

CORE PANEL

Condition	Gene	Population	Carrier frequency before screening (1 in)	Detection rate	Carrier residual risk after negative result (1 in)
Alpha Thalassemia	<i>HBA1, HBA2</i>	African American	30	90%	276
		Asian	20	90%	184
		Caucasian	*500	90%	Reduced
		General population	25	90%	230
Beta Thalassemia	<i>HBB</i>	African-American	8	99%	793
		Chinese	29	99%	2872
		Mediterranean	28	99%	2773
		Middle-Eastern	30	99%	2971
		Thai	20	99%	1981
Bloom Syndrome	<i>BLM</i>	Ashkenazi Jewish	100	99%	9902
Canavan Disease	<i>ASPA</i>	Ashkenazi Jewish	57	99%	5645
		European	3392	99%	Reduced
Cystic Fibrosis	<i>CFTR</i>	General Population	45	97%	1506
Dystrophinopathies (Duchenne/Becker Muscular Dystrophy)	<i>DMD</i>	General Population	*500	90%	Reduced
Familial Dysautonomia	<i>ELP1</i>	Ashkenazi Jewish	31	99%	3070
Fanconi Anemia, Type C	<i>FANCC</i>	Ashkenazi Jewish	89	99%	8813
		General Population	417	99%	41292
Fragile X Syndrome, X-linked	<i>FMR1</i>	African American	267	99%	26439
		Ashkenazi Jewish	102	99%	10101
		Asian	419	99%	41490
		Caucasian	170	99%	16834
		General Population	201	99%	19903
		Hispanic	253	99%	25052
Galactosemia	<i>GALT</i>	African American	87	99%	8615
		Ashkenazi Jewish	156	99%	15448
		General Population	100	99%	9902
		Irish Travellers	11	99%	1090
Gaucher Disease	<i>GBA</i>	Ashkenazi Jewish	15	99%	1486
		General Population	158	99%	15646
Medium Chain Acyl-CoA Dehydrogenase Deficiency	<i>ACADM</i>	General Population	66	99%	6536
Mucopolipidosis, Type IV	<i>MCOLN1</i>	Ashkenazi Jewish	100	97%	3345
		General Population	*500	97%	Reduced
Niemann-Pick Disease, Types A/B	<i>SMPD1</i>	Ashkenazi Jewish	115	99%	11388
		General Population	250	99%	24755
Non-Syndromic Hearing Loss (GJB2-related)	<i>GJB2</i>	Ashkenazi Jewish	13	99%	1288
		General Population	50	99%	4951
Non-Syndromic Hearing Loss (GJB6-related)	<i>GJB6</i>	General Population	423	99%	41886
Phenylalanine Hydroxylase Deficiency	<i>PAH</i>	Ashkenazi Jewish	250	99%	24755
		Caucasian	50	99%	4951
		East Asian	50	99%	4951
		European	50	99%	4951
		Finland	200	99%	19804
		General Population	140	99%	13863
		Ireland	33	99%	3268
		Japanese	200	99%	19804
		Turkish	26	99%	2575
Sickle-Cell Disease	<i>HBB</i>	General Population	NA	99%	Reduced

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Smith-Lemli-Opitz Syndrome	<i>DHCR7</i>	African American	183	99%	18121
		Ashkenazi Jewish	43	99%	4258
		General Population	71	99%	7031
		Hispanic	167	99%	16537
		Northern European	54	99%	5348
		Southern European	66	99%	6536
Spinal Muscular Atrophy	<i>SMN1, SMN2</i>	Caucasian	35	95%	705
		Ashkenazi Jewish	41	90%	414
		Hispanic	117	90%	1180
		Asian	53	90%	535
		African American	66	71%	230
Tay-Sachs Disease	<i>HEXA</i>	Ashkenazi Jewish	25	99%	2476
		General population	250	99%	24755

* <1 in 500