

Avero® Exon Select

13 genes



GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	General Population	1 in 35	99%	1 in 3401
		Asian	1 in 178	99%	1 in 17701
		Caucasian	1 in 64	99%	1 in 6300
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
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
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

GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
<i>G6PC</i>	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
<i>GALT</i>	Galactosemia	General Population	1 in 110	99%	1 in 10901
		African American	1 in 78	99%	1 in 7700
		Ashkenazi Jewish	1 in 172	99%	1 in 17101
		Caucasian	1 in 108	99%	1 in 10701
		East Asian	<1 in 500	99%	1 in 49901
		Hispanic	1 in 305	99%	1 in 30401
		Irish Travellers	1 in 11	99%	1 in 1001
<i>GBA</i>	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
<i>HBA1/HBA2</i>	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
<i>HBB</i>	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

Avero® Exon Select

13 genes



GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
<i>HBB</i>	Beta-hemoglobinopathies	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
<i>HEXA</i>	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
		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
<i>PAH</i>	Phenylalanine hydroxylase deficiency	General Population	1 in 65	99%	1 in 6400
		African American	1 in 143	99%	1 in 14201
		Ashkenazi Jewish	1 in 225	99%	1 in 22401
		Asian	1 in 78	99%	1 in 7700

Avero® Exon Select

13 genes



GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
PAH	Phenylalanine hydroxylase deficiency	Caucasian	1 in 50	99%	1 in 4901
		Irish	1 in 34	99%	1 in 3301
		Sephardic Jewish - Iranian, Bukharian, Kavkazi, Tunisian, Moroccan	1 in 18	99%	1 in 1701
		Sicilian	1 in 26	99%	1 in 2501
		Turkish	1 in 32	99%	1 in 3101
PMM2	Congenital disorder of glycosylation, type IA	General Population	1 in 124	99%	1 in 12301
		Ashkenazi Jewish	1 in 61	99%	1 in 6000
		Asian	1 in 449	99%	1 in 44801
		Caucasian	1 in 42	99%	1 in 4101
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