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HERES-SEQ-DON

SUPPLEMENTAL TABLE

Gene	Mode	Condition	Ethnicity	Carrier frequency	Detection rate	Post test carrier probability*	Post-test probability of having an affected child
ABCD1	XL	Adrenoleukodystrophy, X-linked	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
AR	XL	Androgen Insensitivity Syndrome: Complete	General Population	1 in 14,286	98%	1 in 714,251	1 in 1,428,571
ATP7A	XL	Menkes disease	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
CFTR	AR	Cystic fibrosis	General Population	1 in 32	99%	1 in 3101	1 in 12,404
			African/African American Population	1 in 61	99%	1 in 6001	1 in 24,004
			Ashkenazi Jewish Population	1 in 24	99%	1 in 2301	1 in 9,204
			Caucasian / European Population	1 in 25	99%	1 in 2401	1 in 9,604
			East Asian Population	1 in 94	99%	1 in 9301	1 in 37,204
			Latino Population	1 in 58	99%	1 in 5701	1 in 22,804
CHM	XL	Choroideremia	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
COL4A5	XL	Alport syndrome, COL4A5-related	General Population	1 in 139	98%	1 in 6,901	1 in 27,604
CYBB	XL	Chronic granulomatous disease, X-linked	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
DMD	XL	Duchenne muscular dystrophy	General Population	<1 in 500	93%	<1 in 7,130	<1 in 28,518
EDA	XL	Hypohidrotic ectodermal dysplasia	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
EMD	XL	Emery-Dreifuss muscular dystrophy	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
F8	XL	Hemophilia A	General Population	<1 in 500	48%	<1 in 49,901	<1 in 199,604
F9	XL	Hemophilia B	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
FMR1	XL	Fragile X syndrome	General Population	1 in 151	99%	1 in 15,001	1 in 60,004
			Ashkenazi Jewish Population	1 in 115	99%	1 in 11,401	1 in 45,604
G6PD	XL	Glucose-6-Phosphate Dehydrogenase Deficiency	General Population	1 in 7	98%	1 in 301	1 in 1,204
GJB1	XL	Charcot-Marie-Tooth disease, X-linked type 1	General Population	1 in 667	90%	1 in 6,661	1 in 26,644
GLA	XL	Fabry disease	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
HBA1, HBA2	AR	Alpha thalassemia	General Population	1 in 20	90%	1 in 191	1 in 764
			African/African American Population	1 in 3	90%	1 in 21	1 in 84
			Ashkenazi Jewish Population	1 in 13	90%	1 in 121	1 in 484
			East Asian Population	1 in 8	90%	1 in 71	1 in 284
			Middle-Eastern Population	1 in 3	90%	1 in 21	1 in 84
			South Asian/Indian Population	1 in 5	90%	1 in 41	1 in 164
HBB	AR	Sickle cell disease; Beta thalassemia	General Population	1 in 158	95%	1 in 3,141	1 in 12,564
			African/African American Pop.	1 in 10	95%	1 in 181	1 in 724
			East Asian Population	1 in 50	95%	1 in 981	1 in 3,924
			Latino Population	1 in 128	95%	1 in 2,541	1 in 10,164
			Mediterranean Population	1 in 3	95%	1 in 41	1 in 164
			South Asian/Indian Population	1 in 25	95%	1 in 481	1 in 1,924
IDS	XL	Mucopolysaccharidosis type II (Hunter syndrome)	General Population	<1 in 500	91%	<1 in 5,545	<1 in 22,182
IL2RG	XL	Severe combined immunodeficiency, X-linked	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
MTM1	XL	Myotubular myopathy, X-linked	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
OCRL	XL	Dent disease 2; Lowe syndrome	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
OTC	XL	Ornithine transcarbamylase deficiency	General Population	<1 in 500	90%	<1 in 4,991	<1 in 19,964
PDHA1	XL	Pyruvate dehydrogenase E1-alpha deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
PRPS1	XL	Arts syndrome; Rosenberg-Chutorian syndrome; Phosphoribosylpyrophosphate synthetase superactivity; Non-syndromic hearing loss, PRPS1- related	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
RS1	XL	Juvenile retinoschisis, X-linked	General Population	<1 in 500	96%	<1 in 12,476	<1 in 49,904
SMN1	AR	Spinal Muscular Atrophy	General Population	1 in 54	91%	1 in 590	1 in 2,360
			African/African American Population	1 in 72	71%	1 in 246	1 in 983
			Ashkenazi Jewish Population	1 in 67	91%	1 in 734	1 in 2,937
			Caucasian / European Population	1 in 47	95%	1 in 921	1 in 3,684
			East Asian Population	1 in 59	93%	1 in 830	1 in 3,318
			Latino Population	1 in 68	90%	1 in 671	1 in 2,684
WAS	XL	Wiskott-Aldrich syndrome; Thrombocytopenia, X- linked; Severe Congenital Neutropenia, WAS-related	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604

*For genes that have tested negative
Abbreviations: AR, autosomal recessive; XL, X-linked