

**Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Disorder)**

The detection rates and risks set forth below were derived using publicly available information, including gnomAD (<https://gnomad.broadinstitute.org>) and ClinVar (<https://www.ncbi.nlm.nih.gov/clinvar>), and the rates were extrapolated using appropriate scientific methodologies. Published studies were used to derive individual carrier risks for CYP21A2, HBA1/HBA2 and SMN1. As additional clinical evidence is available, the data in these charts may be updated from time to time. These data are provided for general informational purposes only and are not intended as a substitute for professional medical judgment or medical advice.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

| Disorder                             | Gene     | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|--------------------------------------|----------|---------------------------------|--------------------|-------------------------|--|--|
| Renal tubular acidosis with deafness | ATP6V0A4 | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |          | Latino/Admixed American         | 99                 | 1 in 281                | 1 in 28,000                                    | 1 in 110,000   |
|                                      |          | Ashkenazi Jewish                | 99                 | 1 in 360                | 1 in 36,000                                    | 1 in 140,000   |
|                                      |          | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |          | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |          | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |          | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |          | Other (population not assigned) | 99                 | 1 in 458                | 1 in 46,000                                    | 1 in 180,000   |
|                                      |          | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      | ATP6V1B1 | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |          | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |          | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      |          | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      |          | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |          | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |          | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |          | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |          | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
| Retinitis pigmentosa                 | CERKL    | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |          | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |          | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |          | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |          | Finnish                         | 99                 | 1 in 48                 | 1 in 4,700                                     | 1 in 19,000  |
|                                      |          | Non-Finnish European/Caucasian  | 99                 | 1 in 361                | 1 in 36,000                                    | 1 in 140,000   |
|                                      |          | South Asian                     | 99                 | 1 in 309                | 1 in 31,000                                    | 1 in 120,000   |
|                                      |          | Other (population not assigned) | 99                 | 1 in 305                | 1 in 30,000                                    | 1 in 120,000   |
|                                      |          | US general population           | 99                 | 1 in 423                | 1 in 42,000                                    | 1 in 170,000   |

| Disorder | Gene  | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|-------|---------------------------------|--------------------|-------------------------|--|--|
|          | CNGA1 | African/African American        | 98                 | 1 in 267                | 1 in 13,000                                    | 1 in 52,000  |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 97                 | 1 in 252                | 1 in 8,400                                     | 1 in 34,000  |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 99                 | 1 in 196                | 1 in 20,000                                    | 1 in 80,000  |
|          |       | South Asian                     | 99                 | 1 in 493                | 1 in 49,000                                    | 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | 1 in 186                | 1 in 19,000                                    | 1 in 76,000  |
|          |       | US general population           | 99                 | 1 in 234                | 1 in 23,000                                    | 1 in 92,000  |
|          | CNGB1 | African/African American        | 99                 | 1 in 136                | 1 in 14,000                                    | 1 in 56,000  |
|          |       | Latino/Admixed American         | 99                 | 1 in 245                | 1 in 24,000                                    | 1 in 96,000  |
|          |       | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | East Asian                      | 99                 | 1 in 281                | 1 in 28,000                                    | 1 in 110,000   |
|          |       | Finnish                         | 99                 | 1 in 77                 | 1 in 7,600                                     | 1 in 30,000  |
|          |       | Non-Finnish European/Caucasian  | 99                 | 1 in 220                | 1 in 22,000                                    | 1 in 88,000  |
|          |       | South Asian                     | 99                 | 1 in 402                | 1 in 40,000                                    | 1 in 160,000   |
|          |       | Other (population not assigned) | 99                 | 1 in 195                | 1 in 19,000                                    | 1 in 76,000  |
|          |       | US general population           | 99                 | 1 in 210                | 1 in 21,000                                    | 1 in 84,000  |
|          | CWC27 | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | DHDDS | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 99                 | 1 in 97                 | 1 in 9,600                                     | 1 in 38,000  |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |

| Disorder | Gene    | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|---------|---------------------------------|--------------------|-------------------------|--|--|
|          | EYS     | African/African American        | 97                 | 1 in 71                 | 1 in 2,300                                     | 1 in 9,200   |
|          |         | Latino/Admixed American         | 99                 | 1 in 146                | 1 in 15,000                                    | 1 in 60,000  |
|          |         | Ashkenazi Jewish                | 99                 | 1 in 42                 | 1 in 4,100                                     | 1 in 16,000  |
|          |         | East Asian                      | 99                 | 1 in 62                 | 1 in 6,100                                     | 1 in 24,000  |
|          |         | Finnish                         | 98                 | 1 in 39                 | 1 in 1,900                                     | 1 in 7,600   |
|          |         | Non-Finnish European/Caucasian  | 99                 | 1 in 106                | 1 in 11,000                                    | 1 in 44,000  |
|          |         | South Asian                     | 98                 | 1 in 220                | 1 in 11,000                                    | 1 in 44,000  |
|          |         | Other (population not assigned) | 98                 | 1 in 85                 | 1 in 4,200                                     | 1 in 17,000  |
|          |         | US general population           | 98                 | 1 in 103                | 1 in 5,100                                     | 1 in 20,000  |
|          | FAM161A | African/African American        | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |         | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |         | Ashkenazi Jewish                | 97                 | 1 in 236                | 1 in 7,800                                     | 1 in 31,000  |
|          |         | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |         | Finnish                         | 97                 | 1 in 469                | 1 in 16,000                                    | 1 in 64,000  |
|          |         | Non-Finnish European/Caucasian  | 98                 | 1 in 356                | 1 in 18,000                                    | 1 in 72,000  |
|          |         | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |         | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |         | US general population           | 98                 | 1 in 469                | 1 in 23,000                                    | 1 in 92,000  |
|          | IFT140  | African/African American        | 98                 | 1 in 348                | 1 in 17,000                                    | 1 in 68,000  |
|          |         | Latino/Admixed American         | 98                 | 1 in 498                | 1 in 25,000                                    | 1 in 100,000   |
|          |         | Ashkenazi Jewish                | 99                 | 1 in 171                | 1 in 17,000                                    | 1 in 68,000  |
|          |         | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |         | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |         | Non-Finnish European/Caucasian  | 99                 | 1 in 307                | 1 in 31,000                                    | 1 in 120,000   |
|          |         | South Asian                     | 99                 | 1 in 450                | 1 in 45,000                                    | 1 in 180,000   |
|          |         | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |         | US general population           | 99                 | 1 in 351                | 1 in 35,000                                    | 1 in 140,000   |
|          | MAK     | African/African American        | 98                 | 1 in 315                | 1 in 16,000                                    | 1 in 64,000  |
|          |         | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |         | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |         | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |         | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |         | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |         | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |         | Other (population not assigned) | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |         | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |

| Disorder | Gene  | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|-------|---------------------------------|--------------------|-------------------------|--|--|
|          | PRCD  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | RLBP1 | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 97                 | 1 in 366                | 1 in 12,000                                    | 1 in 48,000  |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | RP2   | African/African American        | 99                 | N/A                     | N/A  | N/A  |
|          |       | Latino/Admixed American         | 99                 | N/A                     | N/A  | N/A  |
|          |       | Ashkenazi Jewish                | 99                 | N/A                     | N/A  | N/A  |
|          |       | East Asian                      | 99                 | N/A                     | N/A  | N/A  |
|          |       | Finnish                         | 99                 | N/A                     | N/A  | N/A  |
|          |       | Non-Finnish European/Caucasian  | 99                 | N/A                     | N/A  | N/A  |
|          |       | South Asian                     | 97                 | N/A                     | N/A  | N/A  |
|          |       | Other (population not assigned) | 99                 | N/A                     | N/A  | N/A  |
|          |       | US general population           | 97                 | N/A                     | N/A  | N/A  |
|          | RPGR  | African/African American        | 99                 | N/A                     | N/A  | N/A  |
|          |       | Latino/Admixed American         | 99                 | N/A                     | N/A  | N/A  |
|          |       | Ashkenazi Jewish                | 99                 | N/A                     | N/A  | N/A  |
|          |       | East Asian                      | 97                 | N/A                     | N/A  | N/A  |
|          |       | Finnish                         | 99                 | N/A                     | N/A  | N/A  |
|          |       | Non-Finnish European/Caucasian  | 97                 | N/A                     | N/A  | N/A  |
|          |       | South Asian                     | 97                 | N/A                     | N/A  | N/A  |
|          |       | Other (population not assigned) | 99                 | N/A                     | N/A  | N/A  |
|          |       | US general population           | 97                 | N/A                     | N/A  | N/A  |

| Disorder                             | Gene  | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|--------------------------------------|-------|---------------------------------|--------------------|-------------------------|--|--|
| Rhizomelic chondrodysplasia punctata | AGPS  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | Non-Finnish European/Caucasian  | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      |       | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | US general population           | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      | GNPAT | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      |       | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |       | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      | PEX7  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | Non-Finnish European/Caucasian  | 99                 | 1 in 429                | 1 in 43,000                                    | 1 in 170,000   |
|                                      |       | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
| Roberts syndrome                     | ESCO2 | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |       | Latino/Admixed American         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      |       | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                      |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                      |       | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |       | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |       | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                      |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |

| Disorder                        | Gene    | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|---------------------------------|---------|---------------------------------|--------------------|-------------------------|--|--|
| Sandhoff disease                | HEXB    | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | Latino/Admixed American         | 98                 | 1 in 325                | 1 in 16,000                                    | 1 in 64,000  |
|                                 |         | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | East Asian                      | 99                 | 1 in 472                | 1 in 47,000                                    | 1 in 190,000   |
|                                 |         | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | Non-Finnish European/Caucasian  | 99                 | 1 in 278                | 1 in 28,000                                    | 1 in 110,000   |
|                                 |         | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | Other (population not assigned) | 98                 | 1 in 467                | 1 in 23,000                                    | 1 in 92,000  |
|                                 |         | US general population           | 99                 | 1 in 325                | 1 in 32,000                                    | 1 in 130,000   |
| Schimke immunoosseous dysplasia | SMARCA1 | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                 |         | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                 |         | Ashkenazi Jewish                | 99                 | 1 in 192                | 1 in 19,000                                    | 1 in 76,000  |
|                                 |         | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                 |         | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                 |         | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
| Schindler disease               | NAGA    | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | Latino/Admixed American         | 99                 | 1 in 410                | 1 in 41,000                                    | 1 in 160,000   |
|                                 |         | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | Finnish                         | 99                 | 1 in 349                | 1 in 35,000                                    | 1 in 140,000   |
|                                 |         | Non-Finnish European/Caucasian  | 99                 | 1 in 115                | 1 in 11,000                                    | 1 in 44,000  |
|                                 |         | South Asian                     | 99                 | 1 in 165                | 1 in 16,000                                    | 1 in 64,000  |
|                                 |         | Other (population not assigned) | 99                 | 1 in 131                | 1 in 13,000                                    | 1 in 52,000  |
|                                 |         | US general population           | 99                 | 1 in 160                | 1 in 16,000                                    | 1 in 64,000  |
| SELENON-related disorders       | SELENON | African/African American        | 97                 | 1 in 239                | 1 in 7,900                                     | 1 in 32,000  |
|                                 |         | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                 |         | Ashkenazi Jewish                | 97                 | 1 in 345                | 1 in 11,000                                    | 1 in 44,000  |
|                                 |         | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | Non-Finnish European/Caucasian  | 99                 | 1 in 409                | 1 in 41,000                                    | 1 in 160,000   |
|                                 |         | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |         | Other (population not assigned) | 98                 | 1 in 474                | 1 in 24,000                                    | 1 in 96,000  |
|                                 |         | US general population           | 98                 | 1 in 441                | 1 in 22,000                                    | 1 in 88,000  |

| Disorder                                | Gene | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|---|------|---------------------------------|--------------------|-------------------------|--|--|
| Severe combined immunodeficiency (SCID) | AK2  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |      | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |      | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |      | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   | CD3D | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |      | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |      | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |      | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   | CD3E | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   | CD3G | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |      | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |      | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |      | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |      | South Asian                     | 97                 | 1 in 450                | 1 in 15,000                                    | 1 in 60,000  |
|   |      | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |      | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |

| Disorder | Gene   | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|--------|---------------------------------|--------------------|-------------------------|--|--|
|          | CD8A   | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |        | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |        | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | CD247  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | CORO1A | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Latino/Admixed American         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |        | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |        | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | DOCK8  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Other (population not assigned) | 99                 | 1 in 458                | 1 in 46,000                                    | 1 in 180,000   |
|          |        | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |



| Disorder | Gene  | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|-------|---------------------------------|--------------------|-------------------------|--|--|
|          | FOXN1 | African/African American        | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | IKBKB | African/African American        | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | IL2RA | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | IL7R  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Other (population not assigned) | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |

| Disorder | Gene                            | Ethnicity                       | Detection Rate (%)       | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|---------------------------------|---------------------------------|--------------------------|-------------------------|--|--|
|          | JAK3                            | African/African American        | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | Latino/Admixed American         | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | Ashkenazi Jewish                | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | East Asian                      | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | Finnish                         | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |                                 | Non-Finnish European/Caucasian  | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | South Asian                     | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | Other (population not assigned) | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | US general population           | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | LCK                             | African/African American | 99                      | < 1 in 500                                     | < 1 in 50,000  |
|          | Latino/Admixed American         |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | Ashkenazi Jewish                |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | East Asian                      |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | Finnish                         |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | Non-Finnish European/Caucasian  |                                 | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          | South Asian                     |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | Other (population not assigned) |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | US general population           |                                 | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          | LIG4                            |                                 | African/African American | 97                      | 1 in 253                                       | 1 in 8,400   |
|          |                                 | Latino/Admixed American         | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |                                 | Ashkenazi Jewish                | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | East Asian                      | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | Finnish                         | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | Non-Finnish European/Caucasian  | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | South Asian                     | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | Other (population not assigned) | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |                                 | US general population           | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | MALT1                           | African/African American | 97                      | < 1 in 500                                     | < 1 in 17,000  |
|          | Latino/Admixed American         |                                 | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          | Ashkenazi Jewish                |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | East Asian                      |                                 | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          | Finnish                         |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | Non-Finnish European/Caucasian  |                                 | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | South Asian                     |                                 | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | Other (population not assigned) |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | US general population           |                                 | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |

| Disorder | Gene          | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|---------------|---------------------------------|--------------------|-------------------------|--|--|
|          | <i>MTHFD1</i> | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Ashkenazi Jewish                | 99                 | 1 in 464                | 1 in 46,000                                    | 1 in 180,000   |
|          |               | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |               | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | <i>NHEJ1</i>  | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |               | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |               | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |               | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Other (population not assigned) | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |               | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | <i>PGM3</i>   | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |               | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |               | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |               | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | <i>PNP</i>    | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |               | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |

| Disorder | Gene  | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|-------|---------------------------------|--------------------|-------------------------|--|--|
|          | PRKDC | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | PTPRC | African/African American        | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Other (population not assigned) | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | STK4  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | TTC7A | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |

| Disorder  | Gene  | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|---|-------|---------------------------------|--------------------|-------------------------|--|--|
|   | ZAP70 | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |       | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |       | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |       | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
| Severe combined Immunodeficiency (SCID), X-linked | IL2RG | African/African American        | 99                 | N/A                     | N/A  | N/A  |
|   |       | Latino/Admixed American         | 99                 | N/A                     | N/A  | N/A  |
|   |       | Ashkenazi Jewish                | 99                 | N/A                     | N/A  | N/A  |
|   |       | East Asian                      | 99                 | N/A                     | N/A  | N/A  |
|   |       | Finnish                         | 99                 | N/A                     | N/A  | N/A  |
|   |       | Non-Finnish European/Caucasian  | 99                 | N/A                     | N/A  | N/A  |
|   |       | South Asian                     | 99                 | N/A                     | N/A  | N/A  |
|   |       | Other (population not assigned) | 99                 | N/A                     | N/A  | N/A  |
|   |       | US general population           | 99                 | N/A                     | N/A  | N/A  |
| Severe congenital neutropenia                     | HAX1  | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |       | Non-Finnish European/Caucasian  | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   | VPS45 | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |       | Latino/Admixed American         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |       | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |

| Disorder   | Gene    | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|--|---------|---------------------------------|--------------------|-------------------------|--|--|
| Short/branched chain acyl-CoA dehydrogenase deficiency | ACADSB  | African/African American        | 97                 | 1 in 316                | 1 in 11,000                                    | 1 in 44,000  |
|  |         | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | East Asian                      | 99                 | 1 in 147                | 1 in 15,000                                    | 1 in 60,000  |
|  |         | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | Non-Finnish European/Caucasian  | 99                 | 1 in 478                | 1 in 48,000                                    | 1 in 190,000   |
|  |         | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | US general population           | 99                 | 1 in 439                | 1 in 44,000                                    | 1 in 180,000   |
| Short-rib thoracic dysplasia                           | DYNC2H1 | African/African American        | 99                 | 1 in 143                | 1 in 14,000                                    | 1 in 56,000  |
|  |         | Latino/Admixed American         | 99                 | 1 in 183                | 1 in 18,000                                    | 1 in 72,000  |
|  |         | Ashkenazi Jewish                | 99                 | 1 in 179                | 1 in 18,000                                    | 1 in 72,000  |
|  |         | East Asian                      | 99                 | 1 in 307                | 1 in 31,000                                    | 1 in 120,000   |
|  |         | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | Non-Finnish European/Caucasian  | 99                 | 1 in 138                | 1 in 14,000                                    | 1 in 56,000  |
|  |         | South Asian                     | 99                 | 1 in 266                | 1 in 27,000                                    | 1 in 110,000   |
|  |         | Other (population not assigned) | 99                 | 1 in 164                | 1 in 16,000                                    | 1 in 64,000  |
|  |         | US general population           | 99                 | 1 in 150                | 1 in 15,000                                    | 1 in 60,000  |
| Sialic acid storage disorders                          | SLC17A5 | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |         | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|  |         | Finnish                         | 99                 | 1 in 83                 | 1 in 8,200                                     | 1 in 33,000  |
|  |         | Non-Finnish European/Caucasian  | 99                 | 1 in 362                | 1 in 36,000                                    | 1 in 140,000   |
|  |         | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | Other (population not assigned) | 98                 | 1 in 479                | 1 in 24,000                                    | 1 in 96,000  |
|  |         | US general population           | 99                 | 1 in 471                | 1 in 47,000                                    | 1 in 190,000   |
| Sialidosis   | NEU1    | African/African American        | 99                 | 1 in 427                | 1 in 43,000                                    | 1 in 170,000   |
|  |         | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|  |         | East Asian                      | 99                 | 1 in 308                | 1 in 31,000                                    | 1 in 120,000   |
|  |         | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |         | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |         | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |

| Disorder                   | Gene    | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------------------------|---------|---------------------------------|--------------------|-------------------------|--|--|
| Sjogren-Larsson syndrome   | ALDH3A2 | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                            |         | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                            |         | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                            |         | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                            |         | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                            |         | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                            |         | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                            |         | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                            |         | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
| Smith-Lemli-Opitz syndrome | DHCR7   | African/African American        | 99                 | 1 in 129                | 1 in 13,000                                    | 1 in 52,000  |
|                            |         | Latino/Admixed American         | 99                 | 1 in 136                | 1 in 14,000                                    | 1 in 56,000  |
|                            |         | Ashkenazi Jewish                | 99                 | 1 in 40                 | 1 in 3,900                                     | 1 in 16,000  |
|                            |         | East Asian                      | 99                 | 1 in 416                | 1 in 42,000                                    | 1 in 170,000   |
|                            |         | Finnish                         | 99                 | 1 in 181                | 1 in 18,000                                    | 1 in 72,000  |
|                            |         | Non-Finnish European/Caucasian  | 99                 | 1 in 51                 | 1 in 5,000                                     | 1 in 20,000  |
|                            |         | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                            |         | Other (population not assigned) | 99                 | 1 in 72                 | 1 in 7,100                                     | 1 in 28,000  |
|                            |         | US general population           | 99                 | 1 in 67                 | 1 in 6,600                                     | 1 in 26,000  |

| Disorder                | Gene | Population                       | Detection rate (%)<br>(Copy number + SNP)  | Pre-test carrier risk | Post-test risk of being a carrier with 2 copies** |                                    | Post-test risk of being a carrier with 3 copies |
|-------------------------|------|----------------------------------|--|-----------------------|---|------------------------------------|---|
|                         |      |                                  |  |                       | POSITIVE for the<br>c.*3+80T>G SNP                | NEGATIVE for the<br>c.*3+80T>G SNP |   |
| Spinal muscular atrophy | SMN1 | African American                 | 90.3   | 1 in 72               | 1 in 34   | 1 in 375                           | 1 in 4200                                       |
|                         |      | Ashkenazi Jewish                 | 92.8   | 1 in 67               | High risk   | 1 in 918                           | 1 in 5400                                       |
|                         |      | Asian                            | 93.6   | 1 in 59               | High risk   | 1 in 907                           | 1 in 5600                                       |
|                         |      | Caucasian                        | 95.0   | 1 in 47               | 1 in 29   | 1 in 921                           | 1 in 5600                                       |
|                         |      | Hispanic                         | 92.6   | 1 in 68               | 1 in 140  | 1 in 906                           | 1 in 5400                                       |
|                         |      | Mixed or other ethnic background | For counseling purposes, consider using the ethnic background with the most conservative risk estimates. |                       |   |                                    |   |

Footnotes:

\*\* includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

Feng, PMID 28125085; Luo, PMID 23788250

| Disorder  | Gene    | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|---|---------|---------------------------------|--------------------|-------------------------|--|--|
| Spinocerebellar ataxia 10   | ANO10   | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |         | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |         | Non-Finnish European/Caucasian  | 97                 | 1 in 340                | 1 in 11,000                                    | 1 in 44,000  |
|   |         | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | US general population           | 98                 | 1 in 457                | 1 in 23,000                                    | 1 in 92,000  |
| Spondylothoracic dysostosis   | MESP2   | African/African American        | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |         | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |         | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |         | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |         | Finnish                         | 97                 | 1 in 124                | 1 in 4,100                                     | 1 in 16,000  |
|   |         | Non-Finnish European/Caucasian  | 97                 | 1 in 233                | 1 in 7,700                                     | 1 in 31,000  |
|   |         | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | Other (population not assigned) | 97                 | 1 in 48                 | 1 in 1,600                                     | 1 in 6,400   |
|   |         | US general population           | 97                 | 1 in 293                | 1 in 9,700                                     | 1 in 39,000  |
| Stüve-Wiedemann syndrome  | LIFR    | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |         | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|   |         | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |         | Other (population not assigned) | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |         | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
| Sulfate transporter-related osteochondrodysplasias, includes achondrogenesis type 1B, atelosteogenesis type 2, diastrophic dysplasia, and recessive multiple epiphyseal dysplasia | SLC26A2 | African/African American        | 98                 | 1 in 433                | 1 in 22,000                                    | 1 in 88,000  |
|   |         | Latino/Admixed American         | 99                 | 1 in 256                | 1 in 26,000                                    | 1 in 100,000   |
|   |         | Ashkenazi Jewish                | 99                 | 1 in 216                | 1 in 22,000                                    | 1 in 88,000  |
|   |         | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|   |         | Finnish                         | 99                 | 1 in 70                 | 1 in 6,900                                     | 1 in 28,000  |
|   |         | Non-Finnish European/Caucasian  | 99                 | 1 in 144                | 1 in 14,000                                    | 1 in 56,000  |
|   |         | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|   |         | Other (population not assigned) | 99                 | 1 in 348                | 1 in 35,000                                    | 1 in 140,000   |
|   |         | US general population           | 99                 | 1 in 184                | 1 in 18,000                                    | 1 in 72,000  |



| Disorder                              | Gene    | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|---------------------------------------|---------|---------------------------------|--------------------|-------------------------|--|--|
| Sulfite oxidase deficiency            | SUOX    | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                       |         | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | East Asian                      | 99                 | 1 in 377                | 1 in 38,000                                    | 1 in 150,000   |
|                                       |         | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                       |         | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                       |         | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
| Systemic primary carnitine deficiency | SLC22A5 | African/African American        | 99                 | 1 in 228                | 1 in 23,000                                    | 1 in 92,000  |
|                                       |         | Latino/Admixed American         | 99                 | 1 in 112                | 1 in 11,000                                    | 1 in 44,000  |
|                                       |         | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | East Asian                      | 99                 | 1 in 75                 | 1 in 7,400                                     | 1 in 30,000  |
|                                       |         | Finnish                         | 99                 | 1 in 227                | 1 in 23,000                                    | 1 in 92,000  |
|                                       |         | Non-Finnish European/Caucasian  | 99                 | 1 in 114                | 1 in 11,000                                    | 1 in 44,000  |
|                                       |         | South Asian                     | 99                 | 1 in 48                 | 1 in 4,700                                     | 1 in 19,000  |
|                                       |         | Other (population not assigned) | 99                 | 1 in 211                | 1 in 21,000                                    | 1 in 84,000  |
|                                       |         | US general population           | 99                 | 1 in 117                | 1 in 12,000                                    | 1 in 48,000  |
| Tay-Sachs disease                     | HEXA    | African/African American        | 99                 | 1 in 317                | 1 in 32,000                                    | 1 in 130,000   |
|                                       |         | Latino/Admixed American         | 99                 | 1 in 288                | 1 in 29,000                                    | 1 in 120,000   |
|                                       |         | Ashkenazi Jewish                | 97                 | 1 in 31                 | 1 in 1,000                                     | 1 in 4,000   |
|                                       |         | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                                       |         | Non-Finnish European/Caucasian  | 99                 | 1 in 265                | 1 in 26,000                                    | 1 in 100,000   |
|                                       |         | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | Other (population not assigned) | 97                 | 1 in 252                | 1 in 8,400                                     | 1 in 34,000  |
|                                       |         | US general population           | 99                 | 1 in 285                | 1 in 28,000                                    | 1 in 110,000   |
| Tetrahydrobiopterin deficiency        | PCBD1   | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | Ashkenazi Jewish                | 99                 | 1 in 399                | 1 in 40,000                                    | 1 in 160,000   |
|                                       |         | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                       |         | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |

| Disorder                     | Gene   | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|------------------------------|--------|---------------------------------|--------------------|-------------------------|--|--|
|                              | PTS    | African/African American        | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                              |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | East Asian                      | 99                 | 1 in 118                | 1 in 12,000                                    | 1 in 48,000  |
|                              |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                              |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              | QDPR   | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
| Trichohepatoenteric syndrome | SKIV2L | African/African American        | 98                 | 1 in 337                | 1 in 17,000                                    | 1 in 68,000  |
|                              |        | Latino/Admixed American         | 98                 | 1 in 342                | 1 in 17,000                                    | 1 in 68,000  |
|                              |        | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                              |        | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Non-Finnish European/Caucasian  | 98                 | 1 in 347                | 1 in 17,000                                    | 1 in 68,000  |
|                              |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | US general population           | 98                 | 1 in 362                | 1 in 18,000                                    | 1 in 72,000  |
|                              | TTC37  | African/African American        | 99                 | 1 in 404                | 1 in 40,000                                    | 1 in 160,000   |
|                              |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Ashkenazi Jewish                | 99                 | 1 in 370                | 1 in 37,000                                    | 1 in 150,000   |
|                              |        | East Asian                      | 99                 | 1 in 121                | 1 in 12,000                                    | 1 in 48,000  |
|                              |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | Non-Finnish European/Caucasian  | 98                 | 1 in 483                | 1 in 24,000                                    | 1 in 96,000  |
|                              |        | South Asian                     | 99                 | 1 in 215                | 1 in 21,000                                    | 1 in 84,000  |
|                              |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                              |        | US general population           | 98                 | 1 in 452                | 1 in 23,000                                    | 1 in 92,000  |

| Disorder                        | Gene | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|---------------------------------|------|---------------------------------|--------------------|-------------------------|--|--|
| Trimethylaminuria               | FMO3 | African/African American        | 98                 | 1 in 183                | 1 in 9,100                                     | 1 in 36,000  |
|                                 |      | Latino/Admixed American         | 99                 | 1 in 374                | 1 in 37,000                                    | 1 in 150,000   |
|                                 |      | Ashkenazi Jewish                | 99                 | 1 in 345                | 1 in 34,000                                    | 1 in 140,000   |
|                                 |      | East Asian                      | 98                 | 1 in 115                | 1 in 5,700                                     | 1 in 23,000  |
|                                 |      | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                 |      | Non-Finnish European/Caucasian  | 99                 | 1 in 134                | 1 in 13,000                                    | 1 in 52,000  |
|                                 |      | South Asian                     | 98                 | 1 in 326                | 1 in 16,000                                    | 1 in 64,000  |
|                                 |      | Other (population not assigned) | 99                 | 1 in 130                | 1 in 13,000                                    | 1 in 52,000  |
|                                 |      | US general population           | 99                 | 1 in 158                | 1 in 16,000                                    | 1 in 64,000  |
| Triple A syndrome               | AAAS | African/African American        | 98                 | 1 in 210                | 1 in 10,000                                    | 1 in 40,000  |
|                                 |      | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                 |      | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                 |      | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                 |      | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | Non-Finnish European/Caucasian  | 99                 | 1 in 422                | 1 in 42,000                                    | 1 in 170,000   |
|                                 |      | South Asian                     | 99                 | 1 in 437                | 1 in 44,000                                    | 1 in 180,000   |
|                                 |      | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                                 |      | US general population           | 98                 | 1 in 405                | 1 in 20,000                                    | 1 in 80,000  |
| Tyrosine hydroxylase deficiency | TH   | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | East Asian                      | 99                 | 1 in 288                | 1 in 29,000                                    | 1 in 120,000   |
|                                 |      | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
| Tyrosinemia type I              | FAH  | African/African American        | 99                 | 1 in 477                | 1 in 48,000                                    | 1 in 190,000   |
|                                 |      | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | Ashkenazi Jewish                | 99                 | 1 in 137                | 1 in 14,000                                    | 1 in 56,000  |
|                                 |      | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | Finnish                         | 99                 | 1 in 327                | 1 in 33,000                                    | 1 in 130,000   |
|                                 |      | Non-Finnish European/Caucasian  | 99                 | 1 in 309                | 1 in 31,000                                    | 1 in 120,000   |
|                                 |      | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                                 |      | Other (population not assigned) | 99                 | 1 in 292                | 1 in 29,000                                    | 1 in 120,000   |
|                                 |      | US general population           | 99                 | 1 in 378                | 1 in 38,000                                    | 1 in 150,000   |

| Disorder   | Gene   | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|--|--------|---------------------------------|--------------------|-------------------------|--|--|
| Tyrosinemia type II                                    | TAT    | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
| Tyrosinemia type III                                   | HPD    | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |        | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |        | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|  |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
| Usher syndrome (hearing loss and retinitis pigmentosa) | ADGRV1 | African/African American        | 98                 | 1 in 191                | 1 in 9,500                                     | 1 in 38,000  |
|  |        | Latino/Admixed American         | 98                 | 1 in 459                | 1 in 23,000                                    | 1 in 92,000  |
|  |        | Ashkenazi Jewish                | 99                 | 1 in 298                | 1 in 30,000                                    | 1 in 120,000   |
|  |        | East Asian                      | 98                 | 1 in 305                | 1 in 15,000                                    | 1 in 60,000  |
|  |        | Finnish                         | 98                 | 1 in 404                | 1 in 20,000                                    | 1 in 80,000  |
|  |        | Non-Finnish European/Caucasian  | 98                 | 1 in 173                | 1 in 8,600                                     | 1 in 34,000  |
|  |        | South Asian                     | 99                 | 1 in 252                | 1 in 25,000                                    | 1 in 100,000   |
|  |        | Other (population not assigned) | 98                 | 1 in 281                | 1 in 14,000                                    | 1 in 56,000  |
|  |        | US general population           | 98                 | 1 in 205                | 1 in 10,000                                    | 1 in 40,000  |
|  | CDH23  | African/African American        | 99                 | 1 in 280                | 1 in 28,000                                    | 1 in 110,000   |
|  |        | Latino/Admixed American         | 98                 | 1 in 421                | 1 in 21,000                                    | 1 in 84,000  |
|  |        | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | East Asian                      | 97                 | 1 in 85                 | 1 in 2,800                                     | 1 in 11,000  |
|  |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |        | Non-Finnish European/Caucasian  | 99                 | 1 in 281                | 1 in 28,000                                    | 1 in 110,000   |
|  |        | South Asian                     | 99                 | 1 in 343                | 1 in 34,000                                    | 1 in 140,000   |
|  |        | Other (population not assigned) | 98                 | 1 in 331                | 1 in 17,000                                    | 1 in 68,000  |
|  |        | US general population           | 98                 | 1 in 277                | 1 in 14,000                                    | 1 in 56,000  |

| Disorder | Gene   | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|--------|---------------------------------|--------------------|-------------------------|--|--|
|          | CIB2   | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | CLR1   | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Ashkenazi Jewish                | 99                 | 1 in 87                 | 1 in 8,600                                     | 1 in 34,000  |
|          |        | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |        | Finnish                         | 99                 | 1 in 71                 | 1 in 7,000                                     | 1 in 28,000  |
|          |        | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Other (population not assigned) | 99                 | 1 in 312                | 1 in 31,000                                    | 1 in 120,000   |
|          |        | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | MYO7A  | African/African American        | 99                 | 1 in 169                | 1 in 17,000                                    | 1 in 68,000  |
|          |        | Latino/Admixed American         | 99                 | 1 in 300                | 1 in 30,000                                    | 1 in 120,000   |
|          |        | Ashkenazi Jewish                | 99                 | 1 in 380                | 1 in 38,000                                    | 1 in 150,000   |
|          |        | East Asian                      | 99                 | 1 in 297                | 1 in 30,000                                    | 1 in 120,000   |
|          |        | Finnish                         | 97                 | 1 in 319                | 1 in 11,000                                    | 1 in 44,000  |
|          |        | Non-Finnish European/Caucasian  | 99                 | 1 in 158                | 1 in 16,000                                    | 1 in 64,000  |
|          |        | South Asian                     | 99                 | 1 in 215                | 1 in 21,000                                    | 1 in 84,000  |
|          |        | Other (population not assigned) | 98                 | 1 in 270                | 1 in 13,000                                    | 1 in 52,000  |
|          |        | US general population           | 99                 | 1 in 181                | 1 in 18,000                                    | 1 in 72,000  |
|          | PCDH15 | African/African American        | 98                 | 1 in 423                | 1 in 21,000                                    | 1 in 84,000  |
|          |        | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Ashkenazi Jewish                | 99                 | 1 in 116                | 1 in 12,000                                    | 1 in 48,000  |
|          |        | East Asian                      | 97                 | 1 in 206                | 1 in 6,800                                     | 1 in 27,000  |
|          |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Non-Finnish European/Caucasian  | 98                 | 1 in 397                | 1 in 20,000                                    | 1 in 80,000  |
|          |        | South Asian                     | 97                 | 1 in 476                | 1 in 16,000                                    | 1 in 64,000  |
|          |        | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | US general population           | 98                 | 1 in 417                | 1 in 21,000                                    | 1 in 84,000  |

| Disorder | Gene  | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|-------|---------------------------------|--------------------|-------------------------|--|--|
|          | USH1C | African/African American        | 98                 | 1 in 498                | 1 in 25,000                                    | 1 in 100,000   |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 97                 | 1 in 235                | 1 in 7,800                                     | 1 in 31,000  |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | USH1G | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | USH2A | African/African American        | 98                 | 1 in 87                 | 1 in 4,300                                     | 1 in 17,000  |
|          |       | Latino/Admixed American         | 99                 | 1 in 54                 | 1 in 5,300                                     | 1 in 21,000  |
|          |       | Ashkenazi Jewish                | 99                 | 1 in 44                 | 1 in 4,300                                     | 1 in 17,000  |
|          |       | East Asian                      | 99                 | 1 in 67                 | 1 in 6,600                                     | 1 in 26,000  |
|          |       | Finnish                         | 98                 | 1 in 142                | 1 in 7,100                                     | 1 in 28,000  |
|          |       | Non-Finnish European/Caucasian  | 99                 | 1 in 55                 | 1 in 5,400                                     | 1 in 22,000  |
|          |       | South Asian                     | 99                 | 1 in 132                | 1 in 13,000                                    | 1 in 52,000  |
|          |       | Other (population not assigned) | 99                 | 1 in 61                 | 1 in 6,000                                     | 1 in 24,000  |
|          |       | US general population           | 99                 | 1 in 59                 | 1 in 5,800                                     | 1 in 23,000  |
|          | WHRN  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Latino/Admixed American         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |

| Disorder   | Gene    | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|--|---------|---------------------------------|--------------------|-------------------------|--|--|
| Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency  | ACADVL  | African/African American        | 99                 | 1 in 216                | 1 in 22,000                                    | 1 in 88,000  |
|  |         | Latino/Admixed American         | 99                 | 1 in 403                | 1 in 40,000                                    | 1 in 160,000   |
|  |         | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | East Asian                      | 99                 | 1 in 471                | 1 in 47,000                                    | 1 in 190,000   |
|  |         | Finnish                         | 99                 | 1 in 261                | 1 in 26,000                                    | 1 in 100,000   |
|  |         | Non-Finnish European/Caucasian  | 99                 | 1 in 122                | 1 in 12,000                                    | 1 in 48,000  |
|  |         | South Asian                     | 99                 | 1 in 432                | 1 in 43,000                                    | 1 in 170,000   |
|  |         | Other (population not assigned) | 99                 | 1 in 118                | 1 in 12,000                                    | 1 in 48,000  |
|  |         | US general population           | 99                 | 1 in 156                | 1 in 15,000                                    | 1 in 60,000  |
| Vitamin D-dependent rickets                                | CYP27B1 | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |         | Latino/Admixed American         | 98                 | 1 in 458                | 1 in 23,000                                    | 1 in 92,000  |
|  |         | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|  |         | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |         | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |         | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|  |         | Other (population not assigned) | 99                 | 1 in 397                | 1 in 40,000                                    | 1 in 160,000   |
|  |         | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
| Walker-Warburg syndrome and other FKTN-related dystrophies | FKTN    | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |         | Latino/Admixed American         | 97                 | 1 in 385                | 1 in 13,000                                    | 1 in 52,000  |
|  |         | Ashkenazi Jewish                | 97                 | 1 in 63                 | 1 in 2,100                                     | 1 in 8,400   |
|  |         | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |         | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|  |         | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |         | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
| Werner syndrome  | WRN     | African/African American        | 98                 | 1 in 408                | 1 in 20,000                                    | 1 in 80,000  |
|  |         | Latino/Admixed American         | 98                 | 1 in 471                | 1 in 24,000                                    | 1 in 96,000  |
|  |         | Ashkenazi Jewish                | 99                 | 1 in 428                | 1 in 43,000                                    | 1 in 170,000   |
|  |         | East Asian                      | 98                 | 1 in 259                | 1 in 13,000                                    | 1 in 52,000  |
|  |         | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | Non-Finnish European/Caucasian  | 98                 | 1 in 335                | 1 in 17,000                                    | 1 in 68,000  |
|  |         | South Asian                     | 98                 | 1 in 354                | 1 in 18,000                                    | 1 in 72,000  |
|  |         | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |         | US general population           | 98                 | 1 in 363                | 1 in 18,000                                    | 1 in 72,000  |

| Disorder                 | Gene  | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|--------------------------|-------|---------------------------------|--------------------|-------------------------|--|--|
| Wilson disease           | ATP7B | African/African American        | 99                 | 1 in 205                | 1 in 20,000                                    | 1 in 80,000  |
|                          |       | Latino/Admixed American         | 99                 | 1 in 83                 | 1 in 8,200                                     | 1 in 33,000  |
|                          |       | Ashkenazi Jewish                | 99                 | 1 in 42                 | 1 in 4,100                                     | 1 in 16,000  |
|                          |       | East Asian                      | 99                 | 1 in 44                 | 1 in 4,300                                     | 1 in 17,000  |
|                          |       | Finnish                         | 99                 | 1 in 215                | 1 in 21,000                                    | 1 in 84,000  |
|                          |       | Non-Finnish European/Caucasian  | 99                 | 1 in 72                 | 1 in 7,100                                     | 1 in 28,000  |
|                          |       | South Asian                     | 99                 | 1 in 114                | 1 in 11,000                                    | 1 in 44,000  |
|                          |       | Other (population not assigned) | 99                 | 1 in 98                 | 1 in 9,700                                     | 1 in 39,000  |
|                          |       | US general population           | 99                 | 1 in 79                 | 1 in 7,800                                     | 1 in 31,000  |
| Wiskott-Aldrich syndrome | WAS   | African/African American        | 99                 | N/A                     | N/A  | N/A  |
|                          |       | Latino/Admixed American         | 99                 | N/A                     | N/A  | N/A  |
|                          |       | Ashkenazi Jewish                | 99                 | N/A                     | N/A  | N/A  |
|                          |       | East Asian                      | 99                 | N/A                     | N/A  | N/A  |
|                          |       | Finnish                         | 99                 | N/A                     | N/A  | N/A  |
|                          |       | Non-Finnish European/Caucasian  | 99                 | N/A                     | N/A  | N/A  |
|                          |       | South Asian                     | 99                 | N/A                     | N/A  | N/A  |
|                          |       | Other (population not assigned) | 99                 | N/A                     | N/A  | N/A  |
|                          |       | US general population           | 99                 | N/A                     | N/A  | N/A  |
| Xeroderma pigmentosum    | DDB2  | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                          |       | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                          |       | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                          |       | Other (population not assigned) | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|                          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                          | ERCC2 | African/African American        | 99                 | 1 in 347                | 1 in 35,000                                    | 1 in 140,000   |
|                          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                          |       | Ashkenazi Jewish                | 99                 | 1 in 107                | 1 in 11,000                                    | 1 in 44,000  |
|                          |       | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                          |       | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|                          |       | Non-Finnish European/Caucasian  | 99                 | 1 in 202                | 1 in 20,000                                    | 1 in 80,000  |
|                          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|                          |       | Other (population not assigned) | 99                 | 1 in 312                | 1 in 31,000                                    | 1 in 120,000   |
|                          |       | US general population           | 99                 | 1 in 256                | 1 in 26,000                                    | 1 in 100,000   |



| Disorder | Gene  | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|-------|---------------------------------|--------------------|-------------------------|--|--|
|          | ERCC3 | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 99                 | 1 in 55                 | 1 in 5,400                                     | 1 in 22,000  |
|          |       | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Non-Finnish European/Caucasian  | 98                 | 1 in 388                | 1 in 19,000                                    | 1 in 76,000  |
|          |       | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Other (population not assigned) | 98                 | 1 in 318                | 1 in 16,000                                    | 1 in 64,000  |
|          |       | US general population           | 98                 | 1 in 455                | 1 in 23,000                                    | 1 in 92,000  |
|          | ERCC4 | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Other (population not assigned) | 98                 | 1 in 441                | 1 in 22,000                                    | 1 in 88,000  |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | ERCC5 | African/African American        | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | South Asian                     | 99                 | 1 in 283                | 1 in 28,000                                    | 1 in 110,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | POLH  | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | East Asian                      | 99                 | 1 in 488                | 1 in 49,000                                    | 1 in 200,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |

| Disorder   | Gene | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|--|------|---------------------------------|--------------------|-------------------------|--|--|
|  | XPA  | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |      | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | Finnish                         | 99                 | 1 in 155                | 1 in 15,000                                    | 1 in 60,000  |
|  |      | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |      | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  | XPC  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |      | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|  |      | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |      | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |      | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |      | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
| <b>Zellweger spectrum disorder/<br/>peroxisome biogenesis disorder</b> | PEX1 | African/African American        | 97                 | 1 in 392                | 1 in 13,000                                    | 1 in 52,000  |
|  |      | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|  |      | Non-Finnish European/Caucasian  | 98                 | 1 in 204                | 1 in 10,000                                    | 1 in 40,000  |
|  |      | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |      | Other (population not assigned) | 98                 | 1 in 393                | 1 in 20,000                                    | 1 in 80,000  |
|  |      | US general population           | 98                 | 1 in 266                | 1 in 13,000                                    | 1 in 52,000  |
|  | PEX2 | African/African American        | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|  |      | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | Ashkenazi Jewish                | 99                 | 1 in 208                | 1 in 21,000                                    | 1 in 84,000  |
|  |      | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |      | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|  |      | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|  |      | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|  |      | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |

| Disorder | Gene  | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|-------|---------------------------------|--------------------|-------------------------|--|--|
|          | PEX3  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | PEX5  | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | PEX6  | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Non-Finnish European/Caucasian  | 98                 | 1 in 470                | 1 in 23,000                                    | 1 in 92,000  |
|          |       | South Asian                     | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | PEX10 | African/African American        | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Ashkenazi Jewish                | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |       | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |       | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |       | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |

| Disorder | Gene   | Ethnicity                       | Detection Rate (%) | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|--------|---------------------------------|--------------------|-------------------------|--|--|
|          | PEX11B | African/African American        | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Ashkenazi Jewish                | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Finnish                         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Other (population not assigned) | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |        | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | PEX12  | African/African American        | 98                 | 1 in 437                | 1 in 22,000                                    | 1 in 88,000  |
|          |        | Latino/Admixed American         | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | East Asian                      | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Non-Finnish European/Caucasian  | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Other (population not assigned) | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | US general population           | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | PEX13  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | East Asian                      | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Finnish                         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | South Asian                     | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | PEX14  | African/African American        | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Latino/Admixed American         | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Ashkenazi Jewish                | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | East Asian                      | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | Finnish                         | 97                 | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |        | Non-Finnish European/Caucasian  | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | South Asian                     | 98                 | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |        | Other (population not assigned) | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |        | US general population           | 99                 | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |

| Disorder | Gene                            | Ethnicity                       | Detection Rate (%)       | Individual Carrier Risk | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|---------------------------------|---------------------------------|--------------------------|-------------------------|--|--|
|          | PEX16                           | African/African American        | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | Latino/Admixed American         | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |                                 | Ashkenazi Jewish                | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | East Asian                      | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | Finnish                         | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | Non-Finnish European/Caucasian  | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | South Asian                     | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | Other (population not assigned) | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | US general population           | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | PEX19                           | African/African American | 97                      | < 1 in 500                                     | < 1 in 17,000  |
|          | Latino/Admixed American         |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | Ashkenazi Jewish                |                                 | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          | East Asian                      |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | Finnish                         |                                 | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          | Non-Finnish European/Caucasian  |                                 | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | South Asian                     |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | Other (population not assigned) |                                 | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          | US general population           |                                 | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          | PEX26                           |                                 | African/African American | 99                      | < 1 in 500                                     | < 1 in 50,000  |
|          |                                 | Latino/Admixed American         | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | Ashkenazi Jewish                | 97                       | < 1 in 500              | < 1 in 17,000                                  | < 1 in 68,000  |
|          |                                 | East Asian                      | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | Finnish                         | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | Non-Finnish European/Caucasian  | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | South Asian                     | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |
|          |                                 | Other (population not assigned) | 99                       | < 1 in 500              | < 1 in 50,000                                  | < 1 in 200,000   |
|          |                                 | US general population           | 98                       | < 1 in 500              | < 1 in 25,000                                  | < 1 in 100,000   |