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

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3M syndrome	***CCDC8***	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
	CUL7	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
		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
	OBSL1	African/African American	99	1 in 43	1 in 4,200	1 in 17,000
		Latino/Admixed American	99	1 in 66	1 in 6,500	1 in 26,000
		Ashkenazi Jewish	99	1 in 33	1 in 3,200	1 in 13,000
		East Asian	97	1 in 43	1 in 1,400	1 in 5,600
		Finnish	99	1 in 113	1 in 11,000	1 in 44,000
		Non-Finnish European/Caucasian	99	1 in 103	1 in 10,000	1 in 40,000
		South Asian	97	1 in 187	1 in 6,200	1 in 25,000
		Other (population not assigned)	99	1 in 61	1 in 6,000	1 in 24,000
3-Methylcrotonyl-CoA carboxylase deficiency	***MCCC1***	African/African American	99	1 in 370	1 in 37,000	1 in 150,000
		Latino/Admixed American	99	1 in 904	1 in 90,000	1 in 360,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 1,157	1 in 120,000	1 in 480,000
		Finnish	99	1 in 3,682	1 in 370,000	1 in 1,500,000
		Non-Finnish European/Caucasian	98	1 in 528	1 in 26,000	1 in 100,000
		South Asian	99	1 in 417	1 in 42,000	1 in 170,000
		Other (population not assigned)	99	1 in 536	1 in 54,000	1 in 220,000
	MCCC2	African/African American	99	1 in 374	1 in 37,000	1 in 150,000
		Latino/Admixed American	99	1 in 124	1 in 12,000	1 in 48,000
		Ashkenazi Jewish	99	1 in 187	1 in 19,000	1 in 76,000
		East Asian	99	1 in 294	1 in 29,000	1 in 120,000
		Finnish	99	1 in 1,358	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	99	1 in 182	1 in 18,000	1 in 72,000
		South Asian	99	1 in 480	1 in 48,000	1 in 190,000
		Other (population not assigned)	99	1 in 128	1 in 13,000	1 in 52,000
Abetalipoproteinemia	***MTTP***	African/African American	99	1 in 181	1 in 18,000	1 in 72,000
		Latino/Admixed American	99	1 in 1,781	1 in 180,000	1 in 720,000
		Ashkenazi Jewish	99	1 in 177	1 in 18,000	1 in 72,000
		East Asian	99	1 in 1,182	1 in 120,000	1 in 480,000
		Finnish	99	1 in 2,157	1 in 220,000	1 in 880,000

Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

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		Non-Finnish European/Caucasian	98	1 in 1,581	1 in 79,000	1 in 320,000
		South Asian	97	1 in 804	1 in 27,000	1 in 110,000
		Other (population not assigned)	97	1 in 3,069	1 in 100,000	1 in 400,000
Acute infantile liver failure	***LARS***	African/African American	99	1 in 728	1 in 73,000	1 in 290,000
		Latino/Admixed American	99	1 in 474	1 in 47,000	1 in 190,000
		Ashkenazi Jewish	99	1 in 2,516	1 in 250,000	1 in 1,000,000
		East Asian	98	1 in 1,007	1 in 50,000	1 in 200,000
		Finnish	99	1 in 1,820	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 1,000	1 in 100,000	1 in 400,000
		South Asian	99	1 in 948	1 in 95,000	1 in 380,000
		Other (population not assigned)	99	1 in 479	1 in 48,000	1 in 190,000
	NBAS	African/African American	99	1 in 232	1 in 23,000	1 in 92,000
		Latino/Admixed American	98	1 in 219	1 in 11,000	1 in 44,000
		Ashkenazi Jewish	97	1 in 5,032	1 in 170,000	1 in 680,000
		East Asian	99	1 in 482	1 in 48,000	1 in 190,000
		Finnish	97	1 in 347	1 in 12,000	1 in 48,000
		Non-Finnish European/Caucasian	98	1 in 228	1 in 11,000	1 in 44,000
		South Asian	99	1 in 436	1 in 44,000	1 in 180,000
		Other (population not assigned)	98	1 in 147	1 in 7,300	1 in 29,000
	TRMU	African/African American	99	1 in 606	1 in 61,000	1 in 240,000
		Latino/Admixed American	98	1 in 1,045	1 in 52,000	1 in 210,000
		Ashkenazi Jewish	99	1 in 1,232	1 in 120,000	1 in 480,000
		East Asian	99	1 in 762	1 in 76,000	1 in 300,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	1 in 739	1 in 37,000	1 in 150,000
		South Asian	99	1 in 355	1 in 35,000	1 in 140,000
		Other (population not assigned)	97	1 in 2,159	1 in 72,000	1 in 290,000
Adenosine deaminase deficiency	***ADA***	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
Adrenoleukodystrophy, X-linked	***ABCD1***	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
Agammaglobulinemia, X-linked	***BTK***	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A

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		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
Aicardi-Goutières syndrome	***RNASEH2A***	African/African American	99	1 in 75	1 in 7,400	1 in 30,000
		Latino/Admixed American	99	1 in 708	1 in 71,000	1 in 280,000
		Ashkenazi Jewish	99	1 in 708	1 in 71,000	1 in 280,000
		East Asian	99	1 in 115	1 in 11,000	1 in 44,000
		Finnish	99	1 in 222	1 in 22,000	1 in 88,000
		Non-Finnish European/Caucasian	99	1 in 192	1 in 19,000	1 in 76,000
		South Asian	99	1 in 505	1 in 50,000	1 in 200,000
		Other (population not assigned)	99	1 in 297	1 in 30,000	1 in 120,000
	RNASEH2B	African/African American	97	1 in 170	1 in 5,600	1 in 22,000
		Latino/Admixed American	98	1 in 132	1 in 6,600	1 in 26,000
		Ashkenazi Jewish	97	1 in 527	1 in 18,000	1 in 72,000
		East Asian	97	1 in 120	1 in 4,000	1 in 16,000
		Finnish	97	1 in 582	1 in 19,000	1 in 76,000
		Non-Finnish European/Caucasian	98	1 in 80	1 in 4,000	1 in 16,000
		South Asian	98	1 in 105	1 in 5,200	1 in 21,000
		Other (population not assigned)	98	1 in 157	1 in 7,800	1 in 31,000
	RNASEH2C	African/African American	98	1 in 2,703	1 in 140,000	1 in 560,000
		Latino/Admixed American	97	1 in 4,272	1 in 140,000	1 in 560,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 2,928	1 in 98,000	1 in 390,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	1 in 1,525	1 in 76,000	1 in 300,000
		South Asian	99	1 in 374	1 in 37,000	1 in 150,000
		Other (population not assigned)	99	1 in 2,742	1 in 270,000	1 in 1,100,000
	SAMHD1	African/African American	99	1 in 2,032	1 in 200,000	1 in 800,000
		Latino/Admixed American	99	1 in 2,428	1 in 240,000	1 in 960,000
		Ashkenazi Jewish	97	1 in 2,491	1 in 83,000	1 in 330,000
		East Asian	97	N/A	N/A	N/A
		Finnish	97	1 in 5,407	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 1,069	1 in 110,000	1 in 440,000
		South Asian	99	1 in 3,837	1 in 380,000	1 in 1,500,000
		Other (population not assigned)	99	1 in 3,062	1 in 310,000	1 in 1,200,000
Allan-Herndon-Dudley syndrome	***SLC16A2***	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	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

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		Other (population not assigned)	98	N/A	N/A	N/A
Alpha-mannosidosis	***MAN2B1***	African/African American	99	1 in 333	1 in 33,000	1 in 130,000
		Latino/Admixed American	99	1 in 138	1 in 14,000	1 in 56,000
		Ashkenazi Jewish	99	1 in 5,037	1 in 500,000	1 in 2,000,000
		East Asian	98	1 in 900	1 in 45,000	1 in 180,000
		Finnish	99	1 in 206	1 in 21,000	1 in 84,000
		Non-Finnish European/Caucasian	99	1 in 475	1 in 47,000	1 in 190,000
		South Asian	99	1 in 831	1 in 83,000	1 in 330,000
		Other (population not assigned)	99	1 in 371	1 in 37,000	1 in 150,000
Alpha-thalassemia	***HBA1/HBA2***	African	90	1 in 3	N/A	N/A
		American	90	1 in 21	N/A	N/A
		Eastern Mediterranean	90	1 in 5	N/A	N/A
		European	90	1 in 44	N/A	N/A
		Southeast Asian	90	1 in 2	N/A	N/A
		Western Pacific	90	1 in 10	N/A	N/A
Alpha-thalassemia X-linked intellectual disability syndrome	***ATRX***	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
Alport syndrome	***COL4A3***	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	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
Alström syndrome	***ALMS1***	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

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		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
Andermann syndrome	***SLC12A6***	African/African American	97	1 in 324	1 in 11,000	1 in 44,000
		Latino/Admixed American	98	1 in 594	1 in 30,000	1 in 120,000
		Ashkenazi Jewish	99	1 in 1,654	1 in 170,000	1 in 680,000
		East Asian	99	1 in 2,933	1 in 290,000	1 in 1,200,000
		Finnish	98	1 in 2,178	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	98	1 in 1,445	1 in 72,000	1 in 290,000
		South Asian	99	1 in 1,392	1 in 140,000	1 in 560,000
		Other (population not assigned)	99	1 in 1,023	1 in 100,000	1 in 400,000
Arginase deficiency	***ARG1***	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
Argininosuccinic aciduria	***ASL***	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
Aromatic l-amino acid decarboxylase deficiency	***DDC***	African/African American	99	1 in 2,535	1 in 250,000	1 in 1,000,000
		Latino/Admixed American	99	1 in 2,137	1 in 210,000	1 in 840,000
		Ashkenazi Jewish	99	1 in 279	1 in 28,000	1 in 110,000
		East Asian	97	1 in 1,839	1 in 61,000	1 in 240,000
		Finnish	99	1 in 1,361	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	99	1 in 976	1 in 98,000	1 in 390,000
		South Asian	99	1 in 1,180	1 in 120,000	1 in 480,000
		Other (population not assigned)	99	1 in 1,527	1 in 150,000	1 in 600,000
Arterial tortuosity syndrome	***SLC2A10***	African/African American	99	1 in 862	1 in 86,000	1 in 340,000
		Latino/Admixed American	99	1 in 2,320	1 in 230,000	1 in 920,000
		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	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 1,113	1 in 110,000	1 in 440,000
		South Asian	98	1 in 1,705	1 in 85,000	1 in 340,000
		Other (population not assigned)	99	1 in 985	1 in 98,000	1 in 390,000
Arthrogryposis, mental retardation, and seizures (AMRS)	***SLC35A3***	African/African American	98	1 in 2,593	1 in 130,000	1 in 520,000
		Latino/Admixed American	99	1 in 1,744	1 in 170,000	1 in 680,000
		Ashkenazi Jewish	99	1 in 368	1 in 37,000	1 in 150,000

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		East Asian	97	1 in 1,780	1 in 59,000	1 in 240,000
		Finnish	98	1 in 2,123	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	98	1 in 2,619	1 in 130,000	1 in 520,000
		South Asian	98	1 in 1,851	1 in 93,000	1 in 370,000
		Other (population not assigned)	99	1 in 2,250	1 in 220,000	1 in 880,000
Asparagine synthetase deficiency	***ASNS***	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
Aspartylglucosaminuria	***AGA***	African/African American	99	1 in 118	1 in 12,000	1 in 48,000
		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
Ataxia with vitamin E deficiency	***TTPA***	African/African American	99	1 in 2,642	1 in 260,000	1 in 1,000,000
		Latino/Admixed American	98	1 in 1,291	1 in 65,000	1 in 260,000
		Ashkenazi Jewish	97	1 in 580	1 in 19,000	1 in 76,000
		East Asian	99	1 in 4,307	1 in 430,000	1 in 1,700,000
		Finnish	98	1 in 3,712	1 in 190,000	1 in 760,000
		Non-Finnish European/Caucasian	99	1 in 94	1 in 9,300	1 in 37,000
		South Asian	99	1 in 3,065	1 in 310,000	1 in 1,200,000
		Other (population not assigned)	97	1 in 2,736	1 in 91,000	1 in 360,000
Ataxia-telangiectasia	***ATM***	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
		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
ATP7A-related copper transport disorders, includes Menkes syndrome	***ATP7A***	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: 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
Autoimmune polyglandular syndrome type 1	***AIRE***	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
Autosomal recessive congenital ichthyosis (ARCI)	***ABCA12***	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
	ALOX12B	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	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
		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
	CASP14	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
	CERS3	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

Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

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		Non-Finnish European/Caucasian	99	1 in 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
	CYP4F22	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
	LIPN	African/African American	99	1 in 115	1 in 11,000	1 in 44,000
		Latino/Admixed American	99	1 in 602	1 in 60,000	1 in 240,000
		Ashkenazi Jewish	97	1 in 143	1 in 4,700	1 in 19,000
		East Asian	99	1 in 2,172	1 in 220,000	1 in 880,000
		Finnish	99	1 in 459	1 in 46,000	1 in 180,000
		Non-Finnish European/Caucasian	99	1 in 335	1 in 33,000	1 in 130,000
		South Asian	97	1 in 2,776	1 in 93,000	1 in 370,000
		Other (population not assigned)	99	1 in 224	1 in 22,000	1 in 88,000
	NIPAL4	African/African American	99	1 in 1,827	1 in 180,000	1 in 720,000
		Latino/Admixed American	98	1 in 687	1 in 34,000	1 in 140,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	98	1 in 660	1 in 33,000	1 in 130,000
		Finnish	99	1 in 1,759	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 372	1 in 37,000	1 in 150,000
		South Asian	99	1 in 841	1 in 84,000	1 in 340,000
		Other (population not assigned)	99	1 in 657	1 in 66,000	1 in 260,000
	PNPLA1	African/African American	97	1 in 1,352	1 in 45,000	1 in 180,000
		Latino/Admixed American	99	1 in 1,801	1 in 180,000	1 in 720,000
		Ashkenazi Jewish	97	1 in 4,365	1 in 150,000	1 in 600,000
		East Asian	99	1 in 511	1 in 51,000	1 in 200,000
		Finnish	99	1 in 10,806	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	99	1 in 1,456	1 in 150,000	1 in 600,000
		South Asian	97	1 in 3,532	1 in 120,000	1 in 480,000
		Other (population not assigned)	99	1 in 577	1 in 58,000	1 in 230,000
	SDR9C7	African/African American	99	1 in 3,924	1 in 390,000	1 in 1,600,000
		Latino/Admixed American	99	1 in 16,779	1 in 1,700,000	1 in 6,800,000
		Ashkenazi Jewish	99	1 in 4,999	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 3,066	1 in 310,000	1 in 1,200,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 1,942	1 in 190,000	1 in 760,000
		South Asian	97	1 in 1,183	1 in 39,000	1 in 160,000
		Other (population not assigned)	99	1 in 1,514	1 in 150,000	1 in 600,000
	SLC27A4	African/African American	98	1 in 3,928	1 in 200,000	1 in 800,000
		Latino/Admixed American	99	1 in 1,616	1 in 160,000	1 in 640,000

Footnotes:

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

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		Ashkenazi Jewish	99	1 in 3,493	1 in 350,000	1 in 1,400,000
		East Asian	98	1 in 920	1 in 46,000	1 in 180,000
		Finnish	99	1 in 705	1 in 70,000	1 in 280,000
		Non-Finnish European/Caucasian	99	1 in 811	1 in 81,000	1 in 320,000
		South Asian	98	1 in 5,103	1 in 260,000	1 in 1,000,000
		Other (population not assigned)	98	1 in 974	1 in 49,000	1 in 200,000
	TGM1	African/African American	99	1 in 329	1 in 33,000	1 in 130,000
		Latino/Admixed American	99	1 in 701	1 in 70,000	1 in 280,000
		Ashkenazi Jewish	99	1 in 495	1 in 49,000	1 in 200,000
		East Asian	99	1 in 258	1 in 26,000	1 in 100,000
		Finnish	99	1 in 185	1 in 18,000	1 in 72,000
		Non-Finnish European/Caucasian	99	1 in 250	1 in 25,000	1 in 100,000
		South Asian	99	1 in 767	1 in 77,000	1 in 310,000
		Other (population not assigned)	99	1 in 387	1 in 39,000	1 in 160,000
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	***SACS***	African/African American	97	1 in 379	1 in 13,000	1 in 52,000
		Latino/Admixed American	98	1 in 284	1 in 14,000	1 in 56,000
		Ashkenazi Jewish	98	1 in 478	1 in 24,000	1 in 96,000
		East Asian	98	1 in 411	1 in 21,000	1 in 84,000
		Finnish	99	1 in 336	1 in 34,000	1 in 140,000
		Non-Finnish European/Caucasian	99	1 in 108	1 in 11,000	1 in 44,000
		South Asian	98	1 in 287	1 in 14,000	1 in 56,000
		Other (population not assigned)	99	1 in 189	1 in 19,000	1 in 76,000
Axonal neuropathy with neuromyotonia, autosomal recessive	***HINT1***	African/African American	99	1 in 1,824	1 in 180,000	1 in 720,000
		Latino/Admixed American	99	1 in 2,188	1 in 220,000	1 in 880,000
		Ashkenazi Jewish	99	1 in 179	1 in 18,000	1 in 72,000
		East Asian	99	1 in 999	1 in 100,000	1 in 400,000
		Finnish	99	1 in 578	1 in 58,000	1 in 230,000
		Non-Finnish European/Caucasian	99	1 in 766	1 in 77,000	1 in 310,000
		South Asian	99	1 in 3,178	1 in 320,000	1 in 1,300,000
		Other (population not assigned)	99	1 in 2,739	1 in 270,000	1 in 1,100,000
Bardet-Biedl syndrome	***ARL6***	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
	BBS1	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

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		Other (population not assigned)	99	1 in 90	1 in 8,900	1 in 36,000
	BBS2	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	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	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
		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	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	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	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

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		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	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
	MKKS	African/African American	99	1 in 260	1 in 26,000	1 in 100,000
		Latino/Admixed American	99	1 in 214	1 in 21,000	1 in 84,000
		Ashkenazi Jewish	99	1 in 458	1 in 46,000	1 in 180,000
		East Asian	99	1 in 920	1 in 92,000	1 in 370,000
		Finnish	99	1 in 118	1 in 12,000	1 in 48,000
		Non-Finnish European/Caucasian	99	1 in 49	1 in 4,800	1 in 19,000
		South Asian	98	1 in 807	1 in 40,000	1 in 160,000
		Other (population not assigned)	99	1 in 133	1 in 13,000	1 in 52,000
	SDCCAG8	African/African American	97	1 in 656	1 in 22,000	1 in 88,000
		Latino/Admixed American	99	1 in 679	1 in 68,000	1 in 270,000
		Ashkenazi Jewish	97	1 in 2,520	1 in 84,000	1 in 340,000
		East Asian	99	1 in 160	1 in 16,000	1 in 64,000
		Finnish	99	1 in 822	1 in 82,000	1 in 330,000
		Non-Finnish European/Caucasian	98	1 in 678	1 in 34,000	1 in 140,000
		South Asian	99	1 in 265	1 in 26,000	1 in 100,000
		Other (population not assigned)	98	1 in 561	1 in 28,000	1 in 110,000
	TTC8	African/African American	99	1 in 2,003	1 in 200,000	1 in 800,000
		Latino/Admixed American	99	1 in 1,076	1 in 110,000	1 in 440,000
		Ashkenazi Jewish	99	1 in 2,397	1 in 240,000	1 in 960,000
		East Asian	98	1 in 868	1 in 43,000	1 in 170,000
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 962	1 in 96,000	1 in 380,000
		South Asian	99	1 in 1,340	1 in 130,000	1 in 520,000
		Other (population not assigned)	99	1 in 1,997	1 in 200,000	1 in 800,000
Bare lymphocyte syndrome type II	***CIITA***	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
	RFX5	African/African American	99	1 in 948	1 in 95,000	1 in 380,000

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: 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	98	1 in 1,572	1 in 79,000	1 in 320,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	98	1 in 2,295	1 in 110,000	1 in 440,000
		Finnish	99	1 in 429	1 in 43,000	1 in 170,000
		Non-Finnish European/Caucasian	99	1 in 2,774	1 in 280,000	1 in 1,100,000
		South Asian	98	1 in 1,701	1 in 85,000	1 in 340,000
		Other (population not assigned)	98	N/A	N/A	N/A
	RFXANK	African/African American	97	1 in 3,377	1 in 110,000	1 in 440,000
		Latino/Admixed American	97	1 in 1,379	1 in 46,000	1 in 180,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 1,839	1 in 61,000	1 in 240,000
		Finnish	97	1 in 5,855	1 in 200,000	1 in 800,000
		Non-Finnish European/Caucasian	98	1 in 1,650	1 in 82,000	1 in 330,000
		South Asian	99	1 in 5,103	1 in 510,000	1 in 2,000,000
		Other (population not assigned)	98	1 in 1,535	1 in 77,000	1 in 310,000
	RFXAP	African/African American	97	1 in 3,855	1 in 130,000	1 in 520,000
		Latino/Admixed American	97	1 in 864	1 in 29,000	1 in 120,000
		Ashkenazi Jewish	97	1 in 1,358	1 in 45,000	1 in 180,000
		East Asian	97	1 in 1,852	1 in 62,000	1 in 250,000
		Finnish	97	1 in 3,448	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	97	1 in 1,913	1 in 64,000	1 in 260,000
		South Asian	98	1 in 6,006	1 in 300,000	1 in 1,200,000
		Other (population not assigned)	97	N/A	N/A	N/A
Barth syndrome	***TAZ***	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	98	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
Bartter syndrome	***BSND***	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
	KCNJ1	African/African American	99	1 in 2,602	1 in 260,000	1 in 1,000,000
		Latino/Admixed American	99	1 in 405	1 in 40,000	1 in 160,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 1,301	1 in 130,000	1 in 520,000
		Finnish	99	1 in 1,389	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	98	1 in 1,167	1 in 58,000	1 in 230,000

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: 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 3,071	1 in 310,000	1 in 1,200,000
		Other (population not assigned)	99	1 in 239	1 in 24,000	1 in 96,000
	SLC12A1	African/African American	99	1 in 327	1 in 33,000	1 in 130,000
		Latino/Admixed American	99	1 in 358	1 in 36,000	1 in 140,000
		Ashkenazi Jewish	98	1 in 1,548	1 in 77,000	1 in 310,000
		East Asian	99	1 in 426	1 in 43,000	1 in 170,000
		Finnish	97	1 in 706	1 in 24,000	1 in 96,000
		Non-Finnish European/Caucasian	98	1 in 365	1 in 18,000	1 in 72,000
		South Asian	99	1 in 413	1 in 41,000	1 in 160,000
		Other (population not assigned)	98	1 in 456	1 in 23,000	1 in 92,000
Beta-hemoglobinopathies, includes sickle cell disease and beta-thalassemias	***HBB***	African/African American	99	1 in 9	1 in 800	1 in 3,200
		Latino/Admixed American	99	1 in 164	1 in 16,000	1 in 64,000
		Ashkenazi Jewish	99	1 in 4,922	1 in 490,000	1 in 2,000,000
		East Asian	98	1 in 117	1 in 5,800	1 in 23,000
		Finnish	99	1 in 140	1 in 14,000	1 in 56,000
		Non-Finnish European/Caucasian	99	1 in 240	1 in 24,000	1 in 96,000
		South Asian	99	1 in 27	1 in 2,600	1 in 10,000
		Other (population not assigned)	99	1 in 77	1 in 7,600	1 in 30,000
Beta-ketothiolase deficiency	***ACAT1***	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
		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
Beta-mannosidosis	***MANBA***	African/African American	98	1 in 345	1 in 17,000	1 in 68,000
		Latino/Admixed American	99	1 in 1,057	1 in 110,000	1 in 440,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 1,513	1 in 150,000	1 in 600,000
		Finnish	99	1 in 2,705	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 684	1 in 68,000	1 in 270,000
		South Asian	98	1 in 1,028	1 in 51,000	1 in 200,000
		Other (population not assigned)	98	1 in 511	1 in 26,000	1 in 100,000
Biotinidase deficiency	***BTD***	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
Bloom syndrome	***BLM***	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

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: 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 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
Brittle cornea syndrome	***PRDM5***	African/African American	98	1 in 1,625	1 in 81,000	1 in 320,000
		Latino/Admixed American	99	1 in 631	1 in 63,000	1 in 250,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 1,078	1 in 110,000	1 in 440,000
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 1,952	1 in 200,000	1 in 800,000
		South Asian	99	1 in 3,061	1 in 310,000	1 in 1,200,000
		Other (population not assigned)	99	1 in 2,111	1 in 210,000	1 in 840,000
	ZNF469	African/African American	99	1 in 227	1 in 23,000	1 in 92,000
		Latino/Admixed American	99	1 in 257	1 in 26,000	1 in 100,000
		Ashkenazi Jewish	99	1 in 50	1 in 4,900	1 in 20,000
		East Asian	97	1 in 1,334	1 in 44,000	1 in 180,000
		Finnish	99	1 in 41	1 in 4,000	1 in 16,000
		Non-Finnish European/Caucasian	99	1 in 243	1 in 24,000	1 in 96,000
		South Asian	99	1 in 867	1 in 87,000	1 in 350,000
		Other (population not assigned)	99	1 in 136	1 in 14,000	1 in 56,000
Canavan disease	***ASPA***	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
Carbamoyl phosphate synthetase I deficiency	***CPS1***	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
		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
Carnitine palmitoyltransferase I deficiency	***CPT1A***	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

Footnotes:

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

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Disorder	Gene	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
Carnitine palmitoyltransferase II deficiency	***CPT2***	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
Carnitine-acylcarnitine translocase deficiency	***SLC25A20***	African/African American	99	1 in 808	1 in 81,000	1 in 320,000
		Latino/Admixed American	99	1 in 1,902	1 in 190,000	1 in 760,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	1 in 485	1 in 48,000	1 in 190,000
		Finnish	99	1 in 10,815	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	99	1 in 1,988	1 in 200,000	1 in 800,000
		South Asian	99	1 in 2,188	1 in 220,000	1 in 880,000
		Other (population not assigned)	99	1 in 2,736	1 in 270,000	1 in 1,100,000
Carpenter syndrome	***MEGF8***	African/African American	99	1 in 719	1 in 72,000	1 in 290,000
		Latino/Admixed American	98	1 in 544	1 in 27,000	1 in 110,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 804	1 in 80,000	1 in 320,000
		Finnish	97	1 in 538	1 in 18,000	1 in 72,000
		Non-Finnish European/Caucasian	99	1 in 375	1 in 37,000	1 in 150,000
		South Asian	99	1 in 783	1 in 78,000	1 in 310,000
		Other (population not assigned)	99	1 in 617	1 in 62,000	1 in 250,000
	RAB23	African/African American	99	1 in 2,549	1 in 250,000	1 in 1,000,000
		Latino/Admixed American	99	1 in 8,390	1 in 840,000	1 in 3,400,000
		Ashkenazi Jewish	99	1 in 2,484	1 in 250,000	1 in 1,000,000
		East Asian	99	1 in 9,197	1 in 920,000	1 in 3,700,000
		Finnish	99	1 in 5,575	1 in 560,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 744	1 in 74,000	1 in 300,000
		South Asian	99	1 in 7,642	1 in 760,000	1 in 3,000,000
		Other (population not assigned)	99	N/A	N/A	N/A
Cartilage-hair hypoplasia	***RMRP***	African/African American	98	1 in 719	1 in 36,000	1 in 140,000
		Latino/Admixed American	99	1 in 303	1 in 30,000	1 in 120,000
		Ashkenazi Jewish	97	1 in 191	1 in 6,300	1 in 25,000
		East Asian	98	1 in 413	1 in 21,000	1 in 84,000
		Finnish	99	1 in 56	1 in 5,500	1 in 22,000
		Non-Finnish European/Caucasian	98	1 in 229	1 in 11,000	1 in 44,000
		South Asian	99	1 in 294	1 in 29,000	1 in 120,000
		Other (population not assigned)	98	1 in 137	1 in 6,800	1 in 27,000
Cerebellar hypoplasia, VLDLR-associated	***VLDLR***	African/African American	99	1 in 812	1 in 81,000	1 in 320,000
		Latino/Admixed American	99	1 in 5,921	1 in 590,000	1 in 2,400,000
		Ashkenazi Jewish	99	1 in 474	1 in 47,000	1 in 190,000
		East Asian	99	1 in 1,314	1 in 130,000	1 in 520,000
		Finnish	98	1 in 5,404	1 in 270,000	1 in 1,100,000

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		Non-Finnish European/Caucasian	99	1 in 2,026	1 in 200,000	1 in 800,000
		South Asian	99	1 in 7,654	1 in 770,000	1 in 3,100,000
		Other (population not assigned)	98	N/A	N/A	N/A
Cerebral creatine deficiency syndromes	***GAMT***	African/African American	98	1 in 897	1 in 45,000	1 in 180,000
		Latino/Admixed American	98	1 in 3,195	1 in 160,000	1 in 640,000
		Ashkenazi Jewish	97	1 in 1,395	1 in 46,000	1 in 180,000
		East Asian	97	1 in 1,356	1 in 45,000	1 in 180,000
		Finnish	97	1 in 9,400	1 in 310,000	1 in 1,200,000
		Non-Finnish European/Caucasian	99	1 in 417	1 in 42,000	1 in 170,000
		South Asian	99	1 in 2,581	1 in 260,000	1 in 1,000,000
		Other (population not assigned)	98	1 in 547	1 in 27,000	1 in 110,000
	GATM	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 2,225	1 in 220,000	1 in 880,000
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 6,747	1 in 670,000	1 in 2,700,000
		South Asian	99	1 in 15,299	1 in 1,500,000	1 in 6,000,000
		Other (population not assigned)	98	N/A	N/A	N/A
	SLC6A8	African/African American	98	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	98	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)	99	N/A	N/A	N/A
Cerebrotendinous xanthomatosis	***CYP27A1***	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
Chronic granulomatous disease	***CYBA***	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	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A

Footnotes:

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a Feng et al., PMID 28125085

b Luo et al., PMID 23788250

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		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
	NCF2	African/African American	97	1 in 8,129	1 in 270,000	1 in 1,100,000
		Latino/Admixed American	98	1 in 556	1 in 28,000	1 in 110,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 2,262	1 in 110,000	1 in 440,000
		Finnish	99	1 in 5,575	1 in 560,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 1,883	1 in 190,000	1 in 760,000
		South Asian	98	1 in 2,189	1 in 110,000	1 in 440,000
		Other (population not assigned)	97	1 in 3,066	1 in 100,000	1 in 400,000
	NCF4	African/African American	99	1 in 1,335	1 in 130,000	1 in 520,000
		Latino/Admixed American	99	1 in 8,597	1 in 860,000	1 in 3,400,000
		Ashkenazi Jewish	99	1 in 369	1 in 37,000	1 in 150,000
		East Asian	99	1 in 1,367	1 in 140,000	1 in 560,000
		Finnish	99	1 in 5,385	1 in 540,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 540	1 in 54,000	1 in 220,000
		South Asian	99	1 in 450	1 in 45,000	1 in 180,000
		Other (population not assigned)	99	1 in 614	1 in 61,000	1 in 240,000
Ciliopathies	***CEP290***	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
	MKS1	African/African American	99	1 in 515	1 in 51,000	1 in 200,000
		Latino/Admixed American	99	1 in 946	1 in 95,000	1 in 380,000
		Ashkenazi Jewish	99	1 in 1,679	1 in 170,000	1 in 680,000
		East Asian	98	1 in 257	1 in 13,000	1 in 52,000
		Finnish	97	1 in 73	1 in 2,400	1 in 9,600
		Non-Finnish European/Caucasian	98	1 in 228	1 in 11,000	1 in 44,000
		South Asian	99	1 in 180	1 in 18,000	1 in 72,000
		Other (population not assigned)	98	1 in 248	1 in 12,000	1 in 48,000
Citrullinemia	***ASS1***	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

Footnotes:

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		Other (population not assigned)	99	1 in 142	1 in 14,000	1 in 56,000
	SLC25A13	African/African American	99	1 in 974	1 in 97,000	1 in 390,000
		Latino/Admixed American	98	1 in 1,065	1 in 53,000	1 in 210,000
		Ashkenazi Jewish	98	1 in 1,676	1 in 84,000	1 in 340,000
		East Asian	98	1 in 58	1 in 2,900	1 in 12,000
		Finnish	99	1 in 4,040	1 in 400,000	1 in 1,600,000
		Non-Finnish European/Caucasian	99	1 in 725	1 in 72,000	1 in 290,000
		South Asian	99	1 in 503	1 in 50,000	1 in 200,000
		Other (population not assigned)	99	1 in 3,039	1 in 300,000	1 in 1,200,000
Coats plus syndrome and dyskeratosis congenita, CTC1-related	***CTC1***	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
Cockayne syndrome	***ERCC6***	African/African American	98	1 in 419	1 in 21,000	1 in 84,000
		Latino/Admixed American	99	1 in 214	1 in 21,000	1 in 84,000
		Ashkenazi Jewish	99	1 in 250	1 in 25,000	1 in 100,000
		East Asian	98	1 in 241	1 in 12,000	1 in 48,000
		Finnish	99	1 in 360	1 in 36,000	1 in 140,000
		Non-Finnish European/Caucasian	99	1 in 335	1 in 33,000	1 in 130,000
		South Asian	99	1 in 431	1 in 43,000	1 in 170,000
		Other (population not assigned)	99	1 in 431	1 in 43,000	1 in 170,000
	ERCC8	African/African American	99	1 in 218	1 in 22,000	1 in 88,000
		Latino/Admixed American	99	1 in 1,557	1 in 160,000	1 in 640,000
		Ashkenazi Jewish	99	1 in 235	1 in 23,000	1 in 92,000
		East Asian	99	1 in 1,006	1 in 100,000	1 in 400,000
		Finnish	99	1 in 3,607	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 1,080	1 in 110,000	1 in 440,000
		South Asian	98	1 in 1,387	1 in 69,000	1 in 280,000
		Other (population not assigned)	99	1 in 3,038	1 in 300,000	1 in 1,200,000
Coffin-Lowry syndrome	***RPS6KA3***	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
Cohen syndrome	***VPS13B***	African/African American	99	1 in 10	1 in 900	1 in 3,600
		Latino/Admixed American	99	1 in 6	1 in 500	1 in 2,000
		Ashkenazi Jewish	99	1 in 6	1 in 500	1 in 2,000
		East Asian	99	1 in 6	1 in 500	1 in 2,000

Footnotes:

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		Finnish	99	1 in 5	1 in 400	1 in 1,600
		Non-Finnish European/Caucasian	99	1 in 7	1 in 600	1 in 2,400
		South Asian	99	1 in 9	1 in 800	1 in 3,200
		Other (population not assigned)	99	1 in 6	1 in 500	1 in 2,000
Cold-induced sweating syndrome, includes Crisponi syndrome	***CLCF1***	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
	CRLF1	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
Combined malonic and methylmalonic aciduria	***ACSF3***	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
Congenital adrenal hyperplasia (CAH)	***CYP11B1***	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
		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	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
		African American	82	1 in 79	1 in 430	1 in 1700

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	CYP21A2 **(Classic form of CAH)**	Ashkenazi Jew	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
	HSD3B2	African/African American	97	1 in 1,151	1 in 38,000	1 in 150,000
		Latino/Admixed American	99	1 in 2,853	1 in 290,000	1 in 1,200,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 2,258	1 in 230,000	1 in 920,000
		Finnish	99	1 in 5,495	1 in 550,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 1,712	1 in 170,000	1 in 680,000
		South Asian	98	1 in 1,183	1 in 59,000	1 in 240,000
		Other (population not assigned)	97	N/A	N/A	N/A
	POR	African/African American	99	1 in 563	1 in 56,000	1 in 220,000
		Latino/Admixed American	98	1 in 278	1 in 14,000	1 in 56,000
		Ashkenazi Jewish	99	1 in 321	1 in 32,000	1 in 130,000
		East Asian	98	1 in 105	1 in 5,200	1 in 21,000
		Finnish	98	1 in 177	1 in 8,800	1 in 35,000
		Non-Finnish European/Caucasian	99	1 in 268	1 in 27,000	1 in 110,000
		South Asian	98	1 in 417	1 in 21,000	1 in 84,000
		Other (population not assigned)	98	1 in 185	1 in 9,200	1 in 37,000
	STAR	African/African American	99	1 in 1,605	1 in 160,000	1 in 640,000
		Latino/Admixed American	99	1 in 940	1 in 94,000	1 in 380,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	1 in 370	1 in 37,000	1 in 150,000
		Finnish	99	1 in 2,706	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 1,425	1 in 140,000	1 in 560,000
		South Asian	99	1 in 1,710	1 in 170,000	1 in 680,000
		Other (population not assigned)	99	1 in 559	1 in 56,000	1 in 220,000
Congenital adrenal hypoplasia, X-linked	***NR0B1***	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
Congenital amegakaryocytic thrombocytopenia	***MPL***	African/African American	99	1 in 599	1 in 60,000	1 in 240,000
		Latino/Admixed American	99	1 in 677	1 in 68,000	1 in 270,000
		Ashkenazi Jewish	99	1 in 60	1 in 5,900	1 in 24,000
		East Asian	99	1 in 292	1 in 29,000	1 in 120,000
		Finnish	99	1 in 1,383	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	99	1 in 284	1 in 28,000	1 in 110,000
		South Asian	98	1 in 529	1 in 26,000	1 in 100,000

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		Other (population not assigned)	98	1 in 391	1 in 20,000	1 in 80,000
Congenital disorder of deglycosylation	***NGLY1***	African/African American	99	1 in 879	1 in 88,000	1 in 350,000
		Latino/Admixed American	98	1 in 1,654	1 in 83,000	1 in 330,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 365	1 in 36,000	1 in 140,000
		Finnish	97	1 in 4,736	1 in 160,000	1 in 640,000
		Non-Finnish European/Caucasian	99	1 in 610	1 in 61,000	1 in 240,000
		South Asian	99	1 in 785	1 in 78,000	1 in 310,000
		Other (population not assigned)	99	1 in 542	1 in 54,000	1 in 220,000
Congenital disorders of glycosylation type 1	***ALG1***	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	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
	MPI	African/African American	99	1 in 496	1 in 50,000	1 in 200,000
		Latino/Admixed American	99	1 in 202	1 in 20,000	1 in 80,000
		Ashkenazi Jewish	99	1 in 5,039	1 in 500,000	1 in 2,000,000
		East Asian	98	1 in 1,079	1 in 54,000	1 in 220,000
		Finnish	98	1 in 1,096	1 in 55,000	1 in 220,000
		Non-Finnish European/Caucasian	99	1 in 467	1 in 47,000	1 in 190,000
		South Asian	98	1 in 921	1 in 46,000	1 in 180,000
		Other (population not assigned)	99	1 in 280	1 in 28,000	1 in 110,000
	PMM2	African/African American	99	1 in 186	1 in 19,000	1 in 76,000
		Latino/Admixed American	99	1 in 102	1 in 10,000	1 in 40,000
		Ashkenazi Jewish	99	1 in 66	1 in 6,500	1 in 26,000
		East Asian	99	1 in 142	1 in 14,000	1 in 56,000
		Finnish	99	1 in 59	1 in 5,800	1 in 23,000
		Non-Finnish European/Caucasian	99	1 in 59	1 in 5,800	1 in 23,000
		South Asian	99	1 in 246	1 in 25,000	1 in 100,000
		Other (population not assigned)	99	1 in 102	1 in 10,000	1 in 40,000
Congenital generalized lipodystrophy	***AGPAT2***	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

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: 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 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
	CAVIN1	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
Congenital insensitivity to pain with anhidrosis	***NTRK1***	African/African American	98	1 in 297	1 in 15,000	1 in 60,000
		Latino/Admixed American	99	1 in 322	1 in 32,000	1 in 130,000
		Ashkenazi Jewish	98	1 in 1,827	1 in 91,000	1 in 360,000
		East Asian	98	1 in 181	1 in 9,000	1 in 36,000
		Finnish	99	1 in 233	1 in 23,000	1 in 92,000
		Non-Finnish European/Caucasian	99	1 in 177	1 in 18,000	1 in 72,000
		South Asian	99	1 in 554	1 in 55,000	1 in 220,000
		Other (population not assigned)	99	1 in 198	1 in 20,000	1 in 80,000
Congenital myasthenic syndrome	***CHAT***	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
	COLQ	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
		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
	DOK7	African/African American	98	1 in 255	1 in 13,000	1 in 52,000
		Latino/Admixed American	98	1 in 178	1 in 8,900	1 in 36,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 255	1 in 13,000	1 in 52,000
		Finnish	97	1 in 117	1 in 3,900	1 in 16,000
		Non-Finnish European/Caucasian	97	1 in 262	1 in 8,700	1 in 35,000
		South Asian	99	1 in 193	1 in 19,000	1 in 76,000
		Other (population not assigned)	98	1 in 356	1 in 18,000	1 in 72,000
	GFPT1	African/African American	99	1 in 1,012	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 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: 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 293	1 in 29,000	1 in 120,000
		Ashkenazi Jewish	99	1 in 2,514	1 in 250,000	1 in 1,000,000
		East Asian	99	1 in 1,729	1 in 170,000	1 in 680,000
		Finnish	99	1 in 974	1 in 97,000	1 in 390,000
		Non-Finnish European/Caucasian	99	1 in 192	1 in 19,000	1 in 76,000
		South Asian	98	1 in 7,652	1 in 380,000	1 in 1,500,000
		Other (population not assigned)	99	1 in 231	1 in 23,000	1 in 92,000
	RAPSN	African/African American	98	1 in 551	1 in 28,000	1 in 110,000
		Latino/Admixed American	99	1 in 405	1 in 40,000	1 in 160,000
		Ashkenazi Jewish	99	1 in 226	1 in 23,000	1 in 92,000
		East Asian	98	1 in 346	1 in 17,000	1 in 68,000
		Finnish	99	1 in 1,036	1 in 100,000	1 in 400,000
		Non-Finnish European/Caucasian	99	1 in 141	1 in 14,000	1 in 56,000
		South Asian	98	1 in 128	1 in 6,400	1 in 26,000
		Other (population not assigned)	99	1 in 274	1 in 27,000	1 in 110,000
Corneal dystrophy and perceptive deafness	***SLC4A11***	African/African American	99	1 in 419	1 in 42,000	1 in 170,000
		Latino/Admixed American	98	1 in 207	1 in 10,000	1 in 40,000
		Ashkenazi Jewish	97	1 in 247	1 in 8,200	1 in 33,000
		East Asian	97	1 in 175	1 in 5,800	1 in 23,000
		Finnish	98	1 in 2,454	1 in 120,000	1 in 480,000
		Non-Finnish European/Caucasian	98	1 in 333	1 in 17,000	1 in 68,000
		South Asian	97	1 in 287	1 in 9,500	1 in 38,000
		Other (population not assigned)	98	1 in 276	1 in 14,000	1 in 56,000
Costeff optic atrophy syndrome, autosomal recessive	***OPA3***	African/African American	97	1 in 6,886	1 in 230,000	1 in 920,000
		Latino/Admixed American	98	1 in 1,889	1 in 94,000	1 in 380,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 3,006	1 in 300,000	1 in 1,200,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	1 in 2,572	1 in 130,000	1 in 520,000
		South Asian	99	1 in 391	1 in 39,000	1 in 160,000
		Other (population not assigned)	99	1 in 2,706	1 in 270,000	1 in 1,100,000
Cutis laxa	***ATP6V0A2***	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
	ATP6V1E1	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 Disorder)

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N/A= Not Available

Disorder	Gene	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
	EFEMP2	African/African American	99	1 in 261	1 in 26,000	1 in 100,000
		Latino/Admixed American	99	1 in 1,232	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	1 in 1,022	1 in 100,000	1 in 400,000
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 2,457	1 in 250,000	1 in 1,000,000
		South Asian	99	1 in 1,833	1 in 180,000	1 in 720,000
		Other (population not assigned)	99	1 in 1,529	1 in 150,000	1 in 600,000
	LTBP4	African/African American	98	1 in 2,394	1 in 120,000	1 in 480,000
		Latino/Admixed American	98	1 in 1,803	1 in 90,000	1 in 360,000
		Ashkenazi Jewish	99	1 in 4,859	1 in 490,000	1 in 2,000,000
		East Asian	99	1 in 229	1 in 23,000	1 in 92,000
		Finnish	98	1 in 2,490	1 in 120,000	1 in 480,000
		Non-Finnish European/Caucasian	99	1 in 64	1 in 6,300	1 in 25,000
		South Asian	99	1 in 907	1 in 91,000	1 in 360,000
		Other (population not assigned)	97	1 in 1,432	1 in 48,000	1 in 190,000
	PYCR1	African/African American	99	1 in 459	1 in 46,000	1 in 180,000
		Latino/Admixed American	99	1 in 1,266	1 in 130,000	1 in 520,000
		Ashkenazi Jewish	99	1 in 613	1 in 61,000	1 in 240,000
		East Asian	98	1 in 299	1 in 15,000	1 in 60,000
		Finnish	99	1 in 3,202	1 in 320,000	1 in 1,300,000
		Non-Finnish European/Caucasian	99	1 in 401	1 in 40,000	1 in 160,000
		South Asian	99	1 in 191	1 in 19,000	1 in 76,000
		Other (population not assigned)	99	1 in 590	1 in 59,000	1 in 240,000
Cystic fibrosis	***CFTR***	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
Cystinosis	***CTNS***	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

Footnotes:

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b Luo et al., PMID 23788250