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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
AAAS	Triple A syndrome	African/African American	98	1 in 221	1 in 11,000	1 in 44,000
		Latino/Admixed American	98	1 in 549	1 in 27,000	1 in 110,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	98	1 in 1,313	1 in 66,000	1 in 260,000
		Finnish	99	1 in 618	1 in 62,000	1 in 250,000
		Non-Finnish European/Caucasian	99	1 in 436	1 in 44,000	1 in 180,000
		South Asian	99	1 in 365	1 in 36,000	1 in 140,000
		Other (population not assigned)	98	1 in 1,534	1 in 77,000	1 in 310,000
ABCA12	Autosomal recessive congenital ichthyosis (ARCI)	African/African American	97	1 in 112	1 in 3,700	1 in 15,000
		Latino/Admixed American	98	1 in 271	1 in 14,000	1 in 56,000
		Ashkenazi Jewish	99	1 in 339	1 in 34,000	1 in 140,000
		East Asian	97	1 in 223	1 in 7,400	1 in 30,000
		Finnish	97	1 in 978	1 in 33,000	1 in 130,000
		Non-Finnish European/Caucasian	97	1 in 194	1 in 6,400	1 in 26,000
		South Asian	97	1 in 89	1 in 2,900	1 in 12,000
		Other (population not assigned)	98	1 in 118	1 in 5,900	1 in 24,000
ABCB11	Progressive familial intrahepatic cholestasis	African/African American	99	1 in 510	1 in 51,000	1 in 200,000
		Latino/Admixed American	99	1 in 512	1 in 51,000	1 in 200,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	1 in 951	1 in 95,000	1 in 380,000
		Finnish	99	1 in 1,180	1 in 120,000	1 in 480,000
		Non-Finnish European/Caucasian	99	1 in 476	1 in 48,000	1 in 190,000
		South Asian	99	1 in 774	1 in 77,000	1 in 310,000
		Other (population not assigned)	99	1 in 317	1 in 32,000	1 in 130,000
ABCB4	Progressive familial intrahepatic cholestasis	African/African American	99	1 in 76	1 in 7,500	1 in 30,000
		Latino/Admixed American	99	1 in 75	1 in 7,400	1 in 30,000
		Ashkenazi Jewish	98	1 in 367	1 in 18,000	1 in 72,000
		East Asian	98	1 in 673	1 in 34,000	1 in 140,000
		Finnish	98	1 in 110	1 in 5,500	1 in 22,000
		Non-Finnish European/Caucasian	99	1 in 60	1 in 5,900	1 in 24,000
		South Asian	98	1 in 113	1 in 5,600	1 in 22,000
		Other (population not assigned)	99	1 in 70	1 in 6,900	1 in 28,000
ABCC8	Familial hyperinsulinism	African/African American	99	1 in 642	1 in 64,000	1 in 260,000
		Latino/Admixed American	99	1 in 342	1 in 34,000	1 in 140,000
		Ashkenazi Jewish	99	1 in 79	1 in 7,800	1 in 31,000
		East Asian	99	1 in 377	1 in 38,000	1 in 150,000
		Finnish	99	1 in 228	1 in 23,000	1 in 92,000
		Non-Finnish European/Caucasian	99	1 in 315	1 in 31,000	1 in 120,000
		South Asian	99	1 in 503	1 in 50,000	1 in 200,000
		Other (population not assigned)	99	1 in 355	1 in 35,000	1 in 140,000
ABCD1	Adrenoleukodystrophy, X-linked	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

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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	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
ABCD4	Methylmalonic acidemia with homocystinuria	African/African American	98	1 in 859	1 in 43,000	1 in 170,000
		Latino/Admixed American	99	1 in 534	1 in 53,000	1 in 210,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 328	1 in 33,000	1 in 130,000
		Finnish	99	1 in 901	1 in 90,000	1 in 360,000
		Non-Finnish European/Caucasian	99	1 in 496	1 in 50,000	1 in 200,000
		South Asian	99	1 in 313	1 in 31,000	1 in 120,000
		Other (population not assigned)	99	1 in 304	1 in 30,000	1 in 120,000
ACAD9	Mitochondrial complex I deficiency	African/African American	97	1 in 67	1 in 2,200	1 in 8,800
		Latino/Admixed American	97	1 in 118	1 in 3,900	1 in 16,000
		Ashkenazi Jewish	98	1 in 356	1 in 18,000	1 in 72,000
		East Asian	97	1 in 1,382	1 in 46,000	1 in 180,000
		Finnish	97	1 in 306	1 in 10,000	1 in 40,000
		Non-Finnish European/Caucasian	97	1 in 67	1 in 2,200	1 in 8,800
		South Asian	97	1 in 110	1 in 3,600	1 in 14,000
		Other (population not assigned)	97	1 in 85	1 in 2,800	1 in 11,000
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	African/African American	99	1 in 155	1 in 15,000	1 in 60,000
		Latino/Admixed American	99	1 in 71	1 in 7,000	1 in 28,000
		Ashkenazi Jewish	99	1 in 57	1 in 5,600	1 in 22,000
		East Asian	98	1 in 130	1 in 6,500	1 in 26,000
		Finnish	99	1 in 134	1 in 13,000	1 in 52,000
		Non-Finnish European/Caucasian	99	1 in 41	1 in 4,000	1 in 16,000
		South Asian	99	1 in 143	1 in 14,000	1 in 56,000
		Other (population not assigned)	99	1 in 45	1 in 4,400	1 in 18,000
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	African/African American	99	1 in 212	1 in 21,000	1 in 84,000
		Latino/Admixed American	99	1 in 291	1 in 29,000	1 in 120,000
		Ashkenazi Jewish	99	1 in 1,238	1 in 120,000	1 in 480,000
		East Asian	99	1 in 432	1 in 43,000	1 in 170,000
		Finnish	99	1 in 348	1 in 35,000	1 in 140,000
		Non-Finnish European/Caucasian	99	1 in 123	1 in 12,000	1 in 48,000
		South Asian	99	1 in 371	1 in 37,000	1 in 150,000
		Other (population not assigned)	99	1 in 124	1 in 12,000	1 in 48,000
ACAT1	Beta-ketothiolase deficiency	African/African American	99	1 in 1,141	1 in 110,000	1 in 440,000
		Latino/Admixed American	99	1 in 167	1 in 17,000	1 in 68,000
		Ashkenazi Jewish	97	1 in 5,040	1 in 170,000	1 in 680,000
		East Asian	98	1 in 413	1 in 21,000	1 in 84,000
		Finnish	98	1 in 5,412	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 1,002	1 in 100,000	1 in 400,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
		South Asian	99	1 in 1,504	1 in 150,000	1 in 600,000
		Other (population not assigned)	99	1 in 522	1 in 52,000	1 in 210,000
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	African/African American	98	1 in 866	1 in 43,000	1 in 170,000
		Latino/Admixed American	98	1 in 1,909	1 in 95,000	1 in 380,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 9,197	1 in 920,000	1 in 3,700,000
		Finnish	99	1 in 2,706	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 504	1 in 50,000	1 in 200,000
		South Asian	97	1 in 1,177	1 in 39,000	1 in 160,000
		Other (population not assigned)	97	1 in 3,069	1 in 100,000	1 in 400,000
ACSF3	Combined malonic and methylmalonic aciduria	African/African American	99	1 in 132	1 in 13,000	1 in 52,000
		Latino/Admixed American	99	1 in 129	1 in 13,000	1 in 52,000
		Ashkenazi Jewish	99	1 in 379	1 in 38,000	1 in 150,000
		East Asian	99	1 in 274	1 in 27,000	1 in 110,000
		Finnish	99	1 in 340	1 in 34,000	1 in 140,000
		Non-Finnish European/Caucasian	99	1 in 67	1 in 6,600	1 in 26,000
		South Asian	99	1 in 61	1 in 6,000	1 in 24,000
		Other (population not assigned)	99	1 in 83	1 in 8,200	1 in 33,000
ADA	Adenosine deaminase deficiency	African/African American	99	1 in 126	1 in 13,000	1 in 52,000
		Latino/Admixed American	99	1 in 247	1 in 25,000	1 in 100,000
		Ashkenazi Jewish	99	1 in 55	1 in 5,400	1 in 22,000
		East Asian	99	1 in 211	1 in 21,000	1 in 84,000
		Finnish	99	1 in 942	1 in 94,000	1 in 380,000
		Non-Finnish European/Caucasian	99	1 in 285	1 in 28,000	1 in 110,000
		South Asian	99	1 in 143	1 in 14,000	1 in 56,000
		Other (population not assigned)	99	1 in 135	1 in 13,000	1 in 52,000
ADAMTS2	Ehlers-Danlos syndrome type VIIC	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	1 in 2,434	1 in 240,000	1 in 960,000
		Ashkenazi Jewish	99	1 in 164	1 in 16,000	1 in 64,000
		East Asian	99	1 in 1,502	1 in 150,000	1 in 600,000
		Finnish	99	1 in 3,510	1 in 350,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 2,017	1 in 200,000	1 in 800,000
		South Asian	99	1 in 1,247	1 in 120,000	1 in 480,000
		Other (population not assigned)	99	1 in 908	1 in 91,000	1 in 360,000
ADGRV1	Usher syndrome (hearing loss and retinitis pigmentosa)	African/African American	98	1 in 64	1 in 3,200	1 in 13,000
		Latino/Admixed American	99	1 in 72	1 in 7,100	1 in 28,000
		Ashkenazi Jewish	99	1 in 113	1 in 11,000	1 in 44,000
		East Asian	98	1 in 134	1 in 6,700	1 in 27,000
		Finnish	99	1 in 52	1 in 5,100	1 in 20,000
		Non-Finnish European/Caucasian	99	1 in 35	1 in 3,400	1 in 14,000
		South Asian	99	1 in 30	1 in 2,900	1 in 12,000
		Other (population not assigned)	99	1 in 49	1 in 4,800	1 in 19,000
AGA	Aspartylglucosaminuria	African/African American	99	1 in 118	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

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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	99	1 in 257	1 in 26,000	1 in 100,000
		Ashkenazi Jewish	99	1 in 630	1 in 63,000	1 in 250,000
		East Asian	99	1 in 1,768	1 in 180,000	1 in 720,000
		Finnish	99	1 in 60	1 in 5,900	1 in 24,000
		Non-Finnish European/Caucasian	99	1 in 396	1 in 40,000	1 in 160,000
		South Asian	99	1 in 464	1 in 46,000	1 in 180,000
		Other (population not assigned)	99	1 in 155	1 in 15,000	1 in 60,000
AGL	Glycogen storage disease type III	African/African American	99	1 in 251	1 in 25,000	1 in 100,000
		Latino/Admixed American	98	1 in 486	1 in 24,000	1 in 96,000
		Ashkenazi Jewish	97	1 in 5,015	1 in 170,000	1 in 680,000
		East Asian	99	1 in 629	1 in 63,000	1 in 250,000
		Finnish	99	1 in 1,585	1 in 160,000	1 in 640,000
		Non-Finnish European/Caucasian	99	1 in 353	1 in 35,000	1 in 140,000
		South Asian	98	1 in 424	1 in 21,000	1 in 84,000
		Other (population not assigned)	99	1 in 347	1 in 35,000	1 in 140,000
AGPAT2	Congenital generalized lipodystrophy	African/African American	99	1 in 97	1 in 9,600	1 in 38,000
		Latino/Admixed American	99	1 in 1,179	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	99	1 in 1,864	1 in 190,000	1 in 760,000
		East Asian	99	1 in 391	1 in 39,000	1 in 160,000
		Finnish	99	1 in 10,817	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	99	1 in 2,032	1 in 200,000	1 in 800,000
		South Asian	99	1 in 956	1 in 96,000	1 in 380,000
		Other (population not assigned)	99	1 in 993	1 in 99,000	1 in 400,000
AGPS	Rhizomelic chondrodysplasia punctata	African/African American	99	1 in 260	1 in 26,000	1 in 100,000
		Latino/Admixed American	99	1 in 151	1 in 15,000	1 in 60,000
		Ashkenazi Jewish	99	1 in 94	1 in 9,300	1 in 37,000
		East Asian	99	1 in 165	1 in 16,000	1 in 64,000
		Finnish	99	1 in 177	1 in 18,000	1 in 72,000
		Non-Finnish European/Caucasian	99	1 in 106	1 in 11,000	1 in 44,000
		South Asian	99	1 in 59	1 in 5,800	1 in 23,000
		Other (population not assigned)	99	1 in 65	1 in 6,400	1 in 26,000
AGXT	Primary hyperoxaluria	African/African American	99	1 in 528	1 in 53,000	1 in 210,000
		Latino/Admixed American	99	1 in 451	1 in 45,000	1 in 180,000
		Ashkenazi Jewish	99	1 in 1,595	1 in 160,000	1 in 640,000
		East Asian	99	1 in 170	1 in 17,000	1 in 68,000
		Finnish	99	1 in 1,963	1 in 200,000	1 in 800,000
		Non-Finnish European/Caucasian	99	1 in 254	1 in 25,000	1 in 100,000
		South Asian	99	1 in 235	1 in 23,000	1 in 92,000
		Other (population not assigned)	99	1 in 289	1 in 29,000	1 in 120,000
AHI1	Joubert syndrome and related disorders, including Meckel-Gruber syndrome	African/African American	99	1 in 172	1 in 17,000	1 in 68,000
		Latino/Admixed American	99	1 in 151	1 in 15,000	1 in 60,000
		Ashkenazi Jewish	99	1 in 166	1 in 17,000	1 in 68,000
		East Asian	98	1 in 255	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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
		Finnish	99	1 in 85	1 in 8,400	1 in 34,000
		Non-Finnish European/Caucasian	99	1 in 48	1 in 4,700	1 in 19,000
		South Asian	98	1 in 63	1 in 3,100	1 in 12,000
		Other (population not assigned)	99	1 in 64	1 in 6,300	1 in 25,000
AIPL1	Leber congenital amaurosis	African/African American	98	1 in 524	1 in 26,000	1 in 100,000
		Latino/Admixed American	99	1 in 101	1 in 10,000	1 in 40,000
		Ashkenazi Jewish	98	1 in 30	1 in 1,500	1 in 6,000
		East Asian	99	1 in 1,149	1 in 110,000	1 in 440,000
		Finnish	97	1 in 1,811	1 in 60,000	1 in 240,000
		Non-Finnish European/Caucasian	99	1 in 160	1 in 16,000	1 in 64,000
		South Asian	98	1 in 31	1 in 1,500	1 in 6,000
		Other (population not assigned)	99	1 in 63	1 in 6,200	1 in 25,000
AIRE	Autoimmune polyglandular syndrome type 1	African/African American	99	1 in 482	1 in 48,000	1 in 190,000
		Latino/Admixed American	97	1 in 430	1 in 14,000	1 in 56,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 106	1 in 11,000	1 in 44,000
		Finnish	99	1 in 92	1 in 9,100	1 in 36,000
		Non-Finnish European/Caucasian	98	1 in 180	1 in 9,000	1 in 36,000
		South Asian	97	1 in 349	1 in 12,000	1 in 48,000
		Other (population not assigned)	98	1 in 161	1 in 8,000	1 in 32,000
AK2	Severe combined immunodeficiency (SCID)	African/African American	99	1 in 309	1 in 31,000	1 in 120,000
		Latino/Admixed American	99	1 in 393	1 in 39,000	1 in 160,000
		Ashkenazi Jewish	99	1 in 807	1 in 81,000	1 in 320,000
		East Asian	99	1 in 166	1 in 17,000	1 in 68,000
		Finnish	99	1 in 324	1 in 32,000	1 in 130,000
		Non-Finnish European/Caucasian	99	1 in 232	1 in 23,000	1 in 92,000
		South Asian	99	1 in 479	1 in 48,000	1 in 190,000
		Other (population not assigned)	99	1 in 284	1 in 28,000	1 in 110,000
ALDH3A2	Sjogren-Larsson syndrome	African/African American	98	1 in 1,121	1 in 56,000	1 in 220,000
		Latino/Admixed American	99	1 in 428	1 in 43,000	1 in 170,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 862	1 in 43,000	1 in 170,000
		Finnish	98	1 in 2,202	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	99	1 in 654	1 in 65,000	1 in 260,000
		South Asian	97	1 in 1,636	1 in 55,000	1 in 220,000
		Other (population not assigned)	99	1 in 2,740	1 in 270,000	1 in 1,100,000
ALDH7A1	Pyridoxine-dependent epilepsy	African/African American	98	1 in 245	1 in 12,000	1 in 48,000
		Latino/Admixed American	99	1 in 121	1 in 12,000	1 in 48,000
		Ashkenazi Jewish	99	1 in 309	1 in 31,000	1 in 120,000
		East Asian	99	1 in 310	1 in 31,000	1 in 120,000
		Finnish	99	1 in 1,295	1 in 130,000	1 in 520,000
		Non-Finnish European/Caucasian	99	1 in 88	1 in 8,700	1 in 35,000
		South Asian	99	1 in 321	1 in 32,000	1 in 130,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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 125	1 in 12,000	1 in 48,000
ALDOB	Hereditary fructose Intolerance	African/African American	99	1 in 336	1 in 34,000	1 in 140,000
		Latino/Admixed American	99	1 in 203	1 in 20,000	1 in 80,000
		Ashkenazi Jewish	99	1 in 137	1 in 14,000	1 in 56,000
		East Asian	98	1 in 676	1 in 34,000	1 in 140,000
		Finnish	99	1 in 95	1 in 9,400	1 in 38,000
		Non-Finnish European/Caucasian	99	1 in 80	1 in 7,900	1 in 32,000
		South Asian	99	1 in 394	1 in 39,000	1 in 160,000
		Other (population not assigned)	99	1 in 96	1 in 9,500	1 in 38,000
ALG1	Congenital disorders of glycosylation type 1	African/African American	98	1 in 90	1 in 4,500	1 in 18,000
		Latino/Admixed American	98	1 in 65	1 in 3,200	1 in 13,000
		Ashkenazi Jewish	98	1 in 66	1 in 3,300	1 in 13,000
		East Asian	98	1 in 121	1 in 6,000	1 in 24,000
		Finnish	98	1 in 71	1 in 3,500	1 in 14,000
		Non-Finnish European/Caucasian	98	1 in 87	1 in 4,300	1 in 17,000
		South Asian	98	1 in 57	1 in 2,800	1 in 11,000
		Other (population not assigned)	98	1 in 94	1 in 4,700	1 in 19,000
ALG6	Congenital disorders of glycosylation type 1	African/African American	98	1 in 318	1 in 16,000	1 in 64,000
		Latino/Admixed American	97	1 in 385	1 in 13,000	1 in 52,000
		Ashkenazi Jewish	98	1 in 1,065	1 in 53,000	1 in 210,000
		East Asian	97	1 in 430	1 in 14,000	1 in 56,000
		Finnish	98	1 in 735	1 in 37,000	1 in 150,000
		Non-Finnish European/Caucasian	99	1 in 274	1 in 27,000	1 in 110,000
		South Asian	98	1 in 571	1 in 29,000	1 in 120,000
		Other (population not assigned)	98	1 in 350	1 in 17,000	1 in 68,000
ALMS1	Alström syndrome	African/African American	98	1 in 124	1 in 6,200	1 in 25,000
		Latino/Admixed American	97	1 in 82	1 in 2,700	1 in 11,000
		Ashkenazi Jewish	98	1 in 683	1 in 34,000	1 in 140,000
		East Asian	97	1 in 66	1 in 2,200	1 in 8,800
		Finnish	98	1 in 635	1 in 32,000	1 in 130,000
		Non-Finnish European/Caucasian	98	1 in 232	1 in 12,000	1 in 48,000
		South Asian	98	1 in 265	1 in 13,000	1 in 52,000
		Other (population not assigned)	98	1 in 219	1 in 11,000	1 in 44,000
ALOX12B	Autosomal recessive congenital ichthyosis (ARCI)	African/African American	98	1 in 1,175	1 in 59,000	1 in 240,000
		Latino/Admixed American	99	1 in 1,213	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	98	1 in 1,641	1 in 82,000	1 in 330,000
		Finnish	98	1 in 833	1 in 42,000	1 in 170,000
		Non-Finnish European/Caucasian	98	1 in 475	1 in 24,000	1 in 96,000
		South Asian	98	1 in 574	1 in 29,000	1 in 120,000
		Other (population not assigned)	98	1 in 552	1 in 28,000	1 in 110,000
ALOXE3	Autosomal recessive congenital ichthyosis (ARCI)	African/African American	99	1 in 1,122	1 in 110,000	1 in 440,000
		Latino/Admixed American	99	1 in 674	1 in 67,000	1 in 270,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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	99	1 in 1,246	1 in 120,000	1 in 480,000
		East Asian	98	1 in 613	1 in 31,000	1 in 120,000
		Finnish	99	1 in 673	1 in 67,000	1 in 270,000
		Non-Finnish European/Caucasian	99	1 in 184	1 in 18,000	1 in 72,000
		South Asian	99	1 in 729	1 in 73,000	1 in 290,000
		Other (population not assigned)	99	1 in 374	1 in 37,000	1 in 150,000
ALPL	Hypophosphatasia	African/African American	98	1 in 956	1 in 48,000	1 in 190,000
		Latino/Admixed American	98	1 in 422	1 in 21,000	1 in 84,000
		Ashkenazi Jewish	99	1 in 827	1 in 83,000	1 in 330,000
		East Asian	99	1 in 145	1 in 14,000	1 in 56,000
		Finnish	99	1 in 29	1 in 2,800	1 in 11,000
		Non-Finnish European/Caucasian	99	1 in 135	1 in 13,000	1 in 52,000
		South Asian	99	1 in 959	1 in 96,000	1 in 380,000
		Other (population not assigned)	99	1 in 154	1 in 15,000	1 in 60,000
AMPD2	Pontocerebellar hypoplasia	African/African American	98	1 in 761	1 in 38,000	1 in 150,000
		Latino/Admixed American	97	1 in 400	1 in 13,000	1 in 52,000
		Ashkenazi Jewish	98	1 in 1,494	1 in 75,000	1 in 300,000
		East Asian	97	1 in 195	1 in 6,500	1 in 26,000
		Finnish	97	1 in 834	1 in 28,000	1 in 110,000
		Non-Finnish European/Caucasian	98	1 in 488	1 in 24,000	1 in 96,000
		South Asian	98	1 in 358	1 in 18,000	1 in 72,000
		Other (population not assigned)	97	1 in 445	1 in 15,000	1 in 60,000
AMT	Glycine encephalopathy	African/African American	98	1 in 847	1 in 42,000	1 in 170,000
		Latino/Admixed American	99	1 in 801	1 in 80,000	1 in 320,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 893	1 in 89,000	1 in 360,000
		Finnish	98	1 in 1,194	1 in 60,000	1 in 240,000
		Non-Finnish European/Caucasian	99	1 in 775	1 in 77,000	1 in 310,000
		South Asian	98	1 in 960	1 in 48,000	1 in 190,000
		Other (population not assigned)	99	1 in 486	1 in 49,000	1 in 200,000
ANTXR2	Hyaline fibromatosis syndrome	African/African American	97	1 in 390	1 in 13,000	1 in 52,000
		Latino/Admixed American	97	1 in 284	1 in 9,400	1 in 38,000
		Ashkenazi Jewish	97	1 in 471	1 in 16,000	1 in 64,000
		East Asian	97	1 in 224	1 in 7,400	1 in 30,000
		Finnish	97	1 in 2,086	1 in 70,000	1 in 280,000
		Non-Finnish European/Caucasian	97	1 in 315	1 in 10,000	1 in 40,000
		South Asian	97	1 in 322	1 in 11,000	1 in 44,000
		Other (population not assigned)	97	1 in 558	1 in 19,000	1 in 76,000
AP3B1	Hermansky-Pudlak syndrome	African/African American	99	1 in 1,599	1 in 160,000	1 in 640,000
		Latino/Admixed American	99	1 in 2,854	1 in 290,000	1 in 1,200,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	98	1 in 4,597	1 in 230,000	1 in 920,000
		Finnish	99	1 in 3,580	1 in 360,000	1 in 1,400,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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	98	1 in 3,747	1 in 190,000	1 in 760,000
		South Asian	98	1 in 1,700	1 in 85,000	1 in 340,000
		Other (population not assigned)	99	1 in 3,071	1 in 310,000	1 in 1,200,000
AP3D1	Hermansky-Pudlak syndrome	African/African American	99	1 in 376	1 in 38,000	1 in 150,000
		Latino/Admixed American	98	1 in 1,519	1 in 76,000	1 in 300,000
		Ashkenazi Jewish	99	1 in 2,464	1 in 250,000	1 in 1,000,000
		East Asian	99	1 in 1,468	1 in 150,000	1 in 600,000
		Finnish	98	1 in 1,014	1 in 51,000	1 in 200,000
		Non-Finnish European/Caucasian	99	1 in 2,302	1 in 230,000	1 in 920,000
		South Asian	99	1 in 3,758	1 in 380,000	1 in 1,500,000
		Other (population not assigned)	99	1 in 1,380	1 in 140,000	1 in 560,000
ARG1	Arginase deficiency	African/African American	99	1 in 775	1 in 77,000	1 in 310,000
		Latino/Admixed American	99	1 in 497	1 in 50,000	1 in 200,000
		Ashkenazi Jewish	99	1 in 556	1 in 56,000	1 in 220,000
		East Asian	98	1 in 4,596	1 in 230,000	1 in 920,000
		Finnish	97	1 in 10,810	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 2,559	1 in 260,000	1 in 1,000,000
		South Asian	97	1 in 7,655	1 in 260,000	1 in 1,000,000
		Other (population not assigned)	97	N/A	N/A	N/A
ARL13B	Joubert syndrome and related disorders, including Meckel-Gruber syndrome	African/African American	97	1 in 41	1 in 1,300	1 in 5,200
		Latino/Admixed American	97	1 in 45	1 in 1,500	1 in 6,000
		Ashkenazi Jewish	97	1 in 63	1 in 2,100	1 in 8,400
		East Asian	97	1 in 65	1 in 2,100	1 in 8,400
		Finnish	97	1 in 140	1 in 4,600	1 in 18,000
		Non-Finnish European/Caucasian	97	1 in 72	1 in 2,400	1 in 9,600
		South Asian	97	1 in 52	1 in 1,700	1 in 6,800
		Other (population not assigned)	97	1 in 52	1 in 1,700	1 in 6,800
ARL6	Bardet-Biedl syndrome	African/African American	99	1 in 4,032	1 in 400,000	1 in 1,600,000
		Latino/Admixed American	97	1 in 17,290	1 in 580,000	1 in 2,300,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 1,724	1 in 170,000	1 in 680,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 2,946	1 in 290,000	1 in 1,200,000
		South Asian	99	1 in 1,922	1 in 190,000	1 in 760,000
		Other (population not assigned)	99	1 in 2,736	1 in 270,000	1 in 1,100,000
ARSA	Metachromatic leukodystrophy	African/African American	99	1 in 336	1 in 34,000	1 in 140,000
		Latino/Admixed American	99	1 in 487	1 in 49,000	1 in 200,000
		Ashkenazi Jewish	99	1 in 808	1 in 81,000	1 in 320,000
		East Asian	99	1 in 152	1 in 15,000	1 in 60,000
		Finnish	99	1 in 206	1 in 21,000	1 in 84,000
		Non-Finnish European/Caucasian	99	1 in 154	1 in 15,000	1 in 60,000
		South Asian	99	1 in 262	1 in 26,000	1 in 100,000
		Other (population not assigned)	99	1 in 128	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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
ARSB	Mucopolysaccharidosis type VI	African/African American	97	1 in 268	1 in 8,900	1 in 36,000
		Latino/Admixed American	99	1 in 4,258	1 in 430,000	1 in 1,700,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 1,484	1 in 150,000	1 in 600,000
		Finnish	98	1 in 1,356	1 in 68,000	1 in 270,000
		Non-Finnish European/Caucasian	99	1 in 378	1 in 38,000	1 in 150,000
		South Asian	98	1 in 902	1 in 45,000	1 in 180,000
		Other (population not assigned)	97	1 in 1,533	1 in 51,000	1 in 200,000
ASL	Argininosuccinic aciduria	African/African American	99	1 in 585	1 in 58,000	1 in 230,000
		Latino/Admixed American	99	1 in 457	1 in 46,000	1 in 180,000
		Ashkenazi Jewish	99	1 in 548	1 in 55,000	1 in 220,000
		East Asian	99	1 in 1,309	1 in 130,000	1 in 520,000
		Finnish	99	1 in 94	1 in 9,300	1 in 37,000
		Non-Finnish European/Caucasian	99	1 in 150	1 in 15,000	1 in 60,000
		South Asian	99	1 in 583	1 in 58,000	1 in 230,000
		Other (population not assigned)	99	1 in 170	1 in 17,000	1 in 68,000
ASNS	Asparagine synthetase deficiency	African/African American	98	1 in 795	1 in 40,000	1 in 160,000
		Latino/Admixed American	97	1 in 16,915	1 in 560,000	1 in 2,200,000
		Ashkenazi Jewish	97	1 in 5,035	1 in 170,000	1 in 680,000
		East Asian	99	1 in 570	1 in 57,000	1 in 230,000
		Finnish	99	1 in 1,836	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 1,143	1 in 110,000	1 in 440,000
		South Asian	99	1 in 765	1 in 76,000	1 in 300,000
		Other (population not assigned)	99	1 in 1,526	1 in 150,000	1 in 600,000
ASPA	Canavan disease	African/African American	99	1 in 963	1 in 96,000	1 in 380,000
		Latino/Admixed American	99	1 in 890	1 in 89,000	1 in 360,000
		Ashkenazi Jewish	99	1 in 51	1 in 5,000	1 in 20,000
		East Asian	99	N/A	N/A	N/A
		Finnish	99	1 in 237	1 in 24,000	1 in 96,000
		Non-Finnish European/Caucasian	99	1 in 564	1 in 56,000	1 in 220,000
		South Asian	99	1 in 3,839	1 in 380,000	1 in 1,500,000
		Other (population not assigned)	99	1 in 271	1 in 27,000	1 in 110,000
ASS1	Citrullinemia	African/African American	99	1 in 347	1 in 35,000	1 in 140,000
		Latino/Admixed American	99	1 in 225	1 in 22,000	1 in 88,000
		Ashkenazi Jewish	99	1 in 717	1 in 72,000	1 in 290,000
		East Asian	98	1 in 427	1 in 21,000	1 in 84,000
		Finnish	99	1 in 1,813	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 284	1 in 28,000	1 in 110,000
		South Asian	99	1 in 122	1 in 12,000	1 in 48,000
		Other (population not assigned)	99	1 in 142	1 in 14,000	1 in 56,000
ATM	Ataxia-telangiectasia	African/African American	98	1 in 183	1 in 9,100	1 in 36,000
		Latino/Admixed American	98	1 in 216	1 in 11,000	1 in 44,000
		Ashkenazi Jewish	97	1 in 680	1 in 23,000	1 in 92,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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	98	1 in 219	1 in 11,000	1 in 44,000
		Finnish	98	1 in 375	1 in 19,000	1 in 76,000
		Non-Finnish European/Caucasian	99	1 in 162	1 in 16,000	1 in 64,000
		South Asian	98	1 in 201	1 in 10,000	1 in 40,000
		Other (population not assigned)	98	1 in 186	1 in 9,300	1 in 37,000
ATP13A2	Neurodegeneration with brain iron accumulation disorder	African/African American	99	1 in 92	1 in 9,100	1 in 36,000
		Latino/Admixed American	98	1 in 751	1 in 38,000	1 in 150,000
		Ashkenazi Jewish	97	1 in 181	1 in 6,000	1 in 24,000
		East Asian	99	1 in 449	1 in 45,000	1 in 180,000
		Finnish	99	1 in 725	1 in 72,000	1 in 290,000
		Non-Finnish European/Caucasian	99	1 in 221	1 in 22,000	1 in 88,000
		South Asian	98	1 in 915	1 in 46,000	1 in 180,000
		Other (population not assigned)	99	1 in 263	1 in 26,000	1 in 100,000
ATP6V0A2	Cutis laxa	African/African American	99	1 in 146	1 in 15,000	1 in 60,000
		Latino/Admixed American	98	1 in 832	1 in 42,000	1 in 170,000
		Ashkenazi Jewish	98	1 in 809	1 in 40,000	1 in 160,000
		East Asian	99	1 in 377	1 in 38,000	1 in 150,000
		Finnish	99	1 in 211	1 in 21,000	1 in 84,000
		Non-Finnish European/Caucasian	98	1 in 600	1 in 30,000	1 in 120,000
		South Asian	99	1 in 1,582	1 in 160,000	1 in 640,000
		Other (population not assigned)	99	1 in 339	1 in 34,000	1 in 140,000
ATP6V0A4	Renal tubular acidosis and deafness	African/African American	99	1 in 1,105	1 in 110,000	1 in 440,000
		Latino/Admixed American	99	1 in 280	1 in 28,000	1 in 110,000
		Ashkenazi Jewish	99	1 in 359	1 in 36,000	1 in 140,000
		East Asian	98	1 in 741	1 in 37,000	1 in 150,000
		Finnish	98	1 in 3,434	1 in 170,000	1 in 680,000
		Non-Finnish European/Caucasian	99	1 in 853	1 in 85,000	1 in 340,000
		South Asian	98	1 in 718	1 in 36,000	1 in 140,000
		Other (population not assigned)	99	1 in 705	1 in 70,000	1 in 280,000
ATP6V1B1	Renal tubular acidosis and deafness	African/African American	99	1 in 742	1 in 74,000	1 in 300,000
		Latino/Admixed American	99	1 in 1,219	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	97	1 in 5,032	1 in 170,000	1 in 680,000
		East Asian	98	1 in 851	1 in 43,000	1 in 170,000
		Finnish	99	1 in 4,324	1 in 430,000	1 in 1,700,000
		Non-Finnish European/Caucasian	99	1 in 815	1 in 81,000	1 in 320,000
		South Asian	98	1 in 1,017	1 in 51,000	1 in 200,000
		Other (population not assigned)	99	1 in 982	1 in 98,000	1 in 390,000
ATP6V1E1	Cutis laxa	African/African American	97	1 in 4,064	1 in 140,000	1 in 560,000
		Latino/Admixed American	99	1 in 5,245	1 in 520,000	1 in 2,100,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 2,435	1 in 120,000	1 in 480,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 28,370	1 in 2,800,000	1 in 11,000,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
		South Asian	97	1 in 7,627	1 in 250,000	1 in 1,000,000
		Other (population not assigned)	97	N/A	N/A	N/A
ATP7A	ATP7A-related copper transport disorders, includes Menkes syndrome	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
ATP7B	Wilson disease	African/African American	99	1 in 143	1 in 14,000	1 in 56,000
		Latino/Admixed American	99	1 in 61	1 in 6,000	1 in 24,000
		Ashkenazi Jewish	99	1 in 26	1 in 2,500	1 in 10,000
		East Asian	99	1 in 44	1 in 4,300	1 in 17,000
		Finnish	99	1 in 105	1 in 10,000	1 in 40,000
		Non-Finnish European/Caucasian	99	1 in 42	1 in 4,100	1 in 16,000
		South Asian	99	1 in 70	1 in 6,900	1 in 28,000
		Other (population not assigned)	99	1 in 48	1 in 4,700	1 in 19,000
ATP8B1	Progressive familial intrahepatic cholestasis	African/African American	99	1 in 1,567	1 in 160,000	1 in 640,000
		Latino/Admixed American	98	1 in 718	1 in 36,000	1 in 140,000
		Ashkenazi Jewish	99	1 in 4,925	1 in 490,000	1 in 2,000,000
		East Asian	99	1 in 1,127	1 in 110,000	1 in 440,000
		Finnish	97	1 in 2,724	1 in 91,000	1 in 360,000
		Non-Finnish European/Caucasian	99	1 in 738	1 in 74,000	1 in 300,000
		South Asian	99	1 in 1,679	1 in 170,000	1 in 680,000
		Other (population not assigned)	99	1 in 767	1 in 77,000	1 in 310,000
ATRX	Alpha-thalassemia X-linked intellectual disability syndrome	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	97	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	97	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
AVPR2	Nephrogenic diabetes insipidus	African/African American	98	N/A	N/A	N/A
		Latino/Admixed American	98	N/A	N/A	N/A
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	98	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
B9D1	Joubert syndrome and related disorders, including	African/African American	99	1 in 643	1 in 64,000	1 in 260,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
	Meckel-Gruber syndrome	Latino/Admixed American	99	1 in 690	1 in 69,000	1 in 280,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 1,146	1 in 38,000	1 in 150,000
		Finnish	97	1 in 266	1 in 8,800	1 in 35,000
		Non-Finnish European/Caucasian	99	1 in 571	1 in 57,000	1 in 230,000
		South Asian	98	1 in 1,108	1 in 55,000	1 in 220,000
		Other (population not assigned)	99	1 in 561	1 in 56,000	1 in 220,000
B9D2	Joubert syndrome and related disorders, including Meckel-Gruber syndrome	African/African American	97	1 in 2,710	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	98	1 in 446	1 in 22,000	1 in 88,000
		East Asian	99	1 in 9,185	1 in 920,000	1 in 3,700,000
		Finnish	98	1 in 5,059	1 in 250,000	1 in 1,000,000
		Non-Finnish European/Caucasian	99	1 in 4,005	1 in 400,000	1 in 1,600,000
		South Asian	98	1 in 956	1 in 48,000	1 in 190,000
		Other (population not assigned)	99	1 in 3,068	1 in 310,000	1 in 1,200,000
BBS1	Bardet-Biedl syndrome	African/African American	99	1 in 64	1 in 6,300	1 in 25,000
		Latino/Admixed American	99	1 in 254	1 in 25,000	1 in 100,000
		Ashkenazi Jewish	99	1 in 455	1 in 45,000	1 in 180,000
		East Asian	99	1 in 145	1 in 14,000	1 in 56,000
		Finnish	99	1 in 79	1 in 7,800	1 in 31,000
		Non-Finnish European/Caucasian	99	1 in 94	1 in 9,300	1 in 37,000
		South Asian	99	1 in 1,097	1 in 110,000	1 in 440,000
		Other (population not assigned)	99	1 in 90	1 in 8,900	1 in 36,000
BBS2	Bardet-Biedl syndrome	African/African American	98	1 in 891	1 in 45,000	1 in 180,000
		Latino/Admixed American	99	1 in 572	1 in 57,000	1 in 230,000
		Ashkenazi Jewish	99	1 in 130	1 in 13,000	1 in 52,000
		East Asian	99	1 in 160	1 in 16,000	1 in 64,000
		Finnish	99	1 in 462	1 in 46,000	1 in 180,000
		Non-Finnish European/Caucasian	99	1 in 351	1 in 35,000	1 in 140,000
		South Asian	98	1 in 544	1 in 27,000	1 in 110,000
		Other (population not assigned)	99	1 in 403	1 in 40,000	1 in 160,000
BBS4	Bardet-Biedl syndrome	African/African American	98	1 in 311	1 in 16,000	1 in 64,000
		Latino/Admixed American	98	1 in 368	1 in 18,000	1 in 72,000
		Ashkenazi Jewish	97	1 in 2,520	1 in 84,000	1 in 340,000
		East Asian	99	1 in 38	1 in 3,700	1 in 15,000
		Finnish	97	1 in 540	1 in 18,000	1 in 72,000
		Non-Finnish European/Caucasian	98	1 in 418	1 in 21,000	1 in 84,000
		South Asian	99	1 in 682	1 in 68,000	1 in 270,000
		Other (population not assigned)	98	1 in 123	1 in 6,100	1 in 24,000
BBS5	Bardet-Biedl syndrome	African/African American	99	1 in 505	1 in 50,000	1 in 200,000
		Latino/Admixed American	99	1 in 221	1 in 22,000	1 in 88,000
		Ashkenazi Jewish	99	1 in 105	1 in 10,000	1 in 40,000
		East Asian	97	1 in 1,533	1 in 51,000	1 in 200,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
		Finnish	99	1 in 98	1 in 9,700	1 in 39,000
		Non-Finnish European/Caucasian	99	1 in 66	1 in 6,500	1 in 26,000
		South Asian	99	1 in 414	1 in 41,000	1 in 160,000
		Other (population not assigned)	99	1 in 140	1 in 14,000	1 in 56,000
BBS7	Bardet-Biedl syndrome	African/African American	98	1 in 896	1 in 45,000	1 in 180,000
		Latino/Admixed American	97	1 in 1,326	1 in 44,000	1 in 180,000
		Ashkenazi Jewish	97	1 in 5,038	1 in 170,000	1 in 680,000
		East Asian	99	1 in 654	1 in 65,000	1 in 260,000
		Finnish	98	1 in 671	1 in 34,000	1 in 140,000
		Non-Finnish European/Caucasian	98	1 in 588	1 in 29,000	1 in 120,000
		South Asian	98	1 in 895	1 in 45,000	1 in 180,000
		Other (population not assigned)	98	1 in 1,446	1 in 72,000	1 in 290,000
BBS9	Bardet-Biedl syndrome	African/African American	99	1 in 533	1 in 53,000	1 in 210,000
		Latino/Admixed American	98	1 in 436	1 in 22,000	1 in 88,000
		Ashkenazi Jewish	99	1 in 623	1 in 62,000	1 in 250,000
		East Asian	97	1 in 2,289	1 in 76,000	1 in 300,000
		Finnish	99	1 in 2,154	1 in 220,000	1 in 880,000
		Non-Finnish European/Caucasian	98	1 in 680	1 in 34,000	1 in 140,000
		South Asian	99	1 in 806	1 in 81,000	1 in 320,000
		Other (population not assigned)	98	1 in 429	1 in 21,000	1 in 84,000
BBS10	Bardet-Biedl syndrome	African/African American	98	1 in 768	1 in 38,000	1 in 150,000
		Latino/Admixed American	98	1 in 1,548	1 in 77,000	1 in 310,000
		Ashkenazi Jewish	97	1 in 329	1 in 11,000	1 in 44,000
		East Asian	97	1 in 599	1 in 20,000	1 in 80,000
		Finnish	97	1 in 5,296	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	97	1 in 269	1 in 8,900	1 in 36,000
		South Asian	98	1 in 581	1 in 29,000	1 in 120,000
		Other (population not assigned)	98	1 in 341	1 in 17,000	1 in 68,000
BBS12	Bardet-Biedl syndrome	African/African American	99	1 in 644	1 in 64,000	1 in 260,000
		Latino/Admixed American	99	1 in 843	1 in 84,000	1 in 340,000
		Ashkenazi Jewish	97	1 in 2,520	1 in 84,000	1 in 340,000
		East Asian	98	1 in 4,450	1 in 220,000	1 in 880,000
		Finnish	97	1 in 10,824	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	98	1 in 653	1 in 33,000	1 in 130,000
		South Asian	99	1 in 438	1 in 44,000	1 in 180,000
		Other (population not assigned)	98	1 in 560	1 in 28,000	1 in 110,000
BCHE	Pseudocholinesterase deficiency	African/African American	99	1 in 72	1 in 7,100	1 in 28,000
		Latino/Admixed American	99	1 in 34	1 in 3,300	1 in 13,000
		Ashkenazi Jewish	99	1 in 15	1 in 1,400	1 in 5,600
		East Asian	99	1 in 67	1 in 6,600	1 in 26,000
		Finnish	99	1 in 36	1 in 3,500	1 in 14,000
		Non-Finnish European/Caucasian	99	1 in 17	1 in 1,600	1 in 6,400
		South Asian	99	1 in 45	1 in 4,400	1 in 18,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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 19	1 in 1,800	1 in 7,200
BCKDHA	Maple syrup urine disease	African/African American	98	1 in 495	1 in 25,000	1 in 100,000
		Latino/Admixed American	99	1 in 270	1 in 27,000	1 in 110,000
		Ashkenazi Jewish	99	1 in 3,410	1 in 340,000	1 in 1,400,000
		East Asian	98	1 in 174	1 in 8,700	1 in 35,000
		Finnish	98	1 in 2,239	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	99	1 in 434	1 in 43,000	1 in 170,000
		South Asian	98	1 in 542	1 in 27,000	1 in 110,000
		Other (population not assigned)	99	1 in 914	1 in 91,000	1 in 360,000
BCKDHB	Maple syrup urine disease	African/African American	99	1 in 861	1 in 86,000	1 in 340,000
		Latino/Admixed American	99	1 in 401	1 in 40,000	1 in 160,000
		Ashkenazi Jewish	99	1 in 85	1 in 8,400	1 in 34,000
		East Asian	99	1 in 751	1 in 75,000	1 in 300,000
		Finnish	99	1 in 167	1 in 17,000	1 in 68,000
		Non-Finnish European/Caucasian	99	1 in 451	1 in 45,000	1 in 180,000
		South Asian	99	1 in 1,366	1 in 140,000	1 in 560,000
		Other (population not assigned)	98	1 in 473	1 in 24,000	1 in 96,000
BCS1L	GRACILE syndrome	African/African American	99	1 in 568	1 in 57,000	1 in 230,000
		Latino/Admixed American	99	1 in 498	1 in 50,000	1 in 200,000
		Ashkenazi Jewish	99	1 in 170	1 in 17,000	1 in 68,000
		East Asian	98	1 in 902	1 in 45,000	1 in 180,000
		Finnish	99	1 in 105	1 in 10,000	1 in 40,000
		Non-Finnish European/Caucasian	99	1 in 454	1 in 45,000	1 in 180,000
		South Asian	99	1 in 639	1 in 64,000	1 in 260,000
		Other (population not assigned)	99	1 in 458	1 in 46,000	1 in 180,000
BLM	Bloom syndrome	African/African American	98	1 in 508	1 in 25,000	1 in 100,000
		Latino/Admixed American	97	1 in 293	1 in 9,700	1 in 39,000
		Ashkenazi Jewish	97	1 in 102	1 in 3,400	1 in 14,000
		East Asian	98	1 in 493	1 in 25,000	1 in 100,000
		Finnish	99	1 in 510	1 in 51,000	1 in 200,000
		Non-Finnish European/Caucasian	98	1 in 215	1 in 11,000	1 in 44,000
		South Asian	98	1 in 338	1 in 17,000	1 in 68,000
		Other (population not assigned)	97	1 in 149	1 in 4,900	1 in 20,000
BLOC1S3	Hermansky-Pudlak syndrome	African/African American	99	1 in 5,317	1 in 530,000	1 in 2,100,000
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	1 in 1,457	1 in 150,000	1 in 600,000
		East Asian	99	1 in 1,515	1 in 150,000	1 in 600,000
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 1,818	1 in 180,000	1 in 720,000
		South Asian	99	1 in 901	1 in 90,000	1 in 360,000
		Other (population not assigned)	99	1 in 405	1 in 40,000	1 in 160,000
BLOC1S6	Hermansky-Pudlak syndrome	African/African American	99	1 in 1,351	1 in 140,000	1 in 560,000
		Latino/Admixed American	99	1 in 836	1 in 84,000	1 in 340,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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	99	1 in 809	1 in 81,000	1 in 320,000
		East Asian	99	1 in 1,052	1 in 110,000	1 in 440,000
		Finnish	99	1 in 339	1 in 34,000	1 in 140,000
		Non-Finnish European/Caucasian	99	1 in 365	1 in 36,000	1 in 140,000
		South Asian	98	1 in 787	1 in 39,000	1 in 160,000
		Other (population not assigned)	99	1 in 673	1 in 67,000	1 in 270,000
BMP1	Osteogenesis imperfecta, autosomal recessive	African/African American	99	1 in 49	1 in 4,800	1 in 19,000
		Latino/Admixed American	99	1 in 394	1 in 39,000	1 in 160,000
		Ashkenazi Jewish	99	1 in 207	1 in 21,000	1 in 84,000
		East Asian	99	1 in 740	1 in 74,000	1 in 300,000
		Finnish	99	1 in 2,688	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 643	1 in 64,000	1 in 260,000
		South Asian	98	1 in 647	1 in 32,000	1 in 130,000
		Other (population not assigned)	99	1 in 740	1 in 74,000	1 in 300,000
BRIP1	Fanconi anemia	African/African American	99	1 in 120	1 in 12,000	1 in 48,000
		Latino/Admixed American	99	1 in 502	1 in 50,000	1 in 200,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 242	1 in 12,000	1 in 48,000
		Finnish	97	1 in 577	1 in 19,000	1 in 76,000
		Non-Finnish European/Caucasian	99	1 in 295	1 in 29,000	1 in 120,000
		South Asian	98	1 in 727	1 in 36,000	1 in 140,000
		Other (population not assigned)	98	1 in 488	1 in 24,000	1 in 96,000
BSND	Bartter syndrome	African/African American	99	1 in 207	1 in 21,000	1 in 84,000
		Latino/Admixed American	99	1 in 2,882	1 in 290,000	1 in 1,200,000
		Ashkenazi Jewish	97	1 in 1,680	1 in 56,000	1 in 220,000
		East Asian	99	1 in 4,447	1 in 440,000	1 in 1,800,000
		Finnish	99	1 in 5,393	1 in 540,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 1,000	1 in 100,000	1 in 400,000
		South Asian	97	1 in 961	1 in 32,000	1 in 130,000
		Other (population not assigned)	99	1 in 1,505	1 in 150,000	1 in 600,000
BTD	Biotinidase deficiency	African/African American	99	1 in 185	1 in 18,000	1 in 72,000
		Latino/Admixed American	99	1 in 114	1 in 11,000	1 in 44,000
		Ashkenazi Jewish	99	1 in 4,924	1 in 490,000	1 in 2,000,000
		East Asian	99	1 in 245	1 in 24,000	1 in 96,000
		Finnish	98	1 in 5,489	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 127	1 in 13,000	1 in 52,000
		South Asian	99	1 in 93	1 in 9,200	1 in 37,000
		Other (population not assigned)	99	1 in 204	1 in 20,000	1 in 80,000
BTK	Agammaglobulinemia, X-linked	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

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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	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
C19orf12	Neurodegeneration with brain iron accumulation disorder	African/African American	99	1 in 702	1 in 70,000	1 in 280,000
		Latino/Admixed American	99	1 in 309	1 in 31,000	1 in 120,000
		Ashkenazi Jewish	99	1 in 187	1 in 19,000	1 in 76,000
		East Asian	99	1 in 4,597	1 in 460,000	1 in 1,800,000
		Finnish	98	1 in 3,680	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 201	1 in 20,000	1 in 80,000
		South Asian	99	1 in 49	1 in 4,800	1 in 19,000
		Other (population not assigned)	99	1 in 185	1 in 18,000	1 in 72,000
CAD	Early infantile epileptic encephalopathy	African/African American	99	1 in 59	1 in 5,800	1 in 23,000
		Latino/Admixed American	99	1 in 842	1 in 84,000	1 in 340,000
		Ashkenazi Jewish	99	1 in 272	1 in 27,000	1 in 110,000
		East Asian	99	1 in 162	1 in 16,000	1 in 64,000
		Finnish	99	1 in 91	1 in 9,000	1 in 36,000
		Non-Finnish European/Caucasian	99	1 in 207	1 in 21,000	1 in 84,000
		South Asian	99	1 in 3,712	1 in 370,000	1 in 1,500,000
		Other (population not assigned)	99	1 in 337	1 in 34,000	1 in 140,000
CAPN3	Limb-girdle muscular dystrophy, autosomal recessive	African/African American	99	1 in 118	1 in 12,000	1 in 48,000
		Latino/Admixed American	99	1 in 125	1 in 12,000	1 in 48,000
		Ashkenazi Jewish	99	1 in 359	1 in 36,000	1 in 140,000
		East Asian	99	1 in 95	1 in 9,400	1 in 38,000
		Finnish	99	1 in 551	1 in 55,000	1 in 220,000
		Non-Finnish European/Caucasian	99	1 in 100	1 in 9,900	1 in 40,000
		South Asian	99	1 in 188	1 in 19,000	1 in 76,000
		Other (population not assigned)	99	1 in 121	1 in 12,000	1 in 48,000
CASP14	Autosomal recessive congenital ichthyosis (ARCI)	African/African American	98	1 in 1,919	1 in 96,000	1 in 380,000
		Latino/Admixed American	97	1 in 178	1 in 5,900	1 in 24,000
		Ashkenazi Jewish	97	1 in 329	1 in 11,000	1 in 44,000
		East Asian	98	1 in 484	1 in 24,000	1 in 96,000
		Finnish	99	1 in 10,799	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	98	1 in 1,323	1 in 66,000	1 in 260,000
		South Asian	98	1 in 4,827	1 in 240,000	1 in 960,000
		Other (population not assigned)	97	1 in 343	1 in 11,000	1 in 44,000
CAVIN1	Congenital generalized lipodystrophy	African/African American	97	1 in 8,117	1 in 270,000	1 in 1,100,000
		Latino/Admixed American	98	1 in 3,416	1 in 170,000	1 in 680,000
		Ashkenazi Jewish	99	1 in 1,951	1 in 200,000	1 in 800,000
		East Asian	97	1 in 8,621	1 in 290,000	1 in 1,200,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	1 in 6,013	1 in 300,000	1 in 1,200,000
		South Asian	98	1 in 3,826	1 in 190,000	1 in 760,000
		Other (population not assigned)	97	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)

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
CBS	Homocystinuria	African/African American	98	1 in 150	1 in 7,500	1 in 30,000
		Latino/Admixed American	98	1 in 95	1 in 4,700	1 in 19,000
		Ashkenazi Jewish	98	1 in 182	1 in 9,100	1 in 36,000
		East Asian	98	1 in 343	1 in 17,000	1 in 68,000
		Finnish	98	1 in 348	1 in 17,000	1 in 68,000
		Non-Finnish European/Caucasian	98	1 in 89	1 in 4,400	1 in 18,000
		South Asian	97	1 in 183	1 in 6,100	1 in 24,000
		Other (population not assigned)	98	1 in 132	1 in 6,600	1 in 26,000
CCDC8	3M syndrome	African/African American	97	1 in 4,011	1 in 130,000	1 in 520,000
		Latino/Admixed American	97	1 in 1,921	1 in 64,000	1 in 260,000
		Ashkenazi Jewish	97	1 in 459	1 in 15,000	1 in 60,000
		East Asian	98	1 in 1,022	1 in 51,000	1 in 200,000
		Finnish	98	1 in 2,681	1 in 130,000	1 in 520,000
		Non-Finnish European/Caucasian	98	1 in 777	1 in 39,000	1 in 160,000
		South Asian	97	1 in 1,269	1 in 42,000	1 in 170,000
		Other (population not assigned)	97	1 in 3,068	1 in 100,000	1 in 400,000
CCN6	Progressive pseudorheumatoid dysplasia	African/African American	98	1 in 643	1 in 32,000	1 in 130,000
		Latino/Admixed American	97	1 in 469	1 in 16,000	1 in 64,000
		Ashkenazi Jewish	97	1 in 885	1 in 29,000	1 in 120,000
		East Asian	98	1 in 706	1 in 35,000	1 in 140,000
		Finnish	98	1 in 2,593	1 in 130,000	1 in 520,000
		Non-Finnish European/Caucasian	98	1 in 674	1 in 34,000	1 in 140,000
		South Asian	99	1 in 303	1 in 30,000	1 in 120,000
		Other (population not assigned)	97	1 in 383	1 in 13,000	1 in 52,000
CD247	Severe combined immunodeficiency (SCID)	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	1 in 1,678	1 in 170,000	1 in 680,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	1 in 653	1 in 65,000	1 in 260,000
		Non-Finnish European/Caucasian	99	1 in 1,295	1 in 130,000	1 in 520,000
		South Asian	99	1 in 2,552	1 in 260,000	1 in 1,000,000
		Other (population not assigned)	99	1 in 2,737	1 in 270,000	1 in 1,100,000
CD3D	Severe combined immunodeficiency (SCID)	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	1 in 694	1 in 23,000	1 in 92,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 3,066	1 in 150,000	1 in 600,000
		Finnish	98	1 in 416	1 in 21,000	1 in 84,000
		Non-Finnish European/Caucasian	99	1 in 944	1 in 94,000	1 in 380,000
		South Asian	99	1 in 1,146	1 in 110,000	1 in 440,000
		Other (population not assigned)	99	1 in 756	1 in 76,000	1 in 300,000
CD3E	Severe combined immunodeficiency (SCID)	African/African American	99	1 in 8,125	1 in 810,000	1 in 3,200,000
		Latino/Admixed American	97	1 in 1,729	1 in 58,000	1 in 230,000
		Ashkenazi Jewish	97	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)

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 4,727	1 in 470,000	1 in 1,900,000
		South Asian	99	1 in 15,308	1 in 1,500,000	1 in 6,000,000
		Other (population not assigned)	99	1 in 3,067	1 in 310,000	1 in 1,200,000
CD3G	Severe combined immunodeficiency (SCID)	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	99	1 in 1,921	1 in 190,000	1 in 760,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 762	1 in 76,000	1 in 300,000
		Finnish	99	1 in 1,359	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	99	1 in 3,171	1 in 320,000	1 in 1,300,000
		South Asian	97	1 in 451	1 in 15,000	1 in 60,000
		Other (population not assigned)	99	1 in 597	1 in 60,000	1 in 240,000
CD8A	Severe combined immunodeficiency (SCID)	African/African American	97	1 in 7,491	1 in 250,000	1 in 1,000,000
		Latino/Admixed American	97	1 in 1,694	1 in 56,000	1 in 220,000
		Ashkenazi Jewish	97	1 in 5,034	1 in 170,000	1 in 680,000
		East Asian	99	1 in 9,091	1 in 910,000	1 in 3,600,000
		Finnish	97	1 in 4,777	1 in 160,000	1 in 640,000
		Non-Finnish European/Caucasian	98	1 in 3,179	1 in 160,000	1 in 640,000
		South Asian	99	1 in 2,534	1 in 250,000	1 in 1,000,000
		Other (population not assigned)	98	1 in 1,531	1 in 77,000	1 in 310,000
CDCA7	Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome	African/African American	97	1 in 1,350	1 in 45,000	1 in 180,000
		Latino/Admixed American	99	1 in 1,316	1 in 130,000	1 in 520,000
		Ashkenazi Jewish	99	1 in 4,880	1 in 490,000	1 in 2,000,000
		East Asian	97	N/A	N/A	N/A
		Finnish	99	1 in 11,136	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	99	1 in 3,526	1 in 350,000	1 in 1,400,000
		South Asian	99	1 in 5,107	1 in 510,000	1 in 2,000,000
		Other (population not assigned)	99	1 in 3,054	1 in 310,000	1 in 1,200,000
CDH23	Usher syndrome (hearing loss and retinitis pigmentosa)	African/African American	99	1 in 121	1 in 12,000	1 in 48,000
		Latino/Admixed American	98	1 in 298	1 in 15,000	1 in 60,000
		Ashkenazi Jewish	99	1 in 613	1 in 61,000	1 in 240,000
		East Asian	98	1 in 43	1 in 2,100	1 in 8,400
		Finnish	98	1 in 1,189	1 in 59,000	1 in 240,000
		Non-Finnish European/Caucasian	99	1 in 244	1 in 24,000	1 in 96,000
		South Asian	99	1 in 264	1 in 26,000	1 in 100,000
		Other (population not assigned)	99	1 in 232	1 in 23,000	1 in 92,000
CEP104	Joubert syndrome and related disorders, including Meckel-Gruber syndrome	African/African American	97	1 in 44	1 in 1,400	1 in 5,600
		Latino/Admixed American	97	1 in 56	1 in 1,800	1 in 7,200
		Ashkenazi Jewish	98	1 in 67	1 in 3,300	1 in 13,000
		East Asian	97	1 in 87	1 in 2,900	1 in 12,000
		Finnish	99	1 in 141	1 in 14,000	1 in 56,000
		Non-Finnish European/Caucasian	98	1 in 108	1 in 5,400	1 in 22,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
		South Asian	98	1 in 93	1 in 4,600	1 in 18,000
		Other (population not assigned)	97	1 in 95	1 in 3,100	1 in 12,000
CEP290	Ciliopathies	African/African American	97	1 in 53	1 in 1,700	1 in 6,800
		Latino/Admixed American	97	1 in 79	1 in 2,600	1 in 10,000
		Ashkenazi Jewish	98	1 in 165	1 in 8,200	1 in 33,000
		East Asian	98	1 in 50	1 in 2,500	1 in 10,000
		Finnish	97	1 in 119	1 in 3,900	1 in 16,000
		Non-Finnish European/Caucasian	97	1 in 63	1 in 2,100	1 in 8,400
		South Asian	97	1 in 40	1 in 1,300	1 in 5,200
		Other (population not assigned)	97	1 in 115	1 in 3,800	1 in 15,000
CERKL	Retinitis pigmentosa	African/African American	99	1 in 861	1 in 86,000	1 in 340,000
		Latino/Admixed American	99	1 in 417	1 in 42,000	1 in 170,000
		Ashkenazi Jewish	97	1 in 4,908	1 in 160,000	1 in 640,000
		East Asian	98	1 in 603	1 in 30,000	1 in 120,000
		Finnish	99	1 in 377	1 in 38,000	1 in 150,000
		Non-Finnish European/Caucasian	99	1 in 482	1 in 48,000	1 in 190,000
		South Asian	99	1 in 305	1 in 30,000	1 in 120,000
		Other (population not assigned)	99	1 in 588	1 in 59,000	1 in 240,000
CERS3	Autosomal recessive congenital ichthyosis (ARCI)	African/African American	99	1 in 8,105	1 in 810,000	1 in 3,200,000
		Latino/Admixed American	99	1 in 16,893	1 in 1,700,000	1 in 6,800,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	1 in 10,814	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 6,225	1 in 620,000	1 in 2,500,000
		South Asian	98	1 in 7,635	1 in 380,000	1 in 1,500,000
		Other (population not assigned)	97	N/A	N/A	N/A
CFTR	Cystic fibrosis	African/African American	99	1 in 32	1 in 3,100	1 in 12,000
		Latino/Admixed American	99	1 in 17	1 in 1,600	1 in 6,400
		Ashkenazi Jewish	99	1 in 15	1 in 1,400	1 in 5,600
		East Asian	99	1 in 95	1 in 9,400	1 in 38,000
		Finnish	99	1 in 40	1 in 3,900	1 in 16,000
		Non-Finnish European/Caucasian	99	1 in 13	1 in 1,200	1 in 4,800
		South Asian	99	1 in 32	1 in 3,100	1 in 12,000
		Other (population not assigned)	99	1 in 17	1 in 1,600	1 in 6,400
CHAT	Congenital myasthenic syndrome	African/African American	97	1 in 30	1 in 970	1 in 3,900
		Latino/Admixed American	98	1 in 209	1 in 10,000	1 in 40,000
		Ashkenazi Jewish	99	1 in 258	1 in 26,000	1 in 100,000
		East Asian	98	1 in 318	1 in 16,000	1 in 64,000
		Finnish	99	1 in 74	1 in 7,300	1 in 29,000
		Non-Finnish European/Caucasian	99	1 in 121	1 in 12,000	1 in 48,000
		South Asian	99	1 in 321	1 in 32,000	1 in 130,000
		Other (population not assigned)	99	1 in 236	1 in 24,000	1 in 96,000
CHMP1A	Pontocerebellar hypoplasia	African/African American	99	1 in 1,214	1 in 120,000	1 in 480,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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	99	1 in 907	1 in 91,000	1 in 360,000
		Ashkenazi Jewish	99	1 in 620	1 in 62,000	1 in 250,000
		East Asian	99	1 in 331	1 in 33,000	1 in 130,000
		Finnish	99	1 in 1,064	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	99	1 in 717	1 in 72,000	1 in 290,000
		South Asian	99	1 in 734	1 in 73,000	1 in 290,000
		Other (population not assigned)	99	1 in 365	1 in 36,000	1 in 140,000
CHRNG	Multiple pterygium syndrome	African/African American	97	1 in 371	1 in 12,000	1 in 48,000
		Latino/Admixed American	97	1 in 546	1 in 18,000	1 in 72,000
		Ashkenazi Jewish	99	1 in 2,517	1 in 250,000	1 in 1,000,000
		East Asian	98	1 in 529	1 in 26,000	1 in 100,000
		Finnish	99	1 in 637	1 in 64,000	1 in 260,000
		Non-Finnish European/Caucasian	97	1 in 400	1 in 13,000	1 in 52,000
		South Asian	98	1 in 384	1 in 19,000	1 in 76,000
		Other (population not assigned)	97	1 in 705	1 in 23,000	1 in 92,000
CIB2	Usher syndrome (hearing loss and retinitis pigmentosa)	African/African American	99	1 in 1,693	1 in 170,000	1 in 680,000
		Latino/Admixed American	99	1 in 1,107	1 in 110,000	1 in 440,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 4,599	1 in 460,000	1 in 1,800,000
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	1 in 1,059	1 in 53,000	1 in 210,000
		South Asian	99	1 in 1,210	1 in 120,000	1 in 480,000
		Other (population not assigned)	98	1 in 1,023	1 in 51,000	1 in 200,000
CIITA	Bare lymphocyte syndrome type II	African/African American	98	1 in 1,322	1 in 66,000	1 in 260,000
		Latino/Admixed American	99	1 in 2,879	1 in 290,000	1 in 1,200,000
		Ashkenazi Jewish	98	1 in 974	1 in 49,000	1 in 200,000
		East Asian	99	1 in 1,002	1 in 100,000	1 in 400,000
		Finnish	99	1 in 10,477	1 in 1,000,000	1 in 4,000,000
		Non-Finnish European/Caucasian	98	1 in 1,505	1 in 75,000	1 in 300,000
		South Asian	98	1 in 2,185	1 in 110,000	1 in 440,000
		Other (population not assigned)	99	1 in 3,069	1 in 310,000	1 in 1,200,000
CLCF1	Cold-induced sweating syndrome, includes Crisponi syndrome	African/African American	97	1 in 2,029	1 in 68,000	1 in 270,000
		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	98	1 in 18,716	1 in 940,000	1 in 3,800,000
		South Asian	97	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
CLCN5	Dent disease	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	97	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)

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
		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
CLN3	Neuronal ceroid-lipofuscinosis	African/African American	99	1 in 540	1 in 54,000	1 in 220,000
		Latino/Admixed American	99	1 in 242	1 in 24,000	1 in 96,000
		Ashkenazi Jewish	99	1 in 202	1 in 20,000	1 in 80,000
		East Asian	99	1 in 373	1 in 37,000	1 in 150,000
		Finnish	98	1 in 513	1 in 26,000	1 in 100,000
		Non-Finnish European/Caucasian	99	1 in 411	1 in 41,000	1 in 160,000
		South Asian	99	1 in 133	1 in 13,000	1 in 52,000
		Other (population not assigned)	99	1 in 304	1 in 30,000	1 in 120,000
CLN5	Neuronal ceroid-lipofuscinosis	African/African American	99	1 in 1,021	1 in 100,000	1 in 400,000
		Latino/Admixed American	98	1 in 644	1 in 32,000	1 in 130,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 714	1 in 24,000	1 in 96,000
		Finnish	99	1 in 142	1 in 14,000	1 in 56,000
		Non-Finnish European/Caucasian	99	1 in 254	1 in 25,000	1 in 100,000
		South Asian	99	1 in 840	1 in 84,000	1 in 340,000
		Other (population not assigned)	99	1 in 335	1 in 33,000	1 in 130,000
CLN6	Neuronal ceroid-lipofuscinosis	African/African American	99	1 in 196	1 in 20,000	1 in 80,000
		Latino/Admixed American	99	1 in 358	1 in 36,000	1 in 140,000
		Ashkenazi Jewish	99	1 in 1,256	1 in 130,000	1 in 520,000
		East Asian	99	1 in 572	1 in 57,000	1 in 230,000
		Finnish	99	1 in 333	1 in 33,000	1 in 130,000
		Non-Finnish European/Caucasian	99	1 in 434	1 in 43,000	1 in 170,000
		South Asian	98	1 in 1,000	1 in 50,000	1 in 200,000
		Other (population not assigned)	99	1 in 435	1 in 43,000	1 in 170,000
CLN8	Neuronal ceroid-lipofuscinosis	African/African American	99	1 in 1,566	1 in 160,000	1 in 640,000
		Latino/Admixed American	97	1 in 8,644	1 in 290,000	1 in 1,200,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 4,599	1 in 460,000	1 in 1,800,000
		Finnish	99	1 in 429	1 in 43,000	1 in 170,000
		Non-Finnish European/Caucasian	99	1 in 1,605	1 in 160,000	1 in 640,000
		South Asian	99	1 in 1,920	1 in 190,000	1 in 760,000
		Other (population not assigned)	99	1 in 2,744	1 in 270,000	1 in 1,100,000
CLP1	Pontocerebellar hypoplasia	African/African American	98	N/A	N/A	N/A
		Latino/Admixed American	99	1 in 5,682	1 in 570,000	1 in 2,300,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	98	1 in 3,066	1 in 150,000	1 in 600,000
		Finnish	99	1 in 10,821	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	99	1 in 2,855	1 in 290,000	1 in 1,200,000
		South Asian	98	1 in 2,162	1 in 110,000	1 in 440,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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)	98	N/A	N/A	N/A
CLRN1	Usher syndrome (hearing loss and retinitis pigmentosa)	African/African American	99	1 in 633	1 in 63,000	1 in 250,000
		Latino/Admixed American	99	1 in 1,628	1 in 160,000	1 in 640,000
		Ashkenazi Jewish	99	1 in 86	1 in 8,500	1 in 34,000
		East Asian	98	1 in 1,314	1 in 66,000	1 in 260,000
		Finnish	99	1 in 67	1 in 6,600	1 in 26,000
		Non-Finnish European/Caucasian	99	1 in 254	1 in 25,000	1 in 100,000
		South Asian	97	1 in 352	1 in 12,000	1 in 48,000
		Other (population not assigned)	99	1 in 304	1 in 30,000	1 in 120,000
CNTNAP2	Pitt-Hopkins-like syndrome 1	African/African American	97	1 in 114	1 in 3,800	1 in 15,000
		Latino/Admixed American	98	1 in 355	1 in 18,000	1 in 72,000
		Ashkenazi Jewish	97	1 in 1,874	1 in 62,000	1 in 250,000
		East Asian	99	1 in 571	1 in 57,000	1 in 230,000
		Finnish	97	1 in 381	1 in 13,000	1 in 52,000
		Non-Finnish European/Caucasian	98	1 in 314	1 in 16,000	1 in 64,000
		South Asian	97	1 in 419	1 in 14,000	1 in 56,000
		Other (population not assigned)	98	1 in 349	1 in 17,000	1 in 68,000
COASY	Neurodegeneration with brain iron accumulation disorder	African/African American	99	1 in 179	1 in 18,000	1 in 72,000
		Latino/Admixed American	98	1 in 906	1 in 45,000	1 in 180,000
		Ashkenazi Jewish	99	1 in 709	1 in 71,000	1 in 280,000
		East Asian	98	1 in 819	1 in 41,000	1 in 160,000
		Finnish	97	1 in 1,846	1 in 62,000	1 in 250,000
		Non-Finnish European/Caucasian	97	1 in 383	1 in 13,000	1 in 52,000
		South Asian	99	1 in 437	1 in 44,000	1 in 180,000
		Other (population not assigned)	97	1 in 234	1 in 7,800	1 in 31,000
COL4A3	Alport syndrome	African/African American	99	1 in 287	1 in 29,000	1 in 120,000
		Latino/Admixed American	99	1 in 90	1 in 8,900	1 in 36,000
		Ashkenazi Jewish	98	1 in 241	1 in 12,000	1 in 48,000
		East Asian	99	1 in 408	1 in 41,000	1 in 160,000
		Finnish	99	1 in 674	1 in 67,000	1 in 270,000
		Non-Finnish European/Caucasian	99	1 in 69	1 in 6,800	1 in 27,000
		South Asian	99	1 in 233	1 in 23,000	1 in 92,000
		Other (population not assigned)	99	1 in 106	1 in 11,000	1 in 44,000
COL4A5	Alport syndrome	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
COLQ	Congenital myasthenic syndrome	African/African American	98	1 in 1,494	1 in 75,000	1 in 300,000
		Latino/Admixed American	99	1 in 674	1 in 67,000	1 in 270,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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	98	N/A	N/A	N/A
		East Asian	99	1 in 653	1 in 65,000	1 in 260,000
		Finnish	98	1 in 5,109	1 in 260,000	1 in 1,000,000
		Non-Finnish European/Caucasian	98	1 in 805	1 in 40,000	1 in 160,000
		South Asian	99	1 in 992	1 in 99,000	1 in 400,000
		Other (population not assigned)	99	1 in 576	1 in 58,000	1 in 230,000
CORO1A	Severe combined immunodeficiency (SCID)	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	98	1 in 3,560	1 in 180,000	1 in 720,000
		Ashkenazi Jewish	97	1 in 5,021	1 in 170,000	1 in 680,000
		East Asian	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 4,038	1 in 400,000	1 in 1,600,000
		South Asian	98	1 in 1,192	1 in 60,000	1 in 240,000
		Other (population not assigned)	97	N/A	N/A	N/A
COX15	Leigh syndrome, autosomal recessive	African/African American	99	1 in 887	1 in 89,000	1 in 360,000
		Latino/Admixed American	99	1 in 719	1 in 72,000	1 in 290,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	98	1 in 1,839	1 in 92,000	1 in 370,000
		Finnish	99	1 in 5,475	1 in 550,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 480	1 in 48,000	1 in 190,000
		South Asian	99	1 in 159	1 in 16,000	1 in 64,000
		Other (population not assigned)	99	1 in 300	1 in 30,000	1 in 120,000
CP	Neurodegeneration with brain iron accumulation disorder	African/African American	99	1 in 442	1 in 44,000	1 in 180,000
		Latino/Admixed American	97	1 in 248	1 in 8,200	1 in 33,000
		Ashkenazi Jewish	98	1 in 642	1 in 32,000	1 in 130,000
		East Asian	97	1 in 220	1 in 7,300	1 in 29,000
		Finnish	99	1 in 597	1 in 60,000	1 in 240,000
		Non-Finnish European/Caucasian	99	1 in 497	1 in 50,000	1 in 200,000
		South Asian	99	1 in 266	1 in 27,000	1 in 110,000
		Other (population not assigned)	98	1 in 1,358	1 in 68,000	1 in 270,000
CPLANE1	Joubert syndrome and related disorders, including Meckel-Gruber syndrome	African/African American	99	1 in 97	1 in 9,600	1 in 38,000
		Latino/Admixed American	99	1 in 53	1 in 5,200	1 in 21,000
		Ashkenazi Jewish	99	1 in 156	1 in 16,000	1 in 64,000
		East Asian	98	1 in 217	1 in 11,000	1 in 44,000
		Finnish	99	1 in 232	1 in 23,000	1 in 92,000
		Non-Finnish European/Caucasian	99	1 in 42	1 in 4,100	1 in 16,000
		South Asian	99	1 in 143	1 in 14,000	1 in 56,000
		Other (population not assigned)	99	1 in 44	1 in 4,300	1 in 17,000
CPS1	Carbamoyl phosphate synthetase I deficiency	African/African American	98	1 in 530	1 in 26,000	1 in 100,000
		Latino/Admixed American	98	1 in 2,141	1 in 110,000	1 in 440,000
		Ashkenazi Jewish	99	1 in 1,002	1 in 100,000	1 in 400,000
		East Asian	98	1 in 525	1 in 26,000	1 in 100,000
		Finnish	99	1 in 568	1 in 57,000	1 in 230,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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 769	1 in 77,000	1 in 310,000
		South Asian	99	1 in 1,916	1 in 190,000	1 in 760,000
		Other (population not assigned)	99	1 in 595	1 in 59,000	1 in 240,000
CPT1A	Carnitine palmitoyltransferase I deficiency	African/African American	99	1 in 2,001	1 in 200,000	1 in 800,000
		Latino/Admixed American	99	1 in 2,798	1 in 280,000	1 in 1,100,000
		Ashkenazi Jewish	99	1 in 492	1 in 49,000	1 in 200,000
		East Asian	99	1 in 1,288	1 in 130,000	1 in 520,000
		Finnish	99	1 in 266	1 in 27,000	1 in 110,000
		Non-Finnish European/Caucasian	99	1 in 1,608	1 in 160,000	1 in 640,000
		South Asian	99	1 in 2,188	1 in 220,000	1 in 880,000
		Other (population not assigned)	99	1 in 458	1 in 46,000	1 in 180,000
CPT2	Carnitine palmitoyltransferase II deficiency	African/African American	99	1 in 322	1 in 32,000	1 in 130,000
		Latino/Admixed American	99	1 in 279	1 in 28,000	1 in 110,000
		Ashkenazi Jewish	99	1 in 30	1 in 2,900	1 in 12,000
		East Asian	99	1 in 406	1 in 41,000	1 in 160,000
		Finnish	99	1 in 251	1 in 25,000	1 in 100,000
		Non-Finnish European/Caucasian	99	1 in 185	1 in 18,000	1 in 72,000
		South Asian	99	1 in 523	1 in 52,000	1 in 210,000
		Other (population not assigned)	99	1 in 152	1 in 15,000	1 in 60,000
CRLF1	Cold-induced sweating syndrome, includes Crisponi syndrome	African/African American	98	1 in 785	1 in 39,000	1 in 160,000
		Latino/Admixed American	98	1 in 576	1 in 29,000	1 in 120,000
		Ashkenazi Jewish	99	1 in 683	1 in 68,000	1 in 270,000
		East Asian	98	1 in 470	1 in 23,000	1 in 92,000
		Finnish	97	1 in 3,168	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	98	1 in 1,151	1 in 58,000	1 in 230,000
		South Asian	98	1 in 909	1 in 45,000	1 in 180,000
		Other (population not assigned)	98	1 in 460	1 in 23,000	1 in 92,000
CRTAP	Osteogenesis imperfecta, autosomal recessive	African/African American	99	1 in 1,122	1 in 110,000	1 in 440,000
		Latino/Admixed American	97	1 in 546	1 in 18,000	1 in 72,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 1,377	1 in 140,000	1 in 560,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 1,416	1 in 140,000	1 in 560,000
		South Asian	98	1 in 1,059	1 in 53,000	1 in 210,000
		Other (population not assigned)	99	1 in 2,126	1 in 210,000	1 in 840,000
CTC1	Coats plus syndrome and dyskeratosis congenita, CTC1-related	African/African American	99	1 in 98	1 in 9,700	1 in 39,000
		Latino/Admixed American	98	1 in 273	1 in 14,000	1 in 56,000
		Ashkenazi Jewish	99	1 in 2,488	1 in 250,000	1 in 1,000,000
		East Asian	99	1 in 273	1 in 27,000	1 in 110,000
		Finnish	98	1 in 54	1 in 2,700	1 in 11,000
		Non-Finnish European/Caucasian	98	1 in 256	1 in 13,000	1 in 52,000
		South Asian	98	1 in 679	1 in 34,000	1 in 140,000
		Other (population not assigned)	97	1 in 130	1 in 4,300	1 in 17,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
CTNS	Cystinosis	African/African American	99	1 in 974	1 in 97,000	1 in 390,000
		Latino/Admixed American	99	1 in 1,480	1 in 150,000	1 in 600,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 673	1 in 67,000	1 in 270,000
		Finnish	99	1 in 493	1 in 49,000	1 in 200,000
		Non-Finnish European/Caucasian	99	1 in 453	1 in 45,000	1 in 180,000
		South Asian	98	1 in 1,001	1 in 50,000	1 in 200,000
		Other (population not assigned)	98	1 in 743	1 in 37,000	1 in 150,000
CTSA	Galactosialidosis	African/African American	97	1 in 127	1 in 4,200	1 in 17,000
		Latino/Admixed American	97	1 in 248	1 in 8,200	1 in 33,000
		Ashkenazi Jewish	97	1 in 65	1 in 2,100	1 in 8,400
		East Asian	99	1 in 2,686	1 in 270,000	1 in 1,100,000
		Finnish	97	1 in 206	1 in 6,800	1 in 27,000
		Non-Finnish European/Caucasian	97	1 in 64	1 in 2,100	1 in 8,400
		South Asian	97	1 in 310	1 in 10,000	1 in 40,000
		Other (population not assigned)	97	1 in 94	1 in 3,100	1 in 12,000
CTSD	Neuronal ceroid-lipofuscinosis	African/African American	98	1 in 1,160	1 in 58,000	1 in 230,000
		Latino/Admixed American	98	1 in 1,591	1 in 80,000	1 in 320,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 9,179	1 in 920,000	1 in 3,700,000
		Finnish	97	1 in 1,801	1 in 60,000	1 in 240,000
		Non-Finnish European/Caucasian	97	1 in 1,003	1 in 33,000	1 in 130,000
		South Asian	99	1 in 1,912	1 in 190,000	1 in 760,000
		Other (population not assigned)	99	1 in 1,153	1 in 120,000	1 in 480,000
CTSF	Neuronal ceroid-lipofuscinosis	African/African American	99	1 in 426	1 in 43,000	1 in 170,000
		Latino/Admixed American	99	1 in 1,448	1 in 140,000	1 in 560,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 365	1 in 36,000	1 in 140,000
		Finnish	99	1 in 10,821	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	99	1 in 1,773	1 in 180,000	1 in 720,000
		South Asian	98	1 in 881	1 in 44,000	1 in 180,000
		Other (population not assigned)	98	1 in 1,449	1 in 72,000	1 in 290,000
CTSK	Pycnodysostosis	African/African American	99	1 in 663	1 in 66,000	1 in 260,000
		Latino/Admixed American	98	1 in 897	1 in 45,000	1 in 180,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 1,435	1 in 48,000	1 in 190,000
		Finnish	99	1 in 2,810	1 in 280,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 1,700	1 in 170,000	1 in 680,000
		South Asian	99	1 in 1,183	1 in 120,000	1 in 480,000
		Other (population not assigned)	99	1 in 948	1 in 95,000	1 in 380,000
CUL7	3M syndrome	African/African American	99	1 in 347	1 in 35,000	1 in 140,000
		Latino/Admixed American	99	1 in 711	1 in 71,000	1 in 280,000
		Ashkenazi Jewish	99	1 in 2,457	1 in 250,000	1 in 1,000,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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 754	1 in 75,000	1 in 300,000
		Finnish	99	1 in 259	1 in 26,000	1 in 100,000
		Non-Finnish European/Caucasian	99	1 in 409	1 in 41,000	1 in 160,000
		South Asian	98	1 in 453	1 in 23,000	1 in 92,000
		Other (population not assigned)	99	1 in 946	1 in 95,000	1 in 380,000
CWC27	Retinitis pigmentosa	African/African American	98	1 in 566	1 in 28,000	1 in 110,000
		Latino/Admixed American	98	1 in 1,273	1 in 64,000	1 in 260,000
		Ashkenazi Jewish	97	1 in 3,742	1 in 120,000	1 in 480,000
		East Asian	98	1 in 846	1 in 42,000	1 in 170,000
		Finnish	97	1 in 2,632	1 in 88,000	1 in 350,000
		Non-Finnish European/Caucasian	98	1 in 910	1 in 45,000	1 in 180,000
		South Asian	97	1 in 1,097	1 in 37,000	1 in 150,000
		Other (population not assigned)	97	1 in 2,935	1 in 98,000	1 in 390,000
CYBA	Chronic granulomatous disease	African/African American	98	1 in 291	1 in 15,000	1 in 60,000
		Latino/Admixed American	99	1 in 385	1 in 38,000	1 in 150,000
		Ashkenazi Jewish	99	1 in 2,015	1 in 200,000	1 in 800,000
		East Asian	99	1 in 1,196	1 in 120,000	1 in 480,000
		Finnish	99	1 in 653	1 in 65,000	1 in 260,000
		Non-Finnish European/Caucasian	99	1 in 731	1 in 73,000	1 in 290,000
		South Asian	99	1 in 572	1 in 57,000	1 in 230,000
		Other (population not assigned)	98	1 in 652	1 in 33,000	1 in 130,000
CYBB	Chronic granulomatous disease	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
CYP1B1	Primary congenital glaucoma	African/African American	98	1 in 1,667	1 in 83,000	1 in 330,000
		Latino/Admixed American	98	1 in 246	1 in 12,000	1 in 48,000
		Ashkenazi Jewish	99	1 in 2,465	1 in 250,000	1 in 1,000,000
		East Asian	99	1 in 399	1 in 40,000	1 in 160,000
		Finnish	97	1 in 2,786	1 in 93,000	1 in 370,000
		Non-Finnish European/Caucasian	99	1 in 196	1 in 20,000	1 in 80,000
		South Asian	99	1 in 325	1 in 32,000	1 in 130,000
		Other (population not assigned)	98	1 in 145	1 in 7,200	1 in 29,000
CYP11B1	Congenital adrenal hyperplasia	African/African American	99	1 in 339	1 in 34,000	1 in 140,000
		Latino/Admixed American	98	1 in 1,055	1 in 53,000	1 in 210,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 1,485	1 in 74,000	1 in 300,000
		Finnish	97	1 in 7,778	1 in 260,000	1 in 1,000,000
		Non-Finnish European/Caucasian	99	1 in 792	1 in 79,000	1 in 320,000

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.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ 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
		South Asian	99	1 in 284	1 in 28,000	1 in 110,000
		Other (population not assigned)	99	1 in 948	1 in 95,000	1 in 380,000
CYP17A1	Congenital adrenal hyperplasia	African/African American	99	1 in 1,313	1 in 130,000	1 in 520,000
		Latino/Admixed American	98	1 in 2,147	1 in 110,000	1 in 440,000
		Ashkenazi Jewish	97	1 in 4,689	1 in 160,000	1 in 640,000
		East Asian	98	1 in 409	1 in 20,000	1 in 80,000
		Finnish	98	1 in 5,495	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 1,592	1 in 160,000	1 in 640,000
		South Asian	98	1 in 1,018	1 in 51,000	1 in 200,000
		Other (population not assigned)	97	N/A	N/A	N/A
CYP21A2	Congenital adrenal hyperplasia (classic form)	African American	82	1 in 79	1 in 430	1 in 1700
		Ashkenazi Jewish	90	1 in 40	1 in 390	1 in 1600
		Asian	92	1 in 62	1 in 760	1 in 3000
		Indian	87	1 in 40	1 in 300	1 in 1200
		European	88	1 in 70	1 in 580	1 in 2300
		Hispanic American	89	1 in 73	1 in 660	1 in 2600
		Other	90	1 in 70	1 in 690	1 in 2800
CYP27A1	Cerebrotendinous xanthomatosis	African/African American	99	1 in 428	1 in 43,000	1 in 170,000
		Latino/Admixed American	99	1 in 330	1 in 33,000	1 in 130,000
		Ashkenazi Jewish	99	1 in 292	1 in 29,000	1 in 120,000
		East Asian	99	1 in 123	1 in 12,000	1 in 48,000
		Finnish	99	1 in 1,222	1 in 120,000	1 in 480,000
		Non-Finnish European/Caucasian	99	1 in 359	1 in 36,000	1 in 140,000
		South Asian	99	1 in 158	1 in 16,000	1 in 64,000
		Other (population not assigned)	99	1 in 319	1 in 32,000	1 in 130,000
CYP4F22	Autosomal recessive congenital ichthyosis (ARCI)	African/African American	99	1 in 1,325	1 in 130,000	1 in 520,000
		Latino/Admixed American	99	1 in 1,402	1 in 140,000	1 in 560,000
		Ashkenazi Jewish	99	1 in 713	1 in 71,000	1 in 280,000
		East Asian	99	1 in 315	1 in 31,000	1 in 120,000
		Finnish	99	1 in 350	1 in 35,000	1 in 140,000
		Non-Finnish European/Caucasian	99	1 in 901	1 in 90,000	1 in 360,000
		South Asian	99	1 in 77	1 in 7,600	1 in 30,000
		Other (population not assigned)	99	1 in 265	1 in 26,000	1 in 100,000
CYP7B1	Hereditary spastic paraplegia	African/African American	99	1 in 901	1 in 90,000	1 in 360,000
		Latino/Admixed American	99	1 in 1,878	1 in 190,000	1 in 760,000
		Ashkenazi Jewish	99	1 in 5,030	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 222	1 in 22,000	1 in 88,000
		Finnish	98	1 in 589	1 in 29,000	1 in 120,000
		Non-Finnish European/Caucasian	99	1 in 324	1 in 32,000	1 in 130,000
		South Asian	99	1 in 804	1 in 80,000	1 in 320,000
		Other (population not assigned)	99	1 in 597	1 in 60,000	1 in 240,000

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