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Residual risks by ethnicity

This table displays residual risks after a negative result for each of the genes and corresponding disorders. If a patient is reported to be a carrier of a disease, their residual risk is 1 and this table does not apply for that disease.

Disease (Inheritance)	Gene	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk	Analytical Detection Rate
Abetalipoproteinemia (AR)	*MTTP*	African	1 in 1354	97%	1 in 45,000	97%
NM_000253.3		Ashkenazi Jewish	1 in 176	97%	1 in 5,800	
		East Asian	1 in 1437	81%	1 in 7,500	
		Caucasian	1 in 655	79%	1 in 3,200	
		Latino	1 in 2131	97%	1 in 71,000	
		South Asian	1 in 3078	97%	1 in 103,000	
		Worldwide	1 in 870	85%	1 in 5,900	
Achromatopsia (AR)	*CNGB3*	African	1 in 50	98%	1 in 2,300	99%
NM_019098.4		Ashkenazi Jewish	1 in 97	99%	1 in 9,600	
		East Asian	1 in 208	99%	1 in 20,700	
		Finnish	1 in 163	99%	1 in 16,200	
		Caucasian	1 in 87	99%	1 in 8,600	
		Latino	1 in 90	99%	1 in 8,900	
		South Asian	1 in 18	99%	1 in 1,700	
		Worldwide	1 in 61	99%	1 in 4,300	
Acrodermatitis Enteropathica (AR)	*SLC39A4*	African	1 in 421	98%	1 in 21,000	98%
NM_130849.3		East Asian	1 in 1248	98%	1 in 62,400	
		Finnish	1 in 216	98%	1 in 10,800	
		Caucasian	1 in 316	97%	1 in 12,100	
		Latino	1 in 1300	90%	1 in 13,500	
		South Asian	1 in 1099	98%	1 in 54,900	
		Worldwide	1 in 403	96%	1 in 11,000	
Acute Infantile Liver Failure (AR)	*TRMU*	African	1 in 624	89%	1 in 5,500	99%
NM_018006.4		Ashkenazi Jewish	1 in 459	99%	1 in 45,900	
		East Asian	1 in 551	99%	1 in 55,000	
		Caucasian	1 in 789	92%	1 in 9,400	
		Latino	1 in 1162	99%	1 in 116,000	
		South Asian	1 in 321	78%	1 in 1,500	
		Worldwide	1 in 730	89%	1 in 6,600	
		Sephardic Jewish - Yemenite	1 in 34	81%	1 in 180	
Acyl-CoA Oxidase I Deficiency (AR)	*ACOX1*	African	1 in 1071	98%	1 in 42,800	98%
NM_004035.6		Caucasian	1 in 2394	93%	1 in 35,800	
		Latino	1 in 3358	98%	1 in 134,000	
		South Asian	1 in 3848	98%	1 in 154,000	
		Worldwide	1 in 2212	96%	1 in 52,000	
Adenosine Deaminase Deficiency (AR)	*ADA*	African	1 in 91	92%	1 in 1,200	99%
NM_000022.2		East Asian	1 in 1275	99%	1 in 127,000	
		Finnish	1 in 4299	99%	1 in 430,000	
		Caucasian	1 in 390	92%	1 in 5,100	
		Latino	1 in 250	96%	1 in 5,700	
		South Asian	1 in 282	86%	1 in 2,100	
		Worldwide	1 in 305	91%	1 in 3,300	
Adrenoleukodystrophy, X-Linked (XL)	*ABCD1*	Worldwide	1 in 10,000	47%	1 in 18,900	89%
NM_000033.3						
Exception: Exons 8 and 9						

Aicardi-Goutières Syndrome (*SAMHD1*-Related) (AR) NM_015474.3	*SAMHD1*	African	1 in 754	99%	1 in 75,300	99%
		Ashkenazi Jewish	1 in 130	99%	1 in 12,900	
		East Asian	1 in 355	87%	1 in 2,700	
		Caucasian	1 in 610	94%	1 in 10,100	
		Latino	1 in 2407	99%	1 in 241,000	
		South Asian	1 in 3848	99%	1 in 385,000	
		Worldwide	1 in 728	95%	1 in 13,200	
Alpha-Mannosidosis (AR) NM_000528.3	*MAN2B1*	African	1 in 290	99%	1 in 29,000	99%
		East Asian	1 in 982	88%	1 in 8,000	
		Finnish	1 in 219	99%	1 in 21,800	
		Caucasian	1 in 439	93%	1 in 6,200	
		Latino	1 in 665	87%	1 in 5,200	
		South Asian	1 in 795	68%	1 in 2,500	
		Worldwide	1 in 425	93%	1 in 6,000	
Alpha-Thalassemia (AR) NM_000558.4 / NM_000517.4	*HBA1 / HBA2*	Caucasian	1 in 500	95%	1 in 10,000	99%
		African American	1 in 30	95%	1 in 580	
		Asian	1 in 20	95%	1 in 380	
		Worldwide	1 in 25	95%	1 in 480	
Alpha-Thalassemia Mental Retardation Syndrome (XL) NM_000489.4	*ATRX*	Worldwide	1 in 20,000	58%	1 in 45,000	98%
Alport Syndrome (*COL4A3*-Related) (AR) NM_000091.4	*COL4A3*	African	1 in 329	85%	1 in 2,200	99%
		Ashkenazi Jewish	1 in 227	99%	1 in 22,600	
		East Asian	1 in 241	86%	1 in 1,700	
		Finnish	1 in 1021	81%	1 in 5,300	
		Caucasian	1 in 218	88%	1 in 1,800	
		Latino	1 in 195	88%	1 in 1,600	
		South Asian	1 in 361	90%	1 in 3,500	
		Worldwide	1 in 237	89%	1 in 2,100	
Alport Syndrome (*COL4A4*-Related) (AR) NM_000092.4	*COL4A4*	African	1 in 369	75%	1 in 1,500	98%
		Ashkenazi Jewish	1 in 1640	98%	1 in 82,000	
		East Asian	1 in 158	69%	1 in 510	
		Finnish	1 in 2841	98%	1 in 142,000	
		Caucasian	1 in 349	81%	1 in 1,800	
		Latino	1 in 359	94%	1 in 5,800	
		South Asian	1 in 415	93%	1 in 5,700	
		Worldwide	1 in 356	81%	1 in 1,800	
Alport Syndrome (*COL4A5*-Related) (XL) NM_000495.3	*COL4A5*	Worldwide	1 in 30,000	80%	1 in 16,400	94%
Alstrom Syndrome (AR) NM_015120.4	*ALMS1*	African	1 in 202	91%	1 in 2,300	99%
		East Asian	1 in 107	97%	1 in 3,100	
		Finnish	1 in 626	99%	1 in 62,500	
		Caucasian	1 in 168	96%	1 in 4,500	
		Latino	1 in 352	99%	1 in 35,100	
		South Asian	1 in 256	92%	1 in 3,400	
		Worldwide	1 in 198	96%	1 in 5,100	

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Andermann Syndrome (AR) NM_133647.1	*SLC12A6*	Ashkenazi Jewish	1 in 1641	99%	1 in 164,000	99%
		East Asian	1 in 2872	99%	1 in 287,000	
		Finnish	1 in 2787	99%	1 in 279,000	
		Caucasian	1 in 1515	99%	1 in 151,000	
		Latino	1 in 764	99%	1 in 76,300	
		South Asian	1 in 2564	99%	1 in 256,000	
		Worldwide	1 in 1615	99%	1 in 161,000	
		French-Canadian - Saguenay Lac-St. Jean	1 in 23	99%	1 in 2,200	
Argininosuccinic Aciduria (AR) NM_000048.3	*ASL*	African	1 in 375	70%	1 in 1,300	99%
		Ashkenazi Jewish	1 in 561	99%	1 in 56,000	
		East Asian	1 in 444	89%	1 in 4,000	
		Finnish	1 in 91	99%	1 in 9,000	
		Caucasian	1 in 117	90%	1 in 1,200	
		Latino	1 in 437	71%	1 in 1,500	
		South Asian	1 in 527	82%	1 in 2,900	
		Worldwide	1 in 161	88%	1 in 1,300	
Aromatase Deficiency (AR) NM_031226.2	*CYP19A1*	African	1 in 671	84%	1 in 4,200	89%
		Ashkenazi Jewish	1 in 634	89%	1 in 5,800	
		East Asian	1 in 559	52%	1 in 1,200	
		Finnish	1 in 809	89%	1 in 7,400	
		Caucasian	1 in 2159	60%	1 in 5,400	
		Latino	1 in 1009	89%	1 in 9,200	
		South Asian	1 in 905	79%	1 in 4,200	
		Worldwide	1 in 863	78%	1 in 3,900	
Arthrogryposis, Mental Retardation, and Seizures (AR) NM_012243.2	*SLC35A3*	African	1 in 3999	99%	1 in 400,000	99%
		Ashkenazi Jewish	1 in 367	99%	1 in 36,600	
		Finnish	1 in 2778	99%	1 in 278,000	
		Caucasian	1 in 4537	99%	1 in 454,000	
		Latino	1 in 3356	99%	1 in 336,000	
		South Asian	1 in 3996	99%	1 in 399,000	
		Worldwide	1 in 2402	99%	1 in 240,000	
Asparagine Synthetase Deficiency (AR) NM_001673.4	*ASNS*	African	1 in 845	99%	1 in 84,400	99%
		East Asian	1 in 1777	99%	1 in 178,000	
		Finnish	1 in 2757	25%	1 in 3,700	
		Caucasian	1 in 2023	92%	1 in 23,900	
		South Asian	1 in 3072	99%	1 in 307,000	
		Worldwide	1 in 2049	90%	1 in 20,900	
		Sephardic Jewish - Iranian	1 in 80	99%	1 in 8,100	
Aspartylglycosaminuria (AR) NM_000027.3	*AGA*	African	1 in 1650	99%	1 in 165,000	99%
		East Asian	1 in 1724	99%	1 in 172,000	
		Finnish	1 in 60	98%	1 in 3,800	
		Caucasian	1 in 975	92%	1 in 13,000	
		Latino	1 in 1526	90%	1 in 15,300	
		South Asian	1 in 2198	99%	1 in 220,000	
		Worldwide	1 in 428	97%	1 in 12,800	
Ataxia with Isolated Vitamin E Deficiency (AR) NM_000370.3	*TTPA*	African	1 in 319	99%	1 in 31,800	99%
		Ashkenazi Jewish	1 in 513	99%	1 in 51,200	
		Finnish	1 in 3101	99%	1 in 310,000	
		Caucasian	1 in 607	99%	1 in 60,600	
		Latino	1 in 1293	99%	1 in 129,000	
		South Asian	1 in 2403	99%	1 in 240,000	
		Worldwide	1 in 196	99%	1 in 19,500	

Ataxia Telangiectasia (AR) NM_000051.3	*ATM*	African	1 in 200	86%	1 in 1,400	95%
		Ashkenazi Jewish	1 in 820	79%	1 in 3,900	
		East Asian	1 in 152	72%	1 in 540	
		Finnish	1 in 484	62%	1 in 1,300	
		Caucasian	1 in 150	88%	1 in 1,300	
		Latino	1 in 240	91%	1 in 2,700	
		South Asian	1 in 211	77%	1 in 900	
		Worldwide	1 in 174	85%	1 in 1,200	
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (AR) NM_014363.5	*SACS*	African	1 in 201	99%	1 in 20,000	99%
		Ashkenazi Jewish	1 in 483	80%	1 in 2,400	
		East Asian	1 in 338	84%	1 in 2,100	
		Finnish	1 in 341	99%	1 in 34,000	
		Caucasian	1 in 100	95%	1 in 2,100	
		Latino	1 in 309	88%	1 in 2,600	
		South Asian	1 in 383	97%	1 in 11,000	
		Worldwide	1 in 148	95%	1 in 2,900	
		French Canadian - Charlevoix-Saguenay	1 in 21	99%	1 in 2,000	
Bardet-Biedl Syndrome (*BBS1*-Related) (AR) NM_024649.4	*BBS1*	African	1 in 243	94%	1 in 3,900	99%
		East Asian	1 in 1725	20%	1 in 2,200	
		Finnish	1 in 272	99%	1 in 27,100	
		Caucasian	1 in 152	97%	1 in 5,400	
		Latino	1 in 417	99%	1 in 41,600	
		South Asian	1 in 185	98%	1 in 8,400	
		Worldwide	1 in 198	97%	1 in 6,200	
		Faroese	1 in 30	99%	1 in 2,900	
Bardet-Biedl Syndrome (*BBS2*-Related) (AR) NM_031885.3	*BBS2*	African	1 in 741	89%	1 in 6,900	99%
		Ashkenazi Jewish	1 in 117	92%	1 in 1,500	
		East Asian	1 in 148	97%	1 in 5,400	
		Finnish	1 in 442	99%	1 in 44,100	
		Caucasian	1 in 333	73%	1 in 1,200	
		Latino	1 in 1126	59%	1 in 2,800	
		South Asian	1 in 855	93%	1 in 13,100	
		Worldwide	1 in 353	82%	1 in 2,000	
		Hutterite	1 in 22	99%	1 in 2,100	
Bardet-Biedl Syndrome (*BBS10*-Related) (AR) NM_024685.3	*BBS10*	African	1 in 470	95%	1 in 9,600	99%
		Ashkenazi Jewish	1 in 298	99%	1 in 29,700	
		East Asian	1 in 428	79%	1 in 2,100	
		Caucasian	1 in 237	91%	1 in 2,700	
		Latino	1 in 1204	78%	1 in 5,400	
		South Asian	1 in 425	69%	1 in 1,400	
		Worldwide	1 in 333	89%	1 in 3,000	
Bardet-Biedl Syndrome (*BBS12*-Related) (AR) NM_152618.2	*BBS12*	African	1 in 1070	85%	1 in 7,100	99%
		East Asian	1 in 2870	99%	1 in 287,000	
		Caucasian	1 in 613	94%	1 in 9,900	
		Latino	1 in 1864	99%	1 in 186,000	
		South Asian	1 in 1705	99%	1 in 170,000	
		Worldwide	1 in 895	95%	1 in 17,200	
Bare Lymphocyte Syndrome, Type II (AR) NM_000246.3	*CIITA*	African	1 in 3361	99%	1 in 336,000	99%
		East Asian	1 in 1290	99%	1 in 129,000	
		Caucasian	1 in 924	97%	1 in 34,800	
		Latino	1 in 2405	99%	1 in 240,000	
		South Asian	1 in 2197	99%	1 in 220,000	
		Worldwide	1 in 1366	98%	1 in 64,700	

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Bartter Syndrome, Type 4A (AR)	*BSND*	African	1 in 186	97%	1 in 5,400	99%
NM_057176.2		Ashkenazi Jewish	1 in 1641	99%	1 in 164,000	
		East Asian	1 in 687	99%	1 in 68,600	
		Caucasian	1 in 916	99%	1 in 91,500	
		Latino	1 in 2856	99%	1 in 286,000	
		South Asian	1 in 733	99%	1 in 73,200	
		Worldwide	1 in 739	98%	1 in 46,300	
Bernard-Soulier Syndrome, Type A1 (AR)	*GP1BA*	African	1 in 2035	99%	1 in 203,000	99%
NM_000173.5		East Asian	1 in 1725	99%	1 in 172,000	
		Finnish	1 in 368	99%	1 in 36,700	
		Caucasian	1 in 1677	96%	1 in 42,200	
		Latino	1 in 4198	99%	1 in 420,000	
		Worldwide	1 in 1418	98%	1 in 66,200	
Bernard-Soulier Syndrome, Type C (AR)	*GP9*	African	1 in 318	21%	1 in 400	99%
NM_000174.4		Finnish	1 in 458	35%	1 in 710	
		Caucasian	1 in 451	86%	1 in 3,300	
		Latino	1 in 4269	74%	1 in 16,300	
		South Asian	1 in 848	99%	1 in 84,700	
		Worldwide	1 in 477	57%	1 in 1,100	
Beta-Globin Related Hemoglobinopathies: Beta-Thalassemia (AR)	*HBB*	African	1 in 97	92%	1 in 1,200	99%
NM_000518.4		Ashkenazi Jewish	1 in 28	99%	1 in 2,700	
		East Asian	1 in 87	93%	1 in 1,200	
		Finnish	1 in 1901	48%	1 in 3,700	
		Caucasian	1 in 214	89%	1 in 2,000	
		Latino	1 in 438	89%	1 in 3,900	
		South Asian	1 in 25	98%	1 in 1,000	
		Worldwide	1 in 81	95%	1 in 1,800	
		Mediterranean	1 in 28	99%	1 in 2,700	
Beta-Globin Related Hemoglobinopathies: HbC Variant (AR)	*HBB*	African	1 in 38	99%	1 in 3,700	99%
NM_000518.4		Caucasian	1 in 21074	99%	1 in 2,107,000	
Variant Tested: c.19G>A, p.E7K		Latino	1 in 2150	99%	1 in 21,500	
		Worldwide	1 in 418	99%	1 in 41,700	
Beta-Globin Related Hemoglobinopathies: HbS Variant (Sickle Cell Disease) (AR)	*HBB*	African	1 in 11	99%	1 in 1,000	99%
NM_000518.4		Caucasian	1 in 7903	99%	1 in 790,000	
		Latino	1 in 232	99%	1 in 23,100	
		South Asian	1 in 810	99%	1 in 80,900	
Variant Tested: c.20A>T, p.E7V		Worldwide	1 in 115	99%	1 in 11,400	
3-Beta-Hydroxysteroid Deficiency (AR)	*HSD3B2*	African	1 in 786	89%	1 in 7,000	99%
NM_000198.3		Ashkenazi Jewish	1 in 1639	99%	1 in 164,000	
		East Asian	1 in 1814	99%	1 in 181,000	
		Caucasian	1 in 862	74%	1 in 3,300	
		Latino	1 in 1686	69%	1 in 5,500	
		South Asian	1 in 1026	86%	1 in 7,200	
		Worldwide	1 in 1005	79%	1 in 4,900	
Beta-Ketothiolase Deficiency (AR)	*ACAT1*	African	1 in 1197	99%	1 in 120,000	99%
NM_000019.3		East Asian	1 in 293	50%	1 in 590	
		Caucasian	1 in 629	82%	1 in 3,500	
		Latino	1 in 173	95%	1 in 3,400	
		South Asian	1 in 1378	46%	1 in 2,500	
		Worldwide	1 in 515	83%	1 in 3,100	

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Bilateral Frontoparietal Polymicrogyria (AR) NM_005682.6	*GPR56 (ADGRG1)*	African	1 in 917	99%	1 in 91,600	99%
		East Asian	1 in 1433	99%	1 in 143,000	
		Finnish	1 in 1371	99%	1 in 137,000	
		Caucasian	1 in 2033	99%	1 in 203,000	
		Latino	1 in 1525	90%	1 in 15,300	
		South Asian	1 in 641	99%	1 in 64,000	
		Worldwide	1 in 1220	98%	1 in 61,500	
Biotinidase Deficiency (AR) NM_000060.3	*BTD* †	African	1 in 52	93%	1 in 790	99%
		Ashkenazi Jewish	1 in 15	99%	1 in 1,400	
		East Asian	1 in 324	92%	1 in 3,800	
		Finnish	1 in 9	99%	1 in 810	
		Caucasian	1 in 12	98%	1 in 500	
		Latino	1 in 24	97%	1 in 740	
		South Asian	1 in 7	98%	1 in 370	
		Worldwide	1 in 13	98%	1 in 550	
Bloom Syndrome (AR) NM_000057.2	*BLM*	African	1 in 532	99%	1 in 53,100	99%
		Ashkenazi Jewish	1 in 117	99%	1 in 11,700	
		East Asian	1 in 337	99%	1 in 33,600	
		Finnish	1 in 712	99%	1 in 71,100	
		Caucasian	1 in 358	95%	1 in 7,400	
		Latino	1 in 495	99%	1 in 49,400	
		South Asian	1 in 636	95%	1 in 12,500	
		Worldwide	1 in 357	97%	1 in 11,800	
Canavan Disease (AR) NM_000049.2	*ASPA*	African	1 in 741	98%	1 in 37,000	98%
		Ashkenazi Jewish	1 in 50	98%	1 in 2,400	
		Finnish	1 in 241	98%	1 in 12,000	
		Caucasian	1 in 486	88%	1 in 4,000	
		Latino	1 in 899	87%	1 in 7,100	
		South Asian	1 in 1923	61%	1 in 5,000	
		Worldwide	1 in 393	92%	1 in 5,200	
Carbamoylphosphate Synthetase I Deficiency (AR) NM_001875.4	*CPS1*	African	1 in 401	54%	1 in 870	98%
		Ashkenazi Jewish	1 in 1640	98%	1 in 82,000	
		East Asian	1 in 221	64%	1 in 610	
		Finnish	1 in 1047	73%	1 in 3,900	
		Caucasian	1 in 343	65%	1 in 990	
		Latino	1 in 740	60%	1 in 1,800	
		South Asian	1 in 1026	46%	1 in 1,900	
		Worldwide	1 in 416	64%	1 in 1,200	
Carnitine Palmitoyltransferase IA Deficiency (AR) NM_001876.3	*CPT1A*	African	1 in 2550	99%	1 in 255,000	99%
		Ashkenazi Jewish	1 in 491	99%	1 in 49,000	
		East Asian	1 in 1435	99%	1 in 143,000	
		Finnish	1 in 267	97%	1 in 7,900	
		Caucasian	1 in 1518	94%	1 in 23,800	
		Latino	1 in 2821	49%	1 in 5,500	
		South Asian	1 in 1924	74%	1 in 7,500	
		Worldwide	1 in 970	87%	1 in 7,200	
		Hutterite	1 in 16	99%	1 in 1,500	
Carnitine Palmitoyltransferase II Deficiency (AR) NM_000098.2	*CPT2*	African	1 in 197	85%	1 in 1,300	99%
		Ashkenazi Jewish	1 in 41	99%	1 in 4,000	
		East Asian	1 in 266	71%	1 in 930	
		Finnish	1 in 248	99%	1 in 24,700	
		Caucasian	1 in 147	78%	1 in 670	
		Latino	1 in 251	93%	1 in 3,700	
		South Asian	1 in 523	96%	1 in 11,900	
		Worldwide	1 in 163	85%	1 in 1,100	

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Carpenter Syndrome (AR) NM_0012786671	*RAB23*	African	1 in 395	98%	1 in 19,700	98%
		Finnish	1 in 4296	98%	1 in 215,000	
		Caucasian	1 in 673	97%	1 in 21,100	
		Worldwide	1 in 726	97%	1 in 28,100	
Cartilage-Hair Hypoplasia (AR) NR_003051.3	*RMRP*	African	1 in 210	63%	1 in 570	99%
		Ashkenazi Jewish	1 in 68	99%	1 in 6,700	
		East Asian	1 in 165	63%	1 in 440	
		Finnish	1 in 49	99%	1 in 4,800	
		Caucasian	1 in 143	85%	1 in 960	
		Latino	1 in 157	94%	1 in 2,500	
		South Asian	1 in 192	84%	1 in 1,200	
		Worldwide	1 in 120	87%	1 in 950	
Cerebral Creatine Deficiency Syndrome 1 (XL) NM_005629.3 *Exception:* Exons 3, 4	*SLC6A8*	Worldwide	< 1 in 50,000	76%	1 in 210,000	96%
Cerebral Creatine Deficiency Syndrome 2 (AR) NM_000156.5	*GAMT*	African	1 in 545	98%	1 in 27,200	98%
		Ashkenazi Jewish	1 in 1406	98%	1 in 70,200	
		East Asian	1 in 1150	98%	1 in 57,500	
		Caucasian	1 in 435	93%	1 in 6,500	
		Latino	1 in 4223	73%	1 in 15,800	
		South Asian	1 in 2601	98%	1 in 130,000	
		Worldwide	1 in 649	94%	1 in 11,400	
		Portuguese	1 in 125	98%	1 in 6,200	
Cerebrotendinous Xanthomatosis (AR) NM_000784.3	*CYP27A1*	African	1 in 285	95%	1 in 6,100	99%
		Ashkenazi Jewish	1 in 331	99%	1 in 33,000	
		East Asian	1 in 122	84%	1 in 750	
		Finnish	1 in 1109	99%	1 in 111,000	
		Caucasian	1 in 275	93%	1 in 3,900	
		Latino	1 in 302	92%	1 in 3,800	
		South Asian	1 in 143	85%	1 in 960	
		Worldwide	1 in 228	91%	1 in 2,600	
		Sephardic Jewish - Moroccan	1 in 76	99%	1 in 2,500	
Charcot-Marie-Tooth Disease, Type 4D (AR) NM_001135242.1	*NDRG1*	East Asian	1 in 2252	99%	1 in 225,000	99%
		Caucasian	1 in 7299	99%	1 in 730,000	
		South Asian	1 in 4789	99%	1 in 479,000	
		Worldwide	1 in 6931	99%	1 in 693,000	
		Roma	1 in 22	99%	1 in 2,100	
Charcot-Marie-Tooth Disease, Type 5 / Arts Syndrome (XL) NM_002764.3	*PRPS1*	Worldwide	< 1 in 50,000	56%	1 in 115,000	99%
Charcot-Marie-Tooth Disease, X-Linked (XL) NM_000166.5	*GJB1*†	Worldwide	1 in 5000	53%	1 in 6,800	99%
Choreoacanthocytosis (AR) NM_033305.2	*VPS13A*	African	1 in 321	90%	1 in 3,100	98%
		Ashkenazi Jewish	1 in 628	98%	1 in 31,300	
		East Asian	1 in 204	96%	1 in 4,700	
		Finnish	1 in 614	98%	1 in 30,700	
		Caucasian	1 in 341	97%	1 in 13,100	
		Latino	1 in 466	82%	1 in 2,500	
		South Asian	1 in 540	95%	1 in 9,900	
		Worldwide	1 in 329	95%	1 in 6,700	
Choroideremia (XL) NM_000390.2	*CHM*	Worldwide	1 in 10,000	92%	1 in 125,000	99%

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Chronic Granulomatous Disease (*CYBA*-Related) (AR) NM_000101.2	*CYBA*	African	1 in 806	78%	1 in 3,600	96%
		Finnish	1 in 636	96%	1 in 15,900	
		Caucasian	1 in 1689	66%	1 in 5,000	
		Latino	1 in 1933	96%	1 in 48,300	
		South Asian	1 in 1896	60%	1 in 4,800	
		Worldwide	1 in 1113	70%	1 in 3,700	
		Sephardic Jewish - Moroccan	1 in 13	83%	1 in 72	
Chronic Granulomatous Disease (*CYBB*-Related) (XL) NM_000397.3	*CYBB*	Worldwide	< 1 in 50,000	83%	1 in 290,000	98%
Citrin Deficiency (AR) NM_014251.2	*SLC25A13*	African	1 in 435	75%	1 in 1,700	99%
		Ashkenazi Jewish	1 in 273	99%	1 in 27,300	
		East Asian	1 in 48	98%	1 in 2,300	
		Caucasian	1 in 619	95%	1 in 11,700	
		Latino	1 in 990	93%	1 in 14,500	
		South Asian	1 in 496	86%	1 in 3,600	
		Worldwide	1 in 329	93%	1 in 4,700	
Citrullinemia, Type I (AR) NM_000050.4	*ASS1*	African	1 in 339	87%	1 in 2,600	99%
		Ashkenazi Jewish	1 in 1669	99%	1 in 167,000	
		East Asian	1 in 809	99%	1 in 80,800	
		Finnish	1 in 2984	99%	1 in 298,000	
		Caucasian	1 in 323	87%	1 in 2,500	
		Latino	1 in 304	95%	1 in 6,600	
		South Asian	1 in 192	85%	1 in 1,300	
		Worldwide	1 in 339	87%	1 in 2,700	
Cohen Syndrome (AR) NM_017890.4	*VPS13B*	African	1 in 219	95%	1 in 4,500	98%
		Ashkenazi Jewish	1 in 260	93%	1 in 3,700	
		East Asian	1 in 255	98%	1 in 12,700	
		Finnish	1 in 121	98%	1 in 6,000	
		Caucasian	1 in 224	97%	1 in 6,400	
		Latino	1 in 432	98%	1 in 21,500	
		South Asian	1 in 313	98%	1 in 15,600	
		Worldwide	1 in 207	97%	1 in 7,000	
Combined Malonic and Methylmalonic Aciduria (AR) NM_001127214.3	*ACSF3*	African	1 in 126	99%	1 in 12,500	99%
		Ashkenazi Jewish	1 in 59	99%	1 in 5,800	
		East Asian	1 in 235	99%	1 in 23,400	
		Finnish	1 in 346	99%	1 in 34,500	
		Caucasian	1 in 71	97%	1 in 2,400	
		Latino	1 in 193	99%	1 in 19,300	
		South Asian	1 in 165	51%	1 in 340	
		Worldwide	1 in 99	94%	1 in 1,700	
Combined Oxidative Phosphorylation Deficiency 1 (AR) NM_024996.5	*GFM1*	African	1 in 515	99%	1 in 51,400	99%
		East Asian	1 in 1113	86%	1 in 8,100	
		Finnish	1 in 841	99%	1 in 84,000	
		Caucasian	1 in 480	96%	1 in 13,500	
		Latino	1 in 1318	99%	1 in 132,000	
		South Asian	1 in 769	99%	1 in 76,800	
		Worldwide	1 in 583	97%	1 in 20,200	
Combined Oxidative Phosphorylation Deficiency 3 (AR) NM_001172696.1	*TSFM*	African	1 in 681	99%	1 in 68,000	99%
		Finnish	1 in 35	99%	1 in 3,400	
		Caucasian	1 in 535	98%	1 in 27,000	
		Latino	1 in 1796	99%	1 in 180,000	
		Worldwide	1 in 258	99%	1 in 21,200	

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Combined Pituitary Hormone Deficiency 2 (AR) NM_006261.4	*PROP1*	Finnish Caucasian Latino Worldwide	1 in 1115 1 in 482 1 in 584 1 in 745	99% 83% 92% 86%	1 in 111,000 1 in 2,800 1 in 7,400 1 in 5,300	99%
Combined Pituitary Hormone Deficiency 3 (AR) NM_014564.3	*LHX3*	East Asian Caucasian Worldwide	1 in 1210 1 in 1398 1 in 1975	99% 99% 99%	1 in 121,000 1 in 140,000 1 in 197,000	99%
Combined SAP Deficiency (AR) NM_002778.2	*PSAP*	African Caucasian Latino Worldwide	1 in 1941 1 in 2039 1 in 884 1 in 2088	99% 95% 99% 97%	1 in 194,000 1 in 44,100 1 in 88,300 1 in 77,800	99%
Congenital Adrenal Hyperplasia due to 17-Alpha-Hydroxylase Deficiency (AR) NM_000102.3	*CYP17A1*	African East Asian Finnish Caucasian Latino South Asian Worldwide	1 in 1133 1 in 229 1 in 1855 1 in 560 1 in 1123 1 in 777 1 in 641	78% 73% 50% 68% 86% 87% 73%	1 in 5,200 1 in 840 1 in 3,700 1 in 1,800 1 in 8,100 1 in 6,000 1 in 2,400	99%
Classic Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (AR) NM_000500.7	*CYP21A2*	Ashkenazi Jewish Caucasian Worldwide	1 in 40 1 in 67 1 in 60	95% 95% 95%	1 in 780 1 in 1,300 1 in 1,200	95%
Non-Classic Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (AR) NM_000500.7	*CYP21A2*	Ashkenazi Jewish Caucasian Worldwide	1 in 7 1 in 11 1 in 16	95% 95% 95%	1 in 120 1 in 200 1 in 300	95%
Congenital Amegakaryocytic Thrombocytopenia (AR) NM_005373.2	*MPL*	African Ashkenazi Jewish East Asian Finnish Caucasian Latino South Asian Worldwide	1 in 496 1 in 60 1 in 681 1 in 1802 1 in 241 1 in 602 1 in 617 1 in 299	91% 99% 99% 99% 92% 85% 99% 94%	1 in 5,400 1 in 5,900 1 in 68,000 1 in 180,000 1 in 3,100 1 in 4,000 1 in 61,600 1 in 4,700	99%
Congenital Disorder of Glycosylation, Type Ia (AR) NM_000303.2	*PMM2*	African Ashkenazi Jewish East Asian Finnish Caucasian Latino South Asian Worldwide	1 in 245 1 in 66 1 in 133 1 in 58 1 in 58 1 in 114 1 in 278 1 in 80	99% 99% 76% 99% 89% 91% 86% 91%	1 in 24,400 1 in 6,500 1 in 550 1 in 5,700 1 in 540 1 in 1,200 1 in 2,000 1 in 840	99%
Congenital Disorder of Glycosylation, Type Ib (AR) NM_002435.2	*MPI*	African East Asian Finnish Caucasian Latino South Asian Worldwide	1 in 688 1 in 442 1 in 1172 1 in 473 1 in 1139 1 in 1924 1 in 615	65% 79% 81% 92% 92% 74% 87%	1 in 2,000 1 in 2,100 1 in 6,200 1 in 5,600 1 in 15,100 1 in 7,500 1 in 4,900	99%

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Congenital Disorder of Glycosylation, Type Ic (AR) NM_013339.3	*ALG6*	African	1 in 432	88%	1 in 3,700	99%
		Ashkenazi Jewish	1 in 1671	66%	1 in 5,000	
		East Asian	1 in 529	77%	1 in 2,300	
		Finnish	1 in 1980	99%	1 in 198,000	
		Caucasian	1 in 301	93%	1 in 4,100	
		Latino	1 in 1405	75%	1 in 5,600	
		South Asian	1 in 809	57%	1 in 1,900	
		Worldwide	1 in 439	87%	1 in 3,500	
Congenital Insensitivity to Pain with Anhidrosis (AR) NM_001012331.1	*NTRK1*	African	1 in 713	83%	1 in 4,100	99%
		East Asian	1 in 280	73%	1 in 1,100	
		Finnish	1 in 929	53%	1 in 2,000	
		Caucasian	1 in 1122	80%	1 in 5,700	
		Latino	1 in 2105	87%	1 in 15,700	
		South Asian	1 in 3539	76%	1 in 14,900	
		Worldwide	1 in 849	76%	1 in 3,600	
		Sephardic Jewish - Moroccan	N/A	99%	N/A	
Congenital Myasthenic Syndrome (*CHRNE*-Related) (AR) NM_000080.3	*CHRNE*	African	1 in 300	99%	1 in 29,900	99%
		Ashkenazi Jewish	1 in 149	99%	1 in 14,800	
		East Asian	1 in 299	99%	1 in 29,800	
		Finnish	1 in 971	90%	1 in 9,300	
		Caucasian	1 in 244	94%	1 in 4,100	
		Latino	1 in 366	93%	1 in 4,900	
		South Asian	1 in 312	89%	1 in 2,800	
		Worldwide	1 in 260	94%	1 in 4,680	
		Southeastern European Roma	1 in 25	99%	1 in 2,400	
Congenital Myasthenic Syndrome (*RAPSN*-Related) (AR) NM_005055.4	*RAPSN*	African	1 in 1255	78%	1 in 5,700	99%
		Ashkenazi Jewish	1 in 253	99%	1 in 25,200	
		East Asian	1 in 471	99%	1 in 47,000	
		Finnish	1 in 989	99%	1 in 98,800	
		Caucasian	1 in 165	94%	1 in 2,900	
		Latino	1 in 429	87%	1 in 3,200	
		South Asian	1 in 549	95%	1 in 12,100	
		Worldwide	1 in 265	94%	1 in 4,400	
		Sephardic Jewish - Iraqi and Iranian	N/A	99%	N/A	
Congenital Neutropenia (*HAX1*-Related) (AR) NM_006118.3	*HAX1*	African	1 in 800	99%	1 in 79,900	99%
		Ashkenazi Jewish	1 in 825	99%	1 in 82,400	
		East Asian	1 in 1263	99%	1 in 126,000	
		Caucasian	1 in 824	99%	1 in 82,300	
		Latino	1 in 2798	99%	1 in 280,000	
		South Asian	1 in 5130	99%	1 in 513,000	
		Worldwide	1 in 1069	99%	1 in 107,000	
Congenital Neutropenia (*VPS45*-Related) (AR) NM_001279354.1	*VPS45*	African	1 in 1120	99%	1 in 112,000	99%
		East Asian	1 in 1099	99%	1 in 110,000	
		Finnish	1 in 2774	49%	1 in 5,500	
		Caucasian	1 in 1634	99%	1 in 163,000	
		Latino	1 in 3351	99%	1 in 335,000	
		South Asian	1 in 1703	99%	1 in 170,000	
		Worldwide	1 in 1530	96%	1 in 43,200	

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Corneal Dystrophy and Perceptive Deafness (AR) NM_032034.3	*SLC4A11*	African	1 in 373	65%	1 in 1,100	99%
		East Asian	1 in 316	82%	1 in 1,800	
		Finnish	1 in 3889	99%	1 in 389,000	
		Caucasian	1 in 806	83%	1 in 4,600	
		Latino	1 in 770	45%	1 in 1,400	
		South Asian	1 in 1183	53%	1 in 2,500	
		Worldwide	1 in 666	69%	1 in 2,200	
Corticosterone Methyloxidase Deficiency (AR) NM_000498.3	*CYP11B2*	African	1 in 502	46%	1 in 940	82%
		East Asian	1 in 1457	14%	1 in 1,700	
		Finnish	1 in 1185	18%	1 in 1,400	
		Caucasian	1 in 825	44%	1 in 1,500	
		Latino	1 in 945	46%	1 in 1,700	
		South Asian	1 in 1917	41%	1 in 3,200	
		Worldwide	1 in 870	41%	1 in 1,500	
Exception: Exons 3 - 7		Sephardic Jewish - Iranian	1 in 30	95%	1 in 580	
Cystic Fibrosis (AR) NM_000492.3	*CFTR*	African	1 in 58	91%	1 in 630	99%
		Ashkenazi Jewish	1 in 24	98%	1 in 1,200	
		East Asian	1 in 277	80%	1 in 1,400	
		Finnish	1 in 75	93%	1 in 1,100	
		Caucasian	1 in 23	95%	1 in 440	
		Latino	1 in 40	96%	1 in 1,000	
		South Asian	1 in 73	91%	1 in 800	
Exception: Exon 10		Worldwide	1 in 33	94%	1 in 500	
Cystinosis (AR) NM_004937.2	*CTNS*	African	1 in 942	68%	1 in 2,900	99%
		East Asian	1 in 393	94%	1 in 7,100	
		Caucasian	1 in 249	97%	1 in 7,700	
		Latino	1 in 1696	89%	1 in 15,400	
		South Asian	1 in 1026	79%	1 in 4,900	
		Worldwide	1 in 775	91%	1 in 8,200	
		French Canadian - Saguenay-Lac St. Jean	1 in 39	90%	1 in 380	
		Sephardic Jewish - Moroccan	1 in 100	92%	1 in 1,200	
D-Bifunctional Protein Deficiency (AR) NM_000414.3	*HSD17B4*	African	1 in 375	83%	1 in 2,200	92%
		East Asian	1 in 516	81%	1 in 2,700	
		Caucasian	1 in 534	89%	1 in 5,000	
		Latino	1 in 1123	80%	1 in 5,500	
		South Asian	1 in 1282	84%	1 in 8,200	
		Worldwide	1 in 628	87%	1 in 4,900	
Deafness, Autosomal Recessive 77 (AR) NM_144612.6	*LOXHD1*	African	1 in 282	86%	1 in 2,000	99%
		Ashkenazi Jewish	1 in 125	99%	1 in 12,500	
		East Asian	1 in 358	87%	1 in 2,800	
		Finnish	1 in 508	99%	1 in 50,700	
		Caucasian	1 in 150	98%	1 in 6,700	
		Latino	1 in 341	96%	1 in 9,100	
		South Asian	1 in 353	99%	1 in 35,200	
		Worldwide	1 in 191	95%	1 in 4,000	
Duchenne Muscular Dystrophy/ Becker Muscular Dystrophy (XL) NM_004006.2	*DMD*	Worldwide	1 in 500	95%	1 in 10,000	99%

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Dyskeratosis Congenita (*RTEL1*-Related) (AR)	*RTEL1*	African	1 in 756	99%	1 in 75,500	99%
NM_001283009.1		Ashkenazi Jewish	1 in 111	99%	1 in 11,000	
		East Asian	1 in 385	90%	1 in 3,900	
		Finnish	1 in 1122	99%	1 in 112,000	
		Caucasian	1 in 800	92%	1 in 9,800	
		Latino	1 in 1385	99%	1 in 138,000	
		South Asian	1 in 730	99%	1 in 72,900	
		Worldwide	1 in 587	95%	1 in 12,200	
Dystrophic Epidermolysis Bullosa (AR)	*COL7A1*	African	1 in 199	71%	1 in 690	99%
NM_000094.3		Ashkenazi Jewish	1 in 182	95%	1 in 3,900	
		East Asian	1 in 262	81%	1 in 1,400	
		Finnish	1 in 33	96%	1 in 780	
		Caucasian	1 in 100	89%	1 in 900	
		Latino	1 in 190	80%	1 in 930	
		South Asian	1 in 95	90%	1 in 980	
		Worldwide	1 in 92	90%	1 in 870	
Ehlers-Danlos Syndrome, Type VIIC (AR)	*ADAMTS2*	Ashkenazi Jewish	1 in 164	99%	1 in 16,300	99%
NM_014244.4		East Asian	1 in 631	99%	1 in 63,000	
		Caucasian	1 in 2432	99%	1 in 243,000	
		Latino	1 in 4193	99%	1 in 419,000	
		South Asian	1 in 3796	99%	1 in 380,000	
Exception: Exon 1		Worldwide	1 in 1423	99%	1 in 142,000	
Ellis-van Creveld Syndrome (*EVC*-Related) (AR)	*EVC*	African	1 in 555	97%	1 in 18,500	97%
NM_153717.2		East Asian	1 in 456	97%	1 in 15,200	
		Finnish	1 in 900	97%	1 in 30,000	
		Caucasian	1 in 370	91%	1 in 4,200	
		Latino	1 in 1199	97%	1 in 39,900	
		South Asian	1 in 1486	84%	1 in 9,500	
		Worldwide	1 in 511	93%	1 in 7,300	
Exception: Exon 1		Lancaster County Amish	1 in 12	97%	1 in 370	
Emery-Dreifuss Myopathy 1 (XL)	*EMD*	Worldwide	< 1 in 50,000	94%	1 in 833,000	98%
NM_000117.2						
Enhanced S-Cone Syndrome (AR)	*NR2E3*	African	1 in 389	46%	1 in 730	99%
NM_014249.3		Ashkenazi Jewish	1 in 81	97%	1 in 3,100	
		East Asian	1 in 488	12%	1 in 550	
		Caucasian	1 in 278	82%	1 in 1,500	
		Latino	1 in 536	96%	1 in 12,000	
		South Asian	1 in 874	58%	1 in 2,100	
		Worldwide	1 in 327	79%	1 in 1,600	
Ethylmalonic Encephalopathy (AR)	*ETHE1*	African	1 in 1897	98%	1 in 94,800	98%
NM_014297.3		Caucasian	1 in 1279	62%	1 in 3,400	
		Latino	1 in 934	93%	1 in 12,500	
		South Asian	1 in 3848	98%	1 in 192,000	
		Worldwide	1 in 1527	77%	1 in 6,600	
Fabry Disease (XL)	*GLA* †	Worldwide	1 in 2000	74%	1 in 7700	99%
NM_000169.2						
Factor IX Deficiency (XL)	*F9* †	Worldwide	1 in 2000	61%	1 in 5000	98%
NM_000133.3						

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Factor XI Deficiency (AR)	*F11*	African	1 in 249	86%	1 in 1,800	99%
NM_000128.3		Ashkenazi Jewish	1 in 12	99%	1 in 730	
		East Asian	1 in 94	79%	1 in 440	
		Finnish	1 in 304	97%	1 in 9,100	
		Caucasian	1 in 180	88%	1 in 1,600	
		Latino	1 in 230	81%	1 in 1,200	
		South Asian	1 in 217	82%	1 in 1,200	
		Worldwide	1 in 117	91%	1 in 1,200	
Familial Dysautonomia (AR)	*IKBKAP*	African	1 in 409	99%	1 in 40,800	99%
NM_003640.3		Ashkenazi Jewish	1 in 35	99%	1 in 3,400	
		East Asian	1 in 784	99%	1 in 78,300	
		Finnish	1 in 707	99%	1 in 70,600	
		Caucasian	1 in 506	99%	1 in 50,500	
		Latino	1 in 801	99%	1 in 80,000	
		South Asian	1 in 855	99%	1 in 85,400	
		Worldwide	1 in 345	99%	1 in 34,400	
Familial Hypercholesterolemia (AR)	*LDLR*	African	1 in 156	65%	1 in 450	96%
NM_000527.4		Ashkenazi Jewish	1 in 705	82%	1 in 4,000	
		East Asian	1 in 66	75%	1 in 260	
		Finnish	1 in 292	63%	1 in 790	
		Caucasian	1 in 118	58%	1 in 280	
		Latino	1 in 183	50%	1 in 370	
		South Asian	1 in 132	51%	1 in 270	
		Worldwide	1 in 127	59%	1 in 310	
		French Canadian	1 in 267	17%	1 in 320	26%
		South African Afrikaner	1 in 70	94%	1 in 1,200	
Familial Hypercholesterolemia, Autosomal Recessive (AR)	*LDLRAP1*	African	1 in 2885	98%	1 in 144,000	98%
		Caucasian	1 in 2721	98%	1 in 136,000	
NM_015627.2		Latino	1 in 2798	98%	1 in 140,000	
		South Asian	1 in 3847	98%	1 in 192,000	
		Worldwide	1 in 3429	98%	1 in 171,000	
		Sardinian	1 in 143	98%	1 in 7,100	
Familial Hyperinsulinism (*ABCC8*-Related) (AR)	*ABCC8*	African	1 in 256	43%	1 in 450	99%
		Ashkenazi Jewish	1 in 62	88%	1 in 510	
NM_000352.4		East Asian	1 in 119	51%	1 in 240	
		Finnish	1 in 213	92%	1 in 2,600	
		Caucasian	1 in 192	55%	1 in 420	
		Latino	1 in 285	80%	1 in 1,400	
		South Asian	1 in 364	56%	1 in 840	
		Worldwide	1 in 185	60%	1 in 460	
Familial Hyperinsulinism (*KCNJ11*-Related) (AR)	*KCNJ11*	African	1 in 2899	99%	1 in 290,000	99%
		Caucasian	1 in 1004	71%	1 in 3,500	
NM_000525.3		Latino	1 in 773	54%	1 in 1,700	
		South Asian	1 in 1924	62%	1 in 5,000	
		Worldwide	1 in 1126	57%	1 in 2,600	

Familial Mediterranean Fever (AR) NM_000243.2	*MEFV* †	African	1 in 230	74%	1 in 870	99%
		Ashkenazi Jewish	1 in 8	99%	1 in 720	
		East Asian	1 in 141	96%	1 in 3,400	
		Finnish	1 in 29	99%	1 in 2,800	
		Caucasian	1 in 40	97%	1 in 1,200	
		Latino	1 in 74	95%	1 in 1,500	
		South Asian	1 in 56	95%	1 in 1,000	
		Worldwide	1 in 40	97%	1 in 1,200	
		Sepharic Jewish	1 in 14	99%	1 in 1,300	
		Armenian	1 in 5	99%	1 in 400	
		Turkish	1 in 5	75%	1 in 17	
Fanconi Anemia, Group A (AR) NM_000135.2	*FANCA*	African	1 in 157	86%	1 in 1,100	95%
		Ashkenazi Jewish	1 in 251	90%	1 in 2,500	
		East Asian	1 in 182	89%	1 in 1,700	
		Finnish	1 in 268	95%	1 in 5,300	
		Caucasian	1 in 148	87%	1 in 1,100	
		Latino	1 in 278	87%	1 in 2,200	
		South Asian	1 in 257	78%	1 in 1,100	
		Worldwide	1 in 165	88%	1 in 1,300	
		Spanish Roma	1 in 64	95%	1 in 1,300	
		Sephardic Jewish - Moroccan and Tunisian	1 in 133	86%	1 in 940	
Fanconi Anemia, Group C (AR) NM_000136.2	*FANCC*	African	1 in 486	87%	1 in 3,700	99%
		Ashkenazi Jewish	1 in 82	99%	1 in 8,100	
		East Asian	1 in 344	99%	1 in 34,300	
		Finnish	1 in 1188	99%	1 in 119,000	
		Caucasian	1 in 431	96%	1 in 11,600	
		Latino	1 in 1121	99%	1 in 112,000	
		South Asian	1 in 1025	99%	1 in 102,000	
		Worldwide	1 in 444	97%	1 in 13,700	
Fanconi Anemia, Group G (AR) NM_004629.1	*FANCG*	African	1 in 494	99%	1 in 49,300	99%
		East Asian	1 in 336	72%	1 in 1,200	
		Finnish	1 in 1220	99%	1 in 122,000	
		Caucasian	1 in 563	98%	1 in 28,100	
		Latino	1 in 1864	99%	1 in 186,000	
		South Asian	1 in 1278	99%	1 in 128,000	
		Worldwide	1 in 602	95%	1 in 12,300	
Fragile X Syndrome (XL) NM_002024.5	*FMR1*	African	1 in 268	99%	1 in 26,700	99%
		Ashkenazi Jewish	1 in 84	99%	1 in 8,300	
		East Asian	1 in 2220	99%	1 in 222,000	
		Caucasian	1 in 187	99%	1 in 18,600	
		Latino	1 in 206	99%	1 in 20,500	
		South Asian	1 in 172	99%	1 in 17,100	
		Worldwide	1 in 181	99%	1 in 18,000	
Fumarase Deficiency (AR) NM_000143.3	*FH*	African	1 in 561	91%	1 in 6,100	98%
		Ashkenazi Jewish	1 in 99	98%	1 in 4,900	
		Finnish	1 in 1109	88%	1 in 9,400	
		Caucasian	1 in 252	93%	1 in 3,700	
		Latino	1 in 801	98%	1 in 40,000	
		South Asian	1 in 3511	31%	1 in 5,100	
		Worldwide	1 in 370	93%	1 in 5,300	

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Galactokinase Deficiency (AR)	*GALK1*	African	1 in 388	57%	1 in 910	98%
NM_000154.1		East Asian	1 in 723	55%	1 in 1,600	
		Finnish	1 in 2578	98%	1 in 129,000	
		Caucasian	1 in 747	72%	1 in 2,700	
		Latino	1 in 663	78%	1 in 3,000	
		South Asian	1 in 400	85%	1 in 2,700	
		Worldwide	1 in 594	74%	1 in 2,300	
		Roma	1 in 47	98%	1 in 2,300	
Galactosemia (AR)	*GALT*	African	1 in 87	86%	1 in 610	99%
NM_000155.3		Ashkenazi Jewish	1 in 181	96%	1 in 4,100	
		East Asian	1 in 208	40%	1 in 350	
		Finnish	1 in 4085	68%	1 in 12,600	
		Caucasian	1 in 123	92%	1 in 1,600	
		Latino	1 in 219	93%	1 in 3,000	
		South Asian	1 in 342	81%	1 in 1,800	
		Worldwide	1 in 156	85%	1 in 1,000	
		Irish Travellers	1 in 11	99%	1 in 1,000	
Gaucher Disease (AR)	*GBA*	Caucasian	1 in 164	87%	1 in 1,300	95%
NM_000157.3		Ashkenazi Jewish	1 in 15	95%	1 in 280	
		Worldwide	1 in 158	86%	1 in 1,100	
Gitelman Syndrome (AR)	*SLC12A3*	African	1 in 138	78%	1 in 620	98%
NM_000339.2		Ashkenazi Jewish	1 in 121	98%	1 in 6,000	
		East Asian	1 in 28	88%	1 in 230	
		Finnish	1 in 239	46%	1 in 450	
		Caucasian	1 in 73	75%	1 in 290	
		Latino	1 in 131	82%	1 in 730	
		South Asian	1 in 145	68%	1 in 460	
		Worldwide	1 in 82	78%	1 in 370	
Glutaric Acidemia, Type I (AR)	*GCDH*	African	1 in 93	76%	1 in 390	99%
NM_000159.3		East Asian	1 in 204	94%	1 in 3,600	
		Finnish	1 in 353	90%	1 in 3,700	
		Caucasian	1 in 201	89%	1 in 1,900	
		Latino	1 in 271	93%	1 in 3,700	
		South Asian	1 in 261	34%	1 in 390	
		Worldwide	1 in 201	81%	1 in 1,000	
		Oji-Cree First Nations (N. Manitoba)	1 in 8	99%	1 in 700	
		Old Order Amish of Pennsylvania	1 in 11	99%	1 in 1,000	
		Lumbee Native American	1 in 16	99%	1 in 1,500	
Glutaric Acidemia, Type IIa (AR)	*ETFA*	African	1 in 939	85%	1 in 6,300	97%
NM_000126.3		East Asian	1 in 1246	41%	1 in 2,100	
		Caucasian	1 in 857	82%	1 in 4,700	
		Latino	1 in 3383	77%	1 in 15,000	
		South Asian	1 in 1099	97%	1 in 36,600	
		Worldwide	1 in 1056	83%	1 in 6,400	
Glutaric Acidemia, Type IIc (AR)	*ETFDH*	African	1 in 343	66%	1 in 1,000	99%
NM_004453.3		Ashkenazi Jewish	1 in 1230	99%	1 in 123,000	
		East Asian	1 in 89	66%	1 in 260	
		Finnish	1 in 941	83%	1 in 5,700	
		Caucasian	1 in 336	80%	1 in 1,700	
		Latino	1 in 586	58%	1 in 1,400	
		South Asian	1 in 733	47%	1 in 1,400	
		Worldwide	1 in 338	71%	1 in 1,200	

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Glycine Encephalopathy (*AMT*-Related) (AR)	*AMT*	East Asian	1 in 1437	33%	1 in 2,100	99%
NM_000481.3		Finnish	1 in 2042	81%	1 in 10,700	
		Caucasian	1 in 779	65%	1 in 2,300	
		Latino	1 in 390	44%	1 in 690	
		South Asian	1 in 905	99%	1 in 90,400	
		Worldwide	1 in 819	64%	1 in 2,300	
Glycine Encephalopathy (*GLDC*-Related) (AR)	*GLDC*	African	1 in 515	49%	1 in 1,000	95%
NM_000170.2		East Asian	1 in 137	58%	1 in 330	
		Finnish	1 in 112	85%	1 in 740	
		Caucasian	1 in 255	70%	1 in 840	
		Latino	1 in 323	64%	1 in 900	
		South Asian	1 in 570	56%	1 in 1,300	
Exception: Exon 1		Worldwide	1 in 246	69%	1 in 780	
Glycogen Storage Disease, Type Ia (AR)	*G6PC*	African	1 in 830	88%	1 in 7,000	99%
NM_000151.3		Ashkenazi Jewish	1 in 75	99%	1 in 7,400	
		East Asian	1 in 116	72%	1 in 410	
		Finnish	1 in 549	99%	1 in 54,800	
		Caucasian	1 in 317	94%	1 in 5,300	
		Latino	1 in 346	89%	1 in 3,100	
		South Asian	1 in 5128	66%	1 in 15,100	
		Worldwide	1 in 308	91%	1 in 3,200	
Glycogen Storage Disease, Type Ib (AR)	*SLC37A4*	African	1 in 1414	99%	1 in 141,000	99%
NM_001164277.1		Ashkenazi Jewish	1 in 1254	99%	1 in 125,000	
		East Asian	1 in 511	87%	1 in 3,900	
		Finnish	1 in 788	99%	1 in 78,700	
		Caucasian	1 in 597	92%	1 in 7,300	
		Latino	1 in 979	92%	1 in 11,700	
		South Asian	1 in 821	94%	1 in 13,000	
		Worldwide	1 in 671	93%	1 in 9,600	
Glycogen Storage Disease, Type II (AR)	*GAA*	African	1 in 71	82%	1 in 380	99%
NM_000152.3		Ashkenazi Jewish	1 in 76	97%	1 in 3,000	
		East Asian	1 in 63	78%	1 in 280	
		Finnish	1 in 366	59%	1 in 890	
		Caucasian	1 in 49	91%	1 in 520	
		Latino	1 in 95	86%	1 in 690	
		South Asian	1 in 133	91%	1 in 1,500	
		Worldwide	1 in 71	87%	1 in 530	
Glycogen Storage Disease, Type III (AR)	*AGL*	African	1 in 191	86%	1 in 1,300	99%
NM_000028.2		East Asian	1 in 549	99%	1 in 54,800	
		Finnish	1 in 1580	99%	1 in 158,000	
		Caucasian	1 in 259	95%	1 in 5,700	
		Latino	1 in 470	96%	1 in 12,700	
		South Asian	1 in 510	73%	1 in 1,900	
		Worldwide	1 in 316	91%	1 in 3,700	
		Sephardic Jewish - Moroccan	1 in 34	99%	1 in 3,300	
		Faroese	1 in 28	99%	1 in 2,700	
Glycogen Storage Disease, Type IV / Adult Polyglucosan Body Disease (AR)	*GBE1*	African	1 in 523	80%	1 in 2,600	98%
		Ashkenazi Jewish	1 in 55	98%	1 in 2,700	
NM_000158.3		East Asian	1 in 1282	98%	1 in 64,000	
		Finnish	1 in 384	95%	1 in 7,700	
		Caucasian	1 in 192	92%	1 in 2,400	
		Latino	1 in 222	95%	1 in 4,500	
		South Asian	1 in 417	84%	1 in 2,600	
		Worldwide	1 in 212	93%	1 in 3,000	

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Glycogen Storage Disease, Type V (AR) NM_005609.2	*PYGM*	African	1 in 220	77%	1 in 940	98%
		Ashkenazi Jewish	1 in 120	72%	1 in 420	
		East Asian	1 in 368	73%	1 in 1,400	
		Finnish	1 in 518	85%	1 in 3,400	
		Caucasian	1 in 116	90%	1 in 1,200	
		Latino	1 in 147	92%	1 in 1,800	
		South Asian	1 in 366	86%	1 in 2,700	
		Worldwide	1 in 158	88%	1 in 1,300	
		Sephardic Jewish - Kurdish	1 in 84	98%	1 in 4,200	
Glycogen Storage Disease, Type VII (AR) NM_000289.5	*PFKM*	African	1 in 387	92%	1 in 10,600	99%
		Ashkenazi Jewish	1 in 100	99%	1 in 9,900	
		East Asian	1 in 870	89%	1 in 7,900	
		Finnish	1 in 1726	46%	1 in 3,200	
		Caucasian	1 in 868	80%	1 in 4,300	
		South Asian	1 in 3078	99%	1 in 308,000	
		Worldwide	1 in 777	88%	1 in 6,300	
GRACILE Syndrome and Other *BCS1L*-Related Disorders (AR) NM_001257342.1	*BCS1L*	African	1 in 457	78%	1 in 2,100	99%
		Ashkenazi Jewish	1 in 169	99%	1 in 16,800	
		East Asian	1 in 822	99%	1 in 82,100	
		Finnish	1 in 95	93%	1 in 1,400	
		Caucasian	1 in 385	90%	1 in 3,900	
		Latino	1 in 552	99%	1 in 55,100	
		South Asian	1 in 616	87%	1 in 4,800	
		Worldwide	1 in 314	92%	1 in 3,900	
Hemochromatosis, Type 2A (AR) NM_213653.3	*HFE2*	African	1 in 1368	43%	1 in 2,400	99%
		East Asian	1 in 527	29%	1 in 740	
		Caucasian	1 in 704	90%	1 in 7,000	
		Latino	1 in 1865	44%	1 in 3,300	
		South Asian	1 in 641	87%	1 in 4,800	
		Worldwide	1 in 857	79%	1 in 4,000	
Hemochromatosis, Type 3 (AR) NM_003227.3	*TFR2*	African	1 in 761	82%	1 in 4,200	99%
		East Asian	1 in 2749	99%	1 in 275,000	
		Caucasian	1 in 604	95%	1 in 11,400	
		Latino	1 in 378	99%	1 in 37,700	
		South Asian	1 in 1259	75%	1 in 5,000	
		Worldwide	1 in 677	91%	1 in 7,400	
Hereditary Fructose Intolerance (AR) NM_000035.3	*ALDOB*	African	1 in 319	98%	1 in 15,900	98%
		Ashkenazi Jewish	1 in 141	98%	1 in 7,000	
		East Asian	1 in 705	98%	1 in 35,200	
		Finnish	1 in 100	98%	1 in 5,000	
		Caucasian	1 in 81	96%	1 in 1,900	
		Latino	1 in 235	94%	1 in 3,900	
		South Asian	1 in 394	95%	1 in 8,700	
		Worldwide	1 in 120	96%	1 in 3,000	
Hereditary Spastic Paraparesis 49 (AR) NM_014844.4	*TECPR2*	African	1 in 1869	99%	1 in 187,000	99%
		Ashkenazi Jewish	1 in 151	99%	1 in 15,000	
		East Asian	1 in 1666	99%	1 in 166,000	
		Finnish	1 in 929	99%	1 in 92,800	
		Caucasian	1 in 1072	91%	1 in 12,400	
		Latino	1 in 5596	99%	1 in 559,000	
		South Asian	1 in 1924	25%	1 in 2,600	
		Worldwide	1 in 1030	91%	1 in 11,100	
		Sephardic Jewish - Bukharian	1 in 27	99%	1 in 2,600	

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Hermansky-Pudlak Syndrome, Type 1 (AR) NM_000195.4	*HPS1*	African	1 in 906	84%	1 in 5,680	99%
		East Asian	1 in 2863	99%	1 in 286,000	
		Finnish	1 in 550	99%	1 in 54,900	
		Caucasian	1 in 493	86%	1 in 3,500	
		Latino	1 in 999	99%	1 in 99,800	
		South Asian	1 in 1539	99%	1 in 154,000	
		Worldwide	1 in 634	90%	1 in 6,300	
		Puerto Rican	1 in 59	99%	1 in 5,800	
Hermansky-Pudlak Syndrome, Type 3 (AR) NM_032383.4	*HPS3*	African	1 in 799	99%	1 in 79,800	99%
		Ashkenazi Jewish	1 in 266	99%	1 in 26,500	
		East Asian	1 in 219	99%	1 in 21,800	
		Caucasian	1 in 491	99%	1 in 49,000	
		Latino	1 in 3365	95%	1 in 67,300	95%
		South Asian	1 in 393	79%	1 in 1,850	
		Worldwide	1 in 518	96%	1 in 11,900	
HMG-CoA Lyase Deficiency (AR) NM_000191.2	*HMGCL*	African	1 in 964	98%	1 in 48,100	98%
		East Asian	1 in 2253	98%	1 in 113,000	
		Finnish	1 in 1330	98%	1 in 66,500	
		Caucasian	1 in 875	67%	1 in 2,700	
		Latino	1 in 1123	98%	1 in 56,100	
		South Asian	1 in 1283	98%	1 in 64,100	
		Worldwide	1 in 995	81%	1 in 5,400	
Holocarboxylase Synthetase Deficiency (AR) NM_000411.6	*HLCS*	African	1 in 570	92%	1 in 6,800	99%
		East Asian	1 in 342	95%	1 in 6,900	
		Finnish	1 in 1433	99%	1 in 143,000	
		Caucasian	1 in 703	87%	1 in 5,500	
		Latino	1 in 706	87%	1 in 5,200	
		South Asian	1 in 1099	99%	1 in 110,000	
		Worldwide	1 in 675	91%	1 in 7,400	
Homocystinuria (*CBS*-Related) (AR) NM_000071.2	*CBS*	African	1 in 188	95%	1 in 3,500	97%
		Ashkenazi Jewish	1 in 330	90%	1 in 3,500	
		East Asian	1 in 589	73%	1 in 2,200	
		Finnish	1 in 336	94%	1 in 5,600	
		Caucasian	1 in 142	90%	1 in 1,400	
		Latino	1 in 202	93%	1 in 3,100	
		South Asian	1 in 523	89%	1 in 5,000	
		Worldwide	1 in 179	91%	1 in 1,900	
		Qatari	1 in 21	86%	1 in 140	
Homocystinuria due to *MTHFR* Deficiency (AR) NM_005957.4 *Variant tested:* p.G158G *(Genotyping only)*	*MTHFR*	Sephardic Jewish - Bukharian	1 in 39	99%	1 in 3,800	99%
Homocystinuria, cblE Type (AR) NM_002454.2	*MTRR*	African	1 in 759	99%	1 in 75,800	99%
		Ashkenazi Jewish	1 in 1658	99%	1 in 166,000	
		Finnish	1 in 1523	99%	1 in 152,000	
		Caucasian	1 in 642	93%	1 in 9,600	
		Latino	1 in 489	96%	1 in 12,600	
		South Asian	1 in 2565	99%	1 in 256,000	
		Worldwide	1 in 735	95%	1 in 16,100	

Hydrolethalus Syndrome (AR) NM_001134793.1	*HYLS1*	African	1 in 1092	99%	1 in 109,000	99%
		East Asian	1 in 2959	99%	1 in 296,000	
		Finnish	1 in 51	99%	1 in 5,000	
		Caucasian	1 in 522	99%	1 in 52,100	
		Latino	1 in 885	99%	1 in 88,400	
		South Asian	1 in 2199	99%	1 in 220,000	
		Worldwide	1 in 317	99%	1 in 31,600	
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome (AR NM_014252.3	*SLC25A15*	East Asian	1 in 302	99%	1 in 30,200	99%
		Finnish	1 in 3224	99%	1 in 322,000	
		Caucasian	1 in 1283	78%	1 in 5,700	
		Latino	1 in 1119	99%	1 in 112,000	
		South Asian	1 in 1924	74%	1 in 7,500	
		Worldwide	1 in 1186	87%	1 in 8,900	
		Metis - Saskatchewan	1 in 19	99%	1 in 1,800	
Hypohidrotic Ectodermal Dysplasia 1 (XL) NM_001399.4	*EDA*	Worldwide	1 in 6000	73%	1 in 22,000	97%
Hypophosphatasia (AR) NM_000478.4	*ALPL*	African	1 in 588	87%	1 in 4,400	99%
		Ashkenazi Jewish	1 in 825	66%	1 in 2,500	
		East Asian	1 in 131	97%	1 in 5,200	
		Finnish	1 in 28	96%	1 in 660	
		Caucasian	1 in 119	85%	1 in 790	
		Latino	1 in 447	49%	1 in 880	
		South Asian	1 in 810	68%	1 in 2,500	
		Worldwide	1 in 117	89%	1 in 1,000	
		Mennonite	1 in 25	99%	1 in 2,400	
Inclusion Body Myopathy 2 (AR) NM_005476.5	*GNE*	African	1 in 379	54%	1 in 820	99%
		Ashkenazi Jewish	1 in 1641	66%	1 in 4,800	
		East Asian	1 in 271	90%	1 in 2,600	
		Finnish	1 in 2989	46%	1 in 5,500	
		Caucasian	1 in 279	86%	1 in 2,000	
		Latino	1 in 765	63%	1 in 2,100	
		South Asian	1 in 36	98%	1 in 1,600	
		Worldwide	1 in 174	89%	1 in 1,500	
		Sephardic Jewish - Iranian and Syrian	1 in 10	99%	1 in 900	
Infantile Cerebral and Cerebellar Atrophy (AR) NM_004268.4	*MED17*	African	1 in 752	99%	1 in 75,100	99%
		Caucasian	1 in 1287	99%	1 in 129,000	
		Latino	1 in 5594	99%	1 in 559,000	
		South Asian	1 in 3078	99%	1 in 308,000	
		Worldwide	1 in 1298	99%	1 in 130,000	
		Sephardic Jewish - Bukharian and Kurdish	1 in 20	99%	1 in 1,900	
Isovaleric Acidemia (AR) NM_002225.3	*IVD*	African	1 in 302	88%	1 in 2,400	99%
		East Asian	1 in 901	78%	1 in 4,200	
		Finnish	1 in 1992	81%	1 in 10,700	
		Caucasian	1 in 250	87%	1 in 2,000	
		Latino	1 in 532	90%	1 in 5,100	
		South Asian	1 in 733	75%	1 in 3,000	
		Worldwide	1 in 339	88%	1 in 2,800	
Joubert Syndrome 2 (AR) NM_001173990.2	*TMEM216*	African	1 in 3364	99%	1 in 336,000	99%
		Ashkenazi Jewish	1 in 137	99%	1 in 13,600	
		Caucasian	1 in 1521	99%	1 in 152,000	
		Latino	1 in 2035	99%	1 in 203,000	
		South Asian	1 in 3526	99%	1 in 353,000	
		Worldwide	1 in 1330	99%	1 in 133,000	

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Joubert Syndrome 7 / Meckel Syndrome 5 / COACH Syndrome (AR)	*RPGRIP1L*	African	1 in 257	99%	1 in 25,600	99%
NM_015272.2		East Asian	1 in 197	82%	1 in 1,100	
		Finnish	1 in 989	99%	1 in 98,800	
		Caucasian	1 in 319	99%	1 in 31,800	
		Latino	1 in 619	95%	1 in 13,200	
		South Asian	1 in 528	92%	1 in 6,800	
Exception: Exon 22		Worldwide	1 in 341	96%	1 in 9,000	
Junctional Epidermolysis Bullosa (*LAMA3*-Related) (AR)	*LAMA3*	African	1 in 782	99%	1 in 78,100	99%
NM_000227.4		East Asian	1 in 495	99%	1 in 49,400	
		Finnish	1 in 891	24%	1 in 1,200	
		Caucasian	1 in 606	97%	1 in 20,900	
		Latino	1 in 1416	99%	1 in 142,000	
		South Asian	1 in 810	99%	1 in 80,900	
		Worldwide	1 in 704	92%	1 in 9,300	
Junctional Epidermolysis Bullosa (*LAMB3*-Related) (AR)	*LAMB3*	African	1 in 268	97%	1 in 8,300	99%
NM_000228.2		Ashkenazi Jewish	1 in 984	99%	1 in 98,300	
		East Asian	1 in 877	90%	1 in 8,600	
		Finnish	1 in 957	99%	1 in 95,600	
		Caucasian	1 in 222	89%	1 in 1,900	
		Latino	1 in 1122	99%	1 in 112,000	
		South Asian	1 in 629	99%	1 in 62,800	
		Worldwide	1 in 334	91%	1 in 3,800	
Junctional Epidermolysis Bullosa (*LAMC2*-Related) (AR)	*LAMC2*	African	1 in 823	99%	1 in 82,200	99%
NM_018891.2		East Asian	1 in 285	99%	1 in 28,400	
		Caucasian	1 in 772	99%	1 in 77,100	
		Latino	1 in 4197	99%	1 in 420,000	
		South Asian	1 in 1707	99%	1 in 171,000	
		Worldwide	1 in 777	99%	1 in 77,600	
Krabbe Disease (AR)	*GALC*	African	1 in 119	38%	1 in 190	99%
NM_000153.3		Ashkenazi Jewish	1 in 532	57%	1 in 1,300	
		East Asian	1 in 40	81%	1 in 200	
		Finnish	1 in 146	99%	1 in 14,500	
		Caucasian	1 in 67	88%	1 in 570	
		Latino	1 in 181	80%	1 in 900	
		South Asian	1 in 35	91%	1 in 370	
		Worldwide	1 in 74	83%	1 in 440	
		Druze Northern Israel	1 in 6	99%	1 in 500	
		Muslim Arab (Jerusalem)	1 in 6	99%	1 in 500	
Lamellar Ichthyosis, Type 1 (AR)	*TGM1*	African	1 in 205	76%	1 in 840	99%
NM_000359.2		Ashkenazi Jewish	1 in 620	99%	1 in 61,900	
		East Asian	1 in 279	96%	1 in 6,600	
		Finnish	1 in 179	92%	1 in 2,300	
		Caucasian	1 in 186	84%	1 in 1,100	
		Latino	1 in 562	86%	1 in 4,000	
		South Asian	1 in 79	15%	1 in 93	
		Worldwide	1 in 181	67%	1 in 540	
		Norwegian	1 in 151	80%	1 in 750	
Leber Congenital Amaurosis 10 and Other *CEP290*-Related Ciliopathies (AR)	*CEP290*	African	1 in 131	90%	1 in 1,300	99%
NM_025114.3		Ashkenazi Jewish	1 in 461	86%	1 in 3,200	
		East Asian	1 in 32	97%	1 in 1,100	
		Finnish	1 in 713	99%	1 in 71,200	
		Caucasian	1 in 97	96%	1 in 2,700	
		Latino	1 in 199	90%	1 in 2,000	
		South Asian	1 in 222	99%	1 in 22,100	
Exception: Exons 13, 32, 40		Worldwide	1 in 120	96%	1 in 2,800	

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Leber Congenital Amaurosis 13 (AR) NM_152443.2	*RDH12*	African	1 in 302	93%	1 in 4,100	99%
		East Asian	1 in 877	99%	1 in 87,600	
		Caucasian	1 in 517	91%	1 in 5,500	
		Latino	1 in 290	89%	1 in 2,600	
		South Asian	1 in 549	46%	1 in 1,000	
		Worldwide	1 in 474	83%	1 in 2,800	
Leber Congenital Amaurosis 2 / Retinitis Pigmentosa 20 (AR) NM_000329.2	*RPE65*	African	1 in 190	97%	1 in 5,400	99%
		East Asian	1 in 289	86%	1 in 2,100	
		Finnish	1 in 684	83%	1 in 4,100	
		Caucasian	1 in 366	85%	1 in 2,500	
		Latino	1 in 345	75%	1 in 1,400	
		South Asian	1 in 265	46%	1 in 490	
		Worldwide	1 in 321	81%	1 in 1,700	
		Sephardic Jewish - North African	1 in 90	99%	1 in 8,900	
Leber Congenital Amaurosis 5 (AR) NM_181714.3	*LCA5*	Ashkenazi Jewish	1 in 234	99%	1 in 23,300	99%
		East Asian	1 in 984	76%	1 in 4,200	
		Caucasian	1 in 1811	87%	1 in 14,200	
		Latino	1 in 1703	60%	1 in 4,200	
		South Asian	1 in 1390	63%	1 in 3,800	
		Worldwide	1 in 1308	85%	1 in 8,800	
Leber Congenital Amaurosis 8 / Retinitis Pigmentosa 12 (AR) NM_201253.2	*CRB1*	African	1 in 116	97%	1 in 3,300	99%
		Ashkenazi Jewish	1 in 389	91%	1 in 4,400	
		East Asian	1 in 187	81%	1 in 960	
		Finnish	1 in 1003	91%	1 in 11,500	
		Caucasian	1 in 158	84%	1 in 990	
		Latino	1 in 263	87%	1 in 2,000	
		South Asian	1 in 531	48%	1 in 1,000	
		Worldwide	1 in 190	85%	1 in 1,300	
Leigh Syndrome, French-Canadian Type (AR) NM_133259.3	*LRPPRC*	African	1 in 655	99%	1 in 65,400	99%
		East Asian	1 in 222	99%	1 in 22,100	
		Finnish	1 in 472	99%	1 in 47,100	
		Caucasian	1 in 768	98%	1 in 32,400	
		Latino	1 in 1786	99%	1 in 178,000	
		South Asian	1 in 758	99%	1 in 75,700	
		Worldwide	1 in 574	96%	1 in 13,500	
		French Canadian - Saguenay-Lac St. Jean	1 in 23	99%	1 in 2,200	
Lethal Congenital Contracture Syndrome 1 / Cell Lethal Arthrogryposis with Anterior Horn Disease (AR) NM_001003722.1	*GLE1*	African	1 in 1148	65%	1 in 3,300	99%
		East Asian	1 in 2302	27%	1 in 3,100	
		Finnish	1 in 40	97%	1 in 1,500	
		Caucasian	1 in 453	90%	1 in 4,800	
		Latino	1 in 1201	57%	1 in 2,800	
		South Asian	1 in 669	85%	1 in 4,800	
		Worldwide	1 in 275	93%	1 in 3,700	
Leukoencephalopathy with Vanishing White Matter (AR) NM_003907.2	*EIF2B5*	African	1 in 940	59%	1 in 2,300	99%
		East Asian	1 in 1502	82%	1 in 8,200	
		Caucasian	1 in 390	83%	1 in 2,300	
		Latino	1 in 458	77%	1 in 2,000	
		South Asian	1 in 3078	59%	1 in 7,600	
		Worldwide	1 in 598	80%	1 in 3,000	

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Limb-Girdle Muscular Dystrophy, Type 2A (AR) NM_000070.2	*CAPN3*	African	1 in 111	64%	1 in 310	99%
		Ashkenazi Jewish	1 in 563	99%	1 in 56,200	
		East Asian	1 in 104	78%	1 in 470	
		Finnish	1 in 411	73%	1 in 1,600	
		Caucasian	1 in 103	86%	1 in 720	
		Latino	1 in 144	91%	1 in 1,700	
		South Asian	1 in 223	80%	1 in 1,100	
		Worldwide	1 in 127	84%	1 in 770	
		Amish	N/A	99%	N/A	
Limb-Girdle Muscular Dystrophy, Type 2B (AR) NM_003494.3	*DYSF*	African	1 in 118	75%	1 in 460	96%
		Ashkenazi Jewish	1 in 310	30%	1 in 440	
		East Asian	1 in 141	87%	1 in 1,000	
		Finnish	1 in 1140	52%	1 in 2,400	
		Caucasian	1 in 199	77%	1 in 870	
		Latino	1 in 182	74%	1 in 700	
		South Asian	1 in 199	65%	1 in 570	
		Worldwide	1 in 184	73%	1 in 680	
		Sephardic Jewish - Libyan, Kavkazi and Yemenite	1 in 14	96%	1 in 330	
Limb-Girdle Muscular Dystrophy, Type 2C (AR) NM_000231.2	*SGCG*	African	1 in 828	86%	1 in 5,800	92%
		Caucasian	1 in 1132	77%	1 in 4,900	
		Latino	1 in 2105	92%	1 in 26,300	
		South Asian	1 in 2955	92%	1 in 36,900	
		Worldwide	1 in 1408	82%	1 in 8,000	
		Moroccan	1 in 250	77%	1 in 1,100	
		Roma	1 in 96	92%	1 in 1,200	
Limb-Girdle Muscular Dystrophy, Type 2D (AR) NM_000023.2	*SGCA*	African	1 in 427	84%	1 in 2,600	99%
		Ashkenazi Jewish	1 in 276	99%	1 in 27,500	
		East Asian	1 in 2202	74%	1 in 8,400	
		Finnish	1 in 257	99%	1 in 25,600	
		Caucasian	1 in 361	90%	1 in 3,500	
		Latino	1 in 951	88%	1 in 7,800	
		South Asian	1 in 1539	69%	1 in 5,000	
		Worldwide	1 in 403	87%	1 in 3,000	
Limb-Girdle Muscular Dystrophy, Type 2E (AR) NM_000232.4	*SGCB*	African	1 in 653	98%	1 in 32,600	98%
		East Asian	1 in 1437	98%	1 in 71,800	
		Finnish	1 in 2092	98%	1 in 105,000	
		Caucasian	1 in 628	98%	1 in 31,400	
		Latino	1 in 3358	98%	1 in 168,000	
		South Asian	1 in 373	98%	1 in 18,600	
		Worldwide	1 in 558	98%	1 in 27,800	
Limb-Girdle Muscular Dystrophy, Type 2I (AR) NM_024301.4	*FKRP*	African	1 in 452	86%	1 in 3,300	99%
		Ashkenazi Jewish	1 in 184	87%	1 in 1,400	
		East Asian	1 in 196	57%	1 in 460	
		Finnish	1 in 229	99%	1 in 22,800	
		Caucasian	1 in 176	86%	1 in 1,300	
		Latino	1 in 239	16%	1 in 280	
		South Asian	1 in 2190	45%	1 in 4,000	
		Worldwide	1 in 220	75%	1 in 880	
		Norwegian	1 in 116	99%	1 in 11,500	

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Lipoamide Dehydrogenase Deficiency (AR) NM_000108.4	*DLD*	Ashkenazi Jewish	1 in 60	99%	1 in 5,900	99%
		East Asian	1 in 2252	99%	1 in 225,000	
		Finnish	1 in 705	99%	1 in 70,400	
		Caucasian	1 in 1506	89%	1 in 13,600	
		Latino	1 in 1684	49%	1 in 3,300	
		South Asian	1 in 1183	99%	1 in 118,000	
		Worldwide	1 in 720	93%	1 in 10,800	
Lipoid Adrenal Hyperplasia (AR) NM_000349.2	*STAR*	African	1 in 964	91%	1 in 10,800	99%
		East Asian	1 in 364	99%	1 in 36,300	
		Finnish	1 in 1841	71%	1 in 6,300	
		Caucasian	1 in 1147	68%	1 in 3,600	
		Latino	1 in 731	69%	1 in 2,400	
		South Asian	1 in 1399	81%	1 in 7,400	
		Worldwide	1 in 917	79%	1 in 4,300	
Lipoprotein Lipase Deficiency (AR) NM_000237.2	*LPL*	African	1 in 308	77%	1 in 1,300	99%
		East Asian	1 in 103	87%	1 in 800	
		Caucasian	1 in 374	84%	1 in 2,400	
		Latino	1 in 373	64%	1 in 1,100	
		South Asian	1 in 452	50%	1 in 900	
		Worldwide	1 in 342	78%	1 in 1,600	
		French Canadian - Saguenay - Lac St. Jean	1 in 46	99%	1 in 4,500	
		French Canadian - Other	1 in 139	99%	1 in 13,800	
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (AR) NM_000182.4	*HADHA*	African	1 in 482	78%	1 in 2,200	99%
		East Asian	1 in 1006	78%	1 in 4,600	
		Finnish	1 in 123	99%	1 in 12,200	
		Caucasian	1 in 216	96%	1 in 5,900	
		Latino	1 in 407	94%	1 in 7,100	
		South Asian	1 in 733	99%	1 in 73,200	
		Worldwide	1 in 262	95%	1 in 4,900	
Lysinuric Protein Intolerance (AR) NM_001126106.2	*SLC7A7*	African	1 in 595	81%	1 in 3,200	99%
		East Asian	1 in 724	99%	1 in 72,300	
		Finnish	1 in 106	99%	1 in 10,500	
		Caucasian	1 in 522	83%	1 in 3,000	
		Latino	1 in 2821	99%	1 in 282,000	
		South Asian	1 in 1283	91%	1 in 13,900	
		Worldwide	1 in 449	91%	1 in 5,100	
		Japanese	1 in 119	88%	1 in 980	
Maple Syrup Urine Disease, Type 1a (AR) NM_000709.3	*BCKDHA*	African	1 in 478	70%	1 in 1,600	98%
		Ashkenazi Jewish	1 in 338	98%	1 in 16,900	
		East Asian	1 in 869	78%	1 in 4,000	
		Finnish	1 in 2771	98%	1 in 138,000	
		Caucasian	1 in 555	89%	1 in 5,100	
		Latino	1 in 837	93%	1 in 12,100	
		South Asian	1 in 1068	98%	1 in 53,300	
		Worldwide	1 in 595	90%	1 in 5,700	
		Mennonite	1 in 10	98%	1 in 450	
		Portuguese Roma	1 in 71	98%	1 in 3,500	

Maple Syrup Urine Disease, Type 1b (AR) NM_000056.3	*BCKDHB*	African	1 in 608	76%	1 in 2,500	99%
		Ashkenazi Jewish	1 in 82	99%	1 in 8,100	
		East Asian	1 in 666	84%	1 in 4,100	
		Finnish	1 in 179	99%	1 in 17,800	
		Caucasian	1 in 306	73%	1 in 1,100	
		Latino	1 in 412	94%	1 in 7,000	
		South Asian	1 in 1665	78%	1 in 7,400	
		Worldwide	1 in 299	85%	1 in 1,900	
Meckel Syndrome 1 / Bardet-Biedl Syndrome 13 (AR) NM_017777.3	*MKS1*	African	1 in 750	80%	1 in 3,700	99%
		Ashkenazi Jewish	1 in 1269	99%	1 in 127,000	
		East Asian	1 in 283	99%	1 in 28,200	
		Finnish	1 in 71	99%	1 in 7,000	
		Caucasian	1 in 246	85%	1 in 1,700	
		Latino	1 in 1066	99%	1 in 106,000	
		South Asian	1 in 355	74%	1 in 1,400	
		Worldwide	1 in 246	90%	1 in 2,500	
Medium Chain Acyl-CoA Dehydrogenase Deficiency (AR) NM_000016.5	*ACADM*	African	1 in 172	77%	1 in 740	99%
		Ashkenazi Jewish	1 in 133	99%	1 in 13,200	
		East Asian	1 in 255	35%	1 in 390	
		Finnish	1 in 383	96%	1 in 8,700	
		Caucasian	1 in 56	95%	1 in 1,100	
		Latino	1 in 92	63%	1 in 250	
		South Asian	1 in 142	51%	1 in 290	
		Worldwide	1 in 82	85%	1 in 560	
Megalencephalic Leukoencephalopathy with Subcortical Cysts (AR) NM_015166.3	*MLC1*	African	1 in 737	82%	1 in 4,200	99%
		Ashkenazi Jewish	1 in 196	99%	1 in 19,500	
		East Asian	1 in 1710	99%	1 in 171,000	
		Finnish	1 in 2785	99%	1 in 278,000	
		Caucasian	1 in 884	79%	1 in 4,300	
		Latino	1 in 5597	99%	1 in 560,000	
		South Asian	1 in 1280	99%	1 in 128,000	
		Worldwide	1 in 825	85%	1 in 5,500	
		Sephardic Jewish - Libyan	1 in 40	99%	1 in 3,900	
Menkes Disease (XL) NM_000052.6	*ATP7A*	Worldwide	< 1 in 50,000	71%	1 in 170,000	99%
Metachromatic Leukodystrophy (AR) NM_000487.5	*ARSA*	African	1 in 239	80%	1 in 1,200	99%
		Ashkenazi Jewish	1 in 823	82%	1 in 4,600	
		East Asian	1 in 364	86%	1 in 2,600	
		Finnish	1 in 258	97%	1 in 7,800	
		Caucasian	1 in 131	87%	1 in 1,000	
		Latino	1 in 503	90%	1 in 5,000	
		South Asian	1 in 371	82%	1 in 2,100	
		Worldwide	1 in 179	86%	1 in 1,300	
		Sephardic Jewish - Yemenite	1 in 46	99%	1 in 4,500	
		Navajo	1 in 25	99%	1 in 2,400	
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC1*-Related) (AR) NM_020166.4	*MCCC1*	African	1 in 266	51%	1 in 540	99%
		East Asian	1 in 204	37%	1 in 330	
		Caucasian	1 in 353	82%	1 in 1,900	
		Latino	1 in 488	91%	1 in 5,100	
		South Asian	1 in 1000	99%	1 in 99,900	
		Worldwide	1 in 423	73%	1 in 1,600	

3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC2*-Related) (AR) NM_022132.4	*MCCC2*	African	1 in 407	81%	1 in 2,200	99%
		Ashkenazi Jewish	1 in 267	99%	1 in 26,600	
		East Asian	1 in 192	62%	1 in 500	
		Finnish	1 in 2230	79%	1 in 10,700	
		Caucasian	1 in 204	83%	1 in 1,200	
		Latino	1 in 125	98%	1 in 5,100	
		South Asian	1 in 308	69%	1 in 1,000	
		Worldwide	1 in 213	83%	1 in 1,300	
3-Methylglutaconic Aciduria, Type III (AR) NM_025136.3	*OPA3*	Caucasian	1 in 4808	90%	1 in 49,700	99%
		Latino	1 in 3349	59%	1 in 8,300	
		Worldwide	1 in 4526	84%	1 in 28,800	
		Sephardic Jewish - Iraqi	1 in 13	99%	1 in 1,200	
Methylmalonic Acidemia (*MMAA*-Related) (AR) NM_172250.2	*MMAA*	East Asian	1 in 2156	99%	1 in 216,000	99%
		Finnish	1 in 3890	99%	1 in 389,000	
		Caucasian	1 in 677	95%	1 in 14,600	
		Latino	1 in 2098	86%	1 in 15,500	
		South Asian	1 in 1167	91%	1 in 13,700	
		Worldwide	1 in 1082	95%	1 in 20,100	
Methylmalonic Acidemia (*MMAB*-Related) (AR) NM_052845.3	*MMAB*	African	1 in 542	56%	1 in 1,200	99%
		Caucasian	1 in 672	94%	1 in 11,800	
		Latino	1 in 1411	41%	1 in 2,400	
		South Asian	1 in 640	49%	1 in 1,300	
		Worldwide	1 in 859	77%	1 in 3,800	
Methylmalonic Acidemia (*MUT*-Related) (AR) NM_000255.3	*MUT*	African	1 in 167	88%	1 in 1,400	99%
		Ashkenazi Jewish	1 in 329	99%	1 in 32,800	
		East Asian	1 in 190	77%	1 in 830	
		Finnish	1 in 572	86%	1 in 4,000	
		Caucasian	1 in 296	77%	1 in 1,300	
		Latino	1 in 195	96%	1 in 4,400	
		South Asian	1 in 265	79%	1 in 1,200	
		Worldwide	1 in 251	84%	1 in 1,500	
Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type (AR) NM_015506.2	*MMACHC*	African	1 in 280	94%	1 in 5,000	99%
		Ashkenazi Jewish	1 in 203	99%	1 in 20,200	
		East Asian	1 in 184	86%	1 in 1,300	
		Caucasian	1 in 173	97%	1 in 6,800	
		Latino	1 in 102	99%	1 in 10,100	
		South Asian	1 in 230	87%	1 in 1,800	
		Worldwide	1 in 181	96%	1 in 4,500	
Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type (AR) NM_015702.2	*MMADHC*	African	1 in 3366	99%	1 in 336,000	99%
		East Asian	1 in 1720	99%	1 in 172,000	
		Caucasian	1 in 2194	99%	1 in 219,000	
		Latino	1 in 5641	99%	1 in 564,000	
		South Asian	1 in 1282	99%	1 in 128,000	
		Worldwide	1 in 2503	99%	1 in 250,000	
Microphthalmia / Anophthalmia (AR) NM_182894.2	*VSX2*	African	1 in 1608	78%	1 in 7,400	99%
		East Asian	1 in 829	99%	1 in 82,800	
		Finnish	1 in 1852	99%	1 in 185,000	
		Caucasian	1 in 1337	97%	1 in 39,600	
		Latino	1 in 2776	99%	1 in 278,000	
		South Asian	1 in 3960	99%	1 in 396,000	
		Worldwide	1 in 1511	97%	1 in 44,000	
		Sephardic Jewish - Iranian and Syrian	1 in 145	99%	1 in 14,400	

Mitochondrial Complex I Deficiency (*ACAD9*-Related) (AR) NM_014049.4	*ACAD9*	African	1 in 784	86%	1 in 5,600	99%
		Ashkenazi Jewish	1 in 1239	99%	1 in 124,000	
		East Asian	1 in 2252	75%	1 in 9,100	
		Finnish	1 in 2094	83%	1 in 12,200	
		Caucasian	1 in 309	83%	1 in 1,900	
		Latino	1 in 741	90%	1 in 7,700	
		South Asian	1 in 810	68%	1 in 2,500	
		Worldwide	1 in 472	84%	1 in 2,900	
Mitochondrial Complex I Deficiency (*NDUFAF5*-Related) (AR) NM_024120.4	*NDUFAF5*	African	1 in 1487	99%	1 in 149,000	99%
		Ashkenazi Jewish	1 in 492	99%	1 in 49,100	
		East Asian	1 in 282	13%	1 in 320	
		Caucasian	1 in 982	99%	1 in 98,100	
		Latino	1 in 841	99%	1 in 84,000	
		South Asian	1 in 1183	99%	1 in 118,000	
		Worldwide	1 in 806	82%	1 in 4,500	
Mitochondrial Complex I Deficiency (*NDUFS6*-Related) (AR) NM_004553.4	*NDUFS6*	East Asian	1 in 2112	99%	1 in 211,000	99%
		Caucasian	1 in 3535	99%	1 in 353,000	
		Latino	1 in 4159	99%	1 in 416,000	
		South Asian	1 in 2162	99%	1 in 216,000	
		Worldwide	1 in 3710	99%	1 in 371,000	
		Sephardic Jewish - Caucasus	1 in 24	99%	1 in 2,300	
Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy (AR) NM_002437.4	*MPV17*	African	1 in 566	99%	1 in 56,500	99%
		Ashkenazi Jewish	1 in 1618	99%	1 in 162,000	
		Caucasian	1 in 612	86%	1 in 4,400	
		South Asian	1 in 1399	90%	1 in 14,000	
		Worldwide	1 in 929	89%	1 in 8,400	
		Navajo	1 in 20	99%	1 in 1,900	
Mitochondrial Myopathy and Sideroblastic Anemia 1 (AR) NM_025215.5	*PUS1*	African	1 in 2039	99%	1 in 204,000	99%
		Finnish	1 in 2001	99%	1 in 200,000	
		Caucasian	1 in 4496	99%	1 in 449,000	
		Latino	1 in 3203	99%	1 in 320,000	
		South Asian	1 in 5130	99%	1 in 513,000	
		Worldwide	1 in 3330	99%	1 in 333,000	
		Sephardic Jewish - Iranian	N/A	99%	N/A	
Mucolipidosis II / IIIA (AR) NM_024312.4	*GNPTAB*	African	1 in 328	99%	1 in 32,700	99%
		Ashkenazi Jewish	1 in 1657	99%	1 in 166,000	
		East Asian	1 in 368	68%	1 in 1,100	
		Finnish	1 in 159	99%	1 in 15,800	
		Caucasian	1 in 222	89%	1 in 2,100	
		Latino	1 in 287	91%	1 in 3,000	
		South Asian	1 in 321	97%	1 in 10,400	
		Worldwide	1 in 240	91%	1 in 2,800	
Mucolipidosis III Gamma (AR) NM_032520.4	*GNPTG*	African	1 in 486	99%	1 in 48,500	99%
		Ashkenazi Jewish	1 in 507	99%	1 in 50,600	
		East Asian	1 in 2133	99%	1 in 213,000	
		Finnish	1 in 1782	99%	1 in 178,000	
		Caucasian	1 in 684	99%	1 in 68,300	
		Latino	1 in 735	99%	1 in 73,500	
		South Asian	1 in 1398	81%	1 in 7,400	
		Worldwide	1 in 734	98%	1 in 33,600	

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Mucolipidosis IV (AR) NM_020533.2	*MCOLN1*	African	1 in 2037	99%	1 in 204,000	99%
		Ashkenazi Jewish	1 in 92	99%	1 in 9,100	
		Caucasian	1 in 1166	88%	1 in 9,400	
		Latino	1 in 1537	63%	1 in 4,100	
		South Asian	1 in 2565	83%	1 in 14,700	
		Worldwide	1 in 926	86%	1 in 6,500	
Mucopolysaccharidosis, Type I (AR) NM_000203.4	*IDUA*	African	1 in 376	90%	1 in 3,900	99%
		Ashkenazi Jewish	1 in 1088	99%	1 in 109,000	
		East Asian	1 in 236	63%	1 in 630	
		Finnish	1 in 184	99%	1 in 18,300	
		Caucasian	1 in 115	97%	1 in 3,300	
		Latino	1 in 416	92%	1 in 5,000	
		South Asian	1 in 114	97%	1 in 4,100	
		Worldwide	1 in 144	95%	1 in 2,700	
Mucopolysaccharidosis, Type II (XL) NM_000202.6 *Exception:* Exon 3	*IDS*	Worldwide	1 in 25,000	67%	1 in 75,000	90%
Mucopolysaccharidosis, Type IIIA (AR) NM_000199.3	*SGSH*	African	1 in 470	76%	1 in 2,000	99%
		East Asian	1 in 216	69%	1 in 700	
		Finnish	1 in 514	99%	1 in 51,300	
		Caucasian	1 in 220	92%	1 in 2,700	
		Latino	1 in 436	73%	1 in 1,600	
		South Asian	1 in 459	58%	1 in 1,100	
		Worldwide	1 in 291	85%	1 in 1,900	
Mucopolysaccharidosis, Type IIIB (AR) NM_000263.3	*NAGLU*	African	1 in 216	83%	1 in 1,300	99%
		Ashkenazi Jewish	1 in 117	89%	1 in 1,100	
		East Asian	1 in 324	64%	1 in 900	
		Finnish	1 in 570	10%	1 in 640	
		Caucasian	1 in 199	79%	1 in 950	
		Latino	1 in 647	72%	1 in 2,300	
		South Asian	1 in 442	62%	1 in 1,200	
		Worldwide	1 in 249	73%	1 in 910	
Mucopolysaccharidosis, Type IIIC (AR) NM_152419.2 *Exception:* Exon 1	*HGSNAT*	African	1 in 604	82%	1 in 3,400	98%
		East Asian	1 in 836	98%	1 in 41,700	
		Finnish	1 in 679	98%	1 in 33,900	
		Caucasian	1 in 443	86%	1 in 3,200	
		Latino	1 in 922	76%	1 in 3,800	
		South Asian	1 in 1483	98%	1 in 74,100	
		Worldwide	1 in 594	87%	1 in 4,600	
Mucopolysaccharidosis, Type IIID (AR) NM_002076.3	*GNS*	Caucasian	1 in 2731	98%	1 in 137,000	98%
		Latino	1 in 4197	98%	1 in 210,000	
		Worldwide	1 in 4022	98%	1 in 201,000	
Mucopolysaccharidosis, Type IVb / GM1 Gangliosidosis (AR) NM_000404.2	*GLB1*	African	1 in 356	76%	1 in 1,500	99%
		East Asian	1 in 305	75%	1 in 1,200	
		Finnish	1 in 246	97%	1 in 7,700	
		Caucasian	1 in 277	83%	1 in 1,700	
		Latino	1 in 431	81%	1 in 2,300	
		South Asian	1 in 285	77%	1 in 1,200	
		Worldwide	1 in 297	83%	1 in 1,800	
		Roma	1 in 50	99%	1 in 4,900	
		South Brazilian	1 in 58	99%	1 in 5,700	

Mucopolysaccharidosis, Type VI (AR) NM_000046.3	*ARSB*	African	1 in 664	58%	1 in 1,600	99%
		East Asian	1 in 1437	99%	1 in 144,000	
		Finnish	1 in 1802	85%	1 in 12,100	
		Caucasian	1 in 314	75%	1 in 1,300	
		Latino	1 in 4195	74%	1 in 16,300	
		South Asian	1 in 2198	85%	1 in 14,500	
		Worldwide	1 in 502	73%	1 in 1,900	
Mucopolysaccharidosis, Type IX (AR) NM_153281.1	*HYAL1*	African	1 in 2536	99%	1 in 254,000	99%
		East Asian	1 in 632	99%	1 in 63,100	
		Caucasian	1 in 1495	99%	1 in 149,000	
		Latino	1 in 2125	99%	1 in 212,000	
		South Asian	1 in 1277	99%	1 in 128,000	
		Worldwide	1 in 1704	99%	1 in 170,000	
Multiple Sulfatase Deficiency (AR) NM_182760.3	*SUMF1*	African	1 in 406	99%	1 in 40,500	99%
		Ashkenazi Jewish	1 in 298	99%	1 in 29,700	
		East Asian	1 in 1437	33%	1 in 2,200	
		Caucasian	1 in 696	73%	1 in 2,500	
		Latino	1 in 1525	99%	1 in 152,000	
		South Asian	1 in 834	94%	1 in 13,100	
		Worldwide	1 in 588	65%	1 in 1,700	
Muscle-Eye-Brain Disease and Other *POMGNT1*-Related Congenital Muscular Dystrophy-Dystroglycanopathies (AR) NM_017739.3	*POMGNT1*	African	1 in 674	47%	1 in 1,300	97%
		East Asian	1 in 581	90%	1 in 6,100	
		Finnish	1 in 216	95%	1 in 4,400	
		Caucasian	1 in 315	93%	1 in 4,200	
		Latino	1 in 544	88%	1 in 4,400	
		South Asian	1 in 727	78%	1 in 3,300	
		Worldwide	1 in 377	89%	1 in 3,500	
Myoneurogastrointestinal Encephalopathy (AR) NM_001113755.2	*TYMP*	African	1 in 287	69%	1 in 920	99%
		Ashkenazi Jewish	1 in 828	99%	1 in 82,700	
		East Asian	1 in 2873	66%	1 in 8,400	
		Finnish	1 in 1053	99%	1 in 105,000	
		Caucasian	1 in 425	79%	1 in 2,100	
		Latino	1 in 647	99%	1 in 64,600	
		South Asian	1 in 1834	64%	1 in 5,000	
		Worldwide	1 in 513	83%	1 in 3,000	
		Sephardic Jewish - Iranian	1 in 158	99%	1 in 15,700	
Myotubular Myopathy 1 (XL) NM_000252.2	*MTM1*	Worldwide	1 in 25,000	87%	1 in 180,000	98%
N-Acetylglutamate Synthase Deficiency (AR) NM_153006.2	*NAGS*	African	1 in 701	84%	1 in 4,300	99%
		Ashkenazi Jewish	1 in 601	99%	1 in 60,000	
		Finnish	1 in 966	99%	1 in 96,500	
		Caucasian	1 in 920	72%	1 in 3,200	
		Latino	1 in 2493	99%	1 in 249,000	
		South Asian	1 in 2850	61%	1 in 7,300	
		Worldwide	1 in 937	84%	1 in 5,700	
Nemaline Myopathy 2 (AR) NM_001271208.1 *Exception:* Exons 82 -105	*NEB*	African	1 in 368	98%	1 in 18,400	98%
		Ashkenazi Jewish	1 in 95	95%	1 in 1,900	
		East Asian	1 in 123	45%	1 in 220	
		Finnish	1 in 118	73%	1 in 430	
		Caucasian	1 in 175	93%	1 in 2,400	
		Latino	1 in 172	86%	1 in 1,200	
		South Asian	1 in 200	84%	1 in 1,300	
		Worldwide	1 in 147	75%	1 in 580	

Nephrogenic Diabetes Insipidus, Type II (AR) NM_000486.5	*AQP2*	African	1 in 864	99%	1 in 86,300	99%
		East Asian	1 in 676	91%	1 in 7,700	
		Finnish	1 in 3853	99%	1 in 385,000	
		Caucasian	1 in 721	79%	1 in 3,400	
		Latino	1 in 458	96%	1 in 12,400	
		South Asian	1 in 3078	59%	1 in 7,600	
		Worldwide	1 in 776	87%	1 in 5,900	
Nephrotic Syndrome (*NPHS1*-Related) / Congenital Finnish Nephrosis (AR) NM_004646.3	*NPHS1*	African	1 in 191	77%	1 in 830	99%
		East Asian	1 in 398	59%	1 in 980	
		Finnish	1 in 41	98%	1 in 1,900	
		Caucasian	1 in 190	79%	1 in 920	
		Latino	1 in 298	68%	1 in 920	
		South Asian	1 in 145	77%	1 in 620	
		Worldwide	1 in 137	84%	1 in 880	
		Groffdale Conference Mennonites	1 in 12	99%	1 in 1,100	
Nephrotic Syndrome (*NPHS2*-Related) / Steroid-Resistant Nephrotic Syndrome (AR) NM_014625.3	*NPHS2*	African	1 in 456	93%	1 in 6,600	99%
		East Asian	1 in 595	65%	1 in 1,700	
		Finnish	1 in 4294	99%	1 in 429,000	
		Caucasian	1 in 226	90%	1 in 2,200	
		Latino	1 in 884	47%	1 in 1,700	
		South Asian	1 in 733	71%	1 in 2,500	
		Worldwide	1 in 356	86%	1 in 2,500	
Neuronal Ceroid-Lipofuscinosis (*CLN3*-Related) (AR) NM_000086.2	*CLN3*	African	1 in 1697	77%	1 in 7,400	99%
		East Asian	1 in 589	99%	1 in 58,800	
		Finnish	1 in 1722	99%	1 in 172,000	
		Caucasian	1 in 242	97%	1 in 9,200	
		Latino	1 in 1538	71%	1 in 5,400	
		South Asian	1 in 2552	99%	1 in 255,000	
		Worldwide	1 in 434	96%	1 in 11,600	
Neuronal Ceroid-Lipofuscinosis (*CLN5*-Related) (AR) NM_006493.2	*CLN5*	African	1 in 1473	99%	1 in 147,000	99%
		East Asian	1 in 748	99%	1 in 74,700	
		Finnish	1 in 542	99%	1 in 54,100	
		Caucasian	1 in 762	82%	1 in 4,300	
		Latino	1 in 794	99%	1 in 79,300	
		South Asian	1 in 4827	68%	1 in 15,000	
		Worldwide	1 in 838	90%	1 in 8,100	
Neuronal Ceroid-Lipofuscinosis (*CLN6*-Related) (AR) NM_017882.2	*CLN6*	African	1 in 1528	79%	1 in 7,300	99%
		East Asian	1 in 909	42%	1 in 1,600	
		Caucasian	1 in 977	81%	1 in 5,100	
		Latino	1 in 698	91%	1 in 7,700	
		South Asian	1 in 733	33%	1 in 1,100	
		Worldwide	1 in 1054	72%	1 in 3,700	
Neuronal Ceroid-Lipofuscinosis (*CLN8*-Related) (AR) NM_018941.3	*CLN8*	African	1 in 1107	56%	1 in 2,500	99%
		East Asian	1 in 1725	40%	1 in 2,900	
		Finnish	1 in 397	92%	1 in 4,900	
		Caucasian	1 in 1250	55%	1 in 2,800	
		Latino	1 in 3358	40%	1 in 5,600	
		South Asian	1 in 1924	74%	1 in 7,500	
		Worldwide	1 in 1125	69%	1 in 3,600	

Neuronal Ceroid-Lipofuscinosis (*MFSD8*-Related) (AR) NM_152778.2	*MFSD8*	African	1 in 1351	82%	1 in 7,300	99%
		East Asian	1 in 869	99%	1 in 86,800	
		Finnish	1 in 681	99%	1 in 68,000	
		Caucasian	1 in 555	90%	1 in 5,600	
		Latino	1 in 1289	76%	1 in 5,400	
		South Asian	1 in 480	12%	1 in 550	
		Worldwide	1 in 606	79%	1 in 2,900	
Neuronal Ceroid-Lipofuscinosis (*PPT1*-Related) (AR) NM_000310.3	*PPT1*	African	1 in 628	67%	1 in 1,900	99%
		East Asian	1 in 918	11%	1 in 1,000	
		Finnish	1 in 74	99%	1 in 7,300	
		Caucasian	1 in 268	88%	1 in 2,200	
		Latino	1 in 1901	33%	1 in 2,800	
		South Asian	1 in 641	12%	1 in 730	
		Worldwide	1 in 281	85%	1 in 1,900	
Neuronal Ceroid-Lipofuscinosis (*TPP1*-Related) (AR) NM_000391.3	*TPP1*	African	1 in 833	60%	1 in 2,100	99%
		Ashkenazi Jewish	1 in 1268	99%	1 in 127,000	
		East Asian	1 in 1480	51%	1 in 3,000	
		Finnish	1 in 354	99%	1 in 35,300	
		Caucasian	1 in 266	96%	1 in 6,300	
		Latino	1 in 568	89%	1 in 5,100	
		South Asian	1 in 2199	99%	1 in 220,000	
		Worldwide	1 in 379	93%	1 in 5,700	
		Newfoundland	1 in 59	99%	1 in 5,800	
Niemann-Pick Disease, Type A/B (AR) NM_000543.4	*SMPD1*	African	1 in 120	90%	1 in 1,100	99%
		Ashkenazi Jewish	1 in 98	99%	1 in 9,700	
		East Asian	1 in 81	94%	1 in 1,300	
		Finnish	1 in 2230	99%	1 in 223,000	
		Caucasian	1 in 350	81%	1 in 1,800	
		Latino	1 in 499	87%	1 in 4,000	
		South Asian	1 in 327	76%	1 in 1,300	
		Worldwide	1 in 240	88%	1 in 1,900	
Niemann-Pick Disease, Type C (*NPC1*-Related) (AR) NM_000271.4	*NPC1*	African	1 in 233	67%	1 in 700	99%
		Ashkenazi Jewish	1 in 262	47%	1 in 500	
		East Asian	1 in 211	80%	1 in 1,100	
		Finnish	1 in 334	73%	1 in 1,200	
		Caucasian	1 in 163	71%	1 in 550	
		Latino	1 in 272	62%	1 in 720	
		South Asian	1 in 334	52%	1 in 690	
		Worldwide	1 in 197	68%	1 in 620	
Niemann-Pick Disease, Type C (*NPC2*-Related) (AR) NM_006432.3	*NPC2*	African	1 in 1214	99%	1 in 121,000	99%
		Finnish	1 in 3734	66%	1 in 10,900	
		Caucasian	1 in 945	86%	1 in 6,600	
		Latino	1 in 3089	99%	1 in 309,000	
		Worldwide	1 in 1293	90%	1 in 12,500	
Nijmegen Breakage Syndrome (AR) NM_002485.4	*NBN*	African	1 in 503	99%	1 in 50,200	99%
		Ashkenazi Jewish	1 in 427	99%	1 in 42,600	
		East Asian	1 in 2137	99%	1 in 214,000	
		Finnish	1 in 384	72%	1 in 1,400	
		Caucasian	1 in 525	96%	1 in 13,800	
		Latino	1 in 1403	99%	1 in 140,000	
		South Asian	1 in 1025	99%	1 in 102,000	
		Worldwide	1 in 531	94%	1 in 9,300	

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Non-Syndromic Hearing Loss (*GJB2*-Related) (AR) NM_004004.5	*GJB2* ‡	African	1 in 56	85%	1 in 360	99%
		Ashkenazi Jewish	1 in 13	94%	1 in 210	
		East Asian	1 in 5	98%	1 in 280	
		Finnish	1 in 16	99%	1 in 1,400	
		Caucasian	1 in 18	97%	1 in 600	
		Latino	1 in 28	96%	1 in 610	
		South Asian	1 in 55	94%	1 in 970	
		Worldwide	1 in 18	97%	1 in 530	
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome (AR) NM_025216.2	*WNT10A*	African	1 in 766	64%	1 in 2,100	99%
		East Asian	1 in 594	34%	1 in 900	
		Finnish	1 in 2037	63%	1 in 5,500	
		Caucasian	1 in 216	89%	1 in 1,900	
		Latino	1 in 869	83%	1 in 5,100	
		South Asian	1 in 952	32%	1 in 1,400	
		Worldwide	1 in 358	80%	1 in 1,800	
Omenn Syndrome (*RAG2*-Related) (AR) NM_000536.2	*RAG2*	African	1 in 953	83%	1 in 5,700	99%
		Ashkenazi Jewish	1 in 821	99%	1 in 82,000	
		Finnish	1 in 810	99%	1 in 80,900	
		Caucasian	1 in 1925	82%	1 in 10,600	
		South Asian	1 in 962	25%	1 in 1,300	
		Worldwide	1 in 1388	77%	1 in 6,000	
		Sephardic Jewish - Iraqi	N/A	88%	N/A	
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type (AR) NM_001033855.1	*DCLRE1C*	African	1 in 511	94%	1 in 8,300	98%
		East Asian	1 in 958	98%	1 in 47,900	
		Finnish	1 in 2881	76%	1 in 12,100	
		Caucasian	1 in 903	84%	1 in 5,500	
		Latino	1 in 1907	87%	1 in 14,500	
		South Asian	1 in 901	69%	1 in 2,900	
		Worldwide	1 in 811	87%	1 in 6,400	
		Navajo and Apache Native American	1 in 48	98%	1 in 2,400	
Ornithine Aminotransferase Deficiency (AR) NM_000274.3	*OAT*	African	1 in 2898	99%	1 in 290,000	99%
		Finnish	1 in 138	98%	1 in 6,200	
		Caucasian	1 in 749	83%	1 in 4,400	
		Latino	1 in 1291	53%	1 in 2,800	
		South Asian	1 in 905	47%	1 in 1,700	
		Worldwide	1 in 595	82%	1 in 3,300	
		Sephardic Jewish - Iraqi and Syrian	1 in 177	99%	1 in 17,600	
Ornithine Transcarbamylase Deficiency (XL) NM_000531.5	*OTC*	Worldwide	1 in 30,000	71%	1 in 100,000	99%
Osteopetrosis 1 (AR) NM_006019.2	*TCIRG1*	African	1 in 418	87%	1 in 3,300	98%
		Ashkenazi Jewish	1 in 491	88%	1 in 4,300	
		East Asian	1 in 323	94%	1 in 5,700	
		Finnish	1 in 1790	98%	1 in 89,500	
		Caucasian	1 in 399	92%	1 in 4,700	
		Latino	1 in 414	98%	1 in 20,600	
		South Asian	1 in 749	88%	1 in 6,500	
		Worldwide	1 in 399	93%	1 in 5,900	
		Costa Rican	1 in 86	98%	1 in 4,300	
		Chuvashiyan	1 in 60	98%	1 in 3,000	

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Pendred Syndrome (AR) NM_000441.1	*SLC26A4*	African	1 in 114	77%	1 in 490	99%
		Ashkenazi Jewish	1 in 50	98%	1 in 2,400	
		East Asian	1 in 31	58%	1 in 72	
		Finnish	1 in 304	97%	1 in 9,100	
		Caucasian	1 in 47	88%	1 in 390	
		Latino	1 in 135	70%	1 in 440	
		South Asian	1 in 60	86%	1 in 430	
		Worldwide	1 in 56	83%	1 in 320	
Phenylalanine Hydroxylase Deficiency (AR) NM_000277.1	*PAH*	African	1 in 143	86%	1 in 1,000	99%
		Ashkenazi Jewish	1 in 17	99%	1 in 1,200	
		East Asian	1 in 68	54%	1 in 150	
		Finnish	1 in 158	76%	1 in 650	
		Caucasian	1 in 37	89%	1 in 340	
		Latino	1 in 70	87%	1 in 550	
		South Asian	1 in 121	81%	1 in 640	
		Worldwide	1 in 50	88%	1 in 400	
		Turkish	1 in 32	63%	1 in 85	
		Irish	1 in 34	91%	1 in 370	
		Sicilian	1 in 26	48%	1 in 49	
		Sephardic Jewish - Iranian, Bukharian, Kavkazi, Tunisian and Moroccan	1 in 18	88%	1 in 140	
3-Phosphoglycerate Dehydrogenase Deficiency (AR) NM_006623.3	*PHGDH*	African	1 in 1639	64%	1 in 4,600	99%
		Ashkenazi Jewish	1 in 298	99%	1 in 29,700	
		East Asian	1 in 1232	99%	1 in 123,000	
		Finnish	1 in 1408	99%	1 in 141,000	
		Caucasian	1 in 631	99%	1 in 63,000	
		Latino	1 in 1311	69%	1 in 4,200	
		South Asian	1 in 1665	78%	1 in 7,400	
		Worldwide	1 in 801	94%	1 in 13,800	
Polycystic Kidney Disease, Autosomal Recessive (AR) NM_138694.3	*PKHD1*	African	1 in 66	80%	1 in 320	99%
		Ashkenazi Jewish	1 in 57	99%	1 in 5,600	
		East Asian	1 in 119	66%	1 in 350	
		Finnish	1 in 36	87%	1 in 270	
		Caucasian	1 in 66	85%	1 in 450	
		Latino	1 in 99	82%	1 in 530	
		South Asian	1 in 154	88%	1 in 1,300	
		Worldwide	1 in 68	85%	1 in 440	
		South African Afrikaner	1 in 52	99%	1 in 5,100	
Polyglandular Autoimmune Syndrome, Type 1 (AR) NM_000383.2	*AIRE*	African	1 in 437	99%	1 in 43,600	99%
		East Asian	1 in 313	92%	1 in 4,100	
		Finnish	1 in 93	96%	1 in 2,100	
		Caucasian	1 in 209	96%	1 in 5,300	
		Latino	1 in 422	82%	1 in 2,300	
		South Asian	1 in 979	67%	1 in 3,000	
		Worldwide	1 in 236	94%	1 in 4,000	
		Sardinian	1 in 60	95%	1 in 1,200	
		Sephardic Jewish - Iranian	1 in 27	99%	1 in 2,600	
Pontocerebellar Hypoplasia, Type 1A (AR) NM_003384.2	*VRK1*	Ashkenazi Jewish	1 in 308	99%	1 in 30,700	99%
		East Asian	1 in 2152	99%	1 in 215,000	
		Caucasian	1 in 2583	90%	1 in 25,400	
		Latino	1 in 843	99%	1 in 84,200	
		South Asian	1 in 7530	99%	1 in 753,000	
		Worldwide	1 in 1859	96%	1 in 46,600	

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Pontocerebellar Hypoplasia, Type 6 (AR) NM_020320.3	*RARS2*	African	1 in 365	99%	1 in 36,400	99%
		East Asian	1 in 496	99%	1 in 49,500	
		Finnish	1 in 306	99%	1 in 30,500	
		Caucasian	1 in 269	84%	1 in 1,700	
		Latino	1 in 175	92%	1 in 2,200	
		South Asian	1 in 375	56%	1 in 840	
		Worldwide	1 in 274	84%	1 in 1,700	
		Sephardic Jewish - Iraqi, Syrian and Tunisian	N/A	99%	N/A	
Primary Carnitine Deficiency (AR) NM_003060.2	*SLC22A5*	African	1 in 98	94%	1 in 1,700	98%
		Ashkenazi Jewish	1 in 1002	98%	1 in 50,000	
		East Asian	1 in 69	89%	1 in 600	
		Finnish	1 in 1042	81%	1 in 5,400	
		Caucasian	1 in 251	83%	1 in 1,500	
		Latino	1 in 268	86%	1 in 1,900	
		South Asian	1 in 51	96%	1 in 1,300	
		Worldwide	1 in 144	91%	1 in 1,500	
		Faroese	1 in 20	98%	1 in 1,000	
Primary Ciliary Dyskinesia (*DNAH5*-Related) (AR) NM_001369.2	*DNAH5*	African	1 in 169	88%	1 in 1,400	99%
		Ashkenazi Jewish	1 in 113	97%	1 in 3,500	
		East Asian	1 in 193	99%	1 in 19,200	
		Finnish	1 in 175	97%	1 in 6,800	
		Caucasian	1 in 145	90%	1 in 1,500	
		Latino	1 in 204	94%	1 in 3,600	
		South Asian	1 in 326	91%	1 in 3,500	
		Worldwide	1 in 157	92%	1 in 2,100	
Primary Ciliary Dyskinesia (*DNAI1*-Related) (AR) NM_012144.3	*DNAI1*	African	1 in 434	95%	1 in 9,500	99%
		Ashkenazi Jewish	1 in 380	99%	1 in 37,900	
		Finnish	1 in 1468	99%	1 in 147,000	
		Caucasian	1 in 323	94%	1 in 5,000	
		Latino	1 in 1140	99%	1 in 114,000	
		South Asian	1 in 1184	99%	1 in 118,000	
		Worldwide	1 in 435	95%	1 in 9,300	
Primary Ciliary Dyskinesia (*DNAI2*-Related) (AR) NM_023036.4	*DNAI2*	African	1 in 414	99%	1 in 41,300	99%
		Ashkenazi Jewish	1 in 81	99%	1 in 8,000	
		East Asian	1 in 1437	99%	1 in 144,000	
		Caucasian	1 in 758	99%	1 in 75,700	
		Latino	1 in 632	99%	1 in 63,100	
		South Asian	1 in 669	99%	1 in 66,800	
		Worldwide	1 in 549	99%	1 in 54,800	
Primary Hyperoxaluria, Type 1 (AR) NM_000030.2	*AGXT*	African	1 in 326	88%	1 in 2,800	99%
		Ashkenazi Jewish	1 in 1215	75%	1 in 4,800	
		East Asian	1 in 134	87%	1 in 1,100	
		Finnish	1 in 581	21%	1 in 740	
		Caucasian	1 in 194	78%	1 in 880	
		Latino	1 in 416	81%	1 in 2,100	
		South Asian	1 in 247	68%	1 in 760	
		Worldwide	1 in 230	77%	1 in 990	
Primary Hyperoxaluria, Type 2 (AR) NM_012203.1	*GRHPR*	African	1 in 605	65%	1 in 1,700	99%
		East Asian	1 in 681	99%	1 in 68,000	
		Finnish	1 in 757	99%	1 in 75,600	
		Caucasian	1 in 433	96%	1 in 10,600	
		Latino	1 in 1881	99%	1 in 188,000	
		South Asian	1 in 327	97%	1 in 10,500	
		Worldwide	1 in 489	92%	1 in 6,500	

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Primary Hyperoxaluria, Type 3 (AR) NM_138413.3	*HOGA1*	African	1 in 401	96%	1 in 9,300	99%
		Ashkenazi Jewish	1 in 37	99%	1 in 3,600	
		East Asian	1 in 122	99%	1 in 12,100	
		Finnish	1 in 513	99%	1 in 51,200	
		Caucasian	1 in 169	93%	1 in 2,400	
		Latino	1 in 296	94%	1 in 4,700	
		South Asian	1 in 727	90%	1 in 7,000	
		Worldwide	1 in 186	95%	1 in 3,800	
Progressive Cerebello-Cerebral Atrophy (AR) NM_016955.3	*SEPSECS*	African	1 in 2156	71%	1 in 7,500	99%
		Ashkenazi Jewish	1 in 1640	99%	1 in 164,000	
		East Asian	1 in 2467	99%	1 in 247,000	
		Finnish	1 in 96	95%	1 in 1,800	
		Caucasian	1 in 656	90%	1 in 6,400	
		Latino	1 in 799	66%	1 in 2,400	
		South Asian	1 in 3848	74%	1 in 14,900	
		Worldwide	1 in 503	90%	1 in 5,000	
		Sephardic Jewish - Moroccan and Iraqi	1 in 41	99%	1 in 4,000	
Progressive Familial Intrahepatic Cholestasis, Type 2 (AR) NM_003742.2	*ABCB11*	African	1 in 295	52%	1 in 610	99%
		East Asian	1 in 153	61%	1 in 390	
		Finnish	1 in 835	52%	1 in 1,700	
		Caucasian	1 in 276	71%	1 in 950	
		Latino	1 in 390	57%	1 in 910	
		South Asian	1 in 654	74%	1 in 2,500	
		Worldwide	1 in 306	65%	1 in 880	
Propionic Acidemia (*PCCA*-Related) (AR) NM_000282.3	*PCCA*	African	1 in 393	71%	1 in 1,400	93%
		Ashkenazi Jewish	1 in 548	83%	1 in 3,200	
		East Asian	1 in 419	84%	1 in 2,600	
		Finnish	1 in 2882	93%	1 in 41,200	
		Caucasian	1 in 636	76%	1 in 2,600	
		Latino	1 in 429	59%	1 in 1,100	
		South Asian	1 in 507	78%	1 in 2,300	
		Worldwide	1 in 492	71%	1 in 1,700	
Propionic Acidemia (*PCCB*-Related) (AR) NM_000532.4	*PCCB*	African	1 in 257	96%	1 in 5,900	99%
		East Asian	1 in 192	79%	1 in 920	
		Finnish	1 in 1080	89%	1 in 10,200	
		Caucasian	1 in 635	95%	1 in 12,200	
		Latino	1 in 688	79%	1 in 3,200	
		South Asian	1 in 1490	77%	1 in 6,500	
		Worldwide	1 in 548	89%	1 in 5,100	
Pycnodysostosis (AR) NM_000396.3	*CTSK*	African	1 in 361	99%	1 in 36,000	99%
		East Asian	1 in 413	85%	1 in 2,700	
		Finnish	1 in 2781	99%	1 in 278,000	
		Caucasian	1 in 1067	79%	1 in 5,100	
		Latino	1 in 542	64%	1 in 1,500	
		South Asian	1 in 350	23%	1 in 450	
		Worldwide	1 in 598	69%	1 in 1,900	
Pyruvate Dehydrogenase E1-Alpha Deficiency (XL) NM_000284.3	*PDHA1*	Worldwide	< 1 in 50,000	64%	1 in 140,000	99%

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Pyruvate Dehydrogenase E1-Beta Deficiency (AR) NM_000925.3	*PDHB*	African	1 in 970	43%	1 in 1,700	99%
		Ashkenazi Jewish	1 in 842	17%	1 in 1,000	
		Finnish	1 in 2775	99%	1 in 277,000	
		Caucasian	1 in 2529	83%	1 in 14,600	
		Latino	1 in 1344	99%	1 in 134,000	
		South Asian	1 in 2063	99%	1 in 206,000	
		Worldwide	1 in 1795	78%	1 in 8,300	
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (AR) NM_000317.2	*PTS*	African	1 in 703	99%	1 in 70,200	99%
		Ashkenazi Jewish	1 in 1559	99%	1 in 156,000	
		East Asian	1 in 156	95%	1 in 2,800	
		Finnish	1 in 363	90%	1 in 3,500	
		Caucasian	1 in 478	74%	1 in 1,800	
		Latino	1 in 533	80%	1 in 2,700	
		South Asian	1 in 343	84%	1 in 2,100	
		Worldwide	1 in 395	81%	1 in 2,100	
Renal Tubular Acidosis and Deafness (AR) NM_001692.3	*ATP6V1B1*	African	1 in 524	92%	1 in 6,700	99%
		East Asian	1 in 719	91%	1 in 7,800	
		Caucasian	1 in 1092	84%	1 in 6,600	
		Latino	1 in 2097	99%	1 in 210,000	
		South Asian	1 in 1282	99%	1 in 128,000	
		Worldwide	1 in 995	85%	1 in 6,500	
		Sephardic Jewish - Syrian	1 in 140	99%	1 in 13,900	
Retinitis Pigmentosa 25 (AR) NM_001142800.1	*EYS*	African	1 in 71	94%	1 in 1,100	97%
		Ashkenazi Jewish	1 in 109	97%	1 in 3,600	
		East Asian	1 in 53	81%	1 in 280	
		Finnish	1 in 39	97%	1 in 1,300	
		Caucasian	1 in 82	92%	1 in 980	
		Latino	1 in 152	96%	1 in 3,600	
		South Asian	1 in 168	58%	1 in 400	
		Worldwide	1 in 77	91%	1 in 810	
		Sephardic Jewish - Moroccan	1 in 42	22%	1 in 50	
Retinitis Pigmentosa 26 (AR) NM_001030311.2	*CERKL*	African	1 in 963	99%	1 in 96,200	99%
		East Asian	1 in 547	86%	1 in 4,000	
		Finnish	1 in 48	99%	1 in 4,700	
		Caucasian	1 in 370	97%	1 in 13,400	
		Latino	1 in 602	95%	1 in 13,200	
		South Asian	1 in 416	64%	1 in 1,200	
		Worldwide	1 in 246	95%	1 in 5,000	
		Sephardic Jewish - Yemenite	1 in 24	99%	1 in 2,300	
Retinitis Pigmentosa 28 (AR) NM_032180.2	*FAM161A*	African	1 in 894	99%	1 in 89,300	99%
		Ashkenazi Jewish	1 in 242	99%	1 in 24,100	
		East Asian	1 in 1450	99%	1 in 145,000	
		Finnish	1 in 656	99%	1 in 65,500	
		Caucasian	1 in 343	99%	1 in 34,200	
		Latino	1 in 442	99%	1 in 44,100	
		South Asian	1 in 795	99%	1 in 79,400	
		Worldwide	1 in 423	99%	1 in 42,200	
		Sephardic Jewish - Libyan, Moroccan, Tunisian and Bulgarian	1 in 41	99%	1 in 4,000	
Retinitis Pigmentosa 59 (AR) NM_001243564.1	*DHDDS*	Ashkenazi Jewish	1 in 100	99%	1 in 9,900	99%
		Caucasian	1 in 6008	99%	1 in 601,000	
		Latino	1 in 4223	99%	1 in 422,000	
		Worldwide	1 in 2009	99%	1 in 201,000	

Rhizomelic Chondrodysplasia Punctata, Type 1 (AR) NM_000288.3	*PEX7*	African	1 in 491	99%	1 in 49,000	99%
		Ashkenazi Jewish	1 in 234	99%	1 in 23,300	
		East Asian	1 in 552	99%	1 in 55,100	
		Caucasian	1 in 371	96%	1 in 10,100	
		Latino	1 in 485	93%	1 in 7,200	
		South Asian	1 in 2285	99%	1 in 228,000	
		Worldwide	1 in 480	97%	1 in 14,400	
Rhizomelic Chondrodysplasia Punctata, Type 3 (AR) NM_003659.3	*AGPS*	Caucasian	1 in 18591	97%	1 in 620,000	97%
		Worldwide	1 in 30731	97%	1 in 1,024,000	
Roberts Syndrome (AR) NM_001017420.2	*ESCO2*	African	1 in 671	99%	1 in 67,000	99%
		Ashkenazi Jewish	1 in 626	99%	1 in 62,500	
		East Asian	1 in 950	99%	1 in 94,900	
		Finnish	1 in 1087	99%	1 in 109,000	
		Caucasian	1 in 1395	99%	1 in 139,000	
		Latino	1 in 3312	99%	1 in 331,000	
		South Asian	1 in 1378	99%	1 in 138,000	
		Worldwide	1 in 1119	99%	1 in 112,000	
Salla Disease (AR) NM_012434.4	*SLC17A5*	African	1 in 853	99%	1 in 85,200	99%
		East Asian	1 in 1723	99%	1 in 172,000	
		Finnish	1 in 85	99%	1 in 8,400	
		Caucasian	1 in 328	96%	1 in 8,400	
		Latino	1 in 777	85%	1 in 5,300	
		South Asian	1 in 3847	74%	1 in 14,900	
		Worldwide	1 in 330	97%	1 in 9,700	
		Swedish	1 in 125	99%	1 in 12,400	
		Canadian Inuit	1 in 129	99%	1 in 12,800	
Sandhoff Disease (AR) NM_000521.3	*HEXB*	African	1 in 895	72%	1 in 3,200	98%
		East Asian	1 in 385	98%	1 in 19,200	
		Finnish	1 in 2913	98%	1 in 146,000	
		Caucasian	1 in 202	95%	1 in 4,100	
		Latino	1 in 248	94%	1 in 3,900	
		South Asian	1 in 513	75%	1 in 2,100	
		Worldwide	1 in 286	91%	1 in 3,200	
		Northern Saskachetwan Metis	1 in 15	75%	1 in 57	
		Argentinian Creole	1 in 26	98%	1 in 1,300	
Schimke Immunoosseous Dysplasia (AR) NM_014140.3	*SMARCAL1*	African	1 in 699	90%	1 in 7,000	99%
		Ashkenazi Jewish	1 in 174	99%	1 in 17,300	
		East Asian	1 in 561	99%	1 in 56,000	
		Finnish	1 in 717	99%	1 in 71,600	
		Caucasian	1 in 451	88%	1 in 3,800	
		Latino	1 in 2123	99%	1 in 212,000	
		South Asian	1 in 2565	99%	1 in 256,000	
		Worldwide	1 in 547	92%	1 in 6,900	
Segawa Syndrome (AR) NM_000360.3	*TH*	African	1 in 809	67%	1 in 2,500	99%
		East Asian	1 in 306	90%	1 in 3,000	
		Caucasian	1 in 856	86%	1 in 6,100	
		Latino	1 in 1121	99%	1 in 112,000	
		South Asian	1 in 2145	99%	1 in 214,000	
		Worldwide	1 in 848	87%	1 in 6,700	

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Sjogren-Larsson Syndrome (AR)	*ALDH3A2*	African	1 in 825	65%	1 in 2,400	99%
NM_000382.2		East Asian	1 in 816	80%	1 in 4,100	
		Finnish	1 in 2578	40%	1 in 4,300	
		Caucasian	1 in 718	83%	1 in 4,300	
		Latino	1 in 672	95%	1 in 13,800	
		South Asian	1 in 1152	92%	1 in 13,700	
		Worldwide	1 in 849	83%	1 in 4,900	
		Swedish	1 in 205	99%	1 in 20,400	
Smith-Lemli-Opitz Syndrome (AR)	*DHCR7*	African	1 in 51	98%	1 in 2,400	99%
NM_001360.2		Ashkenazi Jewish	1 in 39	97%	1 in 1,100	
		East Asian	1 in 357	91%	1 in 3,800	
		Finnish	1 in 141	94%	1 in 2,500	
		Caucasian	1 in 46	94%	1 in 750	
		Latino	1 in 118	93%	1 in 1,800	
		South Asian	1 in 334	71%	1 in 1,200	
		Worldwide	1 in 57	94%	1 in 970	

Spinal Muscular Atrophy (AR) *SMN1/SMN2*

NM_000344.3 / NM_017411.3

	Carrier Frequency	Detection Rate	Residual Risk After Negative Result (2 Copies)*	Detection Rate with *SMN1* c.*3+80T>G (2 Copies)	Residual Risk c.*3+80T>G Negative (2 Copies)	Residual Risk c.*3+80T>G Positive (2 Copies)	Residual Risk with ≥3 Copies of *SMN1*
African American	1 in 85	71%	1 in 160	91%	1 in 455	1 in 49	1 in 4,300
Ashkenazi Jewish	1 in 76	90%	1 in 672	93%	1 in 978	1 in 10	1 in 4,800
East Asian	1 in 53	94%	1 in 864	95%	1 in 901	1 in 12	1 in 4,900
Caucasian	1 in 48	95%	1 in 803	95%	1 in 894	1 in 23	1 in 4,900
Latino	1 in 63	91%	1 in 609	94%	1 in 930	1 in 47	1 in 4,800
South Asian	1 in 103	87%	1 in 637	87%	1 in 637	1 in 608	1 in 4,700
Sephardic Jewish	1 in 34	96%	1 in 696	97%	1 in 884	1 in 12	1 in 4,900

*Residual risk with two copies *SMN1* detected using dosage sensitive methods. The presence of three or more copies of *SMN1* reduces the risk of being an *SMN1* carrier between 5-10 fold, depending on ethnicity.

Spondylothoracic Dysostosis (AR)	*MESP2*	East Asian	1 in 534	99%	1 in 53,300	99%
NM_001039958.1		Caucasian	1 in 3820	99%	1 in 382,000	
		Latino	1 in 2327	99%	1 in 233,000	
		South Asian	1 in 3057	99%	1 in 306,000	
		Worldwide	1 in 2247	99%	1 in 225,000	
		Puerto Rican	1 in 55	99%	1 in 5,400	
Steel Syndrome (AR)	*COL27A1*	Puerto Rican	1 in 40	99%	1 in 3,900	99%
NM_032888.2						
Variant tested: p.G697R (Genotyping only)						
Stuve-Wiedemann Syndrome (AR)	*LIFR*	African	1 in 1444	99%	1 in 144,000	99%
NM_002310.5		Ashkenazi Jewish	1 in 630	99%	1 in 62,900	
		East Asian	1 in 1719	99%	1 in 172,000	
		Caucasian	1 in 848	97%	1 in 29,800	
		Latino	1 in 1670	88%	1 in 14,100	
		South Asian	1 in 512	99%	1 in 51,100	
		Worldwide	1 in 909	97%	1 in 26,500	

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Sulfate Transporter-Related Osteochondrodysplasia (AR) NM_000112.3	*SLC26A2*	African	1 in 341	99%	1 in 34,000	99%
		Ashkenazi Jewish	1 in 220	99%	1 in 21,900	
		East Asian	1 in 510	83%	1 in 3,000	
		Finnish	1 in 69	99%	1 in 6,800	
		Caucasian	1 in 129	93%	1 in 1,800	
		Latino	1 in 248	98%	1 in 10,000	
		South Asian	1 in 853	99%	1 in 85,200	
		Worldwide	1 in 147	95%	1 in 3,000	
Tay-Sachs Disease (AR) NM_000520.4	*HEXA*	African	1 in 216	99%*	1 in 21,500	99%
		Ashkenazi Jewish	1 in 30	99%*	1 in 2,900	
		East Asian	1 in 210	99%*	1 in 20,900	
		Finnish	1 in 399	99%*	1 in 39,800	
		Caucasian	1 in 90	97%*	1 in 3,400	
		Latino	1 in 243	89%*	1 in 2,200	
		South Asian	1 in 416	70%*	1 in 1,400	
		Worldwide	1 in 121	96%*	1 in 3,200	
		French Canadian - Gaspesie	1 in 13	99%*	1 in 1,200	
		French Canadian - Other	1 in 73	99%*	1 in 7,200	
		Irish	1 in 41	90%*	1 in 400	
		Sephardic Jewish – Moroccan and Iraqi	1 in 125	99%*	1 in 12,400	
Tyrosinemia, Type I (AR) NM_000137.2	*FAH*	African	1 in 359	73%	1 in 1,300	99%
		Ashkenazi Jewish	1 in 134	99%	1 in 13,300	
		Finnish	1 in 323	96%	1 in 8,300	
		Caucasian	1 in 259	83%	1 in 1,600	
		Latino	1 in 682	91%	1 in 7,600	
		South Asian	1 in 592	95%	1 in 12,300	
		Worldwide	1 in 321	84%	1 in 2,000	
		French Canadian - Saguenay	1 in 25	99%	1 in 2,400	
		Lac-St. Jean French Canadian - Other	1 in 66	99%	1 in 6,500	
Usher Syndrome, Type IB (AR) NM_000260.3	*MYO7A*	African	1 in 174	79%	1 in 820	99%
		Ashkenazi Jewish	1 in 345	69%	1 in 1,100	
		East Asian	1 in 119	31%	1 in 170	
		Finnish	1 in 285	80%	1 in 1,400	
		Caucasian	1 in 129	84%	1 in 780	
		Latino	1 in 300	79%	1 in 1,400	
		South Asian	1 in 61	93%	1 in 810	
		Worldwide	1 in 119	82%	1 in 650	
Usher Syndrome, Type IC (AR) NM_005709.3	*USH1C*	African	1 in 48	96%	1 in 1,200	97%
		Ashkenazi Jewish	1 in 298	97%	1 in 9,900	
		East Asian	1 in 154	61%	1 in 400	
		Finnish	1 in 1079	97%	1 in 35,900	
		Caucasian	1 in 257	84%	1 in 1,600	
		Latino	1 in 526	91%	1 in 5,900	
		South Asian	1 in 485	48%	1 in 930	
		Worldwide	1 in 204	86%	1 in 1,500	
		French Canadian/Acadian	1 in 227	97%	1 in 7,500	
Usher Syndrome, Type ID (AR) NM_022124.5	*CDH23*	African	1 in 118	78%	1 in 530	99%
		Ashkenazi Jewish	1 in 972	99%	1 in 97,100	
		East Asian	1 in 116	87%	1 in 880	
		Finnish	1 in 395	80%	1 in 2,000	
		Caucasian	1 in 216	85%	1 in 1,400	
		Latino	1 in 222	68%	1 in 690	
		South Asian	1 in 166	74%	1 in 640	
		Worldwide	1 in 186	81%	1 in 960	

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Usher Syndrome, Type IF (AR) NM_001142764.1	*PCDH15*	African	1 in 548	98%	1 in 27,400	98%
		Ashkenazi Jewish	1 in 118	98%	1 in 5,800	
		East Asian	1 in 191	83%	1 in 1,100	
		Finnish	1 in 2286	98%	1 in 114,000	
		Caucasian	1 in 497	87%	1 in 3,800	
		Latino	1 in 545	73%	1 in 2,000	
		South Asian	1 in 805	83%	1 in 4,600	
		Worldwide	1 in 447	87%	1 in 3,500	
Usher Syndrome, Type IIA (AR) NM_206933.2	*USH2A*	African	1 in 69	75%	1 in 280	98%
		Ashkenazi Jewish	1 in 40	95%	1 in 750	
		East Asian	1 in 27	50%	1 in 52	
		Finnish	1 in 142	97%	1 in 4,300	
		Caucasian	1 in 46	80%	1 in 230	
		Latino	1 in 51	84%	1 in 320	
		South Asian	1 in 68	64%	1 in 190	
		Worldwide	1 in 49	77%	1 in 210	
		Sephardic Jewish – Iraqi and Iranian	1 in 36	71%	1 in 120	
Usher Syndrome, Type III (AR) NM_174878.2	*CLRN1*	African	1 in 632	99%	1 in 63,100	99%
		Ashkenazi Jewish	1 in 93	99%	1 in 9,200	
		East Asian	1 in 1263	56%	1 in 2,800	
		Finnish	1 in 69	99%	1 in 6,800	
		Caucasian	1 in 420	67%	1 in 1,300	
		Latino	1 in 1889	99%	1 in 189,000	
		Worldwide	1 in 308	87%	1 in 2,400	
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (AR) NM_000018.3	*ACADVL*	African	1 in 146	76%	1 in 600	98%
		Ashkenazi Jewish	1 in 1259	73%	1 in 4,700	
		East Asian	1 in 201	47%	1 in 380	
		Finnish	1 in 291	94%	1 in 4,500	
		Caucasian	1 in 110	88%	1 in 920	
		Latino	1 in 267	67%	1 in 810	
		South Asian	1 in 372	72%	1 in 1,300	
		Worldwide	1 in 156	83%	1 in 920	
Walker-Warburg Syndrome and Other *FKTN*-Related Dystrophies (AR NM_001079802.1	*FKTN*	African	1 in 736	95%	1 in 14,700	95%
		Ashkenazi Jewish	1 in 62	95%	1 in 1,200	
		East Asian	1 in 288	25%	1 in 390	25%
		Caucasian	1 in 1023	76%	1 in 4,200	
		Latino	1 in 382	95%	1 in 7,600	
		South Asian	1 in 854	90%	1 in 8,300	
		Worldwide	1 in 541	87%	1 in 4,100	
		Japanese	1 in 188	4%	1 in 200	4%
Wilson Disease (AR) NM_000053.3	*ATP7B*	African	1 in 146	73%	1 in 540	99%
		Ashkenazi Jewish	1 in 39	97%	1 in 1,500	
		East Asian	1 in 32	78%	1 in 150	
		Finnish	1 in 114	90%	1 in 1,100	
		Caucasian	1 in 63	82%	1 in 350	
		Latino	1 in 63	74%	1 in 240	
		South Asian	1 in 78	60%	1 in 200	
		Worldwide	1 in 65	81%	1 in 330	
		Canary Islands	1 in 25	88%	1 in 200	
		Sardinian	1 in 42	99%	1 in 4,100	
		Sephardic Jewish - North African, Iraqi, Yemenite, Iranian and Bukharian	1 in 65	99%	1 in 6,100	

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Wolman Disease / Cholesteryl Ester Storage Disease (AR) NM_000235.3	*LIPA*	African	1 in 565	84%	1 in 3,600	98%
		Ashkenazi Jewish	1 in 634	98%	1 in 31,700	
		East Asian	1 in 635	98%	1 in 31,700	
		Finnish	1 in 1250	78%	1 in 5,600	
		Caucasian	1 in 233	93%	1 in 3,200	
		Latino	1 in 329	85%	1 in 2,100	
		South Asian	1 in 769	98%	1 in 38,400	
		Worldwide	1 in 328	92%	1 in 3,900	
		Sephardic Jewish - Iranian	1 in 26	98%	1 in 1,300	
X-Linked Juvenile Retinoschisis (XL) NM_000330.3	*RS1*	Worldwide	1 in 10,000	75%	1 in 40,000	96%
X-Linked Severe Combined Immunodeficiency (XL) NM_000206.2	*IL2RG*	Worldwide	1 in 25,000	90%	1 in 230,000	99%
Zellweger Syndrome Spectrum (*PEX1*-Related) (AR) NM_000466.2	*PEX1*	African	1 in 366	98%	1 in 18,200	98%
		Ashkenazi Jewish	1 in 1188	98%	1 in 59,300	
		East Asian	1 in 153	79%	1 in 740	
		Finnish	1 in 1862	61%	1 in 4,800	
		Caucasian	1 in 191	91%	1 in 2,000	
		Latino	1 in 601	98%	1 in 30,000	
		South Asian	1 in 556	94%	1 in 10,000	
		Worldwide	1 in 269	91%	1 in 2,900	
Zellweger Syndrome Spectrum (*PEX2*-Related) (AR) NM_000318.2	*PEX2*	African	1 in 1741	98%	1 in 87,000	98%
		Ashkenazi Jewish	1 in 195	98%	1 in 9,700	
		East Asian	1 in 2156	98%	1 in 108,000	
		Caucasian	1 in 1542	98%	1 in 77,000	
		Latino	1 in 2798	33%	1 in 4,200	
		South Asian	1 in 1922	98%	1 in 96,000	
		Worldwide	1 in 1191	93%	1 in 17,700	
Zellweger Syndrome Spectrum (*PEX6*-Related) (AR) NM_000287.3	*PEX6*	African	1 in 268	97%	1 in 8,900	97%
		Ashkenazi Jewish	1 in 263	71%	1 in 910	
		East Asian	1 in 595	59%	1 in 1,500	
		Finnish	1 in 205	97%	1 in 6,800	
		Caucasian	1 in 83	95%	1 in 1,600	
		Latino	1 in 239	85%	1 in 1,600	
		South Asian	1 in 105	95%	1 in 2,100	
		Worldwide	1 in 118	93%	1 in 1,800	
		French Canadian	1 in 55	97%	1 in 1,800	
		Sephardic Jewish - Yemenite	1 in 18	97%	1 in 570	
Zellweger Syndrome Spectrum (*PEX10*-Related) (AR) NM_153818.1	*PEX10*	African	1 in 1604	63%	1 in 4,300	99%
		East Asian	1 in 2180	99%	1 in 218,000	
		Caucasian	1 in 1287	80%	1 in 6,300	
		Latino	1 in 2113	99%	1 in 211,000	
		Worldwide	1 in 1739	75%	1 in 7,100	

*Carrier detection by *HEXA* enzyme analysis has a detection rate of approximately 98%.
‡Carrier frequencies include milder and reduced penetrance forms of the disease. Therefore, carrier frequencies may appear higher than reported in the literature.
‡Please note that *GJB2* testing includes testing for the two upstream deletions, del(*GJB6*-D13S1830) and del(*GJB6*-D13S1854) (PMID:11807148 and 15994881).
AR: Autosomal recessive; N/A: Not available; XL: X-linked