UNITY Fetal Risk™ Screen: **Carrier Screen Frequencies**

Disease	Gene	NM#	Ethnicity	Carrier Frequency *Before* Testing	Detection Rate	Carrier Risk *After* Testing
Alpha-Thalassemia[1] "Silent" group indicates a single alpha-globin gene deletion (aa/a-). "Other" group includes other *HBA1*/*HBA2* variants such as double deletions (aa/-- or a-/a-) and nondeletion variants, including Hb Constant Spring.	*HBA1* *HBA2*	NM_000558.5 NM_000517.6	African American	Silent: 1 in 3 Other: 1 in 28	>95%	Silent: 1 in 60 Other: 1 in 560
			Asian*	Silent: 1 in 13 Other: 1 in 41	>95%	Silent: 1 in 260 Other: 1 in 820
			Northern European	Silent: 1 in 51 Other: <1 in 1,000	>95%	Silent: 1 in 1,020 Other: 1 in 20,000
			Hispanic	Silent: 1 in 17 Other: 1 in 398	>95%	Silent: 1 in 340 Other: 1 in 7,960
			General Population	Silent: 1 in 16 Other: 1 in 112	>95%	Silent: 1 in 320 Other: 1 in 2,240
Cystic Fibrosis[2]	*CFTR*	NM_000492.4	African American	1 in 84	>99%	<1 in 8,400
			Ashkenazi Jewish	1 in 29	>99%	<1 in 2,900
			Asian*	1 in 118	>99%	<1 in 11,800
			Northern European	1 in 25	>99%	<1 in 2,500
			Hispanic	1 in 59	>99%	<1 in 5,900
			General Population	1 in 38	>99%	<1 in 3,800
Sickle Cell Disease, Beta-Thalassemia, Hemoglobinopathies[1]	*HBB*	NM_000518.5	African American	1 in 8	>99%	<1 in 800
			Ashkenazi Jewish	1 in 32	>99%	<1 in 3,200
			Asian*	1 in 22	>99%	<1 in 2,200
			Northern European	1 in 164	>99%	<1 in 16,400
			Hispanic	1 in 55	>99%	<1 in 5,000
			General Population	1 in 33	>99%	<1 in 3,300
Spinal Muscular Atrophy[3]	*SMN1*	NM_000344.4	African American	1 in 72	>90.3%	<1 in 375 (2 copies, SNP absent) <1 in 4,200 (3+ copies)
			Ashkenazi Jewish	1 in 67	>92.8%	<1 in 900 (2 copies, SNP absent) <1 in 5,400 (3+ copies)
			Asian*	1 in 59	>93.6%	<1 in 900 (2 copies, SNP absent) <1 in 5,600 (3+ copies)
			Northern European	1 in 47	>95%	<1 in 900 (2 copies, SNP absent) <1 in 5,600 (3+ copies)
			Hispanic	1 in 68	>92.6%	<1 in 900 (2 copies, SNP absent) <1 in 5,400 (3+ copies)
			General Population	1 in 54	>91.2%	<1 in 875 (2 copies, SNP absent) <1 in 5,400 (3+ copies)
Fragile X Syndrome[4*]	*FMR1*	NM_002024.6	African American	1 in 267	>99%	<1 in 26,700
			Ashkenazi Jewish	1 in 102	>99%	<1 in 10,000
			Asian*	1 in 419	>99%	<1 in 41,900
			Northern European	1 in 170	>99%	<1 in 17,000
			Hispanic	1 in 253	>99%	<1 in 25,300
			General Population	1 in 201	>99%	<1 in 20,100

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Disease	Gene	NM#	Ethnicity	Carrier Frequency *Before* Testing	Detection Rate	Carrier Risk *After* Testing
Canavan Disease[1]	*ASPA*	NM_000049.4	African American	1 in 2,126	>99%	<1 in 50,000
			Ashkenazi Jewish	1 in 44	>99%	1 in 4,400
			Southeast Asian	1 in 1,470	>99%	<1 in 50,000
			Northern European	1 in 428	>99%	1 in 42,800
			Hispanic	1 in 1,153	>99%	<1 in 50,000
			General Population	1 in 439	>99%	1 in 43,900
DMD-Associated Dystropinopathies[1*]	*DMD*	NM_004006.3	General Population	1 in 717	>99%	<1 in 50,000
Familial Dysautonomia[1]	*ELP1*	NM_003640.5	African American	1 in 1,063	>99%	<1 in 50,000
			Ashkenazi Jewish	1 in 35	>99%	1 in 3,500
			East Asian	1 in 1,285	>99%	<1 in 50,000
			Southeast Asian	1 in 4,411	>99%	<1 in 50,000
			Northern European	1 in 567	>99%	<1 in 50,000
			Hispanic	1 in 824	>99%	<1 in 50,000
			General Population	1 in 402	>99%	1 in 40,200
Medium-Chain Acyl-CoA Dehydrogenase Deficiency[5,6,7]	*ACADM*	NM_000016.6	African American	1 in 217	>99%	1 in 21,700
			Ashkenazi Jewish	1 in 134	>99%	1 in 13,400
			East Asian	1 in 335	>99%	1 in 33,500
			South Asian	1 in 272	>99%	1 in 27,200
			Northern European	1 in 62	>99%	1 in 6,200
			Hispanic	1 in 92	>99%	1 in 9,200
			General Population	1 in 67	>99%	1 in 6,700
Phenylalanine Hydoxylase Deficiency[5,8,9,10]	*PAH*	NM_000277.3	African American	1 in 145	>99%	1 in 14,500
			Ashkenazi Jewish	1 in 17	>99%	1 in 1,700
			East Asian	1 in 50	>96%	1 in 1,563
			Northern European	1 in 50	>99%	1 in 5,000
			Hispanic	1 in 69	>99%	1 in 16,200
			Turkish	1 in 26	>99%	1 in 2,600
			General Population	1 in 79	>99%	1 in 7,900
PMM2-Congenital Disorder of Gylcosylation[5,11,12,13]	*PMM2*	NM_000303.3	African American	1 in 210	>99%	1 in 21,000
			Ashkenazi Jewish	1 in 61	>99%	1 in 6,100
			East Asian	1 in 235	>99%	1 in 23,500
			Southeast Asian	1 in 309	>99%	1 in 30,900
			Northern European	1 in 72	>99%	1 in 7,200
			Hispanic	1 in 101	>99%	1 in 13,500
			General Population	1 in 70	>99%	1 in 7,000

UNITY Fetal Risk™ Screen: **Carrier Screen Frequencies**

Disease	Gene	NM#	Ethnicity	Carrier Frequency *Before* Testing	Detection Rate	Carrier Risk *After* Testing
Smith-Lemli-Opitz Syndrome[14]	*DHCR7*	NM_001360.3	African American	1 in 183	>99%	1 in 18,300
			Ashkenazi Jewish	1 in 43	>99%	1 in 4,300
			Southeast Asian	1 in 417	>99%	<1 in 50,000
			Northern European	1 in 51	>99%	1 in 5,100
			Hispanic	1 in 167	>99%	1 in 16,700
			General Population	1 in 71	>99%	1 in 7,100
Tay Sachs Disease[1,15,16]	*HEXA*	NM_000520.6	African American	1 in 257	>99%	1 in 25,700
			Ashkenazi Jewish	1 in 28	>99%	1 in 2,800
			East Asian	1 in 857	>99%	<1 in 50,000
			Southeast Asian	1 in 630	>99%	1 in 63,000
			Northern European	1 in 191	>99%	1 in 19,100
			Hispanic	1 in 316	>99%	1 in 31,600
			French Canadian	1 in 53	>99%	1 in 5,300
			Irish	1 in 50	>99%	1 in 5,000
			General Population	1 in 193	>99%	1 in 19,300

*For X-linked conditions, female carrier frequencies are presented.
The reported detection rates are derived from literature-based estimates of the fraction of disease alleles, adjusted by frequency, that are not detectable by this methodology.

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