Avero® Exon Ashkenazi Jewish

35 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
ADAMTS2	Ehlers-Danlos syndrome, type VIIC	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 248	99%	1 in 24701
ASPA	Canavan disease	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 60	99%	1 in 5900
ATP7B	Wilson disease	General Population	1 in 90	99%	1 in 8900
		Ashkenazi Jewish	1 in 70	99%	1 in 6900
		Canary Islands	1 in 25	99%	1 in 2401
		Caucasian	1 in 90	99%	1 in 8900
		East Asian	1 in 27	99%	1 in 2601
		Sardinian	1 in 42	99%	1 in 4101
		Sephardic Jewish - North African, Iraqi, Yemenite, Iranian, Bukharian	1 in 65	99%	1 in 6400
BCKDHB	Maple syrup urine disease, type IB	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

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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
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
CPT2	Carnitine palmitoyl transferase II deficiency	General Population	<1 in 500	99%	1 in 49901
		African American	1 in 308	99%	1 in 30701
		Ashkenazi Jewish	1 in 51	99%	1 in 5001
		Asian	<1 in 500	99%	1 in 49901
		Caucasian	1 in 200	99%	1 in 19901
DHCR7	Smith-Lemli-Opitz syndrome	General Population	1 in 100	99%	1 in 9900
		African American	1 in 93	99%	1 in 9200
		Ashkenazi Jewish	1 in 36	99%	1 in 3501
		Asian	<1 in 500	99%	1 in 49901
		Caucasian	1 in 50	99%	1 in 4901
DHDDS	Retinitis pigmentosa 59	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 333	99%	1 in 11601

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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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GALC	Krabbe disease	General Population	1 in 150	99%	1 in 14901
		Asian	<1 in 500	99%	1 in 49901
		Druze Northern Israel	1 in 6	99%	1 in 500
		Muslim Arab (Jerusalem)	1 in 6	99%	1 in 500
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
GBE1	Glycogen storage disease, type IV	General Population	1 in 387	99%	1 in 38601
		Ashkenazi Jewish	1 in 68	99%	1 in 6701
		Caucasian	1 in 144	99%	1 in 14301
GJB2	Nonsyndromic hearing loss, GJB2-related	General Population	1 in 42	99%	1 in 4101
		Ashkenazi Jewish	1 in 21	99%	1 in 2001
		Caucasian	1 in 30	99%	1 in 2901
		East Asian	1 in 40	99%	1 in 3901
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
		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

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HEXA	Tay-Sachs disease	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
HPS3	Hermansky-Pudlak syndrome, HPS3-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 235	99%	1 in 23401
MCOLN1	Mucolipidosis, type IV	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 89	99%	1 in 8800
MEFV	Familial Mediterranean fever	General Population	1 in 115	99%	1 in 11401
		Armenian	1 in 5	99%	1 in 401
		Ashkenazi Jewish	1 in 13	99%	1 in 1201
		Sephardic Jewish	1 in 14	99%	1 in 1301
		Turkish	1 in 5	99%	1 in 401
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
MTTP	Abetalipoproteinemia	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 186	99%	1 in 18501
		Caucasian	<1 in 500	99%	1 in 49901
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

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PCDH15	Usher syndrome, type IF	General Population	1 in 395	99%	1 in 39401
		Ashkenazi Jewish	1 in 78	99%	1 in 7700
PFKM	Glycogen storage disease, type VII	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 250	99%	1 in 24901
PHGDH	3-Phosphoglycerate dehydrogenase deficiency	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 453	99%	1 in 45201
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
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
VRK1	Pontocerebellar hypoplasia, VRK1-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 225	99%	1 in 22401

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