

Avero® Exon Standard

24 genes



GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
ABCC8	Familial hyperinsulinism, ABCC8-related	General Population	1 in 112	99%	1 in 11101
		Ashkenazi Jewish	1 in 52	99%	1 in 5101
		Finnish	1 in 29	99%	1 in 2801
ASPA	Canavan disease	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 60	99%	1 in 5900
BCKDHA	Maple syrup urine disease, type 1A	General Population	1 in 321	99%	1 in 32001
		Caucasian	1 in 320	99%	1 in 31901
		Mennonite	1 in 10	99%	1 in 900
		Portuguese Roma	1 in 71	99%	1 in 7000
BCKDHB	Maple syrup urine disease, type 1B	General Population	1 in 364	99%	1 in 36301
		Ashkenazi Jewish	1 in 97	99%	1 in 9600
		Asian	1 in 163	99%	1 in 16201
		Caucasian	1 in 433	99%	1 in 43201
BLM	Bloom syndrome	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 140	99%	1 in 13901
CFTR	Cystic fibrosis	General Population	1 in 25	99%	1 in 2401
		African American	1 in 61	99%	1 in 6000
		Ashkenazi Jewish	1 in 25	99%	1 in 2401
		Caucasian	1 in 25	99%	1 in 2401
		East Asian	1 in 90	99%	1 in 8900
		Hispanic	1 in 46	99%	1 in 4501

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CLRN1	Usher syndrome, type III	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 120	99%	1 in 11901
		Finnish	1 in 70	99%	1 in 6900
DLD	Dihydrolipoamide dehydrogenase deficiency	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 94	99%	1 in 9300
ELP1	Familial dysautonomia	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 34	99%	1 in 3301
FANCC	Fanconi anemia, type C	General Population	1 in 1053	99%	1 in 105201
		Ashkenazi Jewish	1 in 98	99%	1 in 9700
FKTN	Walker-Warburg syndrome, FKTN-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 150	99%	1 in 14901
		Japanese	1 in 188	99%	1 in 18701
FMR1	Fragile X syndrome	General Population	1 in 250	99%	1 in 24901
		African American	1 in 251	99%	1 in 25001
		Ashkenazi Jewish	1 in 58	99%	1 in 5700
		Asian	<1 in 500	99%	1 in 49901
		Caucasian	1 in 178	99%	1 in 17701
		Hispanic	<1 in 500	99%	1 in 49901
G6PC	Glycogen storage disease, type IA	General Population	1 in 177	99%	1 in 17601
		Ashkenazi Jewish	1 in 71	99%	1 in 7000
		Asian	1 in 192	99%	1 in 19101
		Caucasian	1 in 177	99%	1 in 17601

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GBA	Gaucher disease	General Population	1 in 153	99%	1 in 15201
		Ashkenazi Jewish	1 in 18	99%	1 in 1701
		Caucasian	1 in 164	99%	1 in 16301
HBA1/HBA2	Alpha-thalassemia	General Population	1 in 25	99%	1 in 2401
		African American	1 in 30	99%	1 in 2901
		Asian	1 in 20	99%	1 in 1901
		Caucasian	<1 in 500	99%	1 in 49901
HBB	Beta-hemoglobinopathies	General Population	1 in 129	99%	1 in 12801
		African American	1 in 10	99%	1 in 900
		Asian	1 in 54	99%	1 in 5300
		Caucasian	1 in 373	99%	1 in 37201
		East Asian	1 in 78	99%	1 in 7700
		Hispanic	1 in 83	99%	1 in 8200
		Mediterranean	1 in 28	99%	1 in 2701
		Middle Eastern	1 in 5	99%	1 in 401
		South Asian	1 in 32	99%	1 in 3101
Southeast Asian	1 in 30	99%	1 in 2901		
HEXA	Tay-Sachs disease	General Population	1 in 250	99%	1 in 24901
		African American	1 in 271	99%	1 in 27001
		Ashkenazi Jewish	1 in 27	99%	1 in 2601
		Asian	1 in 126	99%	1 in 12501

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HEXA	Tay-Sachs disease	Caucasian	1 in 182	99%	1 in 18101
		French Canadian - Gaspesie	1 in 13	99%	1 in 1201
		French Canadian - Other	1 in 73	99%	1 in 7200
		Irish	1 in 41	99%	1 in 4001
		Old Order Amish	1 in 3.4	99%	1 in 241
		Sephardic Jewish - Moroccan, Iraqi	1 in 125	99%	1 in 12401
MCOLN1	Mucopolipidosis, type IV	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 89	99%	1 in 8800
MPL	Congenital amegakaryocytic thrombocytopenia	General Population	1 in 415	99%	1 in 41401
		Ashkenazi Jewish	1 in 57	99%	1 in 5600
		Caucasian	1 in 266	99%	1 in 26501
NEB	Nemaline myopathy 2	General Population	1 in 224	99%	1 in 22301
		Ashkenazi Jewish	1 in 168	99%	1 in 16701
		Finnish	1 in 112	99%	1 in 11101
PCDH15	Usher syndrome, type 1F	General Population	1 in 395	99%	1 in 39401
		Ashkenazi Jewish	1 in 78	99%	1 in 7700
SMN1	Spinal muscular atrophy	General Population	1 in 54	91%	1 in 589
		African American	1 in 72	71%	1 in 245
		Ashkenazi Jewish	1 in 67	99%	1 in 6600
		Asian	1 in 59	99%	1 in 5801
		Caucasian	1 in 47	95%	1 in 920
		Hispanic	1 in 117	90%	1 in 1161

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SMPD1	Niemann-Pick disease, type A/B	General Population	1 in 196	99%	1 in 19501
		Ashkenazi Jewish	1 in 115	99%	1 in 11401
		Caucasian	1 in 244	99%	1 in 24301
TMEM216	Joubert syndrome 2	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 110	99%	1 in 10901