000	98%
NM_000478.4		Ashkenazi Jewish	<1 in 1,000,000	44%	1 in 1,800,000	
		East Asian	1 in 69,000	95%	1 in 1,400,000	
		Finnish	1 in 3,100	92%	1 in 39,000	
		Caucasian	1 in 57,000	72%	1 in 200,000	
		Latino	1 in 800,000	24%	1 in 1,100,000	
		South Asian	<1 in 1,000,000	46%	1 in 1,800,000	
		Worldwide	1 in 55,000	79%	1 in 260,000	
		Mennonite	1 in 2,500	>90%	1 in 25,000	
Immunodeficiency 18 (AR)	*CD3E*	Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_000733.3		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Immunodeficiency 19 (AR)	*CD3D*	East Asian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_000732.4		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	89%	1 in 9,100,000	
		Latino	<1 in 1,000,000	62%	1 in 2,700,000	
		Worldwide	<1 in 1,000,000	91%	1 in 10,700,000	
Isovaleric Acidemia (AR)	*IVD*	African	1 in 370,0	77%	1 in 1,600,000	98%
NM_002225.3		East Asian	<1 in 1,000,0	61%	1 in 2,600,000	
		Finnish	<1 in 1,000,000	66	1 in 3,000,000	
		Caucasian	1 in 250,000	%	1 in 1,100,000	
		Latino	<1 in 1,000,000	80%	1 in 5,100,000	
		South Asian	<1 in ,000,000	57%	1 in 2,300,000	
		Worldwide	in 40 ,000	77%	1 in 2,000,000	
Krabbe Disease (AR)	*GALC*	African	in 100,0	28%	1 in 140,000	98%
NM_000153.3		Ashkenazi Jewis	1 in 1,000,000	70%	1 in 3,400,000	
		East A n	1 in 6,500	78%	1 in 30,000	
		Finni	1 in 86,000	98%	1 in 4,300,000	
		Caucasi	1 in 19,000	85%	1 in 120,000	
		Latino	1 in 160,000	85%	1 in 1,000,000	
		Sou Asian	1 in 5,000	83%	1 in 30,000	
		rldwi	1 in 24,000	79%	1 in 110,000	
Lipoamide Dehydrogenase Deficiency (AR)	*DLD*	Ashken Jewish	1 in 15,000	98%	1 in 730,000	98%
NM_000108.4		East A n	<1 in 1,000,000	98%	1 in 50,300,000	
		ish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	79%	1 in 4,800,000	
		Latino	<1 in 1,000,000	24%	1 in 1,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	87%	1 in 7,800,000	
Lipoid Adrenal Hyperplasia (AR)	*STAR*	African	<1 in 1,000,000	83%	1 in 5,900,000	98%
NM_000349.2		East Asian	1 in 530,000	98%	1 in 26,600,000	
		Finnish	<1 in 1,000,000	50%	1 in 2,000,000	
		Caucasian	<1 in 1,000,000	46%	1 in 1,900,000	
		Latino	<1 in 1,000,000	48%	1 in 1,900,000	
		South Asian	<1 in 1,000,000	66%	1 in 2,900,000	
		Worldwide	<1 in 1,000,000	62%	1 in 2,600,000	
Lipoprotein Lipase Deficiency (AR)	*LPL*	African	1 in 380,000	59%	1 in 930,000	98%
NM_000237.2		East Asian	1 in 43,000	76%	1 in 180,000	
		Caucasian	1 in 560,000	71%	1 in 1,900,000	
		Latino	1 in 560,000	42%	1 in 950,000	
		South Asian	1 in 820,000	25%	1 in 1,100,000	
		Worldwide	1 in 470,000	61%	1 in 1,200,000	
		French Canadian - Saguenay - Lac St. Jean	1 in 8,400	>90%	1 in 84,000	
		French Canadian - Other	1 in 77,000	>90%	1 in 770,000	

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Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (AR) NM_000182.4	*HADHA*	African	1 in 930,000	61%	1 in 2,400,000	98%
		East Asian	<1 in 1,000,000	61%	1 in 2,500,000	
		Finnish	1 in 60,000	98%	1 in 3,000,000	
		Caucasian	1 in 190,000	93%	1 in 2,600,000	
		Latino	1 in 660,000	89%	1 in 5,900,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 280,000	90%	1 in 2,700,000	
Lysinuric Protein Intolerance (AR) NM_001126106.2	*SLC7A7*	African	<1 in 1,000,000	66%	1 in 3,000,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	1 in 45,000	98%	1 in 2,300,000	
		Caucasian	<1 in 1,000,000	69%	1 in 3,200,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	82%	1 in 5,700,000	
		Worldwide	1 in 810,000	83%	1 in 4,800,000	
		Japanese	1 in 57,000	77%	1 in 250,000	
Malonyl-CoA Decarboxylase Deficiency (AR) NM_012213.2	*MLYCD*	African	<1 in 1,000,000	41%	1 in 1,700,000	74%
		Caucasian	<1 in 1,000,000	44%	1 in 1,800,000	
		Latino	<1 in 1,000,000	29%	1 in 1,400,000	
		South Asian	<1 in 1,000,000	74%	1 in 3,800,000	
		Worldwide	<1 in 1,000,000	51%	1 in 2,100,000	
Maple Syrup Urine Disease, Type 1a (AR) NM_000709.3	*BCKDHA*	African	1 in 910,000	9%	1 in 1,800,000	96%
		Ashkenazi Jewish	1 in 460,000	96%	1 in 11,600,000	
		East Asian	<1 in 1,000,000	61%	1 in 2,600,000	
		Finnish	1 in 1,000,000	96%	1 in 25,300,000	
		Caucasian	<1 in 1,000,000	80%	1 in 4,900,000	
		Latino	1 in 1,000,000	87%	1 in 7,500,000	
		South Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Worldwide	<1 in 1,000,000	80%	1 in 5,100,000	
		Mennonite	1 in 400	>90%	1 in 4,000	
		Portuguese Roma	1 in 20,000	>90%	1 in 200,000	
Maple Syrup Urine Disease, Type 1b (AR) NM_000056.3	*BCKDHB*	African	<1 in 1,000,000	58%	1 in 2,400,000	98%
		Ashkenazi Jewish	1 in 27,000	98%	1 in 1,300,000	
		East Asian	<1 in 1,000,000	70%	1 in 3,300,000	
		Finnish	1 in 130,000	98%	1 in 6,400,000	
		Caucasian	1 in 370,000	54%	1 in 800,000	
		Latino	1 in 680,000	89%	1 in 6,000,000	
		South Asian	<1 in 1,000,000	60%	1 in 2,500,000	
		Worldwide	1 in 360,000	72%	1 in 1,300,000	
Maple Syrup Urine Disease, Type 2 (AR) NM_001918.3	*DBT*	African	1 in 490,000	92%	1 in 6,300,000	92%
		Ashkenazi Jewish	<1 in 1,000,000	92%	1 in 12,800,000	
		East Asian	1 in 980,000	14%	1 in 1,100,000	
		Finnish	<1 in 1,000,000	29%	1 in 1,400,000	
		Caucasian	1 in 410,000	83%	1 in 2,400,000	
		Latino	<1 in 1,000,000	74%	1 in 3,800,000	
		South Asian	<1 in 1,000,000	11%	1 in 1,100,000	
		Worldwide	1 in 770,000	74%	1 in 2,900,000	
Marfan Syndrome and Other *FBN1*-Related Disorders (AD) NM_000138.4	*FBN1*	Worldwide	1 in 5,000	85%	1 in 33,000	>95%
Medium Chain Acyl-CoA) Dehydrogenase Deficiency (AR) NM_000016.5	*ACADM*	African	1 in 120,000	69%	1 in 400,000	98%
		Ashkenazi Jewish	1 in 71,000	98%	1 in 3,600,000	
		East Asian	1 in 270,000	70%	1 in 930,000	
		Finnish	1 in 590,000	98%	1 in 29,600,000	
		Caucasian	1 in 13,000	94%	1 in 210,000	
		Latino	1 in 67,000	81%	1 in 350,000	
		South Asian	1 in 120,000	47%	1 in 230,000	
		Worldwide	1 in 31,000	86%	1 in 220,000	

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Metachromatic Leukodystrophy (AR)	*ARSA*	African	1 in 230,000	64%	1 in 640,000	98%
NM_000487.5		Ashkenazi Jewish	<1 in 1,000,000	67%	1 in 3,100,000	
		East Asian	1 in 530,000	74%	1 in 2,100,000	
		Finnish	1 in 270,000	94%	1 in 4,100,000	
		Caucasian	1 in 68,000	76%	1 in 280,000	
		Latino	<1 in 1,000,000	81%	1 in 5,300,000	
		South Asian	1 in 550,000	67%	1 in 1,700,000	
		Worldwide	1 in 130,000	74%	1 in 500,000	
		Sephardic Jewish - Yemenite	1 in 8,000	>90%	1 in 80,000	
		Navajo	1 in 2,500	>90%	1 in 25,000	
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC1*-Related) (AR)	*MCCC1*	African	1 in 280,000	26%	1 in 380,000	98%
NM_020166.4		East Asian	1 in 170,000	61%	1 in 430,000	
		Caucasian	1 in 500,000	81%	1 in 2,600,000	
		Latino	1 in 950,000	82%	1 in 5,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 710,000	69%	1 in 2,300,000	
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC2*-Related) (AR)	*MCCC2*	African	1 in 660,000	66%	1 in 1,900,000	98%
NM_022132.4		Ashkenazi Jewish	1 in 290,00	98%	1 in 14,300,000	
		East Asian	1 in 150,00	38%	1 in 240,000	
		Finnish	<1 in 1,000,000	63	1 in 2,700,000	
		Caucasian	1 in 170,000	%	1 in 530,000	
		Latino	in 63,000	95%	1 in 1,300,000	
		South Asian	1 i 0,000	48%	1 in 730,000	
		Worldwide	1 in 1 000	70%	1 in 600,000	
Methionine Adenosyltransferase I/III Deficiency (AR)	*MAT1A*	African	<1 in 1,000,	47%	1 in 1,900,000	98%
NM_000429.2		East Asian	1 in 1,000,000	29%	1 in 1,400,000	
		Caucasian	<1 in 1,000,000	23%	1 in 1,300,000	
		Latin	<1 in 1,000,000	63%	1 in 2,700,000	
		South As n	1 in 1,000,000	47%	1 in 1,900,000	
		Worldwide	<1 in 1,000,000	34%	1 in 1,500,000	
Methylmalonic Acidemia (*MMAA*-Related) (AR)	*MMAA*	East Asian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_172250.2		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	91%	1 in 11,100,000	
		Latin	<1 in 1,000,000	75%	1 in 4,000,000	
		South Asian	<1 in 1,000,000	84%	1 in 6,100,000	
		Worldwide	<1 in 1,000,000	90%	1 in 9,500,000	
Methylmalonic Acidemia (*MMAB*-Related) (AR)	*MMAB*	African	<1 in 1,000,000	32%	1 in 1,500,000	98%
NM_052845.3		Finnish	<1 in 1,000,000	43%	1 in 1,800,000	
		Caucasian	<1 in 1,000,000	89%	1 in 9,100,000	
		Latino	<1 in 1,000,000	17%	1 in 1,200,000	
		South Asian	<1 in 1,000,000	24%	1 in 1,300,000	
		Worldwide	<1 in 1,000,000	59%	1 in 2,500,000	
Methylmalonic Acidemia (*MUT*-Related) (AR)	*MUT*	African	1 in 110,000	78%	1 in 510,000	98%
NM_000255.3		Ashkenazi Jewish	1 in 430,000	98%	1 in 21,700,000	
		East Asian	1 in 140,000	60%	1 in 360,000	
		Finnish	<1 in 1,000,000	74%	1 in 3,800,000	
		Caucasian	1 in 350,000	59%	1 in 860,000	
		Latino	1 in 150,000	91%	1 in 1,800,000	
		South Asian	1 in 280,000	62%	1 in 730,000	
		Worldwide	1 in 250,000	70%	1 in 840,000	
Methylmalonic Aciduria and, Homocystinuria Cobalamin C Type (AR)	*MMACHC*	African	1 in 310,000	89%	1 in 2,900,000	98%
NM_015506.2		Ashkenazi Jewish	1 in 160,000	98%	1 in 8,300,000	
		East Asian	1 in 140,000	75%	1 in 530,000	
		Caucasian	1 in 120,000	95%	1 in 2,400,000	
		Latino	1 in 42,000	98%	1 in 2,100,000	
		South Asian	1 in 210,000	76%	1 in 880,000	
		Worldwide	1 in 130,000	92%	1 in 1,700,000	

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Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type (AR) NM_015702.2	*MMADHC*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Methylmalonic Aciduria and Homocystinuria, Cobalamin F Type (AR) NM_018368.3	*LMBRD1*	African	1 in 510,000	74%	1 in 1,900,000	90%
		East Asian	<1 in 1,000,000	90%	1 in 10,300,000	
		Finnish	<1 in 1,000,000	90%	1 in 10,300,000	
		Caucasian	1 in 680,000	88%	1 in 5,600,000	
		Latino	<1 in 1,000,000	90%	1 in 10,300,000	
		South Asian	<1 in 1,000,000	90%	1 in 10,300,000	
		Worldwide	<1 in 1,000,000	87%	1 in 7,800,000	
Methylmalonyl-CoA Epimerase Deficiency NM_032601.3	*MCEE*	Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Mitochondrial Trifunctional Protein Deficiency (*HADHB*-Related) (AR) NM_000183.2	*HADHB*	African	<1 in 1,000,000	67%	1 in 3,000,000	90%
		East Asian	<1 in 1,000,000	37%	1 in 1,600,000	
		Finnish	<1 in 1,000,000	36%	1 in 1,600,000	
		Caucasian	<1 in 1,000,000	6[illegible]%	1 in 2,600,000	
		Latino	<1 in 1,000,000	[illegible]2%	1 in 5,600,000	
		South Asian	<1 in 1,000,000	75%	1 in 3,900,000	
		Worldwide	<1 in 1,000,000	62%	1 in 2,600,000	
Mucolipidosis II / IIIA (AR) NM_024312.4	*GNPTAB*	African	1 in 430,000	98%	1 in 21,600,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	1 in 540,000	46%	1 in 1,000,000	
		Finnish	1 in 100,000	98%	1 in 5,100,000	
		Caucasian	1 in 200,000	80%	1 in 970,000	
		Latino	1 in 330,000	82%	1 in 1,800,000	
		South Asian	1 in 410,000	94%	1 in 6,800,000	
		Worldwide	1 in 24,000	83%	1 in 1,500,000	
Mucopolysaccharidosis Type I (AR) NM_000203.4	*IDUA*	African	1 in 560,000	82%	1 in 3,100,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	1 in 220,000	40%	1 in 370,000	
		Finnish	1 in 140,000	98%	1 in 6,800,000	
		Caucasian	1 in 53,000	93%	1 in 780,000	
		Latino	1 in 690,000	84%	1 in 4,300,000	
		South Asian	1 in 52,000	95%	1 in 950,000	
		Worldwide	1 in 83,000	90%	1 in 810,000	
Mucopolysaccharidosis, Type II (XL) NM_000202.6 *Exception:* Exon 3	*IDS*	Worldwide	1 in 50,000	67%	1 in 150,000	90%
Mucopolysaccharidosis Type IVa (AR) NM_000512.4	*GALNS*	African	1 in 470,000	33%	1 in 700,000	92%
		Ashkenazi Jewish	<1 in 1,000,000	52%	1 in 2,100,000	
		East Asian	1 in 330,000	13%	1 in 370,000	
		Finnish	<1 in 1,000,000	74%	1 in 3,900,000	
		Caucasian	1 in 260,000	39%	1 in 430,000	
		Latino	1 in 440,000	54%	1 in 960,000	
		South Asian	1 in 440,000	14%	1 in 520,000	
		Worldwide	1 in 310,000	41%	1 in 520,000	
Mucopolysaccharidosis Type VI (AR) NM_000046.3	*ARSB*	African	<1 in 1,000,000	33%	1 in 1,500,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	73%	1 in 3,600,000	
		Caucasian	1 in 390,000	57%	1 in 910,000	
		Latino	<1 in 1,000,000	55%	1 in 2,200,000	
		South Asian	<1 in 1,000,000	72%	1 in 3,600,000	
		Worldwide	<1 in 1,000,000	54%	1 in 2,200,000	

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N-Acetylglutamate Synthase Deficiency (AR)	*NAGS*	African	<1 in 1,000,000	70%	1 in 3,400,000	98%
NM_153006.2		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	51%	1 in 2,100,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	37%	1 in 1,600,000	
		Worldwide	<1 in 1,000,000	70%	1 in 3,300,000	
Nephrogenic Diabetes Insipidus (*AVPR2*-Related) / Nephrogenic Syndrome of Inappropriate Antidiuresis (XL)	*AVPR2*	Worldwide	1 in 320,000	66%	1 in 940,000	96%
NM_000054.4						
Nephrogenic Diabetes Insipidus, Type II (AD/AR)	*AQP2*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	<1 in 1,000,000	83%	1 in 6,000,000	
NM_000486.5		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	62%	1 in 2,700,000	
		Latino	1 in 840,000	93%	1 in 11,600,000	
		South Asian	<1 in 1,000,000	35%	1 in 1,500,000	
		Worldwide	<1 in 1,000,000	75%	1 in 4,000,000	
Neurodegeneration due to Cerebral Folate Transport Deficiency (AR)	*FOLR1*	African	<1 in 1,000,000	15%	1 in 1,200,000	72%
		Finnish	1 in 140,000	4[illegible]	1 in 280,000	
NM_016725.2		Caucasian	<1 in 1,000,000	[illegible]2%	1 in 2,600,000	
		South Asian	<1 in 1,000,000	41%	1 in 1,700,000	
		Worldwide	<1 in 1,000,000	54%	1 in 2,200,000	
Neuronal Ceroid-Lipofuscinosis (*CLN3*-Related) (AR)	*CLN3*	African	<1 in 1,000,000	59%	1 in 2,500,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
NM_000089.2		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 240,000	95%	1 in 4,500,000	
		Latino	<1 in 1,000,000	51%	1 in 2,000,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 750,000	93%	1 in 10,300,000	
Neuronal Ceroid-Lipofuscinosis (*CLN5*-Related) (AR)	*CLN5*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
NM_006493.2		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	68%	1 in 3,100,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	46%	1 in 1,900,000	
		Worldwide	<1 in 1,000,000	80%	1 in 5,100,000	
Neuronal Ceroid-Lipofuscinosis (*CLN6*-Related) (AR)	*CLN6*	African	<1 in 1,000,000	63%	1 in 2,700,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
NM_017882.2		Caucasian	<1 in 1,000,000	78%	1 in 4,600,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	89%	1 in 9,000,000	
		Worldwide	<1 in 1,000,000	85%	1 in 6,700,000	
Neuronal Ceroid-Lipofuscinosis (*CLN8*-Related) (AR)	*CLN8*	African	<1 in 1,000,000	31%	1 in 1,500,000	98%
		East Asian	<1 in 1,000,000	25%	1 in 1,300,000	
NM_018941.3		Finnish	1 in 630,000	85%	1 in 4,100,000	
		Caucasian	<1 in 1,000,000	33%	1 in 1,500,000	
		Latino	<1 in 1,000,000	44%	1 in 1,800,000	
		South Asian	<1 in 1,000,000	55%	1 in 2,200,000	
		Worldwide	<1 in 1,000,000	52%	1 in 2,100,000	
Neuronal Ceroid-Lipofuscinosis (*MFSD8*-Related) (AR)	*MFSD8*	African	<1 in 1,000,000	66%	1 in 3,000,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
NM_152778.2		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	83%	1 in 5,900,000	
		Latino	<1 in 1,000,000	70%	1 in 3,400,000	
		South Asian	1 in 920,000	86%	1 in 6,700,000	
		Worldwide	<1 in 1,000,000	86%	1 in 7,100,000	

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Neuronal Ceroid-Lipofuscinosis (*PPT1*-Related) (AR) NM_000310.3	*PPT1*	African	<1 in 1,000,000	83%	1 in 5,700,000	98%
		East Asian	<1 in 1,000,000	31%	1 in 1,500,000	
		Finnish	1 in 22,000	98%	1 in 1,100,000	
		Caucasian	1 in 290,000	93%	1 in 4,100,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	90%	1 in 10,000,000	
		Worldwide	1 in 320,000	93%	1 in 4,600,000	
Neuronal Ceroid-Lipofuscinosis (*TPP1*-Related) (AR) NM_000391.3	*TPP1*	African	<1 in 1,000,000	36%	1 in 1,600,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	<1 in 1,000,000	26%	1 in 1,400,000	
		Finnish	1 in 500,000	98%	1 in 25,200,000	
		Caucasian	1 in 280,000	92%	1 in 3,400,000	
		Latino	<1 in 1,000,000	79%	1 in 4,800,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 570,000	87%	1 in 4,500,000	
Niemann-Pick Disease (*SMPD1*-Related) (AR) NM_000543.4	*SMPD1*	African	1 in 57,000	80%	1 in 290,000	98%
		Ashkenazi Jewish	1 in 39,000	98%	1 in 1,900,000	
		East Asian	1 in 26,00	88%	1 in 220,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 490,000	%	1 in 1,400,000	
		Latino	n 1,000,000	76%	1 in 4,200,000	
		South Asian	1 30,000	57%	1 in 1,000,000	
		Worldwide	1 in 2 000	77%	1 in 990,000	
Omenn Syndrome (*RAG2*-Related) (AR) NM_000536.2	*RAG2*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewi	1 in 1,000,000	98%	1 in 50,300,000	
		Finnis	<1 in 1,000,000	98%	1 in 50,300,000	
		Cauca	<1 in 1,000,000	73%	1 in 3,700,000	
		South As	<1 in 1,000,000	6%	1 in 1,100,000	
		Worldwid	<1 in 1,000,000	66%	1 in 2,900,000	
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type (AR) NM_001033855.1	*DCLRE1C*	rican	<1 in 1,000,000	88%	1 in 8,400,000	96%
		st A	<1 in 1,000,000	96%	1 in 25,300,000	
		nish	<1 in 1,000,000	58%	1 in 2,400,000	
		Cauca n	<1 in 1,000,000	70%	1 in 3,300,000	
		tino	<1 in 1,000,000	75%	1 in 4,100,000	
		South Asian	<1 in 1,000,000	48%	1 in 1,900,000	
		Worldwide	<1 in 1,000,000	76%	1 in 4,200,000	
		Navajo and Apache Native American	1 in 9,000	>90%	1 in 90,000	
Omenn Syndrome and other *RAG1*-Related Disorders (AR) NM_000448.2	*RAG1*	African	1 in 610,000	46%	1 in 1,100,000	94%
		Ashkenazi Jewish	<1 in 1,000,000	23%	1 in 1,300,000	
		East Asian	1 in 62,000	10%	1 in 69,000	
		Finnish	<1 in 1,000,000	50%	1 in 2,000,000	
		Caucasian	1 in 420,000	38%	1 in 680,000	
		Latino	1 in 850,000	57%	1 in 2,000,000	
		South Asian	<1 in 1,000,000	33%	1 in 1,500,000	
		Worldwide	1 in 450,000	35%	1 in 690,000	
Ornithine Aminotransferase Deficiency (AR) NM_000274.3	*OAT*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	1 in 76,000	98%	1 in 3,800,000	
		Caucasian	<1 in 1,000,000	78%	1 in 4,600,000	
		Latino	<1 in 1,000,000	47%	1 in 1,900,000	
		South Asian	<1 in 1,000,000	57%	1 in 2,300,000	
		Worldwide	<1 in 1,000,000	83%	1 in 6,000,000	
		Sephardic Jewish - Iraqi and Syrian	1 in 125,000	>90%	1 in 1,300,000	
Ornithine Transcarbamylase Deficiency (XL) NM_000531.5	*OTC*	Worldwide	1 in 60,000	71%	1 in 210,000	99%

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Osteopetrosis 1 (AR) NM_006019.2	*TCIRG1*	African	1 in 700,000	76%	1 in 2,900,000	96%
		Ashkenazi Jewish	1 in 970,000	78%	1 in 4,400,000	
		East Asian	1 in 420,000	89%	1 in 3,800,000	
		Finnish	<1 in 1,000,000	96%	1 in 25,300,000	
		Caucasian	1 in 640,000	84%	1 in 3,900,000	
		Latino	1 in 690,000	96%	1 in 17,300,000	
		South Asian	<1 in 1,000,000	78%	1 in 4,600,000	
		Worldwide	1 in 640,000	87%	1 in 4,800,000	
		Costa Rican	1 in 30,000	>90%	1 in 300,000	
		Chuvashiyan	1 in 14,000	>90%	1 in 140,000	
Permanent Neonatal Diabetes Mellitus (*INS*-Related) (AD) NM_000207.2	*INS*	Worldwide	1 in 220,000	70%	1 in 730,000	97%
Phenylalanine Hydroxylase Deficiency (AR) NM_000277.1	*PAH*	African	1 in 82,000	74%	1 in 320,000	98%
		Ashkenazi Jewish	1 in 1,100	97%	1 in 41,000	
		East Asian	1 in 18,000	29%	1 in 26,000	
		Finnish	1 in 99,000	57%	1 in 230,000	
		Caucasian	1 in 5,60[illegible]	79%	1 in 27,000	
		Latino	1 in 20,000	76%	1 in 84,000	
		South Asian	1 in 59,000	66[illegible]	1 in 170,000	
		Worldwide	1 in 10,000	[illegible]%	1 in 43,000	
		Turkish	[illegible] in 4,000	40%	1 in 6,700	
		Irish	1 [illegible],000	83%	1 in 29,400	
		Sicilian	1 in [illegible]00	23%	1 in 3,900	
		Sephardic Jewish - Iranian, [illegible]ukharian, Kavkazi, Tunisian and [illegible]occan	1 in 1,30[illegible]	77%	1 in 5,600	
3-Phosphoglycerate Dehydrogenase Deficiency (AR) NM_006623.3	*PHGDH*	Afric[illegible]	<1 in 1,000,000	41%	1 in 1,700,000	98%
		Ashkenazi [illegible]h	1 in 360,000	98%	1 in 17,900,000	
		East Asi[illegible]	[illegible] in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Ca[illegible]sian	<1 in 1,000,000	98%	1 in 50,300,000	
		[illegible]atino	<1 in 1,000,000	47%	1 in 1,900,000	
		Sou[illegible] Asian	<1 in 1,000,000	60%	1 in 2,500,000	
		World[illegible]e	<1 in 1,000,000	89%	1 in 8,900,000	
Primary Carnitine Deficiency (AR) NM_003060.2	*SLC22A5*	[illegible]an	1 in 38,000	89%	1 in 340,000	96%
		Ashkenazi Jewish	<1 in 1,000,000	96%	1 in 25,300,000	
		East Asian	1 in 19,000	79%	1 in 88,000	
		Finnish	<1 in 1,000,000	65%	1 in 2,900,000	
		Caucasian	1 in 250,000	69%	1 in 800,000	
		Latino	1 in 290,000	74%	1 in 1,100,000	
		South Asian	1 in 11,000	92%	1 in 140,000	
		Worldwide	1 in 83,000	82%	1 in 460,000	
		Faroese	1 in 1,600	>90%	1 in 16,000	
Primary Hyperoxaluria, Type 1 (AR) NM_000030.2	*AGXT*	African	1 in 430,000	78%	1 in 1,900,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	56%	1 in 2,300,000	
		East Asian	1 in 72,000	89%	1 in 660,000	
		Finnish	<1 in 1,000,000	66%	1 in 3,000,000	
		Caucasian	1 in 180,000	80%	1 in 860,000	
		Latino	1 in 690,000	73%	1 in 2,500,000	
		South Asian	1 in 370,000	70%	1 in 1,200,000	
		Worldwide	1 in 260,000	79%	1 in 1,200,000	
Primary Hyperoxaluria, Type 2 (AR) NM_012203.1	*GRHPR*	African	<1 in 1,000,000	43%	1 in 1,700,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 750,000	92%	1 in 9,400,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	1 in 430,000	94%	1 in 7,000,000	
		Worldwide	1 in 960,000	86%	1 in 6,600,000	

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Primary Hyperoxaluria, Type 3 (AR)	*HOGA1*	African	1 in 640,000	92%	1 in 7,600,000	98%
NM_138413.3		Ashkenazi Jewish	1 in 5,400	98%	1 in 270,000	
		East Asian	1 in 59,000	98%	1 in 3,000,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 110,000	87%	1 in 860,000	
		Latino	1 in 350,000	88%	1 in 2,900,000	
		South Asian	<1 in 1,000,000	80%	1 in 5,100,000	
		Worldwide	1 in 140,000	90%	1 in 1,400,000	
Propionic Acidemia (*PCCA*-Related) (AR)	*PCCA*	African	1 in 620,000	51%	1 in 1,300,000	86%
NM_000282.3		Ashkenazi Jewish	<1 in 1,000,000	68%	1 in 3,200,000	
		East Asian	1 in 700,000	70%	1 in 2,400,000	
		Finnish	<1 in 1,000,000	86%	1 in 7,400,000	
		Caucasian	<1 in 1,000,000	57%	1 in 2,300,000	
		Latino	1 in 740,000	35%	1 in 1,100,000	
		South Asian	<1 in 1,000,000	60%	1 in 2,500,000	
		Worldwide	1 in 970,000	50%	1 in 1,900,000	
Propionic Acidemia (*PCCB*-Related) (AR)	*PCCB*	African	1 in 270,000	92%	1 in 3,100,000	98%
NM_000532.4		East Asian	1 in 150,00	63%	1 in 400,000	
		Finnish	<1 in 1,000,0	80%	1 in 5,000,000	
		Caucasian	<1 in 1,000,000	9	1 in 9,900,000	
		Latino	<1 in 1,000,000	%	1 in 2,600,000	
		South Asian	<1 in 1,000,000	59%	1 in 2,400,000	
		Worldwide	<1 in ,000,000	80%	1 in 4,900,000	
Pyridoxamine 5'-Phosphate Oxidase Deficiency (AR)	*PNPO*	African	in 1, 000	92%	1 in 12,800,000	92%
		East Asian	<1 in 1,000,	72%	1 in 3,600,000	
NM_018129.3		Finnish	1 in 1,000,000	71%	1 in 3,500,000	
		Cauca n	<1 in 1,000,000	78%	1 in 4,500,000	
		Latin	<1 in 1,000,000	92%	1 in 12,800,000	
		South As n	1 in 1,000,000	59%	1 in 2,400,000	
		Worldwide	<1 in 1,000,000	78%	1 in 4,500,000	
Pyridoxine-Dependent Epilepsy (AR)	*ALDH7A1*	an	1 in 200,000	55%	1 in 440,000	92%
NM_001182.4		t Asi	1 in 290,000	56%	1 in 650,000	
		ish	<1 in 1,000,000	21%	1 in 1,300,000	
		Cauca n	1 in 380,000	53%	1 in 800,000	
		ino	1 in 780,000	74%	1 in 3,000,000	
		South Asian	1 in 870,000	41%	1 in 1,500,000	
		Worldwide	1 in 430,000	52%	1 in 900,000	
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (AR)	*PTS*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
NM_000317.2		East Asian	1 in 97,000	89%	1 in 910,000	
		Finnish	1 in 530,000	81%	1 in 2,700,000	
		Caucasian	1 in 920,000	55%	1 in 2,000,000	
		Latino	<1 in 1,000,000	65%	1 in 2,800,000	
		South Asian	1 in 470,000	70%	1 in 1,600,000	
		Worldwide	1 in 630,000	66%	1 in 1,800,000	
Retinoblastoma (AD)	*RB1*	Worldwide	1 in 20,000	80%	1 in 100,000	96%
NM_000321.2						
Segawa Syndrome (AR)	*TH*	African	<1 in 1,000,000	45%	1 in 1,800,000	98%
NM_000360.3		East Asian	1 in 370,000	80%	1 in 1,900,000	
		Caucasian	<1 in 1,000,000	74%	1 in 3,800,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	76%	1 in 4,200,000	
Sepiapterin Reductase Deficiency (AR)	*SPR*	Finnish	<1 in 1,000,000	92%	1 in 12,800,000	92%
NM_003124.4		Caucasian	<1 in 1,000,000	92%	1 in 12,800,000	
		Latino	<1 in 1,000,000	92%	1 in 12,800,000	
		South Asian	<1 in 1,000,000	92%	1 in 12,800,000	
		Worldwide	<1 in 1,000,000	92%	1 in 12,800,000	

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Severe Combined Immunodeficiency (*IL7R*-Related) (AR) NM_002185.3	*IL7R*	African	<1 in 1,000,000	88%	1 in 8,600,000	88%
		East Asian	<1 in 1,000,000	88%	1 in 8,600,000	
		Caucasian	<1 in 1,000,000	88%	1 in 8,600,000	
		Latino	<1 in 1,000,000	88%	1 in 8,600,000	
		South Asian	<1 in 1,000,000	88%	1 in 8,600,000	
		Worldwide	<1 in 1,000,000	88%	1 in 8,600,000	
Severe Combined Immunodeficiency (*JAK3*-Related) (AR) NM_000215.3	*JAK3*	African	<1 in 1,000,000	96%	1 in 25,300,000	96%
		Caucasian	<1 in 1,000,000	21%	1 in 1,300,000	
		Latino	<1 in 1,000,000	26%	1 in 1,300,000	
		South Asian	<1 in 1,000,000	54%	1 in 2,200,000	
		Worldwide	<1 in 1,000,000	52%	1 in 2,100,000	
Severe Combined Immunodeficiency (*PTPRC*-Related) (AR)	*PTPRC*	East Asian	1 in 860,000	64%	1 in 2,400,000	64%
		Caucasian	<1 in 1,000,000	64%	1 in 2,800,000	
		Latino	<1 in 1,000,000	64%	1 in 2,800,000	
		Worldwide	<1 in 1,000,000	64%	1 in 2,800,000	
Severe Neonatal Hyperparathyroidism / Autosomal Dominant Hypocalcemia) (AD/AR) NM_000388.3	*CASR*	African	<1 in 1,000,000	33%	1 in 1,500,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	13%	1 in 1,200,000	
		South Asian	<1 in 1,000,000	55%	1 in 2,200,000	
		Worldwide	**<1 in 1,000,000**	**26%**	**1 in 1,400,000**	
Spherocytosis, Type 1 (AD) NM_000037.3	*ANK1*	Worldwide	1 in 2,000	2%	1 in 11,000	99%
Spherocytosis, Type 5 (AR) NM_000119.2	*EPB42*	African	1 in 20,000	60%	1 in 2,000,000	86%
		Ashkenazi Jewish	1 in 30,000	86%	1 in 220,000	
		East Asian	1 in 690,000	45%	1 in 1,300,000	
		Finnish	1 in 690,000	86%	1 in 5,100,000	
		Caucasian	<1 in 1,000,000	57%	1 in 2,300,000	
		Latino	<1 in 1,000,000	37%	1 in 1,600,000	
		South Asian	<1 in 1,000,000	86%	1 in 7,400,000	
		Worldwide	1 in 680,000	71%	1 in 2,300,000	
Spinal Muscular Atrophy (AR) NM_000344.3	*SMN1*	African	1 in 17,000	>95%	1 in 440,000	>95%
		Ashkenazi Jewish	1 in 7,000	>95%	1 in 140,000	
		Asian	1 in 11,000	>95%	1 in 220,000	
		Caucasian	1 in 5,000	>95%	1 in 100,000	
		Latino	1 in 55,000	>95%	1 in 1,100,000	
		Worldwide	1 in 10,000	>95%	1 in 200,000	
Tay-Sachs Disease (AR) NM_000520.4	*HEXA*	African	1 in 190,000	98%	1 in 9,300,000	98%
		Ashkenazi Jewish	1 in 3,600	98%	1 in 180,000	
		East Asian	1 in 180,000	98%	1 in 8,900,000	
		Finnish	1 in 640,000	98%	1 in 32,000,000	
		Caucasian	1 in 33,000	95%	1 in 640,000	
		Latino	1 in 240,000	79%	1 in 1,100,000	
		South Asian	1 in 690,000	48%	1 in 1,300,000	
		Worldwide	1 in 59,000	93%	1 in 800,000	
Thyroid Dyshormonogenesis 1 (AR) NM_000453.2	*SLC5A5*	African	<1 in 1,000,000	88%	1 in 8,600,000	88%
		Ashkenazi Jewish	1 in 800,000	88%	1 in 6,900,000	
		East Asian	<1 in 1,000,000	57%	1 in 2,300,000	
		Caucasian	<1 in 1,000,000	88%	1 in 8,600,000	
		Latino	1 in 270,000	88%	1 in 2,300,000	
		South Asian	<1 in 1,000,000	88%	1 in 8,600,000	
		Worldwide	<1 in 1,000,000	83%	1 in 6,000,000	
Thyroid Dyshormonogenesis 2A (AR) NM_000547.5	*TPO*	African	1 in 160,000	25%	1 in 210,000	96%
		Ashkenazi Jewish	1 in 160,000	30%	1 in 220,000	
		East Asian	1 in 13,000	71%	1 in 43,000	
		Finnish	1 in 160,000	43%	1 in 290,000	
		Caucasian	1 in 140,000	63%	1 in 380,000	
		Latino	1 in 150,000	57%	1 in 350,000	
		South Asian	<1 in 1,000,000	18%	1 in 1,200,000	
		Worldwide	1 in 120,000	54%	1 in 250,000	

Thyroid Dyshormonogenesis 3 (AR)	*TG*	African	1 in 65,000	75%	1 in 260,000	96%
NM_003235.4		Ashkenazi Jewish	1 in 81,000	91%	1 in 870,000	
		East Asian	1 in 17,000	24%	1 in 22,000	
		Finnish	1 in 220,000	53%	1 in 470,000	
		Caucasian	1 in 120,000	64%	1 in 330,000	
		Latino	1 in 91,000	91%	1 in 1,000,000	
		South Asian	<1 in 1,000,000	82%	1 in 5,500,000	
		Worldwide	1 in 89,000	59%	1 in 220,000	
Thyroid Dyshormonogenesis 4 (AR)	*IYD*	African	1 in 910,000	83%	1 in 5,400,000	98%
NM_203395.2		Caucasian	<1 in 1,000,000	44%	1 in 1,800,000	
		Latino	<1 in 1,000,000	33%	1 in 1,500,000	
		South Asian	<1 in 1,000,000	18%	1 in 1,200,000	
		Worldwide	<1 in 1,000,000	57%	1 in 2,300,000	
Thyroid Dyshormonogenesis 5 (AR)	*DUOXA2*	African	1 in 810,000	88%	1 in 6,900,000	88%
NM_207581.3		East Asian	1 in 33,000	87%	1 in 240,000	
		Caucasian	<1 in 1,000,000	88%	1 in 8,600,000	
		Latino	<1 in 1,000,000	88%	1 in 8,600,000	
		South Asian	<1 in 1,000,000	88%	1 in 8,600,000	
		Worldwide	<1 in 1,000,000	87%	1 in 8,000,000	
Thyroid Dyshormonogenesis 6 (AR)	*DUOX2*	African	1 in 31,000	67%	1 in 95,000	98%
NM_207581.3		Ashkenazi Jewish	<1 in 1,000,000	2%	1 in 2,600,000	
		East Asian	1 in 500	45%	1 in 900	
		Finnish	1 in 6,000	87%	1 in 46,000	
		Caucasian	1 in 1,000	48%	1 in 28,000	
		Latino	1 in 42,000	54%	1 in 91,000	
		South Asian	1 in 1,000,000	18%	1 in 1,200,000	
		Worldwide	1 in 9,900	53%	1 in 21,000	
Exception: Exons 6 and 7						
Tyrosinemia, Type I (AR)	*FAH*	African	1 in 510,000	83%	1 in 2,900,000	98%
NM_000137.2		Ashkenazi Jewish	1 in 71,000	98%	1 in 3,600,000	
		East Asian	<1 in 1,000,000	15%	1 in 1,200,000	
		Finnish	1 in 420,000	98%	1 in 20,900,000	
		Caucasian	1 in 270,000	74%	1 in 1,000,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 410,000	79%	1 in 1,900,000	
		French Canadian - Saguenay	1 in 2,500	>90%	1 in 25,000	
		Lac-St. Jean French Canadian - Other	1 in 17,000	>90%	1 in 170,000	
Tyrosinemia, Type II (AR)	*TAT*	African	<1 in 1,000,000	88%	1 in 8,600,000	88%
NM_000353.2		East Asian	<1 in 1,000,000	10%	1 in 1,100,000	
		Caucasian	<1 in 1,000,000	22%	1 in 1,300,000	
		South Asian	<1 in 1,000,000	53%	1 in 2,100,000	
		Worldwide	<1 in 1,000,000	41%	1 in 1,700,000	
Tyrosinemia, Type III (AR)	*HPD*	Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_002150.2		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	68%	1 in 3,200,000	
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (AR)	*ACADVL*	African	1 in 85,000	57%	1 in 200,000	96%
NM_000018.3		Ashkenazi Jewish	<1 in 1,000,000	53%	1 in 2,100,000	
		East Asian	1 in 160,000	22%	1 in 210,000	
		Finnish	1 in 340,000	87%	1 in 2,700,000	
		Caucasian	1 in 48,000	78%	1 in 220,000	
		Latino	1 in 290,000	45%	1 in 520,000	
		South Asian	1 in 550,000	52%	1 in 1,100,000	
		Worldwide	1 in 97,000	69%	1 in 310,000	
Wilms Tumor, Type 1 and Other *WT1*-Related Disorders (AD)	*WT1*	Worldwide	1 in 200,000	79%	1 in 950,000	99%
NM_024426.4						

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Wolman Disease / Cholesteryl Ester Storage Disease (AR) NM_000235.3	*LIPA*	African	<1 in 1,000,000	71%	1 in 3,400,000	96%
		Ashkenazi Jewish	<1 in 1,000,000	96%	1 in 25,300,000	
		East Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Finnish	<1 in 1,000,000	60%	1 in 2,500,000	
		Caucasian	1 in 220,000	86%	1 in 1,600,000	
		Latino	1 in 430,000	72%	1 in 1,500,000	
		South Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Worldwide	1 in 430,000	84%	1 in 2,700,000	
		Sephardic Jewish - Iranian	1 in 2,700	>90%	1 in 27,000	
X-Linked Severe Combined Immunodeficiency (XL) NM_000206.2	*IL2RG*	Worldwide	1 in 50,000	90%	1 in 500,000	99%

AD: Autosomal Dominant
AR: Autosomal Recessive
XL: X-Linked