

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

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

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Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
Danon disease	LAMP2	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
D-bifunctional protein deficiency	HSD17B4	African/African American	98	1 in 326	1 in 16,000	1 in 64,000
		Latino/Admixed American	99	1 in 638	1 in 64,000	1 in 260,000
		Ashkenazi Jewish	99	1 in 2,490	1 in 250,000	1 in 1,000,000
		East Asian	99	1 in 449	1 in 45,000	1 in 180,000
		Finnish	99	1 in 293	1 in 29,000	1 in 120,000
		Non-Finnish European/Caucasian	99	1 in 424	1 in 42,000	1 in 170,000
		South Asian	99	1 in 122	1 in 12,000	1 in 48,000
		Other (population not assigned)	99	1 in 2,742	1 in 270,000	1 in 1,100,000
Deafness and hearing loss, nonsyndromic	GJB2	African/African American	99	1 in 113	1 in 11,000	1 in 44,000
		Latino/Admixed American	95	1 in 37	1 in 720	1 in 2,900
		Ashkenazi Jewish	99	1 in 57	1 in 5,600	1 in 22,000
		East Asian	97	1 in 58	1 in 1,900	1 in 7,600
		Finnish	97	1 in 58	1 in 1,900	1 in 7,600
		Non-Finnish European/Caucasian	98	1 in 38	1 in 1,900	1 in 7,600
		South Asian	99	1 in 58	1 in 5,700	1 in 23,000
		Other (population not assigned)	98	1 in 47	1 in 2,300	1 in 9,200
	GJB6	African/African American	97	1 in 1,626	1 in 54,000	1 in 220,000
		Latino/Admixed American	71	1 in 2,161	1 in 7,400	1 in 30,000
		Ashkenazi Jewish	99	1 in 5,032	1 in 500,000	1 in 2,000,000
		East Asian	98	1 in 214	1 in 11,000	1 in 44,000
		Finnish	97	1 in 5,411	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	94	1 in 421	1 in 7,000	1 in 28,000
		South Asian	99	1 in 1,531	1 in 150,000	1 in 600,000
		Other (population not assigned)	97	1 in 256	1 in 8,500	1 in 34,000
	LOXHD1	African/African American	98	1 in 276	1 in 14,000	1 in 56,000
		Latino/Admixed American	98	1 in 224	1 in 11,000	1 in 44,000
		Ashkenazi Jewish	99	1 in 122	1 in 12,000	1 in 48,000
		East Asian	99	1 in 252	1 in 25,000	1 in 100,000
		Finnish	99	1 in 323	1 in 32,000	1 in 130,000
		Non-Finnish European/Caucasian	99	1 in 169	1 in 17,000	1 in 68,000
		South Asian	99	1 in 209	1 in 21,000	1 in 84,000
		Other (population not assigned)	99	1 in 359	1 in 36,000	1 in 140,000
	OTOF	African/African American	99	1 in 201	1 in 20,000	1 in 80,000
		Latino/Admixed American	99	1 in 258	1 in 26,000	1 in 100,000
		Ashkenazi Jewish	99	1 in 78	1 in 7,700	1 in 31,000
		East Asian	99	1 in 52	1 in 5,100	1 in 20,000
Finnish		99	1 in 903	1 in 90,000	1 in 360,000	

Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

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Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result		
Disorder		Non-Finnish European/Caucasian	99	1 in 228	1 in 23,000	1 in 92,000		
		South Asian	99	1 in 240	1 in 24,000	1 in 96,000		
		Other (population not assigned)	99	1 in 229	1 in 23,000	1 in 92,000		
	POU3F4	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		
	SYNE4	African/African American	99	1 in 964	1 in 96,000	1 in 380,000		
		Latino/Admixed American	99	1 in 1,594	1 in 160,000	1 in 640,000		
		Ashkenazi Jewish	99	1 in 4,610	1 in 460,000	1 in 1,800,000		
		East Asian	98	1 in 1,085	1 in 54,000	1 in 220,000		
		Finnish	99	1 in 2,670	1 in 270,000	1 in 1,100,000		
		Non-Finnish European/Caucasian	99	1 in 478	1 in 48,000	1 in 190,000		
		South Asian	98	1 in 597	1 in 30,000	1 in 120,000		
	Other (population not assigned)	99	1 in 448	1 in 45,000	1 in 180,000			
	Dent disease	CLCN5	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		
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		
OCRL		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		
		Dihydropyrimidine dehydrogenase deficiency	DLD	African/African American	99	1 in 1,234	1 in 120,000	1 in 480,000
				Latino/Admixed American	99	1 in 1,692	1 in 170,000	1 in 680,000
				Ashkenazi Jewish	99	1 in 61	1 in 6,000	1 in 24,000
				East Asian	98	1 in 2,239	1 in 110,000	1 in 440,000
Finnish	99			1 in 1,796	1 in 180,000	1 in 720,000		
Non-Finnish European/Caucasian	99			1 in 968	1 in 97,000	1 in 390,000		
South Asian	98			1 in 1,293	1 in 65,000	1 in 260,000		
Other (population not assigned)	99			1 in 912	1 in 91,000	1 in 360,000		
Latino/Admixed American	98			1 in 710	1 in 35,000	1 in 140,000		
Dihydropyrimidine dehydrogenase deficiency	DPYD	African/African American	99	1 in 970	1 in 97,000	1 in 390,000		
		Latino/Admixed American	98	1 in 710	1 in 35,000	1 in 140,000		

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		Ashkenazi Jewish	99	1 in 2,146	1 in 210,000	1 in 840,000
		East Asian	99	1 in 196	1 in 20,000	1 in 80,000
		Finnish	98	1 in 2,176	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	98	1 in 558	1 in 28,000	1 in 110,000
		South Asian	97	1 in 248	1 in 8,200	1 in 33,000
		Other (population not assigned)	98	1 in 1,532	1 in 77,000	1 in 310,000
Distal spinal muscular atrophy, autosomal recessive	PLEKHG5	African/African American	97	1 in 45	1 in 1,500	1 in 6,000
		Latino/Admixed American	98	1 in 250	1 in 12,000	1 in 48,000
		Ashkenazi Jewish	97	1 in 2,014	1 in 67,000	1 in 270,000
		East Asian	99	1 in 614	1 in 61,000	1 in 240,000
		Finnish	97	1 in 1,252	1 in 42,000	1 in 170,000
		Non-Finnish European/Caucasian	99	1 in 693	1 in 69,000	1 in 280,000
		South Asian	99	1 in 433	1 in 43,000	1 in 170,000
		Other (population not assigned)	97	1 in 168	1 in 5,600	1 in 22,000
		Donnai-Barrow syndrome	LRP2	African/African American	99	1 in 334
Latino/Admixed American	99			1 in 403	1 in 40,000	1 in 160,000
Ashkenazi Jewish	99			1 in 171	1 in 17,000	1 in 68,000
East Asian	99			1 in 825	1 in 82,000	1 in 330,000
Finnish	99			1 in 408	1 in 41,000	1 in 160,000
Non-Finnish European/Caucasian	99			1 in 260	1 in 26,000	1 in 100,000
South Asian	99			1 in 405	1 in 40,000	1 in 160,000
Other (population not assigned)	99			1 in 254	1 in 25,000	1 in 100,000
Dystrophinopathies, including Duchenne and Becker muscular dystrophy and X-linked cardiomyopathy	DMD			African/African American	99	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
		Early infantile epileptic encephalopathy	CAD	African/African American	99	1 in 59
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
ITPA	African/African American			98	1 in 3,918	1 in 200,000
	Latino/Admixed American		99	1 in 5,707	1 in 570,000	1 in 2,300,000
	Ashkenazi Jewish		97	1 in 4,918	1 in 160,000	1 in 640,000
	East Asian		98	1 in 1,838	1 in 92,000	1 in 370,000
	Finnish		99	1 in 1,195	1 in 120,000	1 in 480,000
	Non-Finnish European/Caucasian		98	1 in 2,093	1 in 100,000	1 in 400,000
	South Asian		99	1 in 7,654	1 in 770,000	1 in 3,100,000

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		Other (population not assigned)	99	1 in 3,068	1 in 310,000	1 in 1,200,000
Ehlers-Danlos syndrome type VIIC	ADAMTS2	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
Emery-Dreifuss muscular dystrophy	EMD	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
	FHL1	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	97	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
Ethylmalonic encephalopathy	ETHE1	African/African American	99	1 in 833	1 in 83,000	1 in 330,000
		Latino/Admixed American	99	1 in 521	1 in 52,000	1 in 210,000
		Ashkenazi Jewish	99	1 in 4,985	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 1,417	1 in 140,000	1 in 560,000
		Finnish	99	1 in 2,006	1 in 200,000	1 in 800,000
		Non-Finnish European/Caucasian	99	1 in 953	1 in 95,000	1 in 380,000
		South Asian	99	1 in 782	1 in 78,000	1 in 310,000
		Other (population not assigned)	99	1 in 505	1 in 50,000	1 in 200,000
Fabry disease	GLA	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
Familial dysautonomia	ELP1	African/African American	99	1 in 594	1 in 59,000	1 in 240,000
		Latino/Admixed American	98	1 in 808	1 in 40,000	1 in 160,000
		Ashkenazi Jewish	99	1 in 37	1 in 3,600	1 in 14,000
		East Asian	99	1 in 657	1 in 66,000	1 in 260,000

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		Finnish	98	1 in 3,586	1 in 180,000	1 in 720,000		
		Non-Finnish European/Caucasian	99	1 in 782	1 in 78,000	1 in 310,000		
		South Asian	99	1 in 853	1 in 85,000	1 in 340,000		
		Other (population not assigned)	98	1 in 410	1 in 20,000	1 in 80,000		
Familial hemophagocytic lymphohistiocytosis	PRF1	African/African American	97	1 in 126	1 in 4,200	1 in 17,000		
		Latino/Admixed American	99	1 in 429	1 in 43,000	1 in 170,000		
		Ashkenazi Jewish	99	1 in 458	1 in 46,000	1 in 180,000		
		East Asian	97	1 in 1,099	1 in 37,000	1 in 150,000		
		Finnish	97	1 in 10,775	1 in 360,000	1 in 1,400,000		
		Non-Finnish European/Caucasian	99	1 in 308	1 in 31,000	1 in 120,000		
		South Asian	99	1 in 226	1 in 23,000	1 in 92,000		
		Other (population not assigned)	99	1 in 320	1 in 32,000	1 in 130,000		
		STX11	African/African American	98	N/A	N/A	N/A	
			Latino/Admixed American	99	1 in 664	1 in 66,000	1 in 260,000	
			Ashkenazi Jewish	98	N/A	N/A	N/A	
			East Asian	98	1 in 4,592	1 in 230,000	1 in 920,000	
	Finnish		98	N/A	N/A	N/A		
	Non-Finnish European/Caucasian		99	1 in 13,964	1 in 1,400,000	1 in 5,600,000		
	South Asian		98	1 in 3,837	1 in 190,000	1 in 760,000		
	Other (population not assigned)		98	N/A	N/A	N/A		
	STXBP2		African/African American	99	1 in 226	1 in 23,000	1 in 92,000	
			Latino/Admixed American	99	1 in 1,505	1 in 150,000	1 in 600,000	
			Ashkenazi Jewish	97	1 in 356	1 in 12,000	1 in 48,000	
			East Asian	99	1 in 1,459	1 in 150,000	1 in 600,000	
		Finnish	99	1 in 10,752	1 in 1,100,000	1 in 4,400,000		
		Non-Finnish European/Caucasian	99	1 in 406	1 in 41,000	1 in 160,000		
		South Asian	99	1 in 735	1 in 73,000	1 in 290,000		
		Other (population not assigned)	99	1 in 773	1 in 77,000	1 in 310,000		
		UNC13D	African/African American	99	1 in 110	1 in 11,000	1 in 44,000	
			Latino/Admixed American	99	1 in 258	1 in 26,000	1 in 100,000	
			Ashkenazi Jewish	99	1 in 829	1 in 83,000	1 in 330,000	
			East Asian	99	1 in 106	1 in 11,000	1 in 44,000	
	Finnish		99	1 in 179	1 in 18,000	1 in 72,000		
	Non-Finnish European/Caucasian		99	1 in 108	1 in 11,000	1 in 44,000		
	South Asian		99	1 in 186	1 in 19,000	1 in 76,000		
	Other (population not assigned)		99	1 in 163	1 in 16,000	1 in 64,000		
	Familial hyperinsulinism		ABCC8	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		
Familial Mediterranean fever		MEFV		African/African American	99	1 in 426	1 in 43,000	1 in 170,000

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		Latino/Admixed American	99	1 in 246	1 in 25,000	1 in 100,000
		Ashkenazi Jewish	99	1 in 13	1 in 1,200	1 in 4,800
		East Asian	99	1 in 114	1 in 11,000	1 in 44,000
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 186	1 in 19,000	1 in 76,000
		South Asian	99	1 in 309	1 in 31,000	1 in 120,000
		Other (population not assigned)	99	1 in 64	1 in 6,300	1 in 25,000
Fanconi anemia	BRIP1	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
	FANCA	African/African American	99	1 in 88	1 in 8,700	1 in 35,000
		Latino/Admixed American	99	1 in 154	1 in 15,000	1 in 60,000
		Ashkenazi Jewish	99	1 in 168	1 in 17,000	1 in 68,000
		East Asian	98	1 in 87	1 in 4,300	1 in 17,000
		Finnish	99	1 in 228	1 in 23,000	1 in 92,000
		Non-Finnish European/Caucasian	99	1 in 108	1 in 11,000	1 in 44,000
		South Asian	99	1 in 104	1 in 10,000	1 in 40,000
		Other (population not assigned)	99	1 in 153	1 in 15,000	1 in 60,000
	FANCB	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
	FANCC	African/African American	98	1 in 659	1 in 33,000	1 in 130,000
		Latino/Admixed American	98	1 in 1,197	1 in 60,000	1 in 240,000
		Ashkenazi Jewish	99	1 in 84	1 in 8,300	1 in 33,000
		East Asian	99	1 in 2,995	1 in 300,000	1 in 1,200,000
		Finnish	98	1 in 5,493	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	98	1 in 548	1 in 27,000	1 in 110,000
		South Asian	99	1 in 960	1 in 96,000	1 in 380,000
		Other (population not assigned)	99	1 in 1,368	1 in 140,000	1 in 560,000
	FANCD2	African/African American	98	1 in 109	1 in 5,400	1 in 22,000
Latino/Admixed American		99	1 in 322	1 in 32,000	1 in 130,000	
Ashkenazi Jewish		98	1 in 414	1 in 21,000	1 in 84,000	
East Asian		98	1 in 372	1 in 19,000	1 in 76,000	
Finnish		98	1 in 256	1 in 13,000	1 in 52,000	
Non-Finnish European/Caucasian		98	1 in 177	1 in 8,800	1 in 35,000	

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 Disorder)

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Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result	
		South Asian	98	1 in 437	1 in 22,000	1 in 88,000	
		Other (population not assigned)	98	1 in 112	1 in 5,600	1 in 22,000	
	<b>FANCE</b>	African/African American	99	1 in 427	1 in 43,000	1 in 170,000	
		Latino/Admixed American	98	1 in 1,434	1 in 72,000	1 in 290,000	
		Ashkenazi Jewish	99	1 in 5,030	1 in 500,000	1 in 2,000,000	
		East Asian	99	1 in 1,148	1 in 110,000	1 in 440,000	
		Finnish	97	N/A	N/A	N/A	
		Non-Finnish European/Caucasian	99	1 in 916	1 in 92,000	1 in 370,000	
		South Asian	98	1 in 874	1 in 44,000	1 in 180,000	
		Other (population not assigned)	97	1 in 984	1 in 33,000	1 in 130,000	
	<b>FANCF</b>	African/African American	98	1 in 1,104	1 in 55,000	1 in 220,000	
		Latino/Admixed American	99	1 in 1,909	1 in 190,000	1 in 760,000	
		Ashkenazi Jewish	97	N/A	N/A	N/A	
		East Asian	98	1 in 1,836	1 in 92,000	1 in 370,000	
		Finnish	97	1 in 10,825	1 in 360,000	1 in 1,400,000	
		Non-Finnish European/Caucasian	98	1 in 909	1 in 45,000	1 in 180,000	
		South Asian	98	1 in 1,707	1 in 85,000	1 in 340,000	
		Other (population not assigned)	99	1 in 1,022	1 in 100,000	1 in 400,000	
	<b>FANCG</b>	African/African American	97	1 in 605	1 in 20,000	1 in 80,000	
		Latino/Admixed American	99	1 in 1,550	1 in 150,000	1 in 600,000	
		Ashkenazi Jewish	97	N/A	N/A	N/A	
		East Asian	99	1 in 1,087	1 in 110,000	1 in 440,000	
		Finnish	97	1 in 1,423	1 in 47,000	1 in 190,000	
		Non-Finnish European/Caucasian	98	1 in 697	1 in 35,000	1 in 140,000	
		South Asian	98	1 in 1,166	1 in 58,000	1 in 230,000	
		Other (population not assigned)	97	1 in 930	1 in 31,000	1 in 120,000	
	<b>FANCI</b>	African/African American	98	1 in 404	1 in 20,000	1 in 80,000	
		Latino/Admixed American	98	1 in 524	1 in 26,000	1 in 100,000	
		Ashkenazi Jewish	99	1 in 5,038	1 in 500,000	1 in 2,000,000	
		East Asian	99	1 in 459	1 in 46,000	1 in 180,000	
		Finnish	98	1 in 113	1 in 5,600	1 in 22,000	
		Non-Finnish European/Caucasian	99	1 in 557	1 in 56,000	1 in 220,000	
		South Asian	98	1 in 373	1 in 19,000	1 in 76,000	
		Other (population not assigned)	98	1 in 511	1 in 26,000	1 in 100,000	
	<b>FANCL</b>	African/African American	97	1 in 295	1 in 9,800	1 in 39,000	
		Latino/Admixed American	97	1 in 79	1 in 2,600	1 in 10,000	
		Ashkenazi Jewish	97	1 in 68	1 in 2,200	1 in 8,800	
		East Asian	98	1 in 1,530	1 in 76,000	1 in 300,000	
		Finnish	97	1 in 263	1 in 8,700	1 in 35,000	
		Non-Finnish European/Caucasian	97	1 in 102	1 in 3,400	1 in 14,000	
		South Asian	97	1 in 288	1 in 9,600	1 in 38,000	
		Other (population not assigned)	97	1 in 82	1 in 2,700	1 in 11,000	
	<b>Fragile X syndrome</b>	<b>FMR1</b>	African/African American	99	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

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 Disorder)

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N/A= Not Available

Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result		
		East Asian	99	N/A	N/A	N/A		
		Finnish	97	N/A	N/A	N/A		
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A		
		South Asian	99	N/A	N/A	N/A		
		Other (population not assigned)	97	N/A	N/A	N/A		
Fraser syndrome	FRAS1	African/African American	98	1 in 259	1 in 13,000	1 in 52,000		
		Latino/Admixed American	97	1 in 111	1 in 3,700	1 in 15,000		
		Ashkenazi Jewish	98	1 in 492	1 in 25,000	1 in 100,000		
		East Asian	98	1 in 156	1 in 7,800	1 in 31,000		
		Finnish	98	1 in 898	1 in 45,000	1 in 180,000		
		Non-Finnish European/Caucasian	98	1 in 300	1 in 15,000	1 in 60,000		
		South Asian	98	1 in 174	1 in 8,700	1 in 35,000		
		Other (population not assigned)	97	1 in 228	1 in 7,600	1 in 30,000		
		FREM2	African/African American	99	1 in 677	1 in 68,000	1 in 270,000	
	Latino/Admixed American		99	1 in 279	1 in 28,000	1 in 110,000		
	Ashkenazi Jewish		99	1 in 315	1 in 31,000	1 in 120,000		
	East Asian		99	1 in 518	1 in 52,000	1 in 210,000		
	Finnish		99	1 in 89	1 in 8,800	1 in 35,000		
	Non-Finnish European/Caucasian		99	1 in 115	1 in 11,000	1 in 44,000		
	South Asian		99	1 in 49	1 in 4,800	1 in 19,000		
	Other (population not assigned)		99	1 in 134	1 in 13,000	1 in 52,000		
	GRIP1		African/African American	98	1 in 669	1 in 33,000	1 in 130,000	
		Latino/Admixed American	99	1 in 187	1 in 19,000	1 in 76,000		
		Ashkenazi Jewish	99	1 in 82	1 in 8,100	1 in 32,000		
		East Asian	99	1 in 1,032	1 in 100,000	1 in 400,000		
		Finnish	97	1 in 901	1 in 30,000	1 in 120,000		
		Non-Finnish European/Caucasian	99	1 in 333	1 in 33,000	1 in 130,000		
		South Asian	99	1 in 314	1 in 31,000	1 in 120,000		
		Other (population not assigned)	99	1 in 137	1 in 14,000	1 in 56,000		
		Fucosidosis	FUCA1	African/African American	98	1 in 2,032	1 in 100,000	1 in 400,000
	Latino/Admixed American			99	1 in 1,233	1 in 120,000	1 in 480,000	
	Ashkenazi Jewish			99	1 in 5,034	1 in 500,000	1 in 2,000,000	
	East Asian			99	1 in 2,875	1 in 290,000	1 in 1,200,000	
Finnish	98			N/A	N/A	N/A		
Non-Finnish European/Caucasian	99			1 in 1,149	1 in 110,000	1 in 440,000		
South Asian	99			1 in 2,554	1 in 260,000	1 in 1,000,000		
Other (population not assigned)	99			1 in 2,056	1 in 210,000	1 in 840,000		
Galactosemia	GALE			African/African American	99	1 in 2,624	1 in 260,000	1 in 1,000,000
				Latino/Admixed American	99	1 in 1,217	1 in 120,000	1 in 480,000
				Ashkenazi Jewish	97	1 in 4,888	1 in 160,000	1 in 640,000
		East Asian	99	1 in 369	1 in 37,000	1 in 150,000		
		Finnish	97	1 in 10,825	1 in 360,000	1 in 1,400,000		
		Non-Finnish European/Caucasian	99	1 in 2,011	1 in 200,000	1 in 800,000		
		South Asian	99	1 in 2,541	1 in 250,000	1 in 1,000,000		
Other (population not assigned)	99	1 in 915	1 in 91,000	1 in 360,000				

Footnotes:

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

a Feng et al., PMID 28125085

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

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N/A= Not Available

Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result	
Disorder	GALK1	African/African American	99	1 in 652	1 in 65,000	1 in 260,000	
		Latino/Admixed American	97	1 in 57	1 in 1,900	1 in 7,600	
		Ashkenazi Jewish	99	1 in 453	1 in 45,000	1 in 180,000	
		East Asian	99	1 in 430	1 in 43,000	1 in 170,000	
		Finnish	99	1 in 2,150	1 in 210,000	1 in 840,000	
		Non-Finnish European/Caucasian	98	1 in 1,165	1 in 58,000	1 in 230,000	
		South Asian	97	1 in 520	1 in 17,000	1 in 68,000	
		Other (population not assigned)	98	1 in 1,523	1 in 76,000	1 in 300,000	
		GALT	African/African American	99	1 in 104	1 in 10,000	1 in 40,000
	Latino/Admixed American		99	1 in 142	1 in 14,000	1 in 56,000	
	Ashkenazi Jewish		99	1 in 2,489	1 in 250,000	1 in 1,000,000	
	East Asian		99	1 in 687	1 in 69,000	1 in 280,000	
	Finnish		99	1 in 5,575	1 in 560,000	1 in 2,200,000	
	Non-Finnish European/Caucasian		99	1 in 132	1 in 13,000	1 in 52,000	
	South Asian		99	1 in 385	1 in 38,000	1 in 150,000	
	Other (population not assigned)		99	1 in 235	1 in 23,000	1 in 92,000	
	Galactosialidosis		CTSA	African/African American	97	1 in 127	1 in 4,200
		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	
Gaucher disease		GBA		African/African American	87	1 in 139	1 in 1,100
	Latino/Admixed American		87	1 in 148	1 in 1,100	1 in 4,400	
	Ashkenazi Jewish		97	1 in 15	1 in 470	1 in 1,900	
	East Asian		87	1 in 166	1 in 1,300	1 in 5,200	
	Finnish		87	1 in 191	1 in 1,500	1 in 6,000	
	Non-Finnish European/Caucasian		87	1 in 138	1 in 1,100	1 in 4,400	
	South Asian		87	1 in 202	1 in 1,500	1 in 6,000	
	Other (population not assigned)		87	1 in 163	1 in 1,200	1 in 4,800	
	Glutaric acidemia type I		GCDH	African/African American	99	1 in 89	1 in 8,800
Latino/Admixed American		99		1 in 234	1 in 23,000	1 in 92,000	
Ashkenazi Jewish		99		1 in 1,680	1 in 170,000	1 in 680,000	
East Asian		99		1 in 212	1 in 21,000	1 in 84,000	
Finnish		99		1 in 507	1 in 51,000	1 in 200,000	
Non-Finnish European/Caucasian		99		1 in 219	1 in 22,000	1 in 88,000	
South Asian		98		1 in 172	1 in 8,600	1 in 34,000	
Other (population not assigned)		99		1 in 254	1 in 25,000	1 in 100,000	
Glutaric acidemia type II		TFFA		African/African American	97	1 in 172	1 in 5,700
	Latino/Admixed American		99	1 in 1,149	1 in 110,000	1 in 440,000	
	Ashkenazi Jewish		99	1 in 2,445	1 in 240,000	1 in 960,000	
	East Asian		99	1 in 561	1 in 56,000	1 in 220,000	
	Finnish		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

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

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		Non-Finnish European/Caucasian	99	1 in 733	1 in 73,000	1 in 290,000		
		South Asian	99	1 in 911	1 in 91,000	1 in 360,000		
		Other (population not assigned)	98	1 in 837	1 in 42,000	1 in 170,000		
	ETFB	African/African American	99	1 in 8,122	1 in 810,000	1 in 3,200,000		
		Latino/Admixed American	98	1 in 821	1 in 41,000	1 in 160,000		
		Ashkenazi Jewish	97	N/A	N/A	N/A		
		East Asian	99	1 in 9,195	1 in 920,000	1 in 3,700,000		
		Finnish	97	1 in 5,406	1 in 180,000	1 in 720,000		
		Non-Finnish European/Caucasian	98	1 in 1,593	1 in 80,000	1 in 320,000		
		South Asian	98	1 in 1,021	1 in 51,000	1 in 200,000		
		Other (population not assigned)	98	1 in 1,448	1 in 72,000	1 in 290,000		
		ETFDH	African/African American	99	1 in 374	1 in 37,000	1 in 150,000	
	Latino/Admixed American		98	1 in 312	1 in 16,000	1 in 64,000		
	Ashkenazi Jewish		99	1 in 406	1 in 41,000	1 in 160,000		
	East Asian		99	1 in 235	1 in 23,000	1 in 92,000		
	Finnish		99	1 in 531	1 in 53,000	1 in 210,000		
	Non-Finnish European/Caucasian		99	1 in 389	1 in 39,000	1 in 160,000		
	South Asian		99	1 in 1,075	1 in 110,000	1 in 440,000		
	Other (population not assigned)		99	1 in 407	1 in 41,000	1 in 160,000		
	Glutathione synthetase deficiency		GSS	African/African American	99	1 in 49	1 in 4,800	1 in 19,000
Latino/Admixed American		99		1 in 618	1 in 62,000	1 in 250,000		
Ashkenazi Jewish		97		1 in 112	1 in 3,700	1 in 15,000		
East Asian		99		1 in 297	1 in 30,000	1 in 120,000		
Finnish		99		1 in 3,680	1 in 370,000	1 in 1,500,000		
Non-Finnish European/Caucasian		99		1 in 992	1 in 99,000	1 in 400,000		
South Asian		98		1 in 1,703	1 in 85,000	1 in 340,000		
Other (population not assigned)		98		1 in 438	1 in 22,000	1 in 88,000		
Glycine encephalopathy		AMT		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		
	GLDC		African/African American	99	1 in 1,141	1 in 110,000	1 in 440,000	
			Latino/Admixed American	99	1 in 584	1 in 58,000	1 in 230,000	
Ashkenazi Jewish		98	1 in 2,518	1 in 130,000	1 in 520,000			
East Asian		99	1 in 460	1 in 46,000	1 in 180,000			
Finnish		99	1 in 118	1 in 12,000	1 in 48,000			
Non-Finnish European/Caucasian		99	1 in 388	1 in 39,000	1 in 160,000			
South Asian		98	1 in 368	1 in 18,000	1 in 72,000			
Other (population not assigned)		99	1 in 457	1 in 46,000	1 in 180,000			
Glycogen storage disease type I		G6PC	African/African American	99	1 in 985	1 in 98,000	1 in 390,000	
	Latino/Admixed American		98	1 in 374	1 in 19,000	1 in 76,000		

Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

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Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result		
Disorder		Ashkenazi Jewish	99	1 in 74	1 in 7,300	1 in 29,000		
		East Asian	99	1 in 212	1 in 21,000	1 in 84,000		
		Finnish	99	1 in 529	1 in 53,000	1 in 210,000		
		Non-Finnish European/Caucasian	99	1 in 276	1 in 28,000	1 in 110,000		
		South Asian	99	1 in 15,385	1 in 1,500,000	1 in 6,000,000		
		Other (population not assigned)	99	1 in 474	1 in 47,000	1 in 190,000		
	SLC37A4	African/African American	99	1 in 139	1 in 14,000	1 in 56,000		
		Latino/Admixed American	98	1 in 833	1 in 42,000	1 in 170,000		
		Ashkenazi Jewish	97	1 in 1,221	1 in 41,000	1 in 160,000		
		East Asian	98	1 in 616	1 in 31,000	1 in 120,000		
		Finnish	99	1 in 1,208	1 in 120,000	1 in 480,000		
		Non-Finnish European/Caucasian	98	1 in 629	1 in 31,000	1 in 120,000		
		South Asian	99	1 in 189	1 in 19,000	1 in 76,000		
		Other (population not assigned)	99	1 in 315	1 in 31,000	1 in 120,000		
		Glycogen storage disease type III	AGL	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		
Glycogen storage disease type IV	GBE1			African/African American	98	1 in 590	1 in 29,000	1 in 120,000
		Latino/Admixed American	98	1 in 220	1 in 11,000	1 in 44,000		
		Ashkenazi Jewish	99	1 in 72	1 in 7,100	1 in 28,000		
		East Asian	99	1 in 1,124	1 in 110,000	1 in 440,000		
		Finnish	99	1 in 511	1 in 51,000	1 in 200,000		
		Non-Finnish European/Caucasian	99	1 in 180	1 in 18,000	1 in 72,000		
		South Asian	99	1 in 411	1 in 41,000	1 in 160,000		
		Other (population not assigned)	99	1 in 139	1 in 14,000	1 in 56,000		
		Glycogen storage disease type IX	PHKA1	African/African American	99	N/A	N/A	N/A
				Latino/Admixed American	99	N/A	N/A	N/A
Ashkenazi Jewish	99			N/A	N/A	N/A		
East Asian	99			N/A	N/A	N/A		
Finnish	99			N/A	N/A	N/A		
Non-Finnish European/Caucasian	99			N/A	N/A	N/A		
South Asian	98			N/A	N/A	N/A		
Other (population not assigned)	98			N/A	N/A	N/A		
PHKA2	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		

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 Disorder)

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

Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result	
		Other (population not assigned)	99	N/A	N/A	N/A	
	PHKB	African/African American	99	1 in 486	1 in 49,000	1 in 200,000	
		Latino/Admixed American	99	1 in 456	1 in 46,000	1 in 180,000	
		Ashkenazi Jewish	99	1 in 5,040	1 in 500,000	1 in 2,000,000	
		East Asian	98	1 in 432	1 in 22,000	1 in 88,000	
		Finnish	99	1 in 3,636	1 in 360,000	1 in 1,400,000	
		Non-Finnish European/Caucasian	99	1 in 310	1 in 31,000	1 in 120,000	
		South Asian	99	1 in 245	1 in 24,000	1 in 96,000	
		Other (population not assigned)	99	1 in 435	1 in 43,000	1 in 170,000	
	PHKG2	African/African American	97	1 in 176	1 in 5,800	1 in 23,000	
		Latino/Admixed American	98	1 in 393	1 in 20,000	1 in 80,000	
		Ashkenazi Jewish	97	1 in 2,514	1 in 84,000	1 in 340,000	
		East Asian	98	1 in 657	1 in 33,000	1 in 130,000	
		Finnish	97	1 in 375	1 in 12,000	1 in 48,000	
		Non-Finnish European/Caucasian	98	1 in 476	1 in 24,000	1 in 96,000	
		South Asian	98	1 in 657	1 in 33,000	1 in 130,000	
		Other (population not assigned)	97	1 in 437	1 in 15,000	1 in 60,000	
	Glycogen storage disease type V	PYGM	African/African American	99	1 in 160	1 in 16,000	1 in 64,000
			Latino/Admixed American	99	1 in 48	1 in 4,700	1 in 19,000
			Ashkenazi Jewish	99	1 in 197	1 in 20,000	1 in 80,000
East Asian			98	1 in 553	1 in 28,000	1 in 110,000	
Finnish			99	1 in 285	1 in 28,000	1 in 110,000	
Non-Finnish European/Caucasian			99	1 in 42	1 in 4,100	1 in 16,000	
South Asian			99	1 in 272	1 in 27,000	1 in 110,000	
Other (population not assigned)			99	1 in 72	1 in 7,100	1 in 28,000	
Glycogen storage disease type VII	PFKM	African/African American	99	1 in 62	1 in 6,100	1 in 24,000	
		Latino/Admixed American	99	1 in 647	1 in 65,000	1 in 260,000	
		Ashkenazi Jewish	98	1 in 108	1 in 5,400	1 in 22,000	
		East Asian	99	1 in 488	1 in 49,000	1 in 200,000	
		Finnish	97	1 in 773	1 in 26,000	1 in 100,000	
		Non-Finnish European/Caucasian	99	1 in 600	1 in 60,000	1 in 240,000	
		South Asian	98	1 in 1,165	1 in 58,000	1 in 230,000	
		Other (population not assigned)	99	1 in 280	1 in 28,000	1 in 110,000	
GM1 gangliosidosis and mucopolysaccharidosis type IVB	GLB1	African/African American	98	1 in 272	1 in 14,000	1 in 56,000	
		Latino/Admixed American	99	1 in 499	1 in 50,000	1 in 200,000	
		Ashkenazi Jewish	98	N/A	N/A	N/A	
		East Asian	99	1 in 489	1 in 49,000	1 in 200,000	
		Finnish	98	1 in 251	1 in 13,000	1 in 52,000	
		Non-Finnish European/Caucasian	99	1 in 343	1 in 34,000	1 in 140,000	
		South Asian	99	1 in 239	1 in 24,000	1 in 96,000	
		Other (population not assigned)	99	1 in 402	1 in 40,000	1 in 160,000	
GRACILE syndrome	BCS1L	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	

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 Disorder)

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N/A= Not Available

Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		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
Gyrate atrophy of choroid and retina	OAT	African/African American	97	1 in 2,600	1 in 87,000	1 in 350,000
		Latino/Admixed American	99	1 in 892	1 in 89,000	1 in 360,000
		Ashkenazi Jewish	99	1 in 615	1 in 61,000	1 in 240,000
		East Asian	99	1 in 2,935	1 in 290,000	1 in 1,200,000
		Finnish	99	1 in 142	1 in 14,000	1 in 56,000
		Non-Finnish European/Caucasian	99	1 in 426	1 in 43,000	1 in 170,000
		South Asian	99	1 in 904	1 in 90,000	1 in 360,000
		Other (population not assigned)	99	1 in 410	1 in 41,000	1 in 160,000
Hepatic venoocclusive disease with immunodeficiency	SP110	African/African American	99	1 in 677	1 in 68,000	1 in 270,000
		Latino/Admixed American	99	1 in 1,153	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	99	1 in 5,039	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 613	1 in 61,000	1 in 240,000
		Finnish	99	1 in 5,412	1 in 540,000	1 in 2,200,000
		Non-Finnish European/Caucasian	98	1 in 941	1 in 47,000	1 in 190,000
		South Asian	99	1 in 289	1 in 29,000	1 in 120,000
		Other (population not assigned)	99	1 in 1,023	1 in 100,000	1 in 400,000
Hereditary folate malabsorption	SLC46A1	African/African American	98	1 in 3,964	1 in 200,000	1 in 800,000
		Latino/Admixed American	99	1 in 2,698	1 in 270,000	1 in 1,100,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	98	1 in 1,773	1 in 89,000	1 in 360,000
		Finnish	99	1 in 8,724	1 in 870,000	1 in 3,500,000
		Non-Finnish European/Caucasian	98	1 in 2,468	1 in 120,000	1 in 480,000
		South Asian	99	1 in 1,256	1 in 130,000	1 in 520,000
		Other (population not assigned)	98	N/A	N/A	N/A
Hereditary fructose Intolerance	ALDOB	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
Hereditary spastic paraplegia	CYP7B1	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
		SPG11	African/African American	98	1 in 64	1 in 3,200

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 Disorder)

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

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N/A= Not Available

Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result	
		Latino/Admixed American	97	1 in 232	1 in 7,700	1 in 31,000	
		Ashkenazi Jewish	99	1 in 358	1 in 36,000	1 in 140,000	
		East Asian	97	1 in 297	1 in 9,900	1 in 40,000	
		Finnish	98	1 in 739	1 in 37,000	1 in 150,000	
		Non-Finnish European/Caucasian	98	1 in 192	1 in 9,600	1 in 38,000	
		South Asian	98	1 in 267	1 in 13,000	1 in 52,000	
		Other (population not assigned)	97	1 in 292	1 in 9,700	1 in 39,000	
	SPG21	African/African American	98	1 in 1,626	1 in 81,000	1 in 320,000	
		Latino/Admixed American	99	1 in 1,668	1 in 170,000	1 in 680,000	
		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	97	N/A	N/A	N/A	
		Non-Finnish European/Caucasian	99	1 in 1,679	1 in 170,000	1 in 680,000	
		South Asian	97	1 in 7,655	1 in 260,000	1 in 1,000,000	
	Other (population not assigned)	99	1 in 3,066	1 in 310,000	1 in 1,200,000		
	TECPR2	African/African American	97	1 in 1,983	1 in 66,000	1 in 260,000	
		Latino/Admixed American	98	1 in 5,649	1 in 280,000	1 in 1,100,000	
		Ashkenazi Jewish	97	1 in 152	1 in 5,000	1 in 20,000	
		East Asian	98	1 in 823	1 in 41,000	1 in 160,000	
		Finnish	98	1 in 1,914	1 in 96,000	1 in 380,000	
		Non-Finnish European/Caucasian	98	1 in 1,946	1 in 97,000	1 in 390,000	
		South Asian	98	1 in 5,103	1 in 260,000	1 in 1,000,000	
	Other (population not assigned)	97	1 in 1,430	1 in 48,000	1 in 190,000		
	Hermansky-Pudlak syndrome	AP3B1	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
			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		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		
BLOC153		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		

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 Disorder)

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Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
	<b>BLOC156</b>	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
		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
		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	
	<b>DTNBP1</b>	African/African American	98	1 in 379	1 in 19,000	1 in 76,000
		Latino/Admixed American	98	1 in 253	1 in 13,000	1 in 52,000
		Ashkenazi Jewish	97	1 in 206	1 in 6,800	1 in 27,000
		East Asian	98	1 in 266	1 in 13,000	1 in 52,000
		Finnish	97	1 in 532	1 in 18,000	1 in 72,000
		Non-Finnish European/Caucasian	98	1 in 295	1 in 15,000	1 in 60,000
		South Asian	98	1 in 226	1 in 11,000	1 in 44,000
		Other (population not assigned)	97	1 in 228	1 in 7,600	1 in 30,000
		African/African American	98	1 in 2,695	1 in 130,000	1 in 520,000
	<b>HPS1</b>	Latino/Admixed American	98	1 in 1,057	1 in 53,000	1 in 210,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 2,279	1 in 110,000	1 in 440,000
		Finnish	99	1 in 10,813	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	97	1 in 1,250	1 in 42,000	1 in 170,000
		South Asian	99	1 in 1,085	1 in 110,000	1 in 440,000
		Other (population not assigned)	97	N/A	N/A	N/A
		African/African American	98	1 in 541	1 in 27,000	1 in 110,000
		Latino/Admixed American	99	1 in 1,150	1 in 110,000	1 in 440,000
	<b>HPS3</b>	Ashkenazi Jewish	99	1 in 242	1 in 24,000	1 in 96,000
		East Asian	99	1 in 289	1 in 29,000	1 in 120,000
		Finnish	98	1 in 524	1 in 26,000	1 in 100,000
		Non-Finnish European/Caucasian	99	1 in 590	1 in 59,000	1 in 240,000
		South Asian	99	1 in 322	1 in 32,000	1 in 130,000
		Other (population not assigned)	99	1 in 703	1 in 70,000	1 in 280,000
		African/African American	98	1 in 1,402	1 in 70,000	1 in 280,000
		Latino/Admixed American	99	1 in 384	1 in 38,000	1 in 150,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
	<b>HPS4</b>	East Asian	98	1 in 2,289	1 in 110,000	1 in 440,000
		Finnish	98	1 in 5,412	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 703	1 in 70,000	1 in 280,000
		South Asian	98	1 in 2,003	1 in 100,000	1 in 400,000
		Other (population not assigned)	97	1 in 745	1 in 25,000	1 in 100,000
		African/African American	97	1 in 69	1 in 2,300	1 in 9,200
		Latino/Admixed American	99	1 in 202	1 in 20,000	1 in 80,000
		Ashkenazi Jewish	97	1 in 275	1 in 9,100	1 in 36,000

Footnotes:

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

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Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		East Asian	98	1 in 471	1 in 24,000	1 in 96,000
		Finnish	99	1 in 145	1 in 14,000	1 in 56,000
		Non-Finnish European/Caucasian	99	1 in 89	1 in 8,800	1 in 35,000
		South Asian	99	1 in 201	1 in 20,000	1 in 80,000
		Other (population not assigned)	99	1 in 134	1 in 13,000	1 in 52,000
	HPS6	African/African American	98	1 in 1,121	1 in 56,000	1 in 220,000
		Latino/Admixed American	97	1 in 1,310	1 in 44,000	1 in 180,000
		Ashkenazi Jewish	97	1 in 4,990	1 in 170,000	1 in 680,000
		East Asian	98	1 in 1,090	1 in 54,000	1 in 220,000
		Finnish	99	1 in 1,352	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	98	1 in 745	1 in 37,000	1 in 150,000
		South Asian	98	1 in 1,767	1 in 88,000	1 in 350,000
		Other (population not assigned)	98	1 in 1,519	1 in 76,000	1 in 300,000
		HMG-CoA lyase deficiency	HMGCL	African/African American	99	1 in 440
Latino/Admixed American	99			1 in 130	1 in 13,000	1 in 52,000
Ashkenazi Jewish	98			1 in 2,244	1 in 110,000	1 in 440,000
East Asian	99			1 in 2,226	1 in 220,000	1 in 880,000
Finnish	99			1 in 490	1 in 49,000	1 in 200,000
Non-Finnish European/Caucasian	99			1 in 102	1 in 10,000	1 in 40,000
South Asian	98			1 in 197	1 in 9,800	1 in 39,000
Other (population not assigned)	99			1 in 136	1 in 14,000	1 in 56,000
Holocarboxylase synthetase deficiency	HLCS	African/African American	97	1 in 985	1 in 33,000	1 in 130,000
		Latino/Admixed American	99	1 in 1,073	1 in 110,000	1 in 440,000
		Ashkenazi Jewish	97	1 in 5,041	1 in 170,000	1 in 680,000
		East Asian	99	1 in 366	1 in 37,000	1 in 150,000
		Finnish	99	1 in 3,717	1 in 370,000	1 in 1,500,000
		Non-Finnish European/Caucasian	99	1 in 920	1 in 92,000	1 in 370,000
		South Asian	98	1 in 958	1 in 48,000	1 in 190,000
		Other (population not assigned)	98	1 in 724	1 in 36,000	1 in 140,000
Homocystinuria	CBS	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
HSD10 disease	HSD17B10	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

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 Disorder)

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Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result	
Hyaline fibromatosis syndrome	ANTXR2	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	
Hydrolethalus syndrome	HYLS1	African/African American	98	1 in 670	1 in 33,000	1 in 130,000	
		Latino/Admixed American	99	1 in 592	1 in 59,000	1 in 240,000	
		Ashkenazi Jewish	99	1 in 4,925	1 in 490,000	1 in 2,000,000	
		East Asian	99	1 in 1,150	1 in 110,000	1 in 440,000	
		Finnish	99	1 in 54	1 in 5,300	1 in 21,000	
		Non-Finnish European/Caucasian	99	1 in 443	1 in 44,000	1 in 180,000	
		South Asian	98	1 in 487	1 in 24,000	1 in 96,000	
		Other (population not assigned)	99	1 in 348	1 in 35,000	1 in 140,000	
Hypomyelination and congenital cataract	FAM126A	African/African American	97	1 in 1,999	1 in 67,000	1 in 270,000	
		Latino/Admixed American	98	1 in 2,458	1 in 120,000	1 in 480,000	
		Ashkenazi Jewish	99	1 in 1,402	1 in 140,000	1 in 560,000	
		East Asian	99	1 in 4,595	1 in 460,000	1 in 1,800,000	
		Finnish	97	1 in 8,219	1 in 270,000	1 in 1,100,000	
		Non-Finnish European/Caucasian	98	1 in 874	1 in 44,000	1 in 180,000	
		South Asian	97	1 in 717	1 in 24,000	1 in 96,000	
		Other (population not assigned)	98	1 in 780	1 in 39,000	1 in 160,000	
Hypophosphatasia	ALPL	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	
Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome	CDCA7	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	
		DNMT3B	African/African American	99	1 in 7,657	1 in 770,000	1 in 3,100,000
			Latino/Admixed American	99	1 in 3,433	1 in 340,000	1 in 1,400,000
	Ashkenazi Jewish		99	1 in 5,041	1 in 500,000	1 in 2,000,000	
	East Asian		99	1 in 8,621	1 in 860,000	1 in 3,400,000	
			Finnish	98	1 in 5,412	1 in 270,000	1 in 1,100,000

Footnotes:

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

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

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N/A= Not Available

Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Non-Finnish European/Caucasian	99	1 in 2,684	1 in 270,000	1 in 1,100,000
		South Asian	99	1 in 1,175	1 in 120,000	1 in 480,000
		Other (population not assigned)	98	1 in 1,448	1 in 72,000	1 in 290,000
	HELLS	African/African American	97	1 in 1,081	1 in 36,000	1 in 140,000
		Latino/Admixed American	97	1 in 481	1 in 16,000	1 in 64,000
		Ashkenazi Jewish	97	1 in 1,222	1 in 41,000	1 in 160,000
		East Asian	99	1 in 413	1 in 41,000	1 in 160,000
		Finnish	98	1 in 1,793	1 in 90,000	1 in 360,000
		Non-Finnish European/Caucasian	98	1 in 1,155	1 in 58,000	1 in 230,000
		South Asian	98	1 in 674	1 in 34,000	1 in 140,000
		Other (population not assigned)	98	1 in 624	1 in 31,000	1 in 120,000
	ZBTB24	African/African American	99	1 in 2,655	1 in 270,000	1 in 1,100,000
		Latino/Admixed American	98	1 in 2,868	1 in 140,000	1 in 560,000
		Ashkenazi Jewish	99	1 in 5,041	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 1,006	1 in 100,000	1 in 400,000
		Finnish	99	1 in 516	1 in 52,000	1 in 210,000
		Non-Finnish European/Caucasian	98	1 in 2,817	1 in 140,000	1 in 560,000
		South Asian	97	1 in 3,023	1 in 100,000	1 in 400,000
		Other (population not assigned)	98	1 in 1,535	1 in 77,000	1 in 310,000
	Immunodysregulation, polyendocrinopathy, and enteropathy	FOXP3	African/African American	98	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			99	N/A	N/A	N/A
Finnish			98	N/A	N/A	N/A
Non-Finnish European/Caucasian			98	N/A	N/A	N/A
South Asian			98	N/A	N/A	N/A
Other (population not assigned)			98	N/A	N/A	N/A
Inclusion body myopathy 2	GNE	African/African American	98	1 in 249	1 in 12,000	1 in 48,000
		Latino/Admixed American	99	1 in 793	1 in 79,000	1 in 320,000
		Ashkenazi Jewish	99	1 in 4,924	1 in 490,000	1 in 2,000,000
		East Asian	99	1 in 101	1 in 10,000	1 in 40,000
		Finnish	98	1 in 1,387	1 in 69,000	1 in 280,000
		Non-Finnish European/Caucasian	99	1 in 368	1 in 37,000	1 in 150,000
		South Asian	99	1 in 36	1 in 3,500	1 in 14,000
		Other (population not assigned)	99	1 in 252	1 in 25,000	1 in 100,000
Isovaleric acidemia	IVD	African/African American	98	1 in 461	1 in 23,000	1 in 92,000
		Latino/Admixed American	99	1 in 360	1 in 36,000	1 in 140,000
		Ashkenazi Jewish	99	1 in 420	1 in 42,000	1 in 170,000
		East Asian	99	1 in 303	1 in 30,000	1 in 120,000
		Finnish	98	1 in 1,099	1 in 55,000	1 in 220,000
		Non-Finnish European/Caucasian	99	1 in 232	1 in 23,000	1 in 92,000
		South Asian	99	1 in 507	1 in 51,000	1 in 200,000
		Other (population not assigned)	99	1 in 207	1 in 21,000	1 in 84,000
Joubert syndrome and related disorders, including Meckel-Gruber syndrome	AH1	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

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**Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Disorder)**

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Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result	
	<b>ARL13B</b>	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	
		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	
		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	
		<b>B9D1</b>	African/African American	99	1 in 643	1 in 64,000	1 in 260,000
		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		
	<b>B9D2</b>	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		
	<b>CEP104</b>	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		
	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		
	<b>CPLANE1</b>	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		

Footnotes:

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

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		Other (population not assigned)	99	1 in 44	1 in 4,300	1 in 17,000
	<b>INPP5E</b>	African/African American	98	1 in 397	1 in 20,000	1 in 80,000
		Latino/Admixed American	99	1 in 1,685	1 in 170,000	1 in 680,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 1,454	1 in 150,000	1 in 600,000
		Finnish	97	1 in 735	1 in 24,000	1 in 96,000
		Non-Finnish European/Caucasian	97	1 in 156	1 in 5,200	1 in 21,000
		South Asian	98	1 in 1,600	1 in 80,000	1 in 320,000
		Other (population not assigned)	99	1 in 1,933	1 in 190,000	1 in 760,000
	<b>KIF14</b>	African/African American	98	1 in 666	1 in 33,000	1 in 130,000
		Latino/Admixed American	98	1 in 1,218	1 in 61,000	1 in 240,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 599	1 in 20,000	1 in 80,000
		Finnish	97	1 in 2,690	1 in 90,000	1 in 360,000
		Non-Finnish European/Caucasian	98	1 in 1,010	1 in 50,000	1 in 200,000
		South Asian	97	1 in 362	1 in 12,000	1 in 48,000
		Other (population not assigned)	98	1 in 767	1 in 38,000	1 in 150,000
	<b>NPHP1</b>	African/African American	98	1 in 218	1 in 11,000	1 in 44,000
		Latino/Admixed American	98	1 in 315	1 in 16,000	1 in 64,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 164	1 in 16,000	1 in 64,000
		Finnish	98	1 in 806	1 in 40,000	1 in 160,000
		Non-Finnish European/Caucasian	98	1 in 418	1 in 21,000	1 in 84,000
		South Asian	98	1 in 536	1 in 27,000	1 in 110,000
		Other (population not assigned)	98	1 in 362	1 in 18,000	1 in 72,000
	<b>NPHP3</b>	African/African American	99	1 in 42	1 in 4,100	1 in 16,000
		Latino/Admixed American	98	1 in 69	1 in 3,400	1 in 14,000
		Ashkenazi Jewish	97	1 in 219	1 in 7,300	1 in 29,000
		East Asian	99	1 in 67	1 in 6,600	1 in 26,000
		Finnish	99	1 in 78	1 in 7,700	1 in 31,000
		Non-Finnish European/Caucasian	99	1 in 73	1 in 7,200	1 in 29,000
		South Asian	97	1 in 57	1 in 1,900	1 in 7,600
		Other (population not assigned)	98	1 in 66	1 in 3,300	1 in 13,000
	<b>RPGRIP1L</b>	African/African American	98	1 in 293	1 in 15,000	1 in 60,000
		Latino/Admixed American	98	1 in 605	1 in 30,000	1 in 120,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 326	1 in 11,000	1 in 44,000
		Finnish	99	1 in 1,538	1 in 150,000	1 in 600,000
		Non-Finnish European/Caucasian	99	1 in 378	1 in 38,000	1 in 150,000
		South Asian	98	1 in 330	1 in 16,000	1 in 64,000
		Other (population not assigned)	98	1 in 490	1 in 24,000	1 in 96,000
	<b>TCTN1</b>	African/African American	99	1 in 320	1 in 32,000	1 in 130,000
		Latino/Admixed American	99	1 in 387	1 in 39,000	1 in 160,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 1,257	1 in 130,000	1 in 520,000

Footnotes:

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b Luo et al., PMID 23788250

**Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Disorder)**

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

Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result	
		Finnish	97	1 in 709	1 in 24,000	1 in 96,000	
		Non-Finnish European/Caucasian	99	1 in 520	1 in 52,000	1 in 210,000	
		South Asian	97	1 in 125	1 in 4,100	1 in 16,000	
		Other (population not assigned)	98	1 in 468	1 in 23,000	1 in 92,000	
	TCTN2	African/African American	98	1 in 572	1 in 29,000	1 in 120,000	
		Latino/Admixed American	99	1 in 859	1 in 86,000	1 in 340,000	
		Ashkenazi Jewish	98	N/A	N/A	N/A	
		East Asian	99	1 in 2,299	1 in 230,000	1 in 920,000	
		Finnish	98	N/A	N/A	N/A	
		Non-Finnish European/Caucasian	99	1 in 1,046	1 in 100,000	1 in 400,000	
		South Asian	98	1 in 766	1 in 38,000	1 in 150,000	
		Other (population not assigned)	98	1 in 1,440	1 in 72,000	1 in 290,000	
		TCTN3	African/African American	99	1 in 489	1 in 49,000	1 in 200,000
			Latino/Admixed American	97	1 in 257	1 in 8,500	1 in 34,000
	Ashkenazi Jewish		99	1 in 410	1 in 41,000	1 in 160,000	
	East Asian		98	1 in 786	1 in 39,000	1 in 160,000	
	Finnish		99	1 in 10,821	1 in 1,100,000	1 in 4,400,000	
	Non-Finnish European/Caucasian		98	1 in 487	1 in 24,000	1 in 96,000	
	South Asian		97	1 in 552	1 in 18,000	1 in 72,000	
	Other (population not assigned)		98	1 in 1,274	1 in 64,000	1 in 260,000	
	TMEM67		African/African American	99	1 in 309	1 in 31,000	1 in 120,000
			Latino/Admixed American	99	1 in 233	1 in 23,000	1 in 92,000
		Ashkenazi Jewish	99	1 in 164	1 in 16,000	1 in 64,000	
		East Asian	99	1 in 132	1 in 13,000	1 in 52,000	
		Finnish	99	1 in 321	1 in 32,000	1 in 130,000	
		Non-Finnish European/Caucasian	99	1 in 147	1 in 15,000	1 in 60,000	
		South Asian	99	1 in 206	1 in 21,000	1 in 84,000	
		Other (population not assigned)	99	1 in 101	1 in 10,000	1 in 40,000	
		TMEM138	African/African American	97	N/A	N/A	N/A
			Latino/Admixed American	99	1 in 4,323	1 in 430,000	1 in 1,700,000
	Ashkenazi Jewish		97	N/A	N/A	N/A	
	East Asian		97	1 in 4,599	1 in 150,000	1 in 600,000	
	Finnish		97	N/A	N/A	N/A	
	Non-Finnish European/Caucasian		98	1 in 5,161	1 in 260,000	1 in 1,000,000	
	South Asian		99	1 in 1,694	1 in 170,000	1 in 680,000	
	Other (population not assigned)		97	N/A	N/A	N/A	
	TMEM216		African/African American	99	1 in 7,745	1 in 770,000	1 in 3,100,000
			Latino/Admixed American	97	1 in 488	1 in 16,000	1 in 64,000
		Ashkenazi Jewish	99	1 in 141	1 in 14,000	1 in 56,000	
		East Asian	99	1 in 8,621	1 in 860,000	1 in 3,400,000	
		Finnish	97	N/A	N/A	N/A	
		Non-Finnish European/Caucasian	99	1 in 934	1 in 93,000	1 in 370,000	
		South Asian	99	1 in 73	1 in 7,200	1 in 29,000	
		Other (population not assigned)	99	1 in 378	1 in 38,000	1 in 150,000	
		TMEM231	African/African American	99	1 in 924	1 in 92,000	1 in 370,000

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 Disorder)**

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

Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result		
Disorder		Latino/Admixed American	99	1 in 869	1 in 87,000	1 in 350,000		
		Ashkenazi Jewish	99	1 in 2,463	1 in 250,000	1 in 1,000,000		
		East Asian	99	1 in 656	1 in 66,000	1 in 260,000		
		Finnish	99	1 in 2,060	1 in 210,000	1 in 840,000		
		Non-Finnish European/Caucasian	99	1 in 560	1 in 56,000	1 in 220,000		
		South Asian	99	1 in 392	1 in 39,000	1 in 160,000		
		Other (population not assigned)	99	1 in 968	1 in 97,000	1 in 390,000		
	TMEM237	African/African American	99	1 in 1,501	1 in 150,000	1 in 600,000		
		Latino/Admixed American	99	1 in 379	1 in 38,000	1 in 150,000		
		Ashkenazi Jewish	99	1 in 4,108	1 in 410,000	1 in 1,600,000		
		East Asian	98	1 in 368	1 in 18,000	1 in 72,000		
		Finnish	98	N/A	N/A	N/A		
		Non-Finnish European/Caucasian	99	1 in 942	1 in 94,000	1 in 380,000		
		South Asian	99	1 in 1,564	1 in 160,000	1 in 640,000		
		Other (population not assigned)	99	1 in 1,369	1 in 140,000	1 in 560,000		
		Junctional epidermolysis bullosa	LAMA3	African/African American	99	1 in 348	1 in 35,000	1 in 140,000
				Latino/Admixed American	99	1 in 288	1 in 29,000	1 in 120,000
	Ashkenazi Jewish			99	1 in 5,030	1 in 500,000	1 in 2,000,000	
	East Asian			99	1 in 70	1 in 6,900	1 in 28,000	
	Finnish			97	1 in 1,502	1 in 50,000	1 in 200,000	
	Non-Finnish European/Caucasian			99	1 in 320	1 in 32,000	1 in 130,000	
South Asian	98			1 in 201	1 in 10,000	1 in 40,000		
Other (population not assigned)	99			1 in 760	1 in 76,000	1 in 300,000		
LAMB3	African/African American		99	1 in 172	1 in 17,000	1 in 68,000		
	Latino/Admixed American		98	1 in 1,012	1 in 51,000	1 in 200,000		
	Ashkenazi Jewish		99	1 in 1,001	1 in 100,000	1 in 400,000		
	East Asian		98	1 in 1,007	1 in 50,000	1 in 200,000		
	Finnish		98	1 in 1,007	1 in 50,000	1 in 200,000		
	Non-Finnish European/Caucasian		99	1 in 252	1 in 25,000	1 in 100,000		
	South Asian		98	1 in 603	1 in 30,000	1 in 120,000		
	Other (population not assigned)		99	1 in 725	1 in 72,000	1 in 290,000		
LAMC2	African/African American		99	1 in 2,653	1 in 270,000	1 in 1,100,000		
	Latino/Admixed American		99	1 in 3,438	1 in 340,000	1 in 1,400,000		
	Ashkenazi Jewish		98	1 in 2,489	1 in 120,000	1 in 480,000		
	East Asian		99	1 in 1,022	1 in 100,000	1 in 400,000		
	Finnish		99	1 in 10,825	1 in 1,100,000	1 in 4,400,000		
	Non-Finnish European/Caucasian	98	1 in 1,067	1 in 53,000	1 in 210,000			
	South Asian	98	1 in 1,093	1 in 55,000	1 in 220,000			
	Other (population not assigned)	98	1 in 1,535	1 in 77,000	1 in 310,000			
Juvenile hereditary hemochromatosis	HAMP	African/African American	98	1 in 4,059	1 in 200,000	1 in 800,000		
		Latino/Admixed American	98	N/A	N/A	N/A		
		Ashkenazi Jewish	98	N/A	N/A	N/A		
		East Asian	99	1 in 1,150	1 in 110,000	1 in 440,000		
		Finnish	98	N/A	N/A	N/A		
		Non-Finnish European/Caucasian	99	1 in 8,124	1 in 810,000	1 in 3,200,000		

Footnotes:

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

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

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Disorder	Gene	Ethnicity (Source: <a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a> except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		South Asian	99	1 in 7,653	1 in 770,000	1 in 3,100,000
		Other (population not assigned)	98	N/A	N/A	N/A
	HJV	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	1 in 3,456	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 2,225	1 in 220,000	1 in 880,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 858	1 in 86,000	1 in 340,000
		South Asian	97	1 in 766	1 in 26,000	1 in 100,000
		Other (population not assigned)	99	1 in 1,449	1 in 140,000	1 in 560,000

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