

### Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
<b>KCNJ1</b>	Bartter syndrome	African/African American	99	1 in 2,602	1 in 260,000	1 in 1,000,000
		Latino/Admixed American	99	1 in 405	1 in 40,000	1 in 160,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 1,301	1 in 130,000	1 in 520,000
		Finnish	99	1 in 1,389	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	98	1 in 1,167	1 in 58,000	1 in 230,000
		South Asian	99	1 in 3,071	1 in 310,000	1 in 1,200,000
		Other (population not assigned)	99	1 in 239	1 in 24,000	1 in 96,000
<b>KCTD7</b>	Neuronal ceroid-lipofuscinosis	African/African American	99	1 in 1,971	1 in 200,000	1 in 800,000
		Latino/Admixed American	99	1 in 1,022	1 in 100,000	1 in 400,000
		Ashkenazi Jewish	98	1 in 2,520	1 in 130,000	1 in 520,000
		East Asian	99	1 in 2,659	1 in 270,000	1 in 1,100,000
		Finnish	99	1 in 5,373	1 in 540,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 1,083	1 in 110,000	1 in 440,000
		South Asian	99	1 in 1,116	1 in 110,000	1 in 440,000
		Other (population not assigned)	98	1 in 301	1 in 15,000	1 in 60,000
<b>KIF14</b>	Joubert syndrome and related disorders, including Meckel-Gruber syndrome	African/African American	98	1 in 666	1 in 33,000	1 in 130,000
		Latino/Admixed American	98	1 in 1,218	1 in 61,000	1 in 240,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 599	1 in 20,000	1 in 80,000
		Finnish	97	1 in 2,690	1 in 90,000	1 in 360,000
		Non-Finnish European/Caucasian	98	1 in 1,010	1 in 50,000	1 in 200,000
		South Asian	97	1 in 362	1 in 12,000	1 in 48,000
		Other (population not assigned)	98	1 in 767	1 in 38,000	1 in 150,000
<b>L1CAM</b>	L1 syndrome	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	98	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	98	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
<b>LAMA2</b>	Muscular dystrophy, LAMA2-related	African/African American	99	1 in 114	1 in 11,000	1 in 44,000
		Latino/Admixed American	98	1 in 159	1 in 7,900	1 in 32,000
		Ashkenazi Jewish	99	1 in 312	1 in 31,000	1 in 120,000
		East Asian	99	1 in 122	1 in 12,000	1 in 48,000
		Finnish	98	1 in 915	1 in 46,000	1 in 180,000
		Non-Finnish European/Caucasian	98	1 in 273	1 in 14,000	1 in 56,000
		South Asian	98	1 in 306	1 in 15,000	1 in 60,000
		Other (population not assigned)	99	1 in 299	1 in 30,000	1 in 120,000
<b>LAMA3</b>	Junctional epidermolysis bullosa	African/African American	99	1 in 348	1 in 35,000	1 in 140,000
		Latino/Admixed American	99	1 in 288	1 in 29,000	1 in 120,000
		Ashkenazi Jewish	99	1 in 5,030	1 in 500,000	1 in 2,000,000

Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		East Asian	99	1 in 70	1 in 6,900	1 in 28,000
		Finnish	97	1 in 1,502	1 in 50,000	1 in 200,000
		Non-Finnish European/Caucasian	99	1 in 320	1 in 32,000	1 in 130,000
		South Asian	98	1 in 201	1 in 10,000	1 in 40,000
		Other (population not assigned)	99	1 in 760	1 in 76,000	1 in 300,000
<b>LAMB3</b>	Junctional epidermolysis bullosa	African/African American	99	1 in 172	1 in 17,000	1 in 68,000
		Latino/Admixed American	98	1 in 1,012	1 in 51,000	1 in 200,000
		Ashkenazi Jewish	99	1 in 1,001	1 in 100,000	1 in 400,000
		East Asian	98	1 in 1,007	1 in 50,000	1 in 200,000
		Finnish	98	1 in 1,007	1 in 50,000	1 in 200,000
		Non-Finnish European/Caucasian	99	1 in 252	1 in 25,000	1 in 100,000
		South Asian	98	1 in 603	1 in 30,000	1 in 120,000
		Other (population not assigned)	99	1 in 725	1 in 72,000	1 in 290,000
		<b>LAMC2</b>	Junctional epidermolysis bullosa	African/African American	99	1 in 2,653
Latino/Admixed American	99			1 in 3,438	1 in 340,000	1 in 1,400,000
Ashkenazi Jewish	98			1 in 2,489	1 in 120,000	1 in 480,000
East Asian	99			1 in 1,022	1 in 100,000	1 in 400,000
Finnish	99			1 in 10,825	1 in 1,100,000	1 in 4,400,000
Non-Finnish European/Caucasian	98			1 in 1,067	1 in 53,000	1 in 210,000
South Asian	98			1 in 1,093	1 in 55,000	1 in 220,000
Other (population not assigned)	98			1 in 1,535	1 in 77,000	1 in 310,000
<b>LAMP2</b>	Danon disease			African/African American	97	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
		<b>LARS</b>	Acute infantile liver failure	African/African American	99	1 in 728
Latino/Admixed American	99			1 in 474	1 in 47,000	1 in 190,000
Ashkenazi Jewish	99			1 in 2,516	1 in 250,000	1 in 1,000,000
East Asian	98			1 in 1,007	1 in 50,000	1 in 200,000
Finnish	99			1 in 1,820	1 in 180,000	1 in 720,000
Non-Finnish European/Caucasian	99			1 in 1,000	1 in 100,000	1 in 400,000
South Asian	99			1 in 948	1 in 95,000	1 in 380,000
Other (population not assigned)	99			1 in 479	1 in 48,000	1 in 190,000
<b>LCA5</b>	Leber congenital amaurosis	African/African American	97	1 in 8,124	1 in 270,000	1 in 1,100,000
		Latino/Admixed American	98	1 in 1,862	1 in 93,000	1 in 370,000
		Ashkenazi Jewish	99	1 in 235	1 in 23,000	1 in 92,000
		East Asian	98	1 in 1,310	1 in 65,000	1 in 260,000
		Finnish	97	1 in 10,822	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	98	1 in 1,955	1 in 98,000	1 in 390,000

Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
LCK	Severe combined immunodeficiency (SCID)	South Asian	98	1 in 1,899	1 in 95,000	1 in 380,000
		Other (population not assigned)	99	1 in 721	1 in 72,000	1 in 290,000
		African/African American	98	1 in 2,186	1 in 110,000	1 in 440,000
		Latino/Admixed American	97	1 in 248	1 in 8,200	1 in 33,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	1 in 10,069	1 in 340,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 3,606	1 in 360,000	1 in 1,400,000
		South Asian	97	N/A	N/A	N/A
LIG4	Severe combined immunodeficiency (SCID)	Other (population not assigned)	98	1 in 982	1 in 49,000	1 in 200,000
		African/African American	97	1 in 310	1 in 10,000	1 in 40,000
		Latino/Admixed American	97	1 in 807	1 in 27,000	1 in 110,000
		Ashkenazi Jewish	99	1 in 5,029	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 812	1 in 81,000	1 in 320,000
		Finnish	98	1 in 5,507	1 in 280,000	1 in 1,100,000
		Non-Finnish European/Caucasian	98	1 in 580	1 in 29,000	1 in 120,000
		South Asian	98	1 in 766	1 in 38,000	1 in 150,000
		Other (population not assigned)	97	1 in 482	1 in 16,000	1 in 64,000
LIPA	Lysosomal acid lipase deficiency	African/African American	99	1 in 319	1 in 32,000	1 in 130,000
		Latino/Admixed American	99	1 in 104	1 in 10,000	1 in 40,000
		Ashkenazi Jewish	99	1 in 451	1 in 45,000	1 in 180,000
		East Asian	99	1 in 279	1 in 28,000	1 in 110,000
		Finnish	99	1 in 781	1 in 78,000	1 in 310,000
		Non-Finnish European/Caucasian	99	1 in 127	1 in 13,000	1 in 52,000
		South Asian	99	1 in 357	1 in 36,000	1 in 140,000
		Other (population not assigned)	99	1 in 180	1 in 18,000	1 in 72,000
		LIPN	Autosomal recessive congenital ichthyosis (ARCI)	African/African American	99	1 in 115
Latino/Admixed American	99			1 in 602	1 in 60,000	1 in 240,000
Ashkenazi Jewish	97			1 in 143	1 in 4,700	1 in 19,000
East Asian	99			1 in 2,172	1 in 220,000	1 in 880,000
Finnish	99			1 in 459	1 in 46,000	1 in 180,000
Non-Finnish European/Caucasian	99			1 in 335	1 in 33,000	1 in 130,000
South Asian	97			1 in 2,776	1 in 93,000	1 in 370,000
Other (population not assigned)	99			1 in 224	1 in 22,000	1 in 88,000
LMBRD1	Methylmalonic acidemia with homocystinuria			African/African American	99	1 in 363
		Latino/Admixed American	98	1 in 1,035	1 in 52,000	1 in 210,000
		Ashkenazi Jewish	97	1 in 5,035	1 in 170,000	1 in 680,000
		East Asian	98	1 in 1,835	1 in 92,000	1 in 370,000
		Finnish	97	1 in 789	1 in 26,000	1 in 100,000
		Non-Finnish European/Caucasian	97	1 in 414	1 in 14,000	1 in 56,000
		South Asian	99	1 in 2,536	1 in 250,000	1 in 1,000,000
		Other (population not assigned)	97	1 in 715	1 in 24,000	1 in 96,000
		LOXHD1	Deafness and hearing loss, nonsyndromic	African/African American	98	1 in 276

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		Latino/Admixed American	98	1 in 224	1 in 11,000	1 in 44,000
		Ashkenazi Jewish	99	1 in 122	1 in 12,000	1 in 48,000
		East Asian	99	1 in 252	1 in 25,000	1 in 100,000
		Finnish	99	1 in 323	1 in 32,000	1 in 130,000
		Non-Finnish European/Caucasian	99	1 in 169	1 in 17,000	1 in 68,000
		South Asian	99	1 in 209	1 in 21,000	1 in 84,000
		Other (population not assigned)	99	1 in 359	1 in 36,000	1 in 140,000
<b>LPL</b>	Lipoprotein lipase deficiency, familial	African/African American	99	1 in 145	1 in 14,000	1 in 56,000
		Latino/Admixed American	99	1 in 68	1 in 6,700	1 in 27,000
		Ashkenazi Jewish	99	1 in 47	1 in 4,600	1 in 18,000
		East Asian	99	1 in 1,452	1 in 150,000	1 in 600,000
		Finnish	99	1 in 25	1 in 2,400	1 in 9,600
		Non-Finnish European/Caucasian	99	1 in 25	1 in 2,400	1 in 9,600
		South Asian	99	1 in 214	1 in 21,000	1 in 84,000
		Other (population not assigned)	99	1 in 33	1 in 3,200	1 in 13,000
		<b>LRAT</b>	Leber congenital amaurosis	African/African American	99	1 in 8,129
Latino/Admixed American	97			1 in 3,459	1 in 120,000	1 in 480,000
Ashkenazi Jewish	97			N/A	N/A	N/A
East Asian	97			N/A	N/A	N/A
Finnish	97			N/A	N/A	N/A
Non-Finnish European/Caucasian	98			1 in 7,035	1 in 350,000	1 in 1,400,000
South Asian	97			1 in 15,307	1 in 510,000	1 in 2,000,000
Other (population not assigned)	97			N/A	N/A	N/A
<b>LRP2</b>	Donnai-Barrow syndrome	African/African American	99	1 in 334	1 in 33,000	1 in 130,000
		Latino/Admixed American	99	1 in 403	1 in 40,000	1 in 160,000
		Ashkenazi Jewish	99	1 in 171	1 in 17,000	1 in 68,000
		East Asian	99	1 in 825	1 in 82,000	1 in 330,000
		Finnish	99	1 in 408	1 in 41,000	1 in 160,000
		Non-Finnish European/Caucasian	99	1 in 260	1 in 26,000	1 in 100,000
		South Asian	99	1 in 405	1 in 40,000	1 in 160,000
		Other (population not assigned)	99	1 in 254	1 in 25,000	1 in 100,000
		<b>LRPPRC</b>	Leigh syndrome, autosomal recessive	African/African American	98	1 in 650
Latino/Admixed American	99			1 in 591	1 in 59,000	1 in 240,000
Ashkenazi Jewish	97			N/A	N/A	N/A
East Asian	97			1 in 225	1 in 7,500	1 in 30,000
Finnish	97			1 in 428	1 in 14,000	1 in 56,000
Non-Finnish European/Caucasian	99			1 in 789	1 in 79,000	1 in 320,000
South Asian	98			1 in 754	1 in 38,000	1 in 150,000
Other (population not assigned)	98			1 in 239	1 in 12,000	1 in 48,000
<b>LTBP4</b>	Cutis laxa			African/African American	98	1 in 2,394
		Latino/Admixed American	98	1 in 1,803	1 in 90,000	1 in 360,000
		Ashkenazi Jewish	99	1 in 4,859	1 in 490,000	1 in 2,000,000
		East Asian	99	1 in 229	1 in 23,000	1 in 92,000

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		Finnish	98	1 in 2,490	1 in 120,000	1 in 480,000
		Non-Finnish European/Caucasian	99	1 in 64	1 in 6,300	1 in 25,000
		South Asian	99	1 in 907	1 in 91,000	1 in 360,000
		Other (population not assigned)	97	1 in 1,432	1 in 48,000	1 in 190,000
<b>MAK</b>	Retinitis pigmentosa	African/African American	98	1 in 793	1 in 40,000	1 in 160,000
		Latino/Admixed American	99	1 in 786	1 in 79,000	1 in 320,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 511	1 in 51,000	1 in 200,000
		Finnish	99	1 in 5,409	1 in 540,000	1 in 2,200,000
		Non-Finnish European/Caucasian	98	1 in 1,010	1 in 50,000	1 in 200,000
		South Asian	98	1 in 812	1 in 41,000	1 in 160,000
		Other (population not assigned)	97	1 in 1,532	1 in 51,000	1 in 200,000
		<b>MALT1</b>	Severe combined immunodeficiency (SCID)	African/African American	97	1 in 2,016
Latino/Admixed American	99			1 in 891	1 in 89,000	1 in 360,000
Ashkenazi Jewish	99			1 in 758	1 in 76,000	1 in 300,000
East Asian	99			1 in 977	1 in 98,000	1 in 390,000
Finnish	99			1 in 709	1 in 71,000	1 in 280,000
Non-Finnish European/Caucasian	99			1 in 680	1 in 68,000	1 in 270,000
South Asian	97			1 in 579	1 in 19,000	1 in 76,000
Other (population not assigned)	99			1 in 2,736	1 in 270,000	1 in 1,100,000
<b>MAN2B1</b>	Alpha-mannosidosis			African/African American	99	1 in 333
		Latino/Admixed American	99	1 in 138	1 in 14,000	1 in 56,000
		Ashkenazi Jewish	99	1 in 5,037	1 in 500,000	1 in 2,000,000
		East Asian	98	1 in 900	1 in 45,000	1 in 180,000
		Finnish	99	1 in 206	1 in 21,000	1 in 84,000
		Non-Finnish European/Caucasian	99	1 in 475	1 in 47,000	1 in 190,000
		South Asian	99	1 in 831	1 in 83,000	1 in 330,000
		Other (population not assigned)	99	1 in 371	1 in 37,000	1 in 150,000
		<b>MANBA</b>	Beta-mannosidosis	African/African American	98	1 in 345
Latino/Admixed American	99			1 in 1,057	1 in 110,000	1 in 440,000
Ashkenazi Jewish	98			N/A	N/A	N/A
East Asian	99			1 in 1,513	1 in 150,000	1 in 600,000
Finnish	99			1 in 2,705	1 in 270,000	1 in 1,100,000
Non-Finnish European/Caucasian	99			1 in 684	1 in 68,000	1 in 270,000
South Asian	98			1 in 1,028	1 in 51,000	1 in 200,000
Other (population not assigned)	98			1 in 511	1 in 26,000	1 in 100,000
<b>MCCC1</b>	3-Methylcrotonyl-CoA carboxylase deficiency			African/African American	99	1 in 370
		Latino/Admixed American	99	1 in 904	1 in 90,000	1 in 360,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 1,157	1 in 120,000	1 in 480,000
		Finnish	99	1 in 3,682	1 in 370,000	1 in 1,500,000
		Non-Finnish European/Caucasian	98	1 in 528	1 in 26,000	1 in 100,000
		South Asian	99	1 in 417	1 in 42,000	1 in 170,000

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MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency	Other (population not assigned)	99	1 in 536	1 in 54,000	1 in 220,000
		African/African American	99	1 in 374	1 in 37,000	1 in 150,000
		Latino/Admixed American	99	1 in 124	1 in 12,000	1 in 48,000
		Ashkenazi Jewish	99	1 in 187	1 in 19,000	1 in 76,000
		East Asian	99	1 in 294	1 in 29,000	1 in 120,000
		Finnish	99	1 in 1,358	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	99	1 in 182	1 in 18,000	1 in 72,000
		South Asian	99	1 in 480	1 in 48,000	1 in 190,000
MCEE	Methylmalonic acidemia	Other (population not assigned)	99	1 in 128	1 in 13,000	1 in 52,000
		African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	99	1 in 1,878	1 in 190,000	1 in 760,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 2,277	1 in 230,000	1 in 920,000
		Finnish	99	1 in 11,161	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	99	1 in 873	1 in 87,000	1 in 350,000
		South Asian	97	1 in 7,646	1 in 250,000	1 in 1,000,000
MCOLN1	Mucopolipidosis type IV	Other (population not assigned)	99	1 in 2,742	1 in 270,000	1 in 1,100,000
		African/African American	99	1 in 1,566	1 in 160,000	1 in 640,000
		Latino/Admixed American	99	1 in 480	1 in 48,000	1 in 190,000
		Ashkenazi Jewish	99	1 in 112	1 in 11,000	1 in 44,000
		East Asian	99	1 in 2,278	1 in 230,000	1 in 920,000
		Finnish	97	1 in 5,411	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 1,059	1 in 110,000	1 in 440,000
		South Asian	99	1 in 366	1 in 37,000	1 in 150,000
MEFV	Familial Mediterranean fever	Other (population not assigned)	99	1 in 612	1 in 61,000	1 in 240,000
		African/African American	99	1 in 426	1 in 43,000	1 in 170,000
		Latino/Admixed American	99	1 in 246	1 in 25,000	1 in 100,000
		Ashkenazi Jewish	99	1 in 13	1 in 1,200	1 in 4,800
		East Asian	99	1 in 114	1 in 11,000	1 in 44,000
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 186	1 in 19,000	1 in 76,000
		South Asian	99	1 in 309	1 in 31,000	1 in 120,000
MEGF8	Carpenter syndrome	Other (population not assigned)	99	1 in 64	1 in 6,300	1 in 25,000
		African/African American	99	1 in 719	1 in 72,000	1 in 290,000
		Latino/Admixed American	98	1 in 544	1 in 27,000	1 in 110,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 804	1 in 80,000	1 in 320,000
		Finnish	97	1 in 538	1 in 18,000	1 in 72,000
		Non-Finnish European/Caucasian	99	1 in 375	1 in 37,000	1 in 150,000
		South Asian	99	1 in 783	1 in 78,000	1 in 310,000
MESP2	Spondylothoracic dysostosis	Other (population not assigned)	99	1 in 617	1 in 62,000	1 in 250,000
		African/African American	97	1 in 216	1 in 7,200	1 in 29,000
		Latino/Admixed American	97	1 in 145	1 in 4,800	1 in 19,000

Footnotes:

\*\* includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

## Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Ashkenazi Jewish	97	1 in 112	1 in 3,700	1 in 15,000
		East Asian	97	1 in 237	1 in 7,900	1 in 32,000
		Finnish	97	1 in 56	1 in 1,800	1 in 7,200
		Non-Finnish European/Caucasian	97	1 in 126	1 in 4,200	1 in 17,000
		South Asian	97	1 in 168	1 in 5,600	1 in 22,000
		Other (population not assigned)	97	1 in 88	1 in 2,900	1 in 12,000
MFSD8	Neuronal ceroid-lipofuscinosis	African/African American	99	1 in 1,587	1 in 160,000	1 in 640,000
		Latino/Admixed American	99	1 in 1,415	1 in 140,000	1 in 560,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 992	1 in 99,000	1 in 400,000
		Finnish	99	1 in 5,411	1 in 540,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 1,661	1 in 170,000	1 in 680,000
		South Asian	99	1 in 3,047	1 in 300,000	1 in 1,200,000
		Other (population not assigned)	98	1 in 611	1 in 31,000	1 in 120,000
MKKS	Bardet-Biedl syndrome	African/African American	99	1 in 260	1 in 26,000	1 in 100,000
		Latino/Admixed American	99	1 in 214	1 in 21,000	1 in 84,000
		Ashkenazi Jewish	99	1 in 458	1 in 46,000	1 in 180,000
		East Asian	99	1 in 920	1 in 92,000	1 in 370,000
		Finnish	99	1 in 118	1 in 12,000	1 in 48,000
		Non-Finnish European/Caucasian	99	1 in 49	1 in 4,800	1 in 19,000
		South Asian	98	1 in 807	1 in 40,000	1 in 160,000
		Other (population not assigned)	99	1 in 133	1 in 13,000	1 in 52,000
MKS1	Ciliopathies	African/African American	99	1 in 515	1 in 51,000	1 in 200,000
		Latino/Admixed American	99	1 in 946	1 in 95,000	1 in 380,000
		Ashkenazi Jewish	99	1 in 1,679	1 in 170,000	1 in 680,000
		East Asian	98	1 in 257	1 in 13,000	1 in 52,000
		Finnish	97	1 in 73	1 in 2,400	1 in 9,600
		Non-Finnish European/Caucasian	98	1 in 228	1 in 11,000	1 in 44,000
		South Asian	99	1 in 180	1 in 18,000	1 in 72,000
		Other (population not assigned)	98	1 in 248	1 in 12,000	1 in 48,000
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts type 1	African/African American	98	1 in 1,404	1 in 70,000	1 in 280,000
		Latino/Admixed American	99	1 in 5,706	1 in 570,000	1 in 2,300,000
		Ashkenazi Jewish	99	1 in 198	1 in 20,000	1 in 80,000
		East Asian	99	1 in 107	1 in 11,000	1 in 44,000
		Finnish	97	1 in 1,201	1 in 40,000	1 in 160,000
		Non-Finnish European/Caucasian	99	1 in 1,234	1 in 120,000	1 in 480,000
		South Asian	99	1 in 326	1 in 33,000	1 in 130,000
		Other (population not assigned)	99	1 in 703	1 in 70,000	1 in 280,000
MMAA	Methylmalonic acidemia	African/African American	99	1 in 7,611	1 in 760,000	1 in 3,000,000
		Latino/Admixed American	99	1 in 2,126	1 in 210,000	1 in 840,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	1 in 1,792	1 in 180,000	1 in 720,000
		Finnish	99	1 in 3,715	1 in 370,000	1 in 1,500,000

### Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

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		Non-Finnish European/Caucasian	99	1 in 665	1 in 66,000	1 in 260,000
		South Asian	99	1 in 1,012	1 in 100,000	1 in 400,000
		Other (population not assigned)	99	1 in 947	1 in 95,000	1 in 380,000
<b>MMAB</b>	Methylmalonic acidemia	African/African American	99	1 in 50	1 in 4,900	1 in 20,000
		Latino/Admixed American	99	1 in 606	1 in 61,000	1 in 240,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	1 in 3,286	1 in 330,000	1 in 1,300,000
		Non-Finnish European/Caucasian	99	1 in 801	1 in 80,000	1 in 320,000
		South Asian	99	1 in 954	1 in 95,000	1 in 380,000
		Other (population not assigned)	98	1 in 317	1 in 16,000	1 in 64,000
		<b>MMACHC</b>	Methylmalonic acidemia with homocystinuria	African/African American	98	1 in 284
Latino/Admixed American	99			1 in 38	1 in 3,700	1 in 15,000
Ashkenazi Jewish	97			1 in 206	1 in 6,800	1 in 27,000
East Asian	98			1 in 229	1 in 11,000	1 in 44,000
Finnish	99			1 in 10,780	1 in 1,100,000	1 in 4,400,000
Non-Finnish European/Caucasian	98			1 in 151	1 in 7,500	1 in 30,000
South Asian	99			1 in 202	1 in 20,000	1 in 80,000
Other (population not assigned)	98			1 in 127	1 in 6,300	1 in 25,000
<b>MMADHC</b>	Methylmalonic acidemia with homocystinuria			African/African American	99	1 in 3,940
		Latino/Admixed American	98	1 in 5,706	1 in 290,000	1 in 1,200,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 1,835	1 in 92,000	1 in 370,000
		Finnish	99	1 in 809	1 in 81,000	1 in 320,000
		Non-Finnish European/Caucasian	99	1 in 1,515	1 in 150,000	1 in 600,000
		South Asian	97	1 in 282	1 in 9,400	1 in 38,000
		Other (population not assigned)	99	1 in 983	1 in 98,000	1 in 390,000
		<b>MMUT</b>	Methylmalonic acidemia	African/African American	99	1 in 182
Latino/Admixed American	99			1 in 185	1 in 18,000	1 in 72,000
Ashkenazi Jewish	99			1 in 353	1 in 35,000	1 in 140,000
East Asian	99			1 in 178	1 in 18,000	1 in 72,000
Finnish	99			1 in 1,010	1 in 100,000	1 in 400,000
Non-Finnish European/Caucasian	99			1 in 409	1 in 41,000	1 in 160,000
South Asian	99			1 in 82	1 in 8,100	1 in 32,000
Other (population not assigned)	99			1 in 343	1 in 34,000	1 in 140,000
<b>MPI</b>	Congenital disorders of glycosylation type 1			African/African American	99	1 in 496
		Latino/Admixed American	99	1 in 202	1 in 20,000	1 in 80,000
		Ashkenazi Jewish	99	1 in 5,039	1 in 500,000	1 in 2,000,000
		East Asian	98	1 in 1,079	1 in 54,000	1 in 220,000
		Finnish	98	1 in 1,096	1 in 55,000	1 in 220,000
		Non-Finnish European/Caucasian	99	1 in 467	1 in 47,000	1 in 190,000
		South Asian	98	1 in 921	1 in 46,000	1 in 180,000
		Other (population not assigned)	99	1 in 280	1 in 28,000	1 in 110,000

Footnotes:

\*\* includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

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b Luo et al., PMID 23788250



## Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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MPL	Congenital amegakaryocytic thrombocytopenia	African/African American	99	1 in 599	1 in 60,000	1 in 240,000
		Latino/Admixed American	99	1 in 677	1 in 68,000	1 in 270,000
		Ashkenazi Jewish	99	1 in 60	1 in 5,900	1 in 24,000
		East Asian	99	1 in 292	1 in 29,000	1 in 120,000
		Finnish	99	1 in 1,383	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	99	1 in 284	1 in 28,000	1 in 110,000
		South Asian	98	1 in 529	1 in 26,000	1 in 100,000
		Other (population not assigned)	98	1 in 391	1 in 20,000	1 in 80,000
MPV17	Mitochondrial DNA depletion syndrome, MVP17-related	African/African American	99	1 in 237	1 in 24,000	1 in 96,000
		Latino/Admixed American	98	1 in 3,588	1 in 180,000	1 in 720,000
		Ashkenazi Jewish	99	1 in 1,656	1 in 170,000	1 in 680,000
		East Asian	97	1 in 126	1 in 4,200	1 in 17,000
		Finnish	97	1 in 83	1 in 2,700	1 in 11,000
		Non-Finnish European/Caucasian	99	1 in 636	1 in 64,000	1 in 260,000
		South Asian	99	1 in 542	1 in 54,000	1 in 220,000
		Other (population not assigned)	99	1 in 697	1 in 70,000	1 in 280,000
MTHFD1	Severe combined immunodeficiency (SCID)	African/African American	98	1 in 880	1 in 44,000	1 in 180,000
		Latino/Admixed American	97	1 in 361	1 in 12,000	1 in 48,000
		Ashkenazi Jewish	99	1 in 382	1 in 38,000	1 in 150,000
		East Asian	98	1 in 782	1 in 39,000	1 in 160,000
		Finnish	97	1 in 220	1 in 7,300	1 in 29,000
		Non-Finnish European/Caucasian	97	1 in 201	1 in 6,700	1 in 27,000
		South Asian	97	1 in 693	1 in 23,000	1 in 92,000
		Other (population not assigned)	98	1 in 201	1 in 10,000	1 in 40,000
MTM1	Myotubular myopathy	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	97	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
MTTP	Abetalipoproteinemia	African/African American	99	1 in 181	1 in 18,000	1 in 72,000
		Latino/Admixed American	99	1 in 1,781	1 in 180,000	1 in 720,000
		Ashkenazi Jewish	99	1 in 177	1 in 18,000	1 in 72,000
		East Asian	99	1 in 1,182	1 in 120,000	1 in 480,000
		Finnish	99	1 in 2,157	1 in 220,000	1 in 880,000
		Non-Finnish European/Caucasian	98	1 in 1,581	1 in 79,000	1 in 320,000
		South Asian	97	1 in 804	1 in 27,000	1 in 110,000
		Other (population not assigned)	97	1 in 3,069	1 in 100,000	1 in 400,000
NAGLU	Mucopolysaccharidosis type III	African/African American	99	1 in 316	1 in 32,000	1 in 130,000
		Latino/Admixed American	99	1 in 653	1 in 65,000	1 in 260,000
		Ashkenazi Jewish	97	1 in 137	1 in 4,500	1 in 18,000

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		East Asian	98	1 in 557	1 in 28,000	1 in 110,000
		Finnish	97	1 in 10,822	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 361	1 in 36,000	1 in 140,000
		South Asian	99	1 in 529	1 in 53,000	1 in 210,000
		Other (population not assigned)	99	1 in 724	1 in 72,000	1 in 290,000
<b>NBAS</b>	Acute infantile liver failure	African/African American	99	1 in 232	1 in 23,000	1 in 92,000
		Latino/Admixed American	98	1 in 219	1 in 11,000	1 in 44,000
		Ashkenazi Jewish	97	1 in 5,032	1 in 170,000	1 in 680,000
		East Asian	99	1 in 482	1 in 48,000	1 in 190,000
		Finnish	97	1 in 347	1 in 12,000	1 in 48,000
		Non-Finnish European/Caucasian	98	1 in 228	1 in 11,000	1 in 44,000
		South Asian	99	1 in 436	1 in 44,000	1 in 180,000
		Other (population not assigned)	98	1 in 147	1 in 7,300	1 in 29,000
		Other (population not assigned)	99	1 in 379	1 in 38,000	1 in 150,000
<b>NBN</b>	Nijmegen breakage syndrome	Latino/Admixed American	99	1 in 213	1 in 21,000	1 in 84,000
		Ashkenazi Jewish	99	1 in 255	1 in 25,000	1 in 100,000
		East Asian	99	1 in 278	1 in 28,000	1 in 110,000
		Finnish	99	1 in 103	1 in 10,000	1 in 40,000
		Non-Finnish European/Caucasian	99	1 in 70	1 in 6,900	1 in 28,000
		South Asian	99	1 in 767	1 in 77,000	1 in 310,000
		Other (population not assigned)	99	1 in 124	1 in 12,000	1 in 48,000
		Other (population not assigned)	97	1 in 8,129	1 in 270,000	1 in 1,100,000
		Other (population not assigned)	98	1 in 556	1 in 28,000	1 in 110,000
<b>NCF2</b>	Chronic granulomatous disease	Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 2,262	1 in 110,000	1 in 440,000
		Finnish	99	1 in 5,575	1 in 560,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 1,883	1 in 190,000	1 in 760,000
		South Asian	98	1 in 2,189	1 in 110,000	1 in 440,000
		Other (population not assigned)	97	1 in 3,066	1 in 100,000	1 in 400,000
		Other (population not assigned)	99	1 in 1,335	1 in 130,000	1 in 520,000
		Other (population not assigned)	99	1 in 8,597	1 in 860,000	1 in 3,400,000
		Other (population not assigned)	99	1 in 369	1 in 37,000	1 in 150,000
<b>NCF4</b>	Chronic granulomatous disease	East Asian	99	1 in 1,367	1 in 140,000	1 in 560,000
		Finnish	99	1 in 5,385	1 in 540,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 540	1 in 54,000	1 in 220,000
		South Asian	99	1 in 450	1 in 45,000	1 in 180,000
		Other (population not assigned)	99	1 in 614	1 in 61,000	1 in 240,000
		Other (population not assigned)	99	1 in 881	1 in 88,000	1 in 350,000
		Other (population not assigned)	99	1 in 796	1 in 80,000	1 in 320,000
		Other (population not assigned)	97	1 in 92	1 in 3,000	1 in 12,000
		Other (population not assigned)	99	1 in 889	1 in 89,000	1 in 360,000
<b>NDUFAF2</b>	Leigh syndrome, autosomal recessive	Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	1 in 1,014	1 in 51,000	1 in 200,000

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NDUFAF5	Leigh syndrome, autosomal recessive	South Asian	97	1 in 1,326	1 in 44,000	1 in 180,000
		Other (population not assigned)	98	1 in 331	1 in 17,000	1 in 68,000
		African/African American	98	1 in 1,625	1 in 81,000	1 in 320,000
		Latino/Admixed American	99	1 in 861	1 in 86,000	1 in 340,000
		Ashkenazi Jewish	99	1 in 157	1 in 16,000	1 in 64,000
		East Asian	99	1 in 309	1 in 31,000	1 in 120,000
		Finnish	98	1 in 5,320	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 417	1 in 42,000	1 in 170,000
		South Asian	99	1 in 1,180	1 in 120,000	1 in 480,000
NDUFS4	Leigh syndrome, autosomal recessive	Other (population not assigned)	99	1 in 611	1 in 61,000	1 in 240,000
		African/African American	98	1 in 1,207	1 in 60,000	1 in 240,000
		Latino/Admixed American	98	1 in 1,103	1 in 55,000	1 in 220,000
		Ashkenazi Jewish	98	1 in 1,640	1 in 82,000	1 in 330,000
		East Asian	99	1 in 4,597	1 in 460,000	1 in 1,800,000
		Finnish	97	1 in 2,229	1 in 74,000	1 in 300,000
		Non-Finnish European/Caucasian	98	1 in 1,738	1 in 87,000	1 in 350,000
		South Asian	99	1 in 1,914	1 in 190,000	1 in 760,000
		Other (population not assigned)	97	N/A	N/A	N/A
NDUFS6	Leigh syndrome, autosomal recessive	African/African American	99	1 in 2,452	1 in 250,000	1 in 1,000,000
		Latino/Admixed American	98	1 in 1,896	1 in 95,000	1 in 380,000
		Ashkenazi Jewish	98	1 in 1,113	1 in 56,000	1 in 220,000
		East Asian	99	1 in 1,473	1 in 150,000	1 in 600,000
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	1 in 3,599	1 in 180,000	1 in 720,000
		South Asian	98	1 in 1,398	1 in 70,000	1 in 280,000
		Other (population not assigned)	98	N/A	N/A	N/A
		NDUFS7	Leigh syndrome, autosomal recessive	African/African American	99	1 in 1,552
Latino/Admixed American	99			1 in 2,327	1 in 230,000	1 in 920,000
Ashkenazi Jewish	97			N/A	N/A	N/A
East Asian	97			1 in 3,320	1 in 110,000	1 in 440,000
Finnish	97			1 in 10,639	1 in 350,000	1 in 1,400,000
Non-Finnish European/Caucasian	99			1 in 839	1 in 84,000	1 in 340,000
South Asian	99			1 in 180	1 in 18,000	1 in 72,000
Other (population not assigned)	99			1 in 578	1 in 58,000	1 in 230,000
NDUFV1	Leigh syndrome, autosomal recessive			African/African American	99	1 in 157
		Latino/Admixed American	99	1 in 441	1 in 44,000	1 in 180,000
		Ashkenazi Jewish	99	1 in 1,680	1 in 170,000	1 in 680,000
		East Asian	98	1 in 2,260	1 in 110,000	1 in 440,000
		Finnish	99	1 in 108	1 in 11,000	1 in 44,000
		Non-Finnish European/Caucasian	99	1 in 469	1 in 47,000	1 in 190,000
		South Asian	99	1 in 319	1 in 32,000	1 in 130,000
		Other (population not assigned)	99	1 in 282	1 in 28,000	1 in 110,000
		NEB	Nemaline myopathy	African/African American	93	1 in 190

Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

### Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Latino/Admixed American	98	1 in 149	1 in 7,400	1 in 30,000
		Ashkenazi Jewish	99	1 in 226	1 in 23,000	1 in 92,000
		East Asian	97	1 in 87	1 in 2,900	1 in 12,000
		Finnish	98	1 in 181	1 in 9,000	1 in 36,000
		Non-Finnish European/Caucasian	92	1 in 146	1 in 1,800	1 in 7,200
		South Asian	98	1 in 159	1 in 7,900	1 in 32,000
		Other (population not assigned)	99	1 in 94	1 in 9,300	1 in 37,000
<b>NEU1</b>	Sialidosis	African/African American	99	1 in 636	1 in 64,000	1 in 260,000
		Latino/Admixed American	99	1 in 2,833	1 in 280,000	1 in 1,100,000
		Ashkenazi Jewish	99	1 in 610	1 in 61,000	1 in 240,000
		East Asian	99	1 in 279	1 in 28,000	1 in 110,000
		Finnish	99	1 in 2,745	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 1,666	1 in 170,000	1 in 680,000
		South Asian	98	1 in 3,045	1 in 150,000	1 in 600,000
Other (population not assigned)	99	1 in 1,431	1 in 140,000	1 in 560,000		
<b>NGLY1</b>	Congenital disorder of deglycosylation	African/African American	99	1 in 879	1 in 88,000	1 in 350,000
		Latino/Admixed American	98	1 in 1,654	1 in 83,000	1 in 330,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 365	1 in 36,000	1 in 140,000
		Finnish	97	1 in 4,736	1 in 160,000	1 in 640,000
		Non-Finnish European/Caucasian	99	1 in 610	1 in 61,000	1 in 240,000
		South Asian	99	1 in 785	1 in 78,000	1 in 310,000
Other (population not assigned)	99	1 in 542	1 in 54,000	1 in 220,000		
<b>NHEJ1</b>	Severe combined immunodeficiency (SCID)	African/African American	99	1 in 1,016	1 in 100,000	1 in 400,000
		Latino/Admixed American	98	1 in 3,438	1 in 170,000	1 in 680,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 459	1 in 46,000	1 in 180,000
		Finnish	97	1 in 4,474	1 in 150,000	1 in 600,000
		Non-Finnish European/Caucasian	98	1 in 2,750	1 in 140,000	1 in 560,000
		South Asian	99	1 in 1,757	1 in 180,000	1 in 720,000
Other (population not assigned)	97	1 in 3,071	1 in 100,000	1 in 400,000		
<b>NIPAL4</b>	Autosomal recessive congenital ichthyosis (ARCI)	African/African American	99	1 in 1,827	1 in 180,000	1 in 720,000
		Latino/Admixed American	98	1 in 687	1 in 34,000	1 in 140,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	98	1 in 660	1 in 33,000	1 in 130,000
		Finnish	99	1 in 1,759	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 372	1 in 37,000	1 in 150,000
		South Asian	99	1 in 841	1 in 84,000	1 in 340,000
Other (population not assigned)	99	1 in 657	1 in 66,000	1 in 260,000		
<b>NONO</b>	X-linked syndromic mental retardation	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A

Footnotes:

\*\* includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

## Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
NPC1	Niemann-Pick disease type C	African/African American	99	1 in 439	1 in 44,000	1 in 180,000
		Latino/Admixed American	99	1 in 325	1 in 32,000	1 in 130,000
		Ashkenazi Jewish	97	1 in 382	1 in 13,000	1 in 52,000
		East Asian	99	1 in 444	1 in 44,000	1 in 180,000
		Finnish	99	1 in 181	1 in 18,000	1 in 72,000
		Non-Finnish European/Caucasian	99	1 in 150	1 in 15,000	1 in 60,000
		South Asian	99	1 in 548	1 in 55,000	1 in 220,000
		Other (population not assigned)	99	1 in 328	1 in 33,000	1 in 130,000
		NPC2	Niemann-Pick disease type C	African/African American	99	1 in 66
Latino/Admixed American	99			1 in 306	1 in 31,000	1 in 120,000
Ashkenazi Jewish	99			1 in 297	1 in 30,000	1 in 120,000
East Asian	99			1 in 969	1 in 97,000	1 in 390,000
Finnish	99			1 in 171	1 in 17,000	1 in 68,000
Non-Finnish European/Caucasian	99			1 in 74	1 in 7,300	1 in 29,000
South Asian	99			1 in 214	1 in 21,000	1 in 84,000
Other (population not assigned)	99			1 in 136	1 in 14,000	1 in 56,000
NPHP1	Joubert syndrome and related disorders, including Meckel-Gruber syndrome			African/African American	98	1 in 218
		Latino/Admixed American	98	1 in 315	1 in 16,000	1 in 64,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 164	1 in 16,000	1 in 64,000
		Finnish	98	1 in 806	1 in 40,000	1 in 160,000
		Non-Finnish European/Caucasian	98	1 in 418	1 in 21,000	1 in 84,000
		South Asian	98	1 in 536	1 in 27,000	1 in 110,000
		Other (population not assigned)	98	1 in 362	1 in 18,000	1 in 72,000
		NPHP3	Joubert syndrome and related disorders, including Meckel-Gruber syndrome	African/African American	99	1 in 42
Latino/Admixed American	98			1 in 69	1 in 3,400	1 in 14,000
Ashkenazi Jewish	97			1 in 219	1 in 7,300	1 in 29,000
East Asian	99			1 in 67	1 in 6,600	1 in 26,000
Finnish	99			1 in 78	1 in 7,700	1 in 31,000
Non-Finnish European/Caucasian	99			1 in 73	1 in 7,200	1 in 29,000
South Asian	97			1 in 57	1 in 1,900	1 in 7,600
Other (population not assigned)	98			1 in 66	1 in 3,300	1 in 13,000
NPHS1	Nephrotic syndrome			African/African American	99	1 in 42
		Latino/Admixed American	98	1 in 91	1 in 4,500	1 in 18,000
		Ashkenazi Jewish	99	1 in 419	1 in 42,000	1 in 170,000
		East Asian	99	1 in 121	1 in 12,000	1 in 48,000
		Finnish	98	1 in 27	1 in 1,300	1 in 5,200
		Non-Finnish European/Caucasian	99	1 in 67	1 in 6,600	1 in 26,000
		South Asian	99	1 in 123	1 in 12,000	1 in 48,000

Footnotes:

\*\* includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

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b Luo et al., PMID 23788250

## Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
NPHS2	Nephrotic syndrome	Other (population not assigned)	99	1 in 63	1 in 6,200	1 in 25,000
		African/African American	98	1 in 370	1 in 18,000	1 in 72,000
		Latino/Admixed American	99	1 in 339	1 in 34,000	1 in 140,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 710	1 in 71,000	1 in 280,000
		Finnish	98	1 in 1,441	1 in 72,000	1 in 290,000
		Non-Finnish European/Caucasian	99	1 in 187	1 in 19,000	1 in 76,000
		South Asian	98	1 in 628	1 in 31,000	1 in 120,000
		Other (population not assigned)	99	1 in 201	1 in 20,000	1 in 80,000
NROB1	Congenital adrenal hypoplasia, X-linked	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	97	N/A	N/A	N/A
		South Asian	97	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
NTRK1	Congenital insensitivity to pain with anhidrosis	African/African American	98	1 in 297	1 in 15,000	1 in 60,000
		Latino/Admixed American	99	1 in 322	1 in 32,000	1 in 130,000
		Ashkenazi Jewish	98	1 in 1,827	1 in 91,000	1 in 360,000
		East Asian	98	1 in 181	1 in 9,000	1 in 36,000
		Finnish	99	1 in 233	1 in 23,000	1 in 92,000
		Non-Finnish European/Caucasian	99	1 in 177	1 in 18,000	1 in 72,000
		South Asian	99	1 in 554	1 in 55,000	1 in 220,000
		Other (population not assigned)	99	1 in 198	1 in 20,000	1 in 80,000
OAT	Gyrate atrophy of choroid and retina	African/African American	97	1 in 2,600	1 in 87,000	1 in 350,000
		Latino/Admixed American	99	1 in 892	1 in 89,000	1 in 360,000
		Ashkenazi Jewish	99	1 in 615	1 in 61,000	1 in 240,000
		East Asian	99	1 in 2,935	1 in 290,000	1 in 1,200,000
		Finnish	99	1 in 142	1 in 14,000	1 in 56,000
		Non-Finnish European/Caucasian	99	1 in 426	1 in 43,000	1 in 170,000
		South Asian	99	1 in 904	1 in 90,000	1 in 360,000
		Other (population not assigned)	99	1 in 410	1 in 41,000	1 in 160,000
		OBSL1	3M syndrome	African/African American	99	1 in 43
Latino/Admixed American	99			1 in 66	1 in 6,500	1 in 26,000
Ashkenazi Jewish	99			1 in 33	1 in 3,200	1 in 13,000
East Asian	97			1 in 43	1 in 1,400	1 in 5,600
Finnish	99			1 in 113	1 in 11,000	1 in 44,000
Non-Finnish European/Caucasian	99			1 in 103	1 in 10,000	1 in 40,000
South Asian	97			1 in 187	1 in 6,200	1 in 25,000
Other (population not assigned)	99			1 in 61	1 in 6,000	1 in 24,000
OCRL	Dent disease	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A

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		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
OPA3	Costeff optic atrophy syndrome, autosomal recessive	African/African American	97	1 in 6,886	1 in 230,000	1 in 920,000
		Latino/Admixed American	98	1 in 1,889	1 in 94,000	1 in 380,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 3,006	1 in 300,000	1 in 1,200,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	1 in 2,572	1 in 130,000	1 in 520,000
		South Asian	99	1 in 391	1 in 39,000	1 in 160,000
		Other (population not assigned)	99	1 in 2,706	1 in 270,000	1 in 1,100,000
OSTM1	Osteopetrosis, autosomal recessive	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	99	1 in 2,097	1 in 210,000	1 in 840,000
		East Asian	99	1 in 9,197	1 in 920,000	1 in 3,700,000
		Finnish	97	1 in 7,821	1 in 260,000	1 in 1,000,000
		Non-Finnish European/Caucasian	99	1 in 6,173	1 in 620,000	1 in 2,500,000
		South Asian	99	1 in 6,619	1 in 660,000	1 in 2,600,000
		Other (population not assigned)	97	N/A	N/A	N/A
OTC	Ornithine transcarbamylase deficiency	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
OTOF	Deafness and hearing loss, nonsyndromic	African/African American	99	1 in 201	1 in 20,000	1 in 80,000
		Latino/Admixed American	99	1 in 258	1 in 26,000	1 in 100,000
		Ashkenazi Jewish	99	1 in 78	1 in 7,700	1 in 31,000
		East Asian	99	1 in 52	1 in 5,100	1 in 20,000
		Finnish	99	1 in 903	1 in 90,000	1 in 360,000
		Non-Finnish European/Caucasian	99	1 in 228	1 in 23,000	1 in 92,000
		South Asian	99	1 in 240	1 in 24,000	1 in 96,000
		Other (population not assigned)	99	1 in 229	1 in 23,000	1 in 92,000
P3H1	Osteogenesis imperfecta, autosomal recessive	African/African American	99	1 in 136	1 in 14,000	1 in 56,000
		Latino/Admixed American	97	1 in 278	1 in 9,200	1 in 37,000
		Ashkenazi Jewish	99	1 in 816	1 in 82,000	1 in 330,000
		East Asian	99	1 in 206	1 in 21,000	1 in 84,000
		Finnish	98	1 in 2,203	1 in 110,000	1 in 440,000

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		Non-Finnish European/Caucasian	98	1 in 567	1 in 28,000	1 in 110,000
		South Asian	98	1 in 765	1 in 38,000	1 in 150,000
		Other (population not assigned)	98	1 in 912	1 in 46,000	1 in 180,000
		African/African American	99	1 in 213	1 in 21,000	1 in 84,000
		Latino/Admixed American	99	1 in 74	1 in 7,300	1 in 29,000
		Ashkenazi Jewish	99	1 in 17	1 in 1,600	1 in 6,400
		East Asian	98	1 in 57	1 in 2,800	1 in 11,000
		Finnish	99	1 in 195	1 in 19,000	1 in 76,000
		Non-Finnish European/Caucasian	99	1 in 41	1 in 4,000	1 in 16,000
		South Asian	99	1 in 76	1 in 7,500	1 in 30,000
PAH	Phenylalanine hydroxylase deficiency, includes phenylketonuria (PKU)	Other (population not assigned)	99	1 in 39	1 in 3,800	1 in 15,000
		African/African American	98	1 in 1,027	1 in 51,000	1 in 200,000
		Latino/Admixed American	98	1 in 845	1 in 42,000	1 in 170,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 655	1 in 65,000	1 in 260,000
		Finnish	98	1 in 1,418	1 in 71,000	1 in 280,000
		Non-Finnish European/Caucasian	98	1 in 700	1 in 35,000	1 in 140,000
		South Asian	97	1 in 427	1 in 14,000	1 in 56,000
		Other (population not assigned)	98	1 in 510	1 in 25,000	1 in 100,000
		PANK2	Pantothenate kinase-associated neurodegeneration	African/African American	98	1 in 169
Latino/Admixed American	99			1 in 1,306	1 in 130,000	1 in 520,000
Ashkenazi Jewish	99			1 in 411	1 in 41,000	1 in 160,000
East Asian	99			1 in 1,314	1 in 130,000	1 in 520,000
Finnish	99			N/A	N/A	N/A
Non-Finnish European/Caucasian	99			1 in 984	1 in 98,000	1 in 390,000
South Asian	99			1 in 7,638	1 in 760,000	1 in 3,000,000
Other (population not assigned)	99			1 in 745	1 in 74,000	1 in 300,000
African/African American	99			1 in 493	1 in 49,000	1 in 200,000
PCBD1	Tetrahydrobiopterin deficiency			Latino/Admixed American	98	1 in 348
		Ashkenazi Jewish	97	1 in 548	1 in 18,000	1 in 72,000
		East Asian	98	1 in 662	1 in 33,000	1 in 130,000
		Finnish	99	1 in 2,722	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 756	1 in 76,000	1 in 300,000
		South Asian	99	1 in 522	1 in 52,000	1 in 210,000
		Other (population not assigned)	97	1 in 846	1 in 28,000	1 in 110,000
		African/African American	98	1 in 238	1 in 12,000	1 in 48,000
		Latino/Admixed American	99	1 in 285	1 in 28,000	1 in 110,000
		PCCA	Propionic acidemia	Ashkenazi Jewish	97	1 in 4,924
East Asian	99			1 in 233	1 in 23,000	1 in 92,000
Finnish	99			1 in 83	1 in 8,200	1 in 33,000
Non-Finnish European/Caucasian	99			1 in 144	1 in 14,000	1 in 56,000
South Asian	99			1 in 842	1 in 84,000	1 in 340,000
Other (population not assigned)	99			1 in 251	1 in 25,000	1 in 100,000
African/African American	98			1 in 238	1 in 12,000	1 in 48,000
Latino/Admixed American	99			1 in 285	1 in 28,000	1 in 110,000
Ashkenazi Jewish	97			1 in 4,924	1 in 160,000	1 in 640,000
East Asian	99			1 in 233	1 in 23,000	1 in 92,000
PCCB	Propionic acidemia	Finnish	99	1 in 83	1 in 8,200	1 in 33,000
		Non-Finnish European/Caucasian	99	1 in 144	1 in 14,000	1 in 56,000
		South Asian	99	1 in 842	1 in 84,000	1 in 340,000
		Other (population not assigned)	99	1 in 251	1 in 25,000	1 in 100,000
		African/African American	98	1 in 238	1 in 12,000	1 in 48,000
		Latino/Admixed American	99	1 in 285	1 in 28,000	1 in 110,000
		Ashkenazi Jewish	97	1 in 4,924	1 in 160,000	1 in 640,000
		East Asian	99	1 in 233	1 in 23,000	1 in 92,000
		Finnish	99	1 in 83	1 in 8,200	1 in 33,000
		Non-Finnish European/Caucasian	99	1 in 144	1 in 14,000	1 in 56,000

Footnotes:

\*\* includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

a Feng et al., PMID 28125085

b Luo et al., PMID 23788250



### Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
PCDH15	Usher syndrome (hearing loss and retinitis pigmentosa)	African/African American	97	1 in 91	1 in 3,000	1 in 12,000
		Latino/Admixed American	98	1 in 96	1 in 4,800	1 in 19,000
		Ashkenazi Jewish	99	1 in 88	1 in 8,700	1 in 35,000
		East Asian	97	1 in 31	1 in 1,000	1 in 4,000
		Finnish	98	1 in 571	1 in 29,000	1 in 120,000
		Non-Finnish European/Caucasian	98	1 in 114	1 in 5,700	1 in 23,000
		South Asian	97	1 in 59	1 in 1,900	1 in 7,600
		Other (population not assigned)	97	1 in 85	1 in 2,800	1 in 11,000
PDHA1	Pyruvate dehydrogenase deficiency	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	97	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
PDHB	Pyruvate dehydrogenase deficiency	African/African American	99	1 in 8,128	1 in 810,000	1 in 3,200,000
		Latino/Admixed American	97	1 in 1,352	1 in 45,000	1 in 180,000
		Ashkenazi Jewish	97	1 in 5,041	1 in 170,000	1 in 680,000
		East Asian	97	1 in 9,197	1 in 310,000	1 in 1,200,000
		Finnish	98	1 in 3,587	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	98	1 in 6,213	1 in 310,000	1 in 1,200,000
		South Asian	98	1 in 1,797	1 in 90,000	1 in 360,000
		Other (population not assigned)	97	N/A	N/A	N/A
PDHX	Pyruvate dehydrogenase deficiency	African/African American	99	1 in 155	1 in 15,000	1 in 60,000
		Latino/Admixed American	98	1 in 4,294	1 in 210,000	1 in 840,000
		Ashkenazi Jewish	97	1 in 4,921	1 in 160,000	1 in 640,000
		East Asian	99	1 in 461	1 in 46,000	1 in 180,000
		Finnish	98	1 in 2,164	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	99	1 in 1,524	1 in 150,000	1 in 600,000
		South Asian	99	1 in 372	1 in 37,000	1 in 150,000
		Other (population not assigned)	99	1 in 1,242	1 in 120,000	1 in 480,000
PDP1	Pyruvate dehydrogenase deficiency	African/African American	99	1 in 4,064	1 in 410,000	1 in 1,600,000
		Latino/Admixed American	98	1 in 8,647	1 in 430,000	1 in 1,700,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 184	1 in 18,000	1 in 72,000
		Finnish	97	1 in 5,324	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	98	1 in 4,651	1 in 230,000	1 in 920,000
		South Asian	97	1 in 15,296	1 in 510,000	1 in 2,000,000
		Other (population not assigned)	97	N/A	N/A	N/A
PEX1	Zellweger spectrum disorder/ peroxisome biogenesis disorder	African/African American	98	1 in 413	1 in 21,000	1 in 84,000
		Latino/Admixed American	99	1 in 441	1 in 44,000	1 in 180,000
		Ashkenazi Jewish	98	1 in 1,201	1 in 60,000	1 in 240,000

Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

### Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result		
PEX2	Zellweger spectrum disorder/ peroxisome biogenesis disorder	East Asian	99	1 in 336	1 in 34,000	1 in 140,000		
		Finnish	97	1 in 2,150	1 in 72,000	1 in 290,000		
		Non-Finnish European/Caucasian	98	1 in 212	1 in 11,000	1 in 44,000		
		South Asian	99	1 in 513	1 in 51,000	1 in 200,000		
		Other (population not assigned)	98	1 in 400	1 in 20,000	1 in 80,000		
		African/African American	97	1 in 2,702	1 in 90,000	1 in 360,000		
		Latino/Admixed American	98	1 in 8,645	1 in 430,000	1 in 1,700,000		
		Ashkenazi Jewish	99	1 in 198	1 in 20,000	1 in 80,000		
		East Asian	99	1 in 2,225	1 in 220,000	1 in 880,000		
		Finnish	97	1 in 10,784	1 in 360,000	1 in 1,400,000		
		Non-Finnish European/Caucasian	98	1 in 2,002	1 in 100,000	1 in 400,000		
		South Asian	98	1 in 1,914	1 in 96,000	1 in 380,000		
		Other (population not assigned)	98	1 in 704	1 in 35,000	1 in 140,000		
		PEX3	Zellweger spectrum disorder/ peroxisome biogenesis disorder	African/African American	99	1 in 2,708	1 in 270,000	1 in 1,100,000
Latino/Admixed American	98			1 in 2,458	1 in 120,000	1 in 480,000		
Ashkenazi Jewish	97			1 in 5,037	1 in 170,000	1 in 680,000		
East Asian	99			1 in 4,599	1 in 460,000	1 in 1,800,000		
Finnish	97			N/A	N/A	N/A		
Non-Finnish European/Caucasian	99			1 in 2,968	1 in 300,000	1 in 1,200,000		
South Asian	99			1 in 2,553	1 in 260,000	1 in 1,000,000		
Other (population not assigned)	97			N/A	N/A	N/A		
PEX5	Zellweger spectrum disorder/ peroxisome biogenesis disorder			African/African American	99	1 in 1,937	1 in 190,000	1 in 760,000
				Latino/Admixed American	98	1 in 2,460	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	97	N/A	N/A	N/A		
		East Asian	97	1 in 202	1 in 6,700	1 in 27,000		
		Finnish	99	1 in 10,813	1 in 1,100,000	1 in 4,400,000		
		Non-Finnish European/Caucasian	99	1 in 914	1 in 91,000	1 in 360,000		
		South Asian	97	1 in 1,024	1 in 34,000	1 in 140,000		
		Other (population not assigned)	98	1 in 1,532	1 in 77,000	1 in 310,000		
		PEX6	Zellweger spectrum disorder/ peroxisome biogenesis disorder	African/African American	98	1 in 322	1 in 16,000	1 in 64,000
				Latino/Admixed American	99	1 in 262	1 in 26,000	1 in 100,000
Ashkenazi Jewish	99			1 in 249	1 in 25,000	1 in 100,000		
East Asian	99			1 in 749	1 in 75,000	1 in 300,000		
Finnish	99			1 in 280	1 in 28,000	1 in 110,000		
Non-Finnish European/Caucasian	99			1 in 97	1 in 9,600	1 in 38,000		
South Asian	99			1 in 97	1 in 9,600	1 in 38,000		
Other (population not assigned)	99			1 in 189	1 in 19,000	1 in 76,000		
PEX7	Rhizomelic chondrodysplasia punctata			African/African American	99	1 in 261	1 in 26,000	1 in 100,000
				Latino/Admixed American	99	1 in 444	1 in 44,000	1 in 180,000
		Ashkenazi Jewish	99	1 in 118	1 in 12,000	1 in 48,000		
		East Asian	98	1 in 272	1 in 14,000	1 in 56,000		
		Finnish	98	1 in 443	1 in 22,000	1 in 88,000		
		Non-Finnish European/Caucasian	99	1 in 146	1 in 15,000	1 in 60,000		

Footnotes:

\*\* includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

a Feng et al., PMID 28125085

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## Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result		
PEX10	Zellweger spectrum disorder/ peroxisome biogenesis disorder	South Asian	99	1 in 371	1 in 37,000	1 in 150,000		
		Other (population not assigned)	98	1 in 183	1 in 9,100	1 in 36,000		
		African/African American	98	1 in 3,744	1 in 190,000	1 in 760,000		
		Latino/Admixed American	97	1 in 952	1 in 32,000	1 in 130,000		
		Ashkenazi Jewish	99	1 in 518	1 in 52,000	1 in 210,000		
		East Asian	98	1 in 2,025	1 in 100,000	1 in 400,000		
		Finnish	98	1 in 567	1 in 28,000	1 in 110,000		
		Non-Finnish European/Caucasian	98	1 in 766	1 in 38,000	1 in 150,000		
		South Asian	98	1 in 917	1 in 46,000	1 in 180,000		
		Other (population not assigned)	98	1 in 342	1 in 17,000	1 in 68,000		
PEX11B	Zellweger spectrum disorder/ peroxisome biogenesis disorder	African/African American	97	1 in 1,799	1 in 60,000	1 in 240,000		
		Latino/Admixed American	99	1 in 1,370	1 in 140,000	1 in 560,000		
		Ashkenazi Jewish	97	N/A	N/A	N/A		
		East Asian	99	1 in 2,991	1 in 300,000	1 in 1,200,000		
		Finnish	97	1 in 301	1 in 10,000	1 in 40,000		
		Non-Finnish European/Caucasian	97	1 in 1,419	1 in 47,000	1 in 190,000		
		South Asian	99	1 in 1,217	1 in 120,000	1 in 480,000		
		Other (population not assigned)	98	1 in 1,499	1 in 75,000	1 in 300,000		
		African/African American	98	1 in 552	1 in 28,000	1 in 110,000		
		Latino/Admixed American	98	1 in 527	1 in 26,000	1 in 100,000		
PEX12	Zellweger spectrum disorder/ peroxisome biogenesis disorder	Ashkenazi Jewish	99	1 in 1,002	1 in 100,000	1 in 400,000		
		East Asian	97	1 in 2,875	1 in 96,000	1 in 380,000		
		Finnish	99	1 in 10,825	1 in 1,100,000	1 in 4,400,000		
		Non-Finnish European/Caucasian	98	1 in 718	1 in 36,000	1 in 140,000		
		South Asian	98	1 in 1,278	1 in 64,000	1 in 260,000		
		Other (population not assigned)	99	1 in 3,056	1 in 310,000	1 in 1,200,000		
		African/African American	99	1 in 396	1 in 40,000	1 in 160,000		
		Latino/Admixed American	99	1 in 610	1 in 61,000	1 in 240,000		
		Ashkenazi Jewish	98	N/A	N/A	N/A		
		East Asian	98	1 in 1,815	1 in 91,000	1 in 360,000		
PEX13	Zellweger spectrum disorder/ peroxisome biogenesis disorder	Finnish	98	N/A	N/A	N/A		
		Non-Finnish European/Caucasian	99	1 in 5,185	1 in 520,000	1 in 2,100,000		
		South Asian	99	1 in 522	1 in 52,000	1 in 210,000		
		Other (population not assigned)	99	1 in 528	1 in 53,000	1 in 210,000		
		African/African American	99	1 in 7,823	1 in 780,000	1 in 3,100,000		
		Latino/Admixed American	99	1 in 16,707	1 in 1,700,000	1 in 6,800,000		
		Ashkenazi Jewish	99	N/A	N/A	N/A		
		East Asian	99	1 in 3,053	1 in 310,000	1 in 1,200,000		
		Finnish	99	1 in 10,699	1 in 1,100,000	1 in 4,400,000		
		Non-Finnish European/Caucasian	99	1 in 8,009	1 in 800,000	1 in 3,200,000		
PEX14	Zellweger spectrum disorder/ peroxisome biogenesis disorder	South Asian	99	1 in 1,873	1 in 190,000	1 in 760,000		
		Other (population not assigned)	99	N/A	N/A	N/A		
		African/African American	97	1 in 2,657	1 in 89,000	1 in 360,000		
		PEX16	Zellweger spectrum disorder/ peroxisome biogenesis disorder	African/African American	97	1 in 2,657	1 in 89,000	1 in 360,000

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## Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
	biogenesis disorder	Latino/Admixed American	97	1 in 3,189	1 in 110,000	1 in 440,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 3,060	1 in 310,000	1 in 1,200,000
		Finnish	99	1 in 4,559	1 in 460,000	1 in 1,800,000
		Non-Finnish European/Caucasian	98	1 in 3,165	1 in 160,000	1 in 640,000
		South Asian	99	1 in 3,061	1 in 310,000	1 in 1,200,000
		Other (population not assigned)	99	1 in 2,744	1 in 270,000	1 in 1,100,000
PEX19	Zellweger spectrum disorder/ peroxisome biogenesis disorder	African/African American	97	1 in 8,129	1 in 270,000	1 in 1,100,000
		Latino/Admixed American	99	1 in 3,459	1 in 350,000	1 in 1,400,000
		Ashkenazi Jewish	98	1 in 2,520	1 in 130,000	1 in 520,000
		East Asian	99	1 in 1,840	1 in 180,000	1 in 720,000
		Finnish	97	1 in 1,804	1 in 60,000	1 in 240,000
		Non-Finnish European/Caucasian	98	1 in 1,921	1 in 96,000	1 in 380,000
		South Asian	99	1 in 1,178	1 in 120,000	1 in 480,000
PEX26	Zellweger spectrum disorder/ peroxisome biogenesis disorder	Other (population not assigned)	97	N/A	N/A	N/A
		African/African American	97	1 in 271	1 in 9,000	1 in 36,000
		Latino/Admixed American	98	1 in 691	1 in 35,000	1 in 140,000
		Ashkenazi Jewish	97	1 in 2,520	1 in 84,000	1 in 340,000
		East Asian	99	1 in 263	1 in 26,000	1 in 100,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	1 in 996	1 in 50,000	1 in 200,000
PFKM	Glycogen storage disease type VII	South Asian	99	1 in 1,860	1 in 190,000	1 in 760,000
		Other (population not assigned)	98	1 in 766	1 in 38,000	1 in 150,000
		African/African American	99	1 in 62	1 in 6,100	1 in 24,000
		Latino/Admixed American	99	1 in 647	1 in 65,000	1 in 260,000
		Ashkenazi Jewish	98	1 in 108	1 in 5,400	1 in 22,000
		East Asian	99	1 in 488	1 in 49,000	1 in 200,000
		Finnish	97	1 in 773	1 in 26,000	1 in 100,000
PGM3	Severe combined immunodeficiency (SCID)	Non-Finnish European/Caucasian	99	1 in 600	1 in 60,000	1 in 240,000
		South Asian	98	1 in 1,165	1 in 58,000	1 in 230,000
		Other (population not assigned)	99	1 in 280	1 in 28,000	1 in 110,000
		African/African American	99	1 in 690	1 in 69,000	1 in 280,000
		Latino/Admixed American	99	1 in 140	1 in 14,000	1 in 56,000
		Ashkenazi Jewish	98	1 in 960	1 in 48,000	1 in 190,000
		East Asian	99	1 in 1,060	1 in 110,000	1 in 440,000
PHGDH	Phosphoglycerate dehydrogenase deficiency	Finnish	98	1 in 5,466	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	98	1 in 801	1 in 40,000	1 in 160,000
		South Asian	98	1 in 645	1 in 32,000	1 in 130,000
		Other (population not assigned)	98	1 in 338	1 in 17,000	1 in 68,000
		African/African American	99	1 in 3,826	1 in 380,000	1 in 1,500,000
		Latino/Admixed American	99	1 in 4,291	1 in 430,000	1 in 1,700,000
		Ashkenazi Jewish	99	1 in 290	1 in 29,000	1 in 120,000
East Asian	99	1 in 1,500	1 in 150,000	1 in 600,000		

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		Finnish	99	1 in 1,859	1 in 190,000	1 in 760,000
		Non-Finnish European/Caucasian	99	1 in 1,575	1 in 160,000	1 in 640,000
		South Asian	99	1 in 1,862	1 in 190,000	1 in 760,000
		Other (population not assigned)	99	N/A	N/A	N/A
PHKA1	Glycogen storage disease type IX	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	98	N/A	N/A	N/A
		Other (population not assigned)	98	N/A	N/A	N/A
PHKA2	Glycogen storage disease type IX	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
PHKB	Glycogen storage disease type IX	African/African American	99	1 in 486	1 in 49,000	1 in 200,000
		Latino/Admixed American	99	1 in 456	1 in 46,000	1 in 180,000
		Ashkenazi Jewish	99	1 in 5,040	1 in 500,000	1 in 2,000,000
		East Asian	98	1 in 432	1 in 22,000	1 in 88,000
		Finnish	99	1 in 3,636	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 310	1 in 31,000	1 in 120,000
		South Asian	99	1 in 245	1 in 24,000	1 in 96,000
		Other (population not assigned)	99	1 in 435	1 in 43,000	1 in 170,000
PHKG2	Glycogen storage disease type IX	African/African American	97	1 in 176	1 in 5,800	1 in 23,000
		Latino/Admixed American	98	1 in 393	1 in 20,000	1 in 80,000
		Ashkenazi Jewish	97	1 in 2,514	1 in 84,000	1 in 340,000
		East Asian	98	1 in 657	1 in 33,000	1 in 130,000
		Finnish	97	1 in 375	1 in 12,000	1 in 48,000
		Non-Finnish European/Caucasian	98	1 in 476	1 in 24,000	1 in 96,000
		South Asian	98	1 in 657	1 in 33,000	1 in 130,000
		Other (population not assigned)	97	1 in 437	1 in 15,000	1 in 60,000
PKHD1	Polycystic kidney disease, autosomal recessive	African/African American	99	1 in 71	1 in 7,000	1 in 28,000
		Latino/Admixed American	99	1 in 61	1 in 6,000	1 in 24,000
		Ashkenazi Jewish	98	1 in 33	1 in 1,600	1 in 6,400
		East Asian	99	1 in 193	1 in 19,000	1 in 76,000
		Finnish	99	1 in 25	1 in 2,400	1 in 9,600
		Non-Finnish European/Caucasian	99	1 in 31	1 in 3,000	1 in 12,000
		South Asian	99	1 in 127	1 in 13,000	1 in 52,000

Footnotes:

\*\* includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

## Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Other (population not assigned)	99	1 in 34	1 in 3,300	1 in 13,000
<b>PLA2G6</b>	Neurodegeneration with brain iron accumulation disorder	African/African American	99	1 in 214	1 in 21,000	1 in 84,000
		Latino/Admixed American	99	1 in 530	1 in 53,000	1 in 210,000
		Ashkenazi Jewish	99	1 in 254	1 in 25,000	1 in 100,000
		East Asian	99	1 in 251	1 in 25,000	1 in 100,000
		Finnish	99	1 in 2,725	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 343	1 in 34,000	1 in 140,000
		South Asian	99	1 in 60	1 in 5,900	1 in 24,000
		Other (population not assigned)	99	1 in 242	1 in 24,000	1 in 96,000
<b>PLEKHG5</b>	Distal spinal muscular atrophy, autosomal recessive	African/African American	97	1 in 45	1 in 1,500	1 in 6,000
		Latino/Admixed American	98	1 in 250	1 in 12,000	1 in 48,000
		Ashkenazi Jewish	97	1 in 2,014	1 in 67,000	1 in 270,000
		East Asian	99	1 in 614	1 in 61,000	1 in 240,000
		Finnish	97	1 in 1,252	1 in 42,000	1 in 170,000
		Non-Finnish European/Caucasian	99	1 in 693	1 in 69,000	1 in 280,000
		South Asian	99	1 in 433	1 in 43,000	1 in 170,000
		Other (population not assigned)	97	1 in 168	1 in 5,600	1 in 22,000
<b>PLOD2</b>	Osteogenesis imperfecta, autosomal recessive	African/African American	97	1 in 54	1 in 1,800	1 in 7,200
		Latino/Admixed American	97	1 in 504	1 in 17,000	1 in 68,000
		Ashkenazi Jewish	97	1 in 4,573	1 in 150,000	1 in 600,000
		East Asian	98	1 in 2,150	1 in 110,000	1 in 440,000
		Finnish	99	1 in 251	1 in 25,000	1 in 100,000
		Non-Finnish European/Caucasian	99	1 in 1,400	1 in 140,000	1 in 560,000
		South Asian	98	1 in 842	1 in 42,000	1 in 170,000
		Other (population not assigned)	98	1 in 456	1 in 23,000	1 in 92,000
<b>PMM2</b>	Congenital disorders of glycosylation type 1	African/African American	99	1 in 186	1 in 19,000	1 in 76,000
		Latino/Admixed American	99	1 in 102	1 in 10,000	1 in 40,000
		Ashkenazi Jewish	99	1 in 66	1 in 6,500	1 in 26,000
		East Asian	99	1 in 142	1 in 14,000	1 in 56,000
		Finnish	99	1 in 59	1 in 5,800	1 in 23,000
		Non-Finnish European/Caucasian	99	1 in 59	1 in 5,800	1 in 23,000
		South Asian	99	1 in 246	1 in 25,000	1 in 100,000
		Other (population not assigned)	99	1 in 102	1 in 10,000	1 in 40,000
<b>PNP</b>	Severe combined immunodeficiency (SCID)	African/African American	99	1 in 7,657	1 in 770,000	1 in 3,100,000
		Latino/Admixed American	98	1 in 2,767	1 in 140,000	1 in 560,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 4,312	1 in 430,000	1 in 1,700,000
		Finnish	97	1 in 10,817	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 1,145	1 in 110,000	1 in 440,000
		South Asian	98	1 in 918	1 in 46,000	1 in 180,000
		Other (population not assigned)	99	1 in 723	1 in 72,000	1 in 290,000
<b>PNPLA1</b>	Autosomal recessive congenital ichthyosis (ARCI)	African/African American	97	1 in 1,352	1 in 45,000	1 in 180,000
		Latino/Admixed American	99	1 in 1,801	1 in 180,000	1 in 720,000

Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

## Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Ashkenazi Jewish	97	1 in 4,365	1 in 150,000	1 in 600,000
		East Asian	99	1 in 511	1 in 51,000	1 in 200,000
		Finnish	99	1 in 10,806	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	99	1 in 1,456	1 in 150,000	1 in 600,000
		South Asian	97	1 in 3,532	1 in 120,000	1 in 480,000
		Other (population not assigned)	99	1 in 577	1 in 58,000	1 in 230,000
PNPO	Pyridoxal 5'-phosphate-dependent epilepsy	African/African American	98	1 in 386	1 in 19,000	1 in 76,000
		Latino/Admixed American	99	1 in 1,846	1 in 180,000	1 in 720,000
		Ashkenazi Jewish	99	1 in 5,039	1 in 500,000	1 in 2,000,000
		East Asian	97	1 in 1,484	1 in 49,000	1 in 200,000
		Finnish	99	1 in 1,193	1 in 120,000	1 in 480,000
		Non-Finnish European/Caucasian	99	1 in 1,107	1 in 110,000	1 in 440,000
		South Asian	99	1 in 1,920	1 in 190,000	1 in 760,000
		Other (population not assigned)	99	1 in 1,449	1 in 140,000	1 in 560,000
POLH	Xeroderma pigmentosum	African/African American	98	1 in 669	1 in 33,000	1 in 130,000
		Latino/Admixed American	98	1 in 1,003	1 in 50,000	1 in 200,000
		Ashkenazi Jewish	99	1 in 4,915	1 in 490,000	1 in 2,000,000
		East Asian	99	1 in 49	1 in 4,800	1 in 19,000
		Finnish	99	1 in 3,654	1 in 370,000	1 in 1,500,000
		Non-Finnish European/Caucasian	99	1 in 1,002	1 in 100,000	1 in 400,000
		South Asian	99	1 in 393	1 in 39,000	1 in 160,000
		Other (population not assigned)	99	1 in 914	1 in 91,000	1 in 360,000
POMGNT1	Limb-girdle muscular dystrophy, autosomal recessive	African/African American	99	1 in 771	1 in 77,000	1 in 310,000
		Latino/Admixed American	99	1 in 486	1 in 49,000	1 in 200,000
		Ashkenazi Jewish	99	1 in 2,449	1 in 240,000	1 in 960,000
		East Asian	98	1 in 587	1 in 29,000	1 in 120,000
		Finnish	99	1 in 198	1 in 20,000	1 in 80,000
		Non-Finnish European/Caucasian	99	1 in 303	1 in 30,000	1 in 120,000
		South Asian	99	1 in 539	1 in 54,000	1 in 220,000
		Other (population not assigned)	99	1 in 913	1 in 91,000	1 in 360,000
POMT1	Limb-girdle muscular dystrophy, autosomal recessive	African/African American	99	1 in 535	1 in 53,000	1 in 210,000
		Latino/Admixed American	99	1 in 390	1 in 39,000	1 in 160,000
		Ashkenazi Jewish	97	1 in 307	1 in 10,000	1 in 40,000
		East Asian	99	1 in 458	1 in 46,000	1 in 180,000
		Finnish	99	1 in 1,846	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	98	1 in 372	1 in 19,000	1 in 76,000
		South Asian	99	1 in 427	1 in 43,000	1 in 170,000
		Other (population not assigned)	99	1 in 264	1 in 26,000	1 in 100,000
POMT2	Limb-girdle muscular dystrophy, autosomal recessive	African/African American	98	1 in 267	1 in 13,000	1 in 52,000
		Latino/Admixed American	99	1 in 908	1 in 91,000	1 in 360,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 1,274	1 in 130,000	1 in 520,000
		Finnish	98	1 in 3,607	1 in 180,000	1 in 720,000

Footnotes:

\*\* includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

## Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Non-Finnish European/Caucasian	99	1 in 833	1 in 83,000	1 in 330,000
		South Asian	99	1 in 833	1 in 83,000	1 in 330,000
		Other (population not assigned)	97	1 in 1,448	1 in 48,000	1 in 190,000
POR	Congenital adrenal hyperplasia	African/African American	99	1 in 563	1 in 56,000	1 in 220,000
		Latino/Admixed American	98	1 in 278	1 in 14,000	1 in 56,000
		Ashkenazi Jewish	99	1 in 321	1 in 32,000	1 in 130,000
		East Asian	98	1 in 105	1 in 5,200	1 in 21,000
		Finnish	98	1 in 177	1 in 8,800	1 in 35,000
		Non-Finnish European/Caucasian	99	1 in 268	1 in 27,000	1 in 110,000
		South Asian	98	1 in 417	1 in 21,000	1 in 84,000
		Other (population not assigned)	98	1 in 185	1 in 9,200	1 in 37,000
		POU3F4	Deafness and hearing loss, nonsyndromic	African/African American	99	N/A
Latino/Admixed American	99			N/A	N/A	N/A
Ashkenazi Jewish	99			N/A	N/A	N/A
East Asian	99			N/A	N/A	N/A
Finnish	99			N/A	N/A	N/A
Non-Finnish European/Caucasian	99			N/A	N/A	N/A
South Asian	99			N/A	N/A	N/A
Other (population not assigned)	99			N/A	N/A	N/A
PPIB	Osteogenesis imperfecta, autosomal recessive	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	98	1 in 1,722	1 in 86,000	1 in 340,000
		Ashkenazi Jewish	97	1 in 5,041	1 in 170,000	1 in 680,000
		East Asian	99	1 in 2,875	1 in 290,000	1 in 1,200,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 2,688	1 in 270,000	1 in 1,100,000
		South Asian	99	1 in 3,061	1 in 310,000	1 in 1,200,000
		Other (population not assigned)	99	1 in 984	1 in 98,000	1 in 390,000
PPT1	Neuronal ceroid-lipofuscinosis	African/African American	99	1 in 856	1 in 86,000	1 in 340,000
		Latino/Admixed American	99	1 in 4,195	1 in 420,000	1 in 1,700,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	1 in 8,621	1 in 860,000	1 in 3,400,000
		Finnish	99	1 in 78	1 in 7,700	1 in 31,000
		Non-Finnish European/Caucasian	99	1 in 350	1 in 35,000	1 in 140,000
		South Asian	99	1 in 1,550	1 in 150,000	1 in 600,000
		Other (population not assigned)	99	1 in 228	1 in 23,000	1 in 92,000
PRCD	Retinitis pigmentosa	African/African American	99	1 in 3,673	1 in 370,000	1 in 1,500,000
		Latino/Admixed American	98	1 in 3,989	1 in 200,000	1 in 800,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 2,738	1 in 270,000	1 in 1,100,000
		Finnish	99	1 in 1,276	1 in 130,000	1 in 520,000
		Non-Finnish European/Caucasian	98	1 in 627	1 in 31,000	1 in 120,000
		South Asian	99	1 in 3,359	1 in 340,000	1 in 1,400,000
		Other (population not assigned)	98	N/A	N/A	N/A

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PRDM5	Brittle cornea syndrome	African/African American	98	1 in 1,625	1 in 81,000	1 in 320,000
		Latino/Admixed American	99	1 in 631	1 in 63,000	1 in 250,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 1,078	1 in 110,000	1 in 440,000
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 1,952	1 in 200,000	1 in 800,000
		South Asian	99	1 in 3,061	1 in 310,000	1 in 1,200,000
		Other (population not assigned)	99	1 in 2,111	1 in 210,000	1 in 840,000
PRF1	Familial hemophagocytic lymphohistiocytosis	African/African American	97	1 in 126	1 in 4,200	1 in 17,000
		Latino/Admixed American	99	1 in 429	1 in 43,000	1 in 170,000
		Ashkenazi Jewish	99	1 in 458	1 in 46,000	1 in 180,000
		East Asian	97	1 in 1,099	1 in 37,000	1 in 150,000
		Finnish	97	1 in 10,775	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 308	1 in 31,000	1 in 120,000
		South Asian	99	1 in 226	1 in 23,000	1 in 92,000
		Other (population not assigned)	99	1 in 320	1 in 32,000	1 in 130,000
PRKDC	Severe combined immunodeficiency (SCID)	African/African American	98	1 in 545	1 in 27,000	1 in 110,000
		Latino/Admixed American	97	1 in 609	1 in 20,000	1 in 80,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 1,140	1 in 57,000	1 in 230,000
		Finnish	99	1 in 477	1 in 48,000	1 in 190,000
		Non-Finnish European/Caucasian	98	1 in 1,716	1 in 86,000	1 in 340,000
		South Asian	97	1 in 1,165	1 in 39,000	1 in 160,000
		Other (population not assigned)	97	1 in 739	1 in 25,000	1 in 100,000
PSAP	Metachromatic leukodystrophy	African/African American	99	1 in 2,030	1 in 200,000	1 in 800,000
		Latino/Admixed American	99	1 in 884	1 in 88,000	1 in 350,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 2,998	1 in 300,000	1 in 1,200,000
		South Asian	99	1 in 15,385	1 in 1,500,000	1 in 6,000,000
		Other (population not assigned)	99	N/A	N/A	N/A
PTPRC	Severe combined immunodeficiency (SCID)	African/African American	99	1 in 1,304	1 in 130,000	1 in 520,000
		Latino/Admixed American	98	1 in 906	1 in 45,000	1 in 180,000
		Ashkenazi Jewish	99	1 in 2,171	1 in 220,000	1 in 880,000
		East Asian	99	1 in 1,022	1 in 100,000	1 in 400,000
		Finnish	98	1 in 3,606	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	98	1 in 3,195	1 in 160,000	1 in 640,000
		South Asian	97	1 in 1,229	1 in 41,000	1 in 160,000
		Other (population not assigned)	97	1 in 3,044	1 in 100,000	1 in 400,000
PTS	Tetrahydrobiopterin deficiency	African/African American	97	1 in 81	1 in 2,700	1 in 11,000
		Latino/Admixed American	98	1 in 337	1 in 17,000	1 in 68,000
		Ashkenazi Jewish	99	1 in 64	1 in 6,300	1 in 25,000

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		East Asian	99	1 in 152	1 in 15,000	1 in 60,000		
		Finnish	99	1 in 470	1 in 47,000	1 in 190,000		
		Non-Finnish European/Caucasian	98	1 in 532	1 in 27,000	1 in 110,000		
		South Asian	98	1 in 254	1 in 13,000	1 in 52,000		
		Other (population not assigned)	99	1 in 251	1 in 25,000	1 in 100,000		
		PUS1	Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia	African/African American	99	1 in 1,297	1 in 130,000	1 in 520,000
				Latino/Admixed American	98	1 in 1,468	1 in 73,000	1 in 290,000
				Ashkenazi Jewish	97	1 in 1,129	1 in 38,000	1 in 150,000
				East Asian	99	1 in 8,209	1 in 820,000	1 in 3,300,000
				Finnish	98	1 in 1,401	1 in 70,000	1 in 280,000
Non-Finnish European/Caucasian	98			1 in 1,551	1 in 78,000	1 in 310,000		
South Asian	99			1 in 178	1 in 18,000	1 in 72,000		
Other (population not assigned)	99			1 in 3,063	1 in 310,000	1 in 1,200,000		
PYCR1	Cutis laxa			African/African American	99	1 in 459	1 in 46,000	1 in 180,000
				Latino/Admixed American	99	1 in 1,266	1 in 130,000	1 in 520,000
		Ashkenazi Jewish	99	1 in 613	1 in 61,000	1 in 240,000		
		East Asian	98	1 in 299	1 in 15,000	1 in 60,000		
		Finnish	99	1 in 3,202	1 in 320,000	1 in 1,300,000		
		Non-Finnish European/Caucasian	99	1 in 401	1 in 40,000	1 in 160,000		
		South Asian	99	1 in 191	1 in 19,000	1 in 76,000		
		Other (population not assigned)	99	1 in 590	1 in 59,000	1 in 240,000		
		PYGM	Glycogen storage disease type V	African/African American	99	1 in 160	1 in 16,000	1 in 64,000
				Latino/Admixed American	99	1 in 48	1 in 4,700	1 in 19,000
Ashkenazi Jewish	99			1 in 197	1 in 20,000	1 in 80,000		
East Asian	98			1 in 553	1 in 28,000	1 in 110,000		
Finnish	99			1 in 285	1 in 28,000	1 in 110,000		
Non-Finnish European/Caucasian	99			1 in 42	1 in 4,100	1 in 16,000		
South Asian	99			1 in 272	1 in 27,000	1 in 110,000		
Other (population not assigned)	99			1 in 72	1 in 7,100	1 in 28,000		
QDPR	Tetrahydrobiopterin deficiency			African/African American	99	1 in 8,124	1 in 810,000	1 in 3,200,000
				Latino/Admixed American	99	1 in 5,766	1 in 580,000	1 in 2,300,000
		Ashkenazi Jewish	99	N/A	N/A	N/A		
		East Asian	99	1 in 9,197	1 in 920,000	1 in 3,700,000		
		Finnish	99	N/A	N/A	N/A		
		Non-Finnish European/Caucasian	99	1 in 2,419	1 in 240,000	1 in 960,000		
		South Asian	99	1 in 2,641	1 in 260,000	1 in 1,000,000		
		Other (population not assigned)	99	N/A	N/A	N/A		

Footnotes:

\*\* includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

a Feng et al., PMID 28125085

b Luo et al., PMID 23788250