# Expanded Carrier Screen: Table of Residual Risks by Self-Reported Ethnicity

Please note: This table displays residual risks after a negative result for each of the genes and corresponding disorders. Please use the highest residual risk for patients with mixed ethnicity. If a patient is reported to be a carrier of a disease, their residual risk is 1 and this table does not apply for that disease.

| Disease (Inheritance) | *Gene* | Ethnicity | Carrier Frequency | Detection Rate | Residual Risk | Analytical Detection Rate |
|---|---|---|---|---|---|---|
| 3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency (AR) NM_000198.3 | *HSD3B2* | Worldwide | 1 in 1005 | 79% | 1 in 4,900 | 99% |
| | | African | 1 in 786 | 89% | 1 in 7,000 | |
| | | Ashkenazi Jewish | 1 in 1639 | 99% | 1 in 164,000 | |
| | | East Asian | 1 in 1814 | 99% | 1 in 181,000 | |
| | | European (Non-Finnish) | 1 in 862 | 74% | 1 in 3,300 | |
| | | Native American | 1 in 1686 | 69% | 1 in 5,500 | |
| | | South Asian | 1 in 1026 | 86% | 1 in 7,200 | |
| 3-Methylcrotonyl-CoA Carboxylase Deficiency (MCCC1-Related) (AR) NM_020166.4 | *MCCC1* | Worldwide | 1 in 423 | 83% | 1 in 2,500 | 99% |
| | | African | 1 in 266 | 51% | 1 in 540 | |
| | | East Asian | 1 in 204 | 78% | 1 in 930 | |
| | | European (Non-Finnish) | 1 in 353 | 90% | 1 in 3,400 | |
| | | Native American | 1 in 488 | 91% | 1 in 5,100 | |
| | | South Asian | 1 in 1000 | 99% | 1 in 100,000 | |
| 3-Methylcrotonyl-CoA Carboxylase Deficiency (MCCC2-Related) (AR) NM_022132.4 | *MCCC2* | Worldwide | 1 in 213 | 83% | 1 in 1,300 | 99% |
| | | African | 1 in 407 | 81% | 1 in 2,200 | |
| | | Ashkenazi Jewish | 1 in 267 | 99% | 1 in 27,000 | |
| | | East Asian | 1 in 192 | 62% | 1 in 500 | |
| | | Finnish | 1 in 2230 | 79% | 1 in 11,000 | |
| | | European (Non-Finnish) | 1 in 204 | 83% | 1 in 1,200 | |
| | | Native American | 1 in 125 | 98% | 1 in 5,100 | |
| | | South Asian | 1 in 308 | 69% | 1 in 1,000 | |
| 3-Methylglutaconic Aciduria, Type III (AR) NM_025136.3 | *OPA3* | Worldwide | 1 in 4526 | 84% | 1 in 29,000 | 99% |
| | | European (Non-Finnish) | 1 in 4808 | 90% | 1 in 50,000 | |
| | | Native American | 1 in 3349 | 59% | 1 in 8,300 | |
| 3-Phosphoglycerate Dehydrogenase Deficiency (AR) NM_006623.3 | *PHGDH* | Worldwide | 1 in 801 | 94% | 1 in 14,000 | 99% |
| | | African | 1 in 1639 | 64% | 1 in 4,600 | |
| | | Ashkenazi Jewish | 1 in 298 | 99% | 1 in 30,000 | |
| | | East Asian | 1 in 1232 | 99% | 1 in 123,000 | |
| | | Finnish | 1 in 1408 | 99% | 1 in 141,000 | |
| | | European (Non-Finnish) | 1 in 631 | 99% | 1 in 63,000 | |
| | | Native American | 1 in 1311 | 69% | 1 in 4,200 | |
| | | South Asian | 1 in 1665 | 78% | 1 in 7,400 | |
| 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (AR) NM_000317.2 | *PTS* | Worldwide | 1 in 395 | 81% | 1 in 2,100 | 99% |
| | | African | 1 in 703 | 99% | 1 in 70,000 | |
| | | Ashkenazi Jewish | 1 in 1559 | 99% | 1 in 156,000 | |
| | | East Asian | 1 in 156 | 95% | 1 in 2,800 | |
| | | Finnish | 1 in 363 | 90% | 1 in 3,500 | |
| | | European (Non-Finnish) | 1 in 478 | 74% | 1 in 1,800 | |
| | | Native American | 1 in 533 | 80% | 1 in 2,700 | |
| | | South Asian | 1 in 343 | 84% | 1 in 2,100 | |
| Abetalipoproteinemia (AR) NM_000253.3 | *MTTP* | Worldwide | 1 in 870 | 85% | 1 in 5,900 | 97% |
| | | African | 1 in 1354 | 97% | 1 in 45,000 | |
| | | Ashkenazi Jewish | 1 in 176 | 97% | 1 in 5,800 | |
| | | East Asian | 1 in 1437 | 81% | 1 in 7,500 | |
| | | European (Non-Finnish) | 1 in 655 | 79% | 1 in 3,200 | |
| | | Native American | 1 in 2131 | 97% | 1 in 71,000 | |
| | | South Asian | 1 in 3078 | 97% | 1 in 103,000 | |
| Achromatopsia / Progressive Cone Dystrophy (AR) NM_019098.4 | *CNGB3* | Worldwide | 1 in 61 | 99% | 1 in 4,300 | 99% |
| | | African | 1 in 50 | 98% | 1 in 2,300 | |
| | | Ashkenazi Jewish | 1 in 97 | 99% | 1 in 9,600 | |
| | | East Asian | 1 in 208 | 99% | 1 in 21,000 | |
| | | Finnish | 1 in 163 | 99% | 1 in 16,000 | |
| | | European (Non-Finnish) | 1 in 87 | 99% | 1 in 8,600 | |
| | | Native American | 1 in 90 | 99% | 1 in 8,900 | |
| | | South Asian | 1 in 18 | 99% | 1 in 1,700 | |
| Acrodermatitis Enteropathica (AR) NM_130849.3 | *SLC39A4* | Worldwide | 1 in 403 | 96% | 1 in 11,000 | 98% |
| | | African | 1 in 421 | 98% | 1 in 21,000 | |
| | | East Asian | 1 in 1248 | 98% | 1 in 62,000 | |
| | | Finnish | 1 in 216 | 98% | 1 in 11,000 | |
| | | European (Non-Finnish) | 1 in 316 | 97% | 1 in 12,000 | |
| | | Native American | 1 in 1300 | 90% | 1 in 14,000 | |
| | | South Asian | 1 in 1099 | 98% | 1 in 55,000 | |
| Acute Infantile Liver Failure (AR) NM_018006.4 | *TRMU* | Worldwide | 1 in 730 | 89% | 1 in 6,600 | 99% |
| | | African | 1 in 624 | 89% | 1 in 5,500 | |
| | | Ashkenazi Jewish | 1 in 459 | 99% | 1 in 46,000 | |
| | | East Asian | 1 in 551 | 99% | 1 in 55,000 | |
| | | European (Non-Finnish) | 1 in 789 | 92% | 1 in 9,400 | |
| | | Native American | 1 in 1162 | 99% | 1 in 116,000 | |
| | | South Asian | 1 in 321 | 78% | 1 in 1,500 | |
| Acyl-CoA Oxidase I Deficiency (AR) NM_004035.6 | *ACOX1* | Worldwide | 1 in 2212 | 96% | 1 in 59,000 | 98% |
| | | African | 1 in 1071 | 98% | 1 in 54,000 | |
| | | European (Non-Finnish) | 1 in 2394 | 94% | 1 in 39,000 | |
| | | Native American | 1 in 3358 | 98% | 1 in 168,000 | |
| | | South Asian | 1 in 3848 | 98% | 1 in 192,000 | |

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| Usher Syndrome, Type IF (AR)<br>NM_001142764.1 | *PCDH15* | Worldwide | 1 in 447 | 87% | 1 in 3,500 | 98% |
| | | African | 1 in 548 | 98% | 1 in 27,000 | |
| | | Ashkenazi Jewish | 1 in 118 | 98% | 1 in 5,800 | |
| | | East Asian | 1 in 191 | 83% | 1 in 1,100 | |
| | | Finnish | 1 in 2286 | 98% | 1 in 114,000 | |
| | | European (Non-Finnish) | 1 in 497 | 87% | 1 in 3,800 | |
| | | Native American | 1 in 545 | 73% | 1 in 2,000 | |
| | | South Asian | 1 in 805 | 83% | 1 in 4,600 | |
| Usher Syndrome, Type IIA (AR)<br>NM_206933.2 | *USH2A* | Worldwide | 1 in 49 | 77% | 1 in 210 | 98% |
| | | African | 1 in 69 | 75% | 1 in 280 | |
| | | Ashkenazi Jewish | 1 in 40 | 95% | 1 in 750 | |
| | | East Asian | 1 in 27 | 50% | 1 in 52 | |
| | | Finnish | 1 in 142 | 97% | 1 in 4,300 | |
| | | European (Non-Finnish) | 1 in 46 | 80% | 1 in 230 | |
| | | Native American | 1 in 51 | 84% | 1 in 320 | |
| | | South Asian | 1 in 68 | 64% | 1 in 190 | |
| Usher Syndrome, Type III (AR)<br>NM_174878.2 | *CLRN1* | Worldwide | 1 in 308 | 87% | 1 in 2,400 | 99% |
| | | African | 1 in 632 | 99% | 1 in 63,000 | |
| | | Ashkenazi Jewish | 1 in 93 | 99% | 1 in 9,200 | |
| | | East Asian | 1 in 1263 | 56% | 1 in 2,800 | |
| | | Finnish | 1 in 69 | 99% | 1 in 6,800 | |
| | | European (Non-Finnish) | 1 in 420 | 67% | 1 in 1,300 | |
| | | Native American | 1 in 1889 | 99% | 1 in 189,000 | |
| Very Long Chain Acyl-CoA Dehydrogenase Deficiency (AR)<br>NM_000018.3 | *ACADVL* | Worldwide | 1 in 156 | 83% | 1 in 920 | 98% |
| | | African | 1 in 146 | 76% | 1 in 600 | |
| | | Ashkenazi Jewish | 1 in 1259 | 73% | 1 in 4,700 | |
| | | East Asian | 1 in 201 | 47% | 1 in 380 | |
| | | Finnish | 1 in 291 | 94% | 1 in 4,500 | |
| | | European (Non-Finnish) | 1 in 110 | 88% | 1 in 920 | |
| | | Native American | 1 in 267 | 67% | 1 in 810 | |
| | | South Asian | 1 in 372 | 72% | 1 in 1,300 | |
| Walker-Warburg Syndrome and Other FKTN-Related Dystrophies (AR)<br>NM_001079802.1 | *FKTN* | Worldwide | 1 in 541 | 87% | 1 in 4,000 | 95% |
| | | African | 1 in 736 | 95% | 1 in 15,000 | |
| | | Ashkenazi Jewish | 1 in 62 | 95% | 1 in 1,200 | |
| | | East Asian | 1 in 288 | 25% | 1 in 390 | |
| | | European (Non-Finnish) | 1 in 1023 | 76% | 1 in 4,200 | |
| | | Native American | 1 in 382 | 95% | 1 in 7,600 | |
| | | South Asian | 1 in 854 | 90% | 1 in 8,300 | |
| Wilson Disease (AR)<br>NM_000053.3 | *ATP7B* | Worldwide | 1 in 65 | 81% | 1 in 330 | 99% |
| | | African | 1 in 146 | 73% | 1 in 540 | |
| | | Ashkenazi Jewish | 1 in 39 | 97% | 1 in 1,500 | |
| | | East Asian | 1 in 32 | 78% | 1 in 150 | |
| | | Finnish | 1 in 114 | 90% | 1 in 1,100 | |
| | | European (Non-Finnish) | 1 in 63 | 82% | 1 in 350 | |
| | | Native American | 1 in 63 | 74% | 1 in 240 | |
| | | South Asian | 1 in 78 | 60% | 1 in 200 | |
| Wolman Disease / Cholesteryl Ester Storage Disease (AR)<br>NM_000235.3 | *LIPA* | Worldwide | 1 in 328 | 92% | 1 in 3,900 | 98% |
| | | African | 1 in 565 | 84% | 1 in 3,600 | |
| | | Ashkenazi Jewish | 1 in 634 | 98% | 1 in 32,000 | |
| | | East Asian | 1 in 635 | 98% | 1 in 32,000 | |
| | | Finnish | 1 in 1250 | 78% | 1 in 5,600 | |
| | | European (Non-Finnish) | 1 in 233 | 93% | 1 in 3,200 | |
| | | Native American | 1 in 329 | 85% | 1 in 2,100 | |
| | | South Asian | 1 in 769 | 98% | 1 in 38,000 | |
| X-Linked Juvenile Retinoschisis (XL)<br>NM_000330.3 | *RS1* | Worldwide | 1 in 10000 | 72% | 1 in 36,000 | 96% |
| X-Linked Severe Combined Immunodeficiency (XL)<br>NM_000206.2 | *IL2RG* | Worldwide | 1 in 25000 | 89% | 1 in 229,000 | 99% |
| Zellweger Syndrome Spectrum (PEX10-Related) (AR)<br>NM_153818.1 | *PEX10* | Worldwide | 1 in 1739 | 75% | 1 in 7,100 | 99% |
| | | African | 1 in 1604 | 63% | 1 in 4,300 | |
| | | East Asian | 1 in 2180 | 99% | 1 in 218,000 | |
| | | European (Non-Finnish) | 1 in 1287 | 80% | 1 in 6,300 | |
| | | Native American | 1 in 2113 | 99% | 1 in 211,000 | |
| Zellweger Syndrome Spectrum (PEX1-Related) (AR)<br>NM_000466.2 | *PEX1* | Worldwide | 1 in 269 | 91% | 1 in 2,900 | 98% |
| | | African | 1 in 366 | 98% | 1 in 18,000 | |
| | | Ashkenazi Jewish | 1 in 1188 | 98% | 1 in 59,000 | |
| | | East Asian | 1 in 153 | 79% | 1 in 740 | |
| | | Finnish | 1 in 1862 | 61% | 1 in 4,800 | |
| | | European (Non-Finnish) | 1 in 191 | 91% | 1 in 2,000 | |
| | | Native American | 1 in 601 | 98% | 1 in 30,000 | |
| | | South Asian | 1 in 556 | 94% | 1 in 10,000 | |
| Zellweger Syndrome Spectrum (PEX2-Related) (AR)<br>NM_000318.2 | *PEX2* | Worldwide | 1 in 1191 | 93% | 1 in 18,000 | 98% |
| | | African | 1 in 1741 | 98% | 1 in 87,000 | |
| | | Ashkenazi Jewish | 1 in 195 | 98% | 1 in 9,700 | |
| | | East Asian | 1 in 2156 | 98% | 1 in 108,000 | |
| | | European (Non-Finnish) | 1 in 1542 | 98% | 1 in 77,000 | |
| | | Native American | 1 in 2798 | 33% | 1 in 4,200 | |
| | | South Asian | 1 in 1922 | 98% | 1 in 96,000 | |
| Zellweger Syndrome Spectrum (PEX6-Related) (AR)<br>NM_000287.3 | *PEX6* | Worldwide | 1 in 118 | 93% | 1 in 1,800 | 97% |
| | | African | 1 in 268 | 97% | 1 in 8,900 | |
| | | Ashkenazi Jewish | 1 in 263 | 71% | 1 in 910 | |
| | | East Asian | 1 in 595 | 59% | 1 in 1,500 | |
| | | Finnish | 1 in 205 | 97% | 1 in 6,800 | |
| | | European (Non-Finnish) | 1 in 83 | 95% | 1 in 1,600 | |
| | | Native American | 1 in 239 | 85% | 1 in 1,600 | |
| | | South Asian | 1 in 105 | 95% | 1 in 2,100 | |

*Carrier detection by HEXA enzyme analysis has a detection rate of approximately 98%.
†Carrier frequencies include milder and reduced penetrance forms of the disease. Therefore, carrier frequencies may appear higher than reported in the literature.
‡Please note that GJB2 testing includes testing for the two upstream deletions, del(GJB6-D13S1830) and del(GJB6-D13S1854) (PMID:11807148 and 15994881).
AR: Autosomal recessive; N/A: Not available; XL: X-linked