

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

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

N/A= Not Available

| Disorder                   | Gene  | Ethnicity<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|----------------------------|-------|---|---|---|---|---|
| Krabbe disease             | GALC  | African/African American  | 99  | 1 in 164  | 1 in 16,000                                       | 1 in 64,000   |
|                            |       | Latino/Admixed American   | 99  | 1 in 200  | 1 in 20,000                                       | 1 in 80,000   |
|                            |       | Ashkenazi Jewish  | 99  | 1 in 533  | 1 in 53,000                                       | 1 in 210,000  |
|                            |       | East Asian  | 99  | 1 in 46   | 1 in 4,500  | 1 in 18,000   |
|                            |       | Finnish   | 99  | 1 in 152  | 1 in 15,000                                       | 1 in 60,000   |
|                            |       | Non-Finnish European/Caucasian  | 99  | 1 in 80   | 1 in 7,900  | 1 in 32,000   |
|                            |       | South Asian   | 99  | 1 in 35   | 1 in 3,400  | 1 in 14,000   |
|                            |       | Other (population not assigned)   | 99  | 1 in 88   | 1 in 8,700  | 1 in 35,000   |
| L1 syndrome                | L1CAM | African/African American  | 99  | N/A   | N/A   | N/A   |
|                            |       | Latino/Admixed American   | 98  | N/A   | N/A   | N/A   |
|                            |       | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|                            |       | East Asian  | 99  | N/A   | N/A   | N/A   |
|                            |       | Finnish   | 97  | N/A   | N/A   | N/A   |
|                            |       | Non-Finnish European/Caucasian  | 99  | N/A   | N/A   | N/A   |
|                            |       | South Asian   | 98  | N/A   | N/A   | N/A   |
|                            |       | Other (population not assigned)   | 99  | N/A   | N/A   | N/A   |
| Leber congenital amaurosis | AIPL1 | African/African American  | 98  | 1 in 524  | 1 in 26,000                                       | 1 in 100,000  |
|                            |       | Latino/Admixed American   | 99  | 1 in 101  | 1 in 10,000                                       | 1 in 40,000   |
|                            |       | Ashkenazi Jewish  | 98  | 1 in 30   | 1 in 1,500  | 1 in 6,000  |
|                            |       | East Asian  | 99  | 1 in 1,149  | 1 in 110,000                                      | 1 in 440,000  |
|                            |       | Finnish   | 97  | 1 in 1,811  | 1 in 60,000                                       | 1 in 240,000  |
|                            |       | Non-Finnish European/Caucasian  | 99  | 1 in 160  | 1 in 16,000                                       | 1 in 64,000   |
|                            |       | South Asian   | 98  | 1 in 31   | 1 in 1,500  | 1 in 6,000  |
|                            |       | Other (population not assigned)   | 99  | 1 in 63   | 1 in 6,200  | 1 in 25,000   |
|                            | LCA5  | African/African American  | 97  | 1 in 8,124  | 1 in 270,000                                      | 1 in 1,100,000  |
|                            |       | Latino/Admixed American   | 98  | 1 in 1,862  | 1 in 93,000                                       | 1 in 370,000  |
|                            |       | Ashkenazi Jewish  | 99  | 1 in 235  | 1 in 23,000                                       | 1 in 92,000   |
|                            |       | East Asian  | 98  | 1 in 1,310  | 1 in 65,000                                       | 1 in 260,000  |
|                            |       | Finnish   | 97  | 1 in 10,822   | 1 in 360,000                                      | 1 in 1,400,000  |
|                            |       | Non-Finnish European/Caucasian  | 98  | 1 in 1,955  | 1 in 98,000                                       | 1 in 390,000  |
|                            |       | South Asian   | 98  | 1 in 1,899  | 1 in 95,000                                       | 1 in 380,000  |
|                            |       | Other (population not assigned)   | 99  | 1 in 721  | 1 in 72,000                                       | 1 in 290,000  |
|                            | LRAT  | African/African American  | 99  | 1 in 8,129  | 1 in 810,000                                      | 1 in 3,200,000  |
|                            |       | Latino/Admixed American   | 97  | 1 in 3,459  | 1 in 120,000                                      | 1 in 480,000  |
|                            |       | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|                            |       | East Asian  | 97  | N/A   | N/A   | N/A   |
|                            |       | Finnish   | 97  | N/A   | N/A   | N/A   |
|                            |       | Non-Finnish European/Caucasian  | 98  | 1 in 7,035  | 1 in 350,000                                      | 1 in 1,400,000  |
|                            |       | South Asian   | 97  | 1 in 15,307   | 1 in 510,000                                      | 1 in 2,000,000  |
|                            |       | Other (population not assigned)   | 97  | N/A   | N/A   | N/A   |
|                            | RD3   | African/African American  | 97  | 1 in 7,736  | 1 in 260,000                                      | 1 in 1,000,000  |
|                            |       | Latino/Admixed American   | 97  | N/A   | N/A   | N/A   |
|                            |       | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|                            |       | East Asian  | 98  | 1 in 4,575  | 1 in 230,000                                      | 1 in 920,000  |
|                            |       | Finnish   | 97  | N/A   | N/A   | N/A   |

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

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| Disorder | Gene                                | Ethnicity<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |              |
|----------|-------------------------------------|---|---|---|---|---|--------------|
| Disorder |                                     | Non-Finnish European/Caucasian  | 99  | 1 in 4,247  | 1 in 420,000                                      | 1 in 1,700,000  |              |
|          |                                     | South Asian   | 97  | N/A   | N/A   | N/A   |              |
|          |                                     | Other (population not assigned)   | 97  | N/A   | N/A   | N/A   |              |
|          | RDH12                               | African/African American  | 99  | 1 in 1,297  | 1 in 130,000                                      | 1 in 520,000  |              |
|          |                                     | Latino/Admixed American   | 98  | 1 in 581  | 1 in 29,000                                       | 1 in 120,000  |              |
|          |                                     | Ashkenazi Jewish  | 98  | N/A   | N/A   | N/A   |              |
|          |                                     | East Asian  | 99  | 1 in 1,786  | 1 in 180,000                                      | 1 in 720,000  |              |
|          |                                     | Finnish   | 99  | 1 in 9,941  | 1 in 990,000                                      | 1 in 4,000,000  |              |
|          |                                     | Non-Finnish European/Caucasian  | 98  | 1 in 685  | 1 in 34,000                                       | 1 in 140,000  |              |
|          |                                     | South Asian   | 99  | 1 in 3,068  | 1 in 310,000                                      | 1 in 1,200,000  |              |
|          |                                     | Other (population not assigned)   | 98  | 1 in 910  | 1 in 45,000                                       | 1 in 180,000  |              |
|          | RPE65                               | African/African American  | 99  | 1 in 431  | 1 in 43,000                                       | 1 in 170,000  |              |
|          |                                     | Latino/Admixed American   | 99  | 1 in 286  | 1 in 29,000                                       | 1 in 120,000  |              |
|          |                                     | Ashkenazi Jewish  | 99  | 1 in 4,925  | 1 in 490,000                                      | 1 in 2,000,000  |              |
|          |                                     | East Asian  | 98  | 1 in 1,766  | 1 in 88,000                                       | 1 in 350,000  |              |
|          |                                     | Finnish   | 99  | 1 in 1,545  | 1 in 150,000                                      | 1 in 600,000  |              |
|          |                                     | Non-Finnish European/Caucasian  | 99  | 1 in 500  | 1 in 50,000                                       | 1 in 200,000  |              |
|          |                                     | South Asian   | 99  | 1 in 698  | 1 in 70,000                                       | 1 in 280,000  |              |
|          |                                     | Other (population not assigned)   | 99  | 1 in 473  | 1 in 47,000                                       | 1 in 190,000  |              |
|          | RPGRI1                              | African/African American  | 98  | 1 in 397  | 1 in 20,000                                       | 1 in 80,000   |              |
|          |                                     | Latino/Admixed American   | 99  | 1 in 401  | 1 in 40,000                                       | 1 in 160,000  |              |
|          |                                     | Ashkenazi Jewish  | 97  | 1 in 542  | 1 in 18,000                                       | 1 in 72,000   |              |
|          |                                     | East Asian  | 99  | 1 in 210  | 1 in 21,000                                       | 1 in 84,000   |              |
|          |                                     | Finnish   | 99  | 1 in 2,680  | 1 in 270,000                                      | 1 in 1,100,000  |              |
|          |                                     | Non-Finnish European/Caucasian  | 99  | 1 in 608  | 1 in 61,000                                       | 1 in 240,000  |              |
|          |                                     | South Asian   | 97  | 1 in 66   | 1 in 2,200  | 1 in 8,800  |              |
|          |                                     | Other (population not assigned)   | 98  | 1 in 271  | 1 in 14,000                                       | 1 in 56,000   |              |
|          | SPATA7                              | African/African American  | 99  | 1 in 447  | 1 in 45,000                                       | 1 in 180,000  |              |
|          |                                     | Latino/Admixed American   | 99  | 1 in 662  | 1 in 66,000                                       | 1 in 260,000  |              |
|          |                                     | Ashkenazi Jewish  | 99  | 1 in 4,916  | 1 in 490,000                                      | 1 in 2,000,000  |              |
|          |                                     | East Asian  | 97  | 1 in 74   | 1 in 2,400  | 1 in 9,600  |              |
|          |                                     | Finnish   | 99  | 1 in 1,074  | 1 in 110,000                                      | 1 in 440,000  |              |
|          |                                     | Non-Finnish European/Caucasian  | 99  | 1 in 433  | 1 in 43,000                                       | 1 in 170,000  |              |
|          |                                     | South Asian   | 99  | 1 in 264  | 1 in 26,000                                       | 1 in 100,000  |              |
|          |                                     | Other (population not assigned)   | 97  | 1 in 611  | 1 in 20,000                                       | 1 in 80,000   |              |
|          | TULP1                               | African/African American  | 98  | 1 in 1,267  | 1 in 63,000                                       | 1 in 250,000  |              |
|          |                                     | Latino/Admixed American   | 99  | 1 in 1,978  | 1 in 200,000                                      | 1 in 800,000  |              |
|          |                                     | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |              |
|          |                                     | East Asian  | 99  | 1 in 710  | 1 in 71,000                                       | 1 in 280,000  |              |
|          |                                     | Finnish   | 97  | 1 in 140  | 1 in 4,600  | 1 in 18,000   |              |
|          |                                     | Non-Finnish European/Caucasian  | 99  | 1 in 1,285  | 1 in 130,000                                      | 1 in 520,000  |              |
|          |                                     | South Asian   | 99  | 1 in 3,046  | 1 in 300,000                                      | 1 in 1,200,000  |              |
|          |                                     | Other (population not assigned)   | 98  | 1 in 1,444  | 1 in 72,000                                       | 1 in 290,000  |              |
|          | Leigh syndrome, autosomal recessive | COX15   | African/African American  | 99  | 1 in 887  | 1 in 89,000   | 1 in 360,000 |
|          |                                     |   | Latino/Admixed American   | 99  | 1 in 719  | 1 in 72,000   | 1 in 290,000 |

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|----------|--------------------------------|---|---|---|--|--|
|          |                                | Ashkenazi Jewish  | 98  | N/A   | N/A  | N/A  |
|          |                                | East Asian  | 98  | 1 in 1,839  | 1 in 92,000                                    | 1 in 370,000   |
|          |                                | Finnish   | 99  | 1 in 5,475  | 1 in 550,000                                   | 1 in 2,200,000   |
|          |                                | Non-Finnish European/Caucasian  | 99  | 1 in 480  | 1 in 48,000                                    | 1 in 190,000   |
|          |                                | South Asian   | 99  | 1 in 159  | 1 in 16,000                                    | 1 in 64,000  |
|          |                                | Other (population not assigned)   | 99  | 1 in 300  | 1 in 30,000                                    | 1 in 120,000   |
|          | FBXL4                          | African/African American  | 99  | 1 in 697  | 1 in 70,000                                    | 1 in 280,000   |
|          |                                | Latino/Admixed American   | 98  | 1 in 727  | 1 in 36,000                                    | 1 in 140,000   |
|          |                                | Ashkenazi Jewish  | 97  | 1 in 1,642  | 1 in 55,000                                    | 1 in 220,000   |
|          |                                | East Asian  | 98  | 1 in 1,278  | 1 in 64,000                                    | 1 in 260,000   |
|          |                                | Finnish   | 97  | N/A   | N/A  | N/A  |
|          |                                | Non-Finnish European/Caucasian  | 99  | 1 in 913  | 1 in 91,000                                    | 1 in 360,000   |
|          | FOXRED1                        | South Asian   | 99  | 1 in 2,180  | 1 in 220,000                                   | 1 in 880,000   |
|          |                                | Other (population not assigned)   | 99  | 1 in 2,734  | 1 in 270,000                                   | 1 in 1,100,000   |
|          |                                | African/African American  | 99  | 1 in 2,646  | 1 in 260,000                                   | 1 in 1,000,000   |
|          |                                | Latino/Admixed American   | 99  | 1 in 482  | 1 in 48,000                                    | 1 in 190,000   |
|          |                                | Ashkenazi Jewish  | 97  | 1 in 549  | 1 in 18,000                                    | 1 in 72,000  |
|          |                                | East Asian  | 99  | 1 in 180  | 1 in 18,000                                    | 1 in 72,000  |
|          |                                | Finnish   | 99  | 1 in 1,326  | 1 in 130,000                                   | 1 in 520,000   |
|          |                                | Non-Finnish European/Caucasian  | 98  | 1 in 439  | 1 in 22,000                                    | 1 in 88,000  |
|          |                                | South Asian   | 99  | 1 in 145  | 1 in 14,000                                    | 1 in 56,000  |
|          |                                | Other (population not assigned)   | 98  | 1 in 552  | 1 in 28,000                                    | 1 in 110,000   |
|          | LRPPRC                         | African/African American  | 98  | 1 in 650  | 1 in 32,000                                    | 1 in 130,000   |
|          |                                | Latino/Admixed American   | 99  | 1 in 591  | 1 in 59,000                                    | 1 in 240,000   |
|          |                                | Ashkenazi Jewish  | 97  | N/A   | N/A  | N/A  |
|          |                                | East Asian  | 97  | 1 in 225  | 1 in 7,500                                     | 1 in 30,000  |
|          |                                | Finnish   | 97  | 1 in 428  | 1 in 14,000                                    | 1 in 56,000  |
|          |                                | Non-Finnish European/Caucasian  | 99  | 1 in 789  | 1 in 79,000                                    | 1 in 320,000   |
|          |                                | South Asian   | 98  | 1 in 754  | 1 in 38,000                                    | 1 in 150,000   |
|          |                                | Other (population not assigned)   | 98  | 1 in 239  | 1 in 12,000                                    | 1 in 48,000  |
|          | NDUFAF2                        | African/African American  | 99  | 1 in 881  | 1 in 88,000                                    | 1 in 350,000   |
|          |                                | Latino/Admixed American   | 99  | 1 in 796  | 1 in 80,000                                    | 1 in 320,000   |
|          |                                | Ashkenazi Jewish  | 97  | 1 in 92   | 1 in 3,000                                     | 1 in 12,000  |
|          |                                | East Asian  | 99  | 1 in 889  | 1 in 89,000                                    | 1 in 360,000   |
|          |                                | Finnish   | 97  | N/A   | N/A  | N/A  |
|          |                                | Non-Finnish European/Caucasian  | 98  | 1 in 1,014  | 1 in 51,000                                    | 1 in 200,000   |
|          |                                | South Asian   | 97  | 1 in 1,326  | 1 in 44,000                                    | 1 in 180,000   |
|          |                                | Other (population not assigned)   | 98  | 1 in 331  | 1 in 17,000                                    | 1 in 68,000  |
|          |                                | NDUFAF5   | African/African American  | 98  | 1 in 1,625                                     | 1 in 81,000  |
|          | Latino/Admixed American        |   | 99  | 1 in 861  | 1 in 86,000                                    | 1 in 340,000   |
|          | Ashkenazi Jewish               |   | 99  | 1 in 157  | 1 in 16,000                                    | 1 in 64,000  |
|          | East Asian                     |   | 99  | 1 in 309  | 1 in 31,000                                    | 1 in 120,000   |
|          | Finnish                        |   | 98  | 1 in 5,320  | 1 in 270,000                                   | 1 in 1,100,000   |
|          | Non-Finnish European/Caucasian |   | 99  | 1 in 417  | 1 in 42,000                                    | 1 in 170,000   |
|          | South Asian                    |   | 99  | 1 in 1,180  | 1 in 120,000                                   | 1 in 480,000   |

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|----------|---|---|---|---|---|---|--------------|
| Disorder |   | Other (population not assigned)   | 99  | 1 in 611  | 1 in 61,000                                       | 1 in 240,000  |              |
|          | NDUFS4  | African/African American  | 98  | 1 in 1,207  | 1 in 60,000                                       | 1 in 240,000  |              |
|          |   | Latino/Admixed American   | 98  | 1 in 1,103  | 1 in 55,000                                       | 1 in 220,000  |              |
|          |   | Ashkenazi Jewish  | 98  | 1 in 1,640  | 1 in 82,000                                       | 1 in 330,000  |              |
|          |   | East Asian  | 99  | 1 in 4,597  | 1 in 460,000                                      | 1 in 1,800,000  |              |
|          |   | Finnish   | 97  | 1 in 2,229  | 1 in 74,000                                       | 1 in 300,000  |              |
|          |   | Non-Finnish European/Caucasian  | 98  | 1 in 1,738  | 1 in 87,000                                       | 1 in 350,000  |              |
|          |   | South Asian   | 99  | 1 in 1,914  | 1 in 190,000                                      | 1 in 760,000  |              |
|          |   | Other (population not assigned)   | 97  | N/A   | N/A   | N/A   |              |
|          | NDUFS6  | African/African American  | 99  | 1 in 2,452  | 1 in 250,000                                      | 1 in 1,000,000  |              |
|          |   | Latino/Admixed American   | 98  | 1 in 1,896  | 1 in 95,000                                       | 1 in 380,000  |              |
|          |   | Ashkenazi Jewish  | 98  | 1 in 1,113  | 1 in 56,000                                       | 1 in 220,000  |              |
|          |   | East Asian  | 99  | 1 in 1,473  | 1 in 150,000                                      | 1 in 600,000  |              |
|          |   | Finnish   | 98  | N/A   | N/A   | N/A   |              |
|          |   | Non-Finnish European/Caucasian  | 98  | 1 in 3,599  | 1 in 180,000                                      | 1 in 720,000  |              |
|          |   | South Asian   | 98  | 1 in 1,398  | 1 in 70,000                                       | 1 in 280,000  |              |
|          |   | Other (population not assigned)   | 98  | N/A   | N/A   | N/A   |              |
|          | NDUFS7  | African/African American  | 99  | 1 in 1,552  | 1 in 160,000                                      | 1 in 640,000  |              |
|          |   | Latino/Admixed American   | 99  | 1 in 2,327  | 1 in 230,000                                      | 1 in 920,000  |              |
|          |   | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |              |
|          |   | East Asian  | 97  | 1 in 3,320  | 1 in 110,000                                      | 1 in 440,000  |              |
|          |   | Finnish   | 97  | 1 in 10,639   | 1 in 350,000                                      | 1 in 1,400,000  |              |
|          |   | Non-Finnish European/Caucasian  | 99  | 1 in 839  | 1 in 84,000                                       | 1 in 340,000  |              |
|          |   | South Asian   | 99  | 1 in 180  | 1 in 18,000                                       | 1 in 72,000   |              |
|          |   | Other (population not assigned)   | 99  | 1 in 578  | 1 in 58,000                                       | 1 in 230,000  |              |
|          | NDUFV1  | African/African American  | 99  | 1 in 157  | 1 in 16,000                                       | 1 in 64,000   |              |
|          |   | Latino/Admixed American   | 99  | 1 in 441  | 1 in 44,000                                       | 1 in 180,000  |              |
|          |   | Ashkenazi Jewish  | 99  | 1 in 1,680  | 1 in 170,000                                      | 1 in 680,000  |              |
|          |   | East Asian  | 98  | 1 in 2,260  | 1 in 110,000                                      | 1 in 440,000  |              |
|          |   | Finnish   | 99  | 1 in 108  | 1 in 11,000                                       | 1 in 44,000   |              |
|          |   | Non-Finnish European/Caucasian  | 99  | 1 in 469  | 1 in 47,000                                       | 1 in 190,000  |              |
|          |   | South Asian   | 99  | 1 in 319  | 1 in 32,000                                       | 1 in 130,000  |              |
|          |   | Other (population not assigned)   | 99  | 1 in 282  | 1 in 28,000                                       | 1 in 110,000  |              |
|          | SURF1   | African/African American  | 99  | 1 in 89   | 1 in 8,800  | 1 in 35,000   |              |
|          |   | Latino/Admixed American   | 99  | 1 in 256  | 1 in 26,000                                       | 1 in 100,000  |              |
|          |   | Ashkenazi Jewish  | 99  | 1 in 170  | 1 in 17,000                                       | 1 in 68,000   |              |
|          |   | East Asian  | 99  | 1 in 61   | 1 in 6,000  | 1 in 24,000   |              |
|          |   | Finnish   | 99  | 1 in 148  | 1 in 15,000                                       | 1 in 60,000   |              |
|          |   | Non-Finnish European/Caucasian  | 99  | 1 in 191  | 1 in 19,000                                       | 1 in 76,000   |              |
|          |   | South Asian   | 98  | 1 in 302  | 1 in 15,000                                       | 1 in 60,000   |              |
|          |   | Other (population not assigned)   | 99  | 1 in 255  | 1 in 25,000                                       | 1 in 100,000  |              |
|          | Leukoencephalopathy with vanishing white matter | EIF2B1  | African/African American  | 99  | 1 in 712  | 1 in 71,000   | 1 in 280,000 |
|          |   |   | Latino/Admixed American   | 99  | 1 in 524  | 1 in 52,000   | 1 in 210,000 |
|          |   |   | Ashkenazi Jewish  | 99  | N/A   | N/A   | N/A          |
|          |   |   | East Asian  | 99  | 1 in 919  | 1 in 92,000   | 1 in 370,000 |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |              |
|----------|---------------------------------|---|---|---|---|---|--------------|
| Disorder |                                 | Finnish   | 99  | 1 in 704  | 1 in 70,000                                       | 1 in 280,000  |              |
|          |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 153  | 1 in 15,000                                       | 1 in 60,000   |              |
|          |                                 | South Asian   | 99  | 1 in 283  | 1 in 28,000                                       | 1 in 110,000  |              |
|          |                                 | Other (population not assigned)   | 99  | 1 in 337  | 1 in 34,000                                       | 1 in 140,000  |              |
|          | EIF2B2                          | African/African American  | 99  | 1 in 259  | 1 in 26,000                                       | 1 in 100,000  |              |
|          |                                 | Latino/Admixed American   | 99  | 1 in 482  | 1 in 48,000                                       | 1 in 190,000  |              |
|          |                                 | Ashkenazi Jewish  | 98  | 1 in 993  | 1 in 50,000                                       | 1 in 200,000  |              |
|          |                                 | East Asian  | 99  | 1 in 212  | 1 in 21,000                                       | 1 in 84,000   |              |
|          |                                 | Finnish   | 99  | 1 in 113  | 1 in 11,000                                       | 1 in 44,000   |              |
|          |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 227  | 1 in 23,000                                       | 1 in 92,000   |              |
|          |                                 | South Asian   | 99  | 1 in 543  | 1 in 54,000                                       | 1 in 220,000  |              |
|          |                                 | Other (population not assigned)   | 99  | 1 in 221  | 1 in 22,000                                       | 1 in 88,000   |              |
|          |                                 | EIF2B3  | African/African American  | 97  | 1 in 527  | 1 in 18,000   | 1 in 72,000  |
|          |                                 |   | Latino/Admixed American   | 97  | 1 in 147  | 1 in 4,900  | 1 in 20,000  |
|          | Ashkenazi Jewish                |   | 97  | 1 in 161  | 1 in 5,300  | 1 in 21,000   |              |
|          | East Asian                      |   | 97  | 1 in 186  | 1 in 6,200  | 1 in 25,000   |              |
|          | Finnish                         |   | 97  | 1 in 932  | 1 in 31,000                                       | 1 in 120,000  |              |
|          | Non-Finnish European/Caucasian  |   | 97  | 1 in 414  | 1 in 14,000                                       | 1 in 56,000   |              |
|          | South Asian                     |   | 97  | 1 in 306  | 1 in 10,000                                       | 1 in 40,000   |              |
|          | Other (population not assigned) |   | 97  | 1 in 184  | 1 in 6,100  | 1 in 24,000   |              |
|          | EIF2B4                          |   | African/African American  | 98  | 1 in 2,323  | 1 in 120,000  | 1 in 480,000 |
|          |                                 |   | Latino/Admixed American   | 99  | 1 in 672  | 1 in 67,000   | 1 in 270,000 |
|          |                                 | Ashkenazi Jewish  | 98  | N/A   | N/A   | N/A   |              |
|          |                                 | East Asian  | 99  | 1 in 569  | 1 in 57,000                                       | 1 in 230,000  |              |
|          |                                 | Finnish   | 98  | N/A   | N/A   | N/A   |              |
|          |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 2,121  | 1 in 210,000                                      | 1 in 840,000  |              |
|          |                                 | South Asian   | 98  | 1 in 3,062  | 1 in 150,000                                      | 1 in 600,000  |              |
|          |                                 | Other (population not assigned)   | 99  | 1 in 2,744  | 1 in 270,000                                      | 1 in 1,100,000  |              |
|          |                                 | EIF2B5  | African/African American  | 98  | 1 in 50   | 1 in 2,500  | 1 in 10,000  |
|          |                                 |   | Latino/Admixed American   | 98  | 1 in 61   | 1 in 3,000  | 1 in 12,000  |
|          | Ashkenazi Jewish                |   | 99  | 1 in 107  | 1 in 11,000                                       | 1 in 44,000   |              |
|          | East Asian                      |   | 99  | 1 in 105  | 1 in 10,000                                       | 1 in 40,000   |              |
|          | Finnish                         |   | 99  | 1 in 50   | 1 in 4,900  | 1 in 20,000   |              |
|          | Non-Finnish European/Caucasian  |   | 98  | 1 in 63   | 1 in 3,100  | 1 in 12,000   |              |
|          | South Asian                     |   | 98  | 1 in 82   | 1 in 4,100  | 1 in 16,000   |              |
|          | Other (population not assigned) |   | 98  | 1 in 67   | 1 in 3,300  | 1 in 13,000   |              |
|          | CAPN3                           |   | African/African American  | 99  | 1 in 118  | 1 in 12,000   | 1 in 48,000  |
|          |                                 |   | Latino/Admixed American   | 99  | 1 in 125  | 1 in 12,000   | 1 in 48,000  |
|          |                                 | Ashkenazi Jewish  | 99  | 1 in 359  | 1 in 36,000                                       | 1 in 140,000  |              |
|          |                                 | East Asian  | 99  | 1 in 95   | 1 in 9,400  | 1 in 38,000   |              |
|          |                                 | Finnish   | 99  | 1 in 551  | 1 in 55,000                                       | 1 in 220,000  |              |
|          |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 100  | 1 in 9,900  | 1 in 40,000   |              |
|          |                                 | South Asian   | 99  | 1 in 188  | 1 in 19,000                                       | 1 in 76,000   |              |
|          |                                 | Other (population not assigned)   | 99  | 1 in 121  | 1 in 12,000                                       | 1 in 48,000   |              |
|          |                                 | DYSF  | African/African American  | 99  | 1 in 77   | 1 in 7,600  | 1 in 30,000  |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|----------|---------|---|---|---|---|---|
|          |         | Latino/Admixed American   | 99  | 1 in 81   | 1 in 8,000  | 1 in 32,000   |
|          |         | Ashkenazi Jewish  | 99  | 1 in 103  | 1 in 10,000                                       | 1 in 40,000   |
|          |         | East Asian  | 99  | 1 in 135  | 1 in 13,000                                       | 1 in 52,000   |
|          |         | Finnish   | 99  | 1 in 31   | 1 in 3,000  | 1 in 12,000   |
|          |         | Non-Finnish European/Caucasian  | 99  | 1 in 48   | 1 in 4,700  | 1 in 19,000   |
|          |         | South Asian   | 99  | 1 in 79   | 1 in 7,800  | 1 in 31,000   |
|          |         | Other (population not assigned)   | 99  | 1 in 65   | 1 in 6,400  | 1 in 26,000   |
|          | FKRP    | African/African American  | 99  | 1 in 3,032  | 1 in 300,000                                      | 1 in 1,200,000  |
|          |         | Latino/Admixed American   | 99  | 1 in 424  | 1 in 42,000                                       | 1 in 170,000  |
|          |         | Ashkenazi Jewish  | 99  | 1 in 4,031  | 1 in 400,000                                      | 1 in 1,600,000  |
|          |         | East Asian  | 99  | 1 in 248  | 1 in 25,000                                       | 1 in 100,000  |
|          |         | Finnish   | 99  | 1 in 318  | 1 in 32,000                                       | 1 in 130,000  |
|          |         | Non-Finnish European/Caucasian  | 99  | 1 in 193  | 1 in 19,000                                       | 1 in 76,000   |
|          |         | South Asian   | 99  | 1 in 1,429  | 1 in 140,000                                      | 1 in 560,000  |
|          | POMGNT1 | Other (population not assigned)   | 99  | 1 in 231  | 1 in 23,000                                       | 1 in 92,000   |
|          |         | African/African American  | 99  | 1 in 771  | 1 in 77,000                                       | 1 in 310,000  |
|          |         | Latino/Admixed American   | 99  | 1 in 486  | 1 in 49,000                                       | 1 in 200,000  |
|          |         | Ashkenazi Jewish  | 99  | 1 in 2,449  | 1 in 240,000                                      | 1 in 960,000  |
|          |         | East Asian  | 98  | 1 in 587  | 1 in 29,000                                       | 1 in 120,000  |
|          |         | Finnish   | 99  | 1 in 198  | 1 in 20,000                                       | 1 in 80,000   |
|          |         | Non-Finnish European/Caucasian  | 99  | 1 in 303  | 1 in 30,000                                       | 1 in 120,000  |
|          | POMT1   | South Asian   | 99  | 1 in 539  | 1 in 54,000                                       | 1 in 220,000  |
|          |         | Other (population not assigned)   | 99  | 1 in 913  | 1 in 91,000                                       | 1 in 360,000  |
|          |         | African/African American  | 99  | 1 in 535  | 1 in 53,000                                       | 1 in 210,000  |
|          |         | Latino/Admixed American   | 99  | 1 in 390  | 1 in 39,000                                       | 1 in 160,000  |
|          |         | Ashkenazi Jewish  | 97  | 1 in 307  | 1 in 10,000                                       | 1 in 40,000   |
|          |         | East Asian  | 99  | 1 in 458  | 1 in 46,000                                       | 1 in 180,000  |
|          |         | Finnish   | 99  | 1 in 1,846  | 1 in 180,000                                      | 1 in 720,000  |
|          | POMT2   | Non-Finnish European/Caucasian  | 98  | 1 in 372  | 1 in 19,000                                       | 1 in 76,000   |
|          |         | South Asian   | 99  | 1 in 427  | 1 in 43,000                                       | 1 in 170,000  |
|          |         | Other (population not assigned)   | 99  | 1 in 264  | 1 in 26,000                                       | 1 in 100,000  |
|          |         | African/African American  | 98  | 1 in 267  | 1 in 13,000                                       | 1 in 52,000   |
|          |         | Latino/Admixed American   | 99  | 1 in 908  | 1 in 91,000                                       | 1 in 360,000  |
|          |         | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|          |         | East Asian  | 99  | 1 in 1,274  | 1 in 130,000                                      | 1 in 520,000  |
|          | SGCA    | Finnish   | 98  | 1 in 3,607  | 1 in 180,000                                      | 1 in 720,000  |
|          |         | Non-Finnish European/Caucasian  | 99  | 1 in 833  | 1 in 83,000                                       | 1 in 330,000  |
|          |         | South Asian   | 99  | 1 in 833  | 1 in 83,000                                       | 1 in 330,000  |
|          |         | Other (population not assigned)   | 97  | 1 in 1,448  | 1 in 48,000                                       | 1 in 190,000  |
|          |         | African/African American  | 98  | 1 in 91   | 1 in 4,500  | 1 in 18,000   |
|          |         | Latino/Admixed American   | 99  | 1 in 317  | 1 in 32,000                                       | 1 in 130,000  |
|          |         | Ashkenazi Jewish  | 99  | 1 in 256  | 1 in 26,000                                       | 1 in 100,000  |
|          |         | East Asian  | 99  | 1 in 774  | 1 in 77,000                                       | 1 in 310,000  |
|          |         | Finnish   | 99  | 1 in 214  | 1 in 21,000                                       | 1 in 84,000   |
|          |         | Non-Finnish European/Caucasian  | 99  | 1 in 191  | 1 in 19,000                                       | 1 in 76,000   |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |             |
|----------|---|---|---|---|---|---|-------------|
|          |   | South Asian   | 99  | 1 in 901  | 1 in 90,000                                       | 1 in 360,000  |             |
|          |   | Other (population not assigned)   | 99  | 1 in 189  | 1 in 19,000                                       | 1 in 76,000   |             |
|          | SGCB                                    | African/African American  | 97  | 1 in 1,087  | 1 in 36,000                                       | 1 in 140,000  |             |
|          |   | Latino/Admixed American   | 99  | 1 in 4,977  | 1 in 500,000                                      | 1 in 2,000,000  |             |
|          |   | Ashkenazi Jewish  | 97  | 1 in 1,008  | 1 in 34,000                                       | 1 in 140,000  |             |
|          |   | East Asian  | 99  | 1 in 1,347  | 1 in 130,000                                      | 1 in 520,000  |             |
|          |   | Finnish   | 99  | 1 in 2,230  | 1 in 220,000                                      | 1 in 880,000  |             |
|          |   | Non-Finnish European/Caucasian  | 99  | 1 in 671  | 1 in 67,000                                       | 1 in 270,000  |             |
|          |   | South Asian   | 99  | 1 in 2,560  | 1 in 260,000                                      | 1 in 1,000,000  |             |
|          |   | Other (population not assigned)   | 97  | 1 in 534  | 1 in 18,000                                       | 1 in 72,000   |             |
|          | SGCD                                    | African/African American  | 99  | 1 in 1,897  | 1 in 190,000                                      | 1 in 760,000  |             |
|          |   | Latino/Admixed American   | 99  | 1 in 3,799  | 1 in 380,000                                      | 1 in 1,500,000  |             |
|          |   | Ashkenazi Jewish  | 99  | 1 in 2,513  | 1 in 250,000                                      | 1 in 1,000,000  |             |
|          |   | East Asian  | 99  | 1 in 6,454  | 1 in 650,000                                      | 1 in 2,600,000  |             |
|          |   | Finnish   | 98  | N/A   | N/A   | N/A   |             |
|          |   | Non-Finnish European/Caucasian  | 99  | 1 in 3,766  | 1 in 380,000                                      | 1 in 1,500,000  |             |
|          |   | South Asian   | 98  | 1 in 6,537  | 1 in 330,000                                      | 1 in 1,300,000  |             |
|          |   | Other (population not assigned)   | 99  | 1 in 1,504  | 1 in 150,000                                      | 1 in 600,000  |             |
|          | SGCG                                    | African/African American  | 99  | 1 in 969  | 1 in 97,000                                       | 1 in 390,000  |             |
|          |   | Latino/Admixed American   | 97  | 1 in 2,114  | 1 in 70,000                                       | 1 in 280,000  |             |
|          |   | Ashkenazi Jewish  | 98  | 1 in 2,315  | 1 in 120,000                                      | 1 in 480,000  |             |
|          |   | East Asian  | 97  | 1 in 4,450  | 1 in 150,000                                      | 1 in 600,000  |             |
|          |   | Finnish   | 99  | 1 in 5,410  | 1 in 540,000                                      | 1 in 2,200,000  |             |
|          |   | Non-Finnish European/Caucasian  | 98  | 1 in 1,558  | 1 in 78,000                                       | 1 in 310,000  |             |
|          |   | South Asian   | 98  | 1 in 2,465  | 1 in 120,000                                      | 1 in 480,000  |             |
|          |   | Other (population not assigned)   | 97  | 1 in 2,742  | 1 in 91,000                                       | 1 in 360,000  |             |
|          | TRAPPC11                                | African/African American  | 99  | 1 in 728  | 1 in 73,000                                       | 1 in 290,000  |             |
|          |   | Latino/Admixed American   | 99  | 1 in 404  | 1 in 40,000                                       | 1 in 160,000  |             |
|          |   | Ashkenazi Jewish  | 99  | 1 in 153  | 1 in 15,000                                       | 1 in 60,000   |             |
|          |   | East Asian  | 99  | 1 in 377  | 1 in 38,000                                       | 1 in 150,000  |             |
|          |   | Finnish   | 99  | 1 in 1,092  | 1 in 110,000                                      | 1 in 440,000  |             |
|          |   | Non-Finnish European/Caucasian  | 99  | 1 in 271  | 1 in 27,000                                       | 1 in 110,000  |             |
|          |   | South Asian   | 99  | 1 in 837  | 1 in 84,000                                       | 1 in 340,000  |             |
|          |   | Other (population not assigned)   | 99  | 1 in 511  | 1 in 51,000                                       | 1 in 200,000  |             |
|          | TRIM32                                  | African/African American  | 98  | 1 in 674  | 1 in 34,000                                       | 1 in 140,000  |             |
|          |   | Latino/Admixed American   | 99  | 1 in 665  | 1 in 66,000                                       | 1 in 260,000  |             |
|          |   | Ashkenazi Jewish  | 99  | 1 in 82   | 1 in 8,100  | 1 in 32,000   |             |
|          |   | East Asian  | 98  | 1 in 920  | 1 in 46,000                                       | 1 in 180,000  |             |
|          |   | Finnish   | 98  | 1 in 5,489  | 1 in 270,000                                      | 1 in 1,100,000  |             |
|          |   | Non-Finnish European/Caucasian  | 99  | 1 in 226  | 1 in 23,000                                       | 1 in 92,000   |             |
|          |   | South Asian   | 99  | 1 in 300  | 1 in 30,000                                       | 1 in 120,000  |             |
|          |   | Other (population not assigned)   | 99  | 1 in 162  | 1 in 16,000                                       | 1 in 64,000   |             |
|          | Lipoprotein lipase deficiency, familial | LPL   | African/African American  | 99  | 1 in 145  | 1 in 14,000   | 1 in 56,000 |
|          |   |   | Latino/Admixed American   | 99  | 1 in 68   | 1 in 6,700  | 1 in 27,000 |
|          |   |   | Ashkenazi Jewish  | 99  | 1 in 47   | 1 in 4,600  | 1 in 18,000 |

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

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

N/A= Not Available

| Disorder  | Gene                            | Ethnicity<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2) | Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2) | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|---|---------------------------------|---|---|---|--|--|
|   |                                 | East Asian  | 99  | 1 in 1,452  | 1 in 150,000                                   | 1 in 600,000   |
|   |                                 | Finnish   | 99  | 1 in 25   | 1 in 2,400                                     | 1 in 9,600   |
|   |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 25   | 1 in 2,400                                     | 1 in 9,600   |
|   |                                 | South Asian   | 99  | 1 in 214  | 1 in 21,000                                    | 1 in 84,000  |
|   |                                 | Other (population not assigned)   | 99  | 1 in 33   | 1 in 3,200                                     | 1 in 13,000  |
| Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency | HADHA                           | African/African American  | 99  | 1 in 463  | 1 in 46,000                                    | 1 in 180,000   |
|   |                                 | Latino/Admixed American   | 99  | 1 in 393  | 1 in 39,000                                    | 1 in 160,000   |
|   |                                 | Ashkenazi Jewish  | 97  | N/A   | N/A  | N/A  |
|   |                                 | East Asian  | 97  | 1 in 537  | 1 in 18,000                                    | 1 in 72,000  |
|   |                                 | Finnish   | 99  | 1 in 117  | 1 in 12,000                                    | 1 in 48,000  |
|   |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 212  | 1 in 21,000                                    | 1 in 84,000  |
|   |                                 | South Asian   | 99  | 1 in 698  | 1 in 70,000                                    | 1 in 280,000   |
|   |                                 | Other (population not assigned)   | 99  | 1 in 196  | 1 in 20,000                                    | 1 in 80,000  |
|   |                                 | Lysinuric protein intolerance   | SLC7A7  | African/African American  | 97   | 1 in 885   |
| Latino/Admixed American                                       | 98                              |   |   | 1 in 2,314  | 1 in 120,000                                   | 1 in 480,000   |
| Ashkenazi Jewish  | 99                              |   |   | 1 in 210  | 1 in 21,000                                    | 1 in 84,000  |
| East Asian  | 99                              |   |   | 1 in 789  | 1 in 79,000                                    | 1 in 320,000   |
| Finnish   | 99                              |   |   | 1 in 109  | 1 in 11,000                                    | 1 in 44,000  |
| Non-Finnish European/Caucasian                                | 99                              |   |   | 1 in 985  | 1 in 98,000                                    | 1 in 390,000   |
| South Asian   | 98                              |   |   | 1 in 1,117  | 1 in 56,000                                    | 1 in 220,000   |
| Other (population not assigned)                               | 99                              |   |   | 1 in 1,529  | 1 in 150,000                                   | 1 in 600,000   |
| Lysosomal acid lipase deficiency                              | LIPA                            |   |   | African/African American  | 99   | 1 in 319   |
|   |                                 | Latino/Admixed American   | 99  | 1 in 104  | 1 in 10,000                                    | 1 in 40,000  |
|   |                                 | Ashkenazi Jewish  | 99  | 1 in 451  | 1 in 45,000                                    | 1 in 180,000   |
|   |                                 | East Asian  | 99  | 1 in 279  | 1 in 28,000                                    | 1 in 110,000   |
|   |                                 | Finnish   | 99  | 1 in 781  | 1 in 78,000                                    | 1 in 310,000   |
|   |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 127  | 1 in 13,000                                    | 1 in 52,000  |
|   |                                 | South Asian   | 99  | 1 in 357  | 1 in 36,000                                    | 1 in 140,000   |
|   |                                 | Other (population not assigned)   | 99  | 1 in 180  | 1 in 18,000                                    | 1 in 72,000  |
|   |                                 | Maple syrup urine disease   | BCKDHA  | African/African American  | 98   | 1 in 495   |
| Latino/Admixed American                                       | 99                              |   |   | 1 in 270  | 1 in 27,000                                    | 1 in 110,000   |
| Ashkenazi Jewish  | 99                              |   |   | 1 in 3,410  | 1 in 340,000                                   | 1 in 1,400,000   |
| East Asian  | 98                              |   |   | 1 in 174  | 1 in 8,700                                     | 1 in 35,000  |
| Finnish   | 98                              |   |   | 1 in 2,239  | 1 in 110,000                                   | 1 in 440,000   |
| Non-Finnish European/Caucasian                                | 99                              |   |   | 1 in 434  | 1 in 43,000                                    | 1 in 170,000   |
| South Asian   | 98                              |   |   | 1 in 542  | 1 in 27,000                                    | 1 in 110,000   |
| Other (population not assigned)                               | 99                              |   |   | 1 in 914  | 1 in 91,000                                    | 1 in 360,000   |
| BCKDHB  | African/African American        |   |   | 99  | 1 in 861                                       | 1 in 86,000  |
|   | Latino/Admixed American         |   | 99  | 1 in 401  | 1 in 40,000                                    | 1 in 160,000   |
|   | Ashkenazi Jewish                |   | 99  | 1 in 85   | 1 in 8,400                                     | 1 in 34,000  |
|   | East Asian                      |   | 99  | 1 in 751  | 1 in 75,000                                    | 1 in 300,000   |
|   | Finnish                         |   | 99  | 1 in 167  | 1 in 17,000                                    | 1 in 68,000  |
|   | Non-Finnish European/Caucasian  |   | 99  | 1 in 451  | 1 in 45,000                                    | 1 in 180,000   |
|   | South Asian                     |   | 99  | 1 in 1,366  | 1 in 140,000                                   | 1 in 560,000   |
|   | Other (population not assigned) |   | 98  | 1 in 473  | 1 in 24,000                                    | 1 in 96,000  |



### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |              |
|--|---------------------------------|---|---|---|---|---|--------------|
| Disorder   | DBT                             | African/African American  | 99  | 1 in 404  | 1 in 40,000                                       | 1 in 160,000  |              |
|  |                                 | Latino/Admixed American   | 98  | 1 in 1,132  | 1 in 57,000                                       | 1 in 230,000  |              |
|  |                                 | Ashkenazi Jewish  | 97  | 1 in 1,640  | 1 in 55,000                                       | 1 in 220,000  |              |
|  |                                 | East Asian  | 98  | 1 in 1,277  | 1 in 64,000                                       | 1 in 260,000  |              |
|  |                                 | Finnish   | 99  | 1 in 1,099  | 1 in 110,000                                      | 1 in 440,000  |              |
|  |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 294  | 1 in 29,000                                       | 1 in 120,000  |              |
|  |                                 | South Asian   | 99  | 1 in 1,090  | 1 in 110,000                                      | 1 in 440,000  |              |
|  |                                 | Other (population not assigned)   | 99  | 1 in 489  | 1 in 49,000                                       | 1 in 200,000  |              |
| Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency                | ACADM                           | African/African American  | 99  | 1 in 155  | 1 in 15,000                                       | 1 in 60,000   |              |
|  |                                 | Latino/Admixed American   | 99  | 1 in 71   | 1 in 7,000  | 1 in 28,000   |              |
|  |                                 | Ashkenazi Jewish  | 99  | 1 in 57   | 1 in 5,600  | 1 in 22,000   |              |
|  |                                 | East Asian  | 98  | 1 in 130  | 1 in 6,500  | 1 in 26,000   |              |
|  |                                 | Finnish   | 99  | 1 in 134  | 1 in 13,000                                       | 1 in 52,000   |              |
|  |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 41   | 1 in 4,000  | 1 in 16,000   |              |
|  |                                 | South Asian   | 99  | 1 in 143  | 1 in 14,000                                       | 1 in 56,000   |              |
|  |                                 | Other (population not assigned)   | 99  | 1 in 45   | 1 in 4,400  | 1 in 18,000   |              |
| Megalencephalic leukoencephalopathy with subcortical<br>cysts type 1 | MLC1                            | African/African American  | 98  | 1 in 1,404  | 1 in 70,000                                       | 1 in 280,000  |              |
|  |                                 | Latino/Admixed American   | 99  | 1 in 5,706  | 1 in 570,000                                      | 1 in 2,300,000  |              |
|  |                                 | Ashkenazi Jewish  | 99  | 1 in 198  | 1 in 20,000                                       | 1 in 80,000   |              |
|  |                                 | East Asian  | 99  | 1 in 107  | 1 in 11,000                                       | 1 in 44,000   |              |
|  |                                 | Finnish   | 97  | 1 in 1,201  | 1 in 40,000                                       | 1 in 160,000  |              |
|  |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 1,234  | 1 in 120,000                                      | 1 in 480,000  |              |
|  |                                 | South Asian   | 99  | 1 in 326  | 1 in 33,000                                       | 1 in 130,000  |              |
|  |                                 | Other (population not assigned)   | 99  | 1 in 703  | 1 in 70,000                                       | 1 in 280,000  |              |
| Metachromatic leukodystrophy   | ARSA                            | African/African American  | 99  | 1 in 336  | 1 in 34,000                                       | 1 in 140,000  |              |
|  |                                 | Latino/Admixed American   | 99  | 1 in 487  | 1 in 49,000                                       | 1 in 200,000  |              |
|  |                                 | Ashkenazi Jewish  | 99  | 1 in 808  | 1 in 81,000                                       | 1 in 320,000  |              |
|  |                                 | East Asian  | 99  | 1 in 152  | 1 in 15,000                                       | 1 in 60,000   |              |
|  |                                 | Finnish   | 99  | 1 in 206  | 1 in 21,000                                       | 1 in 84,000   |              |
|  |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 154  | 1 in 15,000                                       | 1 in 60,000   |              |
|  |                                 | South Asian   | 99  | 1 in 262  | 1 in 26,000                                       | 1 in 100,000  |              |
|  |                                 | Other (population not assigned)   | 99  | 1 in 128  | 1 in 13,000                                       | 1 in 52,000   |              |
|  |                                 | PSAP  | African/African American  | 99  | 1 in 2,030  | 1 in 200,000  | 1 in 800,000 |
|  | Latino/Admixed American         |   | 99  | 1 in 884  | 1 in 88,000                                       | 1 in 350,000  |              |
|  | 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  | 1 in 2,998  | 1 in 300,000                                      | 1 in 1,200,000  |              |
|  | South Asian                     |   | 99  | 1 in 15,385   | 1 in 1,500,000                                    | 1 in 6,000,000  |              |
|  | Other (population not assigned) |   | 99  | N/A   | N/A   | N/A   |              |
|  | Methylmalonic acidemia          |   | MCEE  | African/African American  | 97  | N/A   | N/A          |
|  |                                 | Latino/Admixed American   |   | 99  | 1 in 1,878  | 1 in 190,000  | 1 in 760,000 |
| Ashkenazi Jewish   |                                 | 97  |   | N/A   | N/A   | N/A   |              |
| East Asian   |                                 | 99  |   | 1 in 2,277  | 1 in 230,000                                      | 1 in 920,000  |              |
| Finnish  |                                 | 99  |   | 1 in 11,161   | 1 in 1,100,000                                    | 1 in 4,400,000  |              |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |              |
|---------------------------------|---------------------------------|---|---|---|---|---|--------------|
| Disorder                        |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 873  | 1 in 87,000                                       | 1 in 350,000  |              |
|                                 |                                 | South Asian   | 97  | 1 in 7,646  | 1 in 250,000                                      | 1 in 1,000,000  |              |
|                                 |                                 | Other (population not assigned)   | 99  | 1 in 2,742  | 1 in 270,000                                      | 1 in 1,100,000  |              |
|                                 | MMAA                            | African/African American  | 99  | 1 in 7,611  | 1 in 760,000                                      | 1 in 3,000,000  |              |
|                                 |                                 | Latino/Admixed American   | 99  | 1 in 2,126  | 1 in 210,000                                      | 1 in 840,000  |              |
|                                 |                                 | Ashkenazi Jewish  | 99  | N/A   | N/A   | N/A   |              |
|                                 |                                 | East Asian  | 99  | 1 in 1,792  | 1 in 180,000                                      | 1 in 720,000  |              |
|                                 |                                 | Finnish   | 99  | 1 in 3,715  | 1 in 370,000                                      | 1 in 1,500,000  |              |
|                                 |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 665  | 1 in 66,000                                       | 1 in 260,000  |              |
|                                 |                                 | South Asian   | 99  | 1 in 1,012  | 1 in 100,000                                      | 1 in 400,000  |              |
|                                 |                                 | Other (population not assigned)   | 99  | 1 in 947  | 1 in 95,000                                       | 1 in 380,000  |              |
|                                 |                                 | MMAB  | African/African American  | 99  | 1 in 50   | 1 in 4,900  | 1 in 20,000  |
|                                 | Latino/Admixed American         |   | 99  | 1 in 606  | 1 in 61,000                                       | 1 in 240,000  |              |
|                                 | Ashkenazi Jewish                |   | 99  | N/A   | N/A   | N/A   |              |
|                                 | East Asian                      |   | 99  | N/A   | N/A   | N/A   |              |
|                                 | Finnish                         |   | 99  | 1 in 3,286  | 1 in 330,000                                      | 1 in 1,300,000  |              |
|                                 | Non-Finnish European/Caucasian  |   | 99  | 1 in 801  | 1 in 80,000                                       | 1 in 320,000  |              |
|                                 | South Asian                     |   | 99  | 1 in 954  | 1 in 95,000                                       | 1 in 380,000  |              |
|                                 | Other (population not assigned) |   | 98  | 1 in 317  | 1 in 16,000                                       | 1 in 64,000   |              |
|                                 | MMUT                            |   | African/African American  | 99  | 1 in 182  | 1 in 18,000   | 1 in 72,000  |
|                                 |                                 | Latino/Admixed American   | 99  | 1 in 185  | 1 in 18,000                                       | 1 in 72,000   |              |
|                                 |                                 | Ashkenazi Jewish  | 99  | 1 in 353  | 1 in 35,000                                       | 1 in 140,000  |              |
|                                 |                                 | East Asian  | 99  | 1 in 178  | 1 in 18,000                                       | 1 in 72,000   |              |
|                                 |                                 | Finnish   | 99  | 1 in 1,010  | 1 in 100,000                                      | 1 in 400,000  |              |
|                                 |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 409  | 1 in 41,000                                       | 1 in 160,000  |              |
|                                 |                                 | South Asian   | 99  | 1 in 82   | 1 in 8,100  | 1 in 32,000   |              |
|                                 |                                 | Other (population not assigned)   | 99  | 1 in 343  | 1 in 34,000                                       | 1 in 140,000  |              |
|                                 |                                 | Methylmalonic acidemia with homocystinuria  | ABCD4   | African/African American  | 98  | 1 in 859  | 1 in 43,000  |
|                                 | Latino/Admixed American         |   |   | 99  | 1 in 534  | 1 in 53,000   | 1 in 210,000 |
|                                 | Ashkenazi Jewish                |   |   | 98  | N/A   | N/A   | N/A          |
| East Asian                      | 99                              |   |   | 1 in 328  | 1 in 33,000                                       | 1 in 130,000  |              |
| Finnish                         | 99                              |   |   | 1 in 901  | 1 in 90,000                                       | 1 in 360,000  |              |
| Non-Finnish European/Caucasian  | 99                              |   |   | 1 in 496  | 1 in 50,000                                       | 1 in 200,000  |              |
| South Asian                     | 99                              |   |   | 1 in 313  | 1 in 31,000                                       | 1 in 120,000  |              |
| Other (population not assigned) | 99                              |   |   | 1 in 304  | 1 in 30,000                                       | 1 in 120,000  |              |
| HCFC1                           | African/African American        |   |   | 97  | N/A   | N/A   | N/A          |
|                                 | Latino/Admixed American         |   | 97  | N/A   | N/A   | N/A   |              |
|                                 | Ashkenazi Jewish                |   | 97  | N/A   | N/A   | N/A   |              |
|                                 | East Asian                      |   | 97  | N/A   | N/A   | N/A   |              |
|                                 | Finnish                         |   | 97  | 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) |   | 97  | N/A   | N/A   | N/A   |              |
|                                 | LMBRD1                          |   | African/African American  | 99  | 1 in 363  | 1 in 36,000   | 1 in 140,000 |
| Latino/Admixed American         |                                 |   | 98  | 1 in 1,035  | 1 in 52,000                                       | 1 in 210,000  |              |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |              |
|---|------------------------------------|---|---|---|---|---|--------------|
|   |                                    | Ashkenazi Jewish  | 97  | 1 in 5,035  | 1 in 170,000                                      | 1 in 680,000  |              |
|   |                                    | East Asian  | 98  | 1 in 1,835  | 1 in 92,000                                       | 1 in 370,000  |              |
|   |                                    | Finnish   | 97  | 1 in 789  | 1 in 26,000                                       | 1 in 100,000  |              |
|   |                                    | Non-Finnish European/Caucasian  | 97  | 1 in 414  | 1 in 14,000                                       | 1 in 56,000   |              |
|   |                                    | South Asian   | 99  | 1 in 2,536  | 1 in 250,000                                      | 1 in 1,000,000  |              |
|   |                                    | Other (population not assigned)   | 97  | 1 in 715  | 1 in 24,000                                       | 1 in 96,000   |              |
|   | MMACHC                             | African/African American  | 98  | 1 in 284  | 1 in 14,000                                       | 1 in 56,000   |              |
|   |                                    | Latino/Admixed American   | 99  | 1 in 38   | 1 in 3,700  | 1 in 15,000   |              |
|   |                                    | Ashkenazi Jewish  | 97  | 1 in 206  | 1 in 6,800  | 1 in 27,000   |              |
|   |                                    | East Asian  | 98  | 1 in 229  | 1 in 11,000                                       | 1 in 44,000   |              |
|   |                                    | Finnish   | 99  | 1 in 10,780   | 1 in 1,100,000                                    | 1 in 4,400,000  |              |
|   |                                    | Non-Finnish European/Caucasian  | 98  | 1 in 151  | 1 in 7,500  | 1 in 30,000   |              |
|   | MMADHC                             | South Asian   | 99  | 1 in 202  | 1 in 20,000                                       | 1 in 80,000   |              |
|   |                                    | Other (population not assigned)   | 98  | 1 in 127  | 1 in 6,300  | 1 in 25,000   |              |
|   |                                    | African/African American  | 99  | 1 in 3,940  | 1 in 390,000                                      | 1 in 1,600,000  |              |
|   |                                    | Latino/Admixed American   | 98  | 1 in 5,706  | 1 in 290,000                                      | 1 in 1,200,000  |              |
|   |                                    | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |              |
|   |                                    | East Asian  | 98  | 1 in 1,835  | 1 in 92,000                                       | 1 in 370,000  |              |
|   | Mitochondrial complex I deficiency | ACAD9   | Finnish   | 99  | 1 in 809  | 1 in 81,000   | 1 in 320,000 |
|   |                                    |   | Non-Finnish European/Caucasian  | 99  | 1 in 1,515  | 1 in 150,000  | 1 in 600,000 |
|   |                                    |   | South Asian   | 97  | 1 in 282  | 1 in 9,400  | 1 in 38,000  |
|   |                                    |   | Other (population not assigned)   | 99  | 1 in 983  | 1 in 98,000   | 1 in 390,000 |
|   |                                    |   | African/African American  | 97  | 1 in 67   | 1 in 2,200  | 1 in 8,800   |
|   |                                    |   | Latino/Admixed American   | 97  | 1 in 118  | 1 in 3,900  | 1 in 16,000  |
| Ashkenazi Jewish                                    |                                    |   | 98  | 1 in 356  | 1 in 18,000                                       | 1 in 72,000   |              |
| East Asian  |                                    |   | 97  | 1 in 1,382  | 1 in 46,000                                       | 1 in 180,000  |              |
| Finnish   |                                    |   | 97  | 1 in 306  | 1 in 10,000                                       | 1 in 40,000   |              |
| Non-Finnish European/Caucasian                      |                                    |   | 97  | 1 in 67   | 1 in 2,200  | 1 in 8,800  |              |
| Mitochondrial complex V deficiency                  | TMEM70                             | South Asian   | 97  | 1 in 110  | 1 in 3,600  | 1 in 14,000   |              |
|   |                                    | Other (population not assigned)   | 97  | 1 in 85   | 1 in 2,800  | 1 in 11,000   |              |
|   |                                    | African/African American  | 98  | 1 in 1,912  | 1 in 96,000                                       | 1 in 380,000  |              |
|   |                                    | Latino/Admixed American   | 99  | 1 in 1,408  | 1 in 140,000                                      | 1 in 560,000  |              |
|   |                                    | Ashkenazi Jewish  | 99  | 1 in 4,925  | 1 in 490,000                                      | 1 in 2,000,000  |              |
|   |                                    | East Asian  | 99  | 1 in 4,571  | 1 in 460,000                                      | 1 in 1,800,000  |              |
|   |                                    | Finnish   | 97  | 1 in 3,678  | 1 in 120,000                                      | 1 in 480,000  |              |
|   |                                    | Non-Finnish European/Caucasian  | 98  | 1 in 632  | 1 in 32,000                                       | 1 in 130,000  |              |
|   |                                    | South Asian   | 99  | 1 in 1,922  | 1 in 190,000                                      | 1 in 760,000  |              |
|   |                                    | Other (population not assigned)   | 99  | 1 in 1,369  | 1 in 140,000                                      | 1 in 560,000  |              |
| Mitochondrial DNA depletion syndrome, MVP17-related | MPV17                              | African/African American  | 99  | 1 in 237  | 1 in 24,000                                       | 1 in 96,000   |              |
|   |                                    | Latino/Admixed American   | 98  | 1 in 3,588  | 1 in 180,000                                      | 1 in 720,000  |              |
|   |                                    | Ashkenazi Jewish  | 99  | 1 in 1,656  | 1 in 170,000                                      | 1 in 680,000  |              |
|   |                                    | East Asian  | 97  | 1 in 126  | 1 in 4,200  | 1 in 17,000   |              |
|   |                                    | Finnish   | 97  | 1 in 83   | 1 in 2,700  | 1 in 11,000   |              |
|   |                                    | Non-Finnish European/Caucasian  | 99  | 1 in 636  | 1 in 64,000                                       | 1 in 260,000  |              |
| South Asian   | 99                                 | 1 in 542  | 1 in 54,000   | 1 in 220,000  |   |   |              |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|---|--------|---|---|---|---|---|
|   |        | Other (population not assigned)   | 99  | 1 in 697  | 1 in 70,000                                       | 1 in 280,000  |
| Mitochondrial DNA depletion syndrome, TK2-related                 | TK2    | African/African American  | 99  | 1 in 707  | 1 in 71,000                                       | 1 in 280,000  |
|   |        | Latino/Admixed American   | 99  | 1 in 431  | 1 in 43,000                                       | 1 in 170,000  |
|   |        | Ashkenazi Jewish  | 99  | 1 in 704  | 1 in 70,000                                       | 1 in 280,000  |
|   |        | East Asian  | 99  | 1 in 8,621  | 1 in 860,000                                      | 1 in 3,400,000  |
|   |        | Finnish   | 99  | 1 in 285  | 1 in 28,000                                       | 1 in 110,000  |
|   |        | Non-Finnish European/Caucasian  | 99  | 1 in 730  | 1 in 73,000                                       | 1 in 290,000  |
|   |        | South Asian   | 99  | 1 in 48   | 1 in 4,700  | 1 in 19,000   |
|   |        | Other (population not assigned)   | 99  | 1 in 344  | 1 in 34,000                                       | 1 in 140,000  |
| Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia | PUS1   | African/African American  | 99  | 1 in 1,297  | 1 in 130,000                                      | 1 in 520,000  |
|   |        | Latino/Admixed American   | 98  | 1 in 1,468  | 1 in 73,000                                       | 1 in 290,000  |
|   |        | Ashkenazi Jewish  | 97  | 1 in 1,129  | 1 in 38,000                                       | 1 in 150,000  |
|   |        | East Asian  | 99  | 1 in 8,209  | 1 in 820,000                                      | 1 in 3,300,000  |
|   |        | Finnish   | 98  | 1 in 1,401  | 1 in 70,000                                       | 1 in 280,000  |
|   |        | Non-Finnish European/Caucasian  | 98  | 1 in 1,551  | 1 in 78,000                                       | 1 in 310,000  |
|   |        | South Asian   | 99  | 1 in 178  | 1 in 18,000                                       | 1 in 72,000   |
|   |        | Other (population not assigned)   | 99  | 1 in 3,063  | 1 in 310,000                                      | 1 in 1,200,000  |
| Mucopolipidosis type II and III                                   | GNPTAB | African/African American  | 97  | 1 in 114  | 1 in 3,800  | 1 in 15,000   |
|   |        | Latino/Admixed American   | 97  | 1 in 123  | 1 in 4,100  | 1 in 16,000   |
|   |        | Ashkenazi Jewish  | 97  | 1 in 281  | 1 in 9,300  | 1 in 37,000   |
|   |        | East Asian  | 98  | 1 in 265  | 1 in 13,000                                       | 1 in 52,000   |
|   |        | Finnish   | 97  | 1 in 99   | 1 in 3,300  | 1 in 13,000   |
|   |        | Non-Finnish European/Caucasian  | 97  | 1 in 163  | 1 in 5,400  | 1 in 22,000   |
|   |        | South Asian   | 97  | 1 in 114  | 1 in 3,800  | 1 in 15,000   |
|   |        | Other (population not assigned)   | 97  | 1 in 139  | 1 in 4,600  | 1 in 18,000   |
| Mucopolipidosis type IV   | MCOLN1 | African/African American  | 99  | 1 in 1,566  | 1 in 160,000                                      | 1 in 640,000  |
|   |        | Latino/Admixed American   | 99  | 1 in 480  | 1 in 48,000                                       | 1 in 190,000  |
|   |        | Ashkenazi Jewish  | 99  | 1 in 112  | 1 in 11,000                                       | 1 in 44,000   |
|   |        | East Asian  | 99  | 1 in 2,278  | 1 in 230,000                                      | 1 in 920,000  |
|   |        | Finnish   | 97  | 1 in 5,411  | 1 in 180,000                                      | 1 in 720,000  |
|   |        | Non-Finnish European/Caucasian  | 99  | 1 in 1,059  | 1 in 110,000                                      | 1 in 440,000  |
|   |        | South Asian   | 99  | 1 in 366  | 1 in 37,000                                       | 1 in 150,000  |
|   |        | Other (population not assigned)   | 99  | 1 in 612  | 1 in 61,000                                       | 1 in 240,000  |
| Mucopolysaccharidosis type I                                      | IDUA   | African/African American  | 99  | 1 in 515  | 1 in 51,000                                       | 1 in 200,000  |
|   |        | Latino/Admixed American   | 98  | 1 in 119  | 1 in 5,900  | 1 in 24,000   |
|   |        | Ashkenazi Jewish  | 97  | 1 in 224  | 1 in 7,400  | 1 in 30,000   |
|   |        | East Asian  | 99  | 1 in 55   | 1 in 5,400  | 1 in 22,000   |
|   |        | Finnish   | 98  | 1 in 88   | 1 in 4,400  | 1 in 18,000   |
|   |        | Non-Finnish European/Caucasian  | 99  | 1 in 111  | 1 in 11,000                                       | 1 in 44,000   |
|   |        | South Asian   | 98  | 1 in 214  | 1 in 11,000                                       | 1 in 44,000   |
|   |        | Other (population not assigned)   | 99  | 1 in 165  | 1 in 16,000                                       | 1 in 64,000   |
| Mucopolysaccharidosis type II                                     | IDS    | African/African American  | 99  | N/A   | N/A   | N/A   |
|   |        | Latino/Admixed American   | 97  | N/A   | N/A   | N/A   |
|   |        | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|   |        | East Asian  | 97  | N/A   | N/A   | N/A   |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |              |              |
|---------------------------------|---------------------------------|---|---|---|---|---|--------------|--------------|
|                                 |                                 | Finnish   | 97  | 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)   | 97  | N/A   | N/A   | N/A   |              |              |
| Mucopolysaccharidosis type III  | GNS                             | African/African American  | 98  | 1 in 3,825  | 1 in 190,000                                      | 1 in 760,000  |              |              |
|                                 |                                 | Latino/Admixed American   | 99  | 1 in 793  | 1 in 79,000                                       | 1 in 320,000  |              |              |
|                                 |                                 | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |              |              |
|                                 |                                 | East Asian  | 99  | 1 in 4,588  | 1 in 460,000                                      | 1 in 1,800,000  |              |              |
|                                 |                                 | Finnish   | 97  | 1 in 10,825   | 1 in 360,000                                      | 1 in 1,400,000  |              |              |
|                                 |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 1,961  | 1 in 200,000                                      | 1 in 800,000  |              |              |
|                                 |                                 | South Asian   | 98  | 1 in 4,145  | 1 in 210,000                                      | 1 in 840,000  |              |              |
|                                 |                                 | Other (population not assigned)   | 99  | 1 in 3,066  | 1 in 310,000                                      | 1 in 1,200,000  |              |              |
|                                 | HGSNAT                          | African/African American  | 99  | 1 in 696  | 1 in 70,000                                       | 1 in 280,000  |              |              |
|                                 |                                 | Latino/Admixed American   | 99  | 1 in 730  | 1 in 73,000                                       | 1 in 290,000  |              |              |
|                                 |                                 | Ashkenazi Jewish  | 97  | 1 in 1,480  | 1 in 49,000                                       | 1 in 200,000  |              |              |
|                                 |                                 | East Asian  | 99  | 1 in 858  | 1 in 86,000                                       | 1 in 340,000  |              |              |
|                                 |                                 | Finnish   | 98  | 1 in 775  | 1 in 39,000                                       | 1 in 160,000  |              |              |
|                                 |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 770  | 1 in 77,000                                       | 1 in 310,000  |              |              |
|                                 |                                 | South Asian   | 99  | 1 in 976  | 1 in 98,000                                       | 1 in 390,000  |              |              |
|                                 |                                 | Other (population not assigned)   | 99  | 1 in 924  | 1 in 92,000                                       | 1 in 370,000  |              |              |
|                                 |                                 | NAGLU   | African/African American  | 99  | 1 in 316  | 1 in 32,000   | 1 in 130,000 |              |
|                                 |                                 |   | Latino/Admixed American   | 99  | 1 in 653  | 1 in 65,000   | 1 in 260,000 |              |
|                                 |                                 |   | Ashkenazi Jewish  | 97  | 1 in 137  | 1 in 4,500  | 1 in 18,000  |              |
|                                 |                                 |   | East Asian  | 98  | 1 in 557  | 1 in 28,000   | 1 in 110,000 |              |
|                                 | Finnish                         |   | 97  | 1 in 10,822   | 1 in 360,000                                      | 1 in 1,400,000  |              |              |
|                                 | Non-Finnish European/Caucasian  |   | 99  | 1 in 361  | 1 in 36,000                                       | 1 in 140,000  |              |              |
|                                 | South Asian                     |   | 99  | 1 in 529  | 1 in 53,000                                       | 1 in 210,000  |              |              |
|                                 | Other (population not assigned) |   | 99  | 1 in 724  | 1 in 72,000                                       | 1 in 290,000  |              |              |
|                                 | SGSH                            | African/African American  | 99  | 1 in 598  | 1 in 60,000                                       | 1 in 240,000  |              |              |
|                                 |                                 | Latino/Admixed American   | 99  | 1 in 410  | 1 in 41,000                                       | 1 in 160,000  |              |              |
|                                 |                                 | Ashkenazi Jewish  | 99  | 1 in 963  | 1 in 96,000                                       | 1 in 380,000  |              |              |
|                                 |                                 | East Asian  | 99  | 1 in 582  | 1 in 58,000                                       | 1 in 230,000  |              |              |
|                                 |                                 | Finnish   | 99  | 1 in 621  | 1 in 62,000                                       | 1 in 250,000  |              |              |
|                                 |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 227  | 1 in 23,000                                       | 1 in 92,000   |              |              |
|                                 |                                 | South Asian   | 99  | 1 in 311  | 1 in 31,000                                       | 1 in 120,000  |              |              |
|                                 |                                 | Other (population not assigned)   | 99  | 1 in 675  | 1 in 67,000                                       | 1 in 270,000  |              |              |
|                                 |                                 | Mucopolysaccharidosis type IVA  | GALNS   | African/African American  | 99  | 1 in 548  | 1 in 55,000  | 1 in 220,000 |
|                                 |                                 |   |   | Latino/Admixed American   | 99  | 1 in 317  | 1 in 32,000  | 1 in 130,000 |
|                                 |                                 |   |   | Ashkenazi Jewish  | 99  | 1 in 824  | 1 in 82,000  | 1 in 330,000 |
|                                 |                                 |   |   | East Asian  | 99  | 1 in 263  | 1 in 26,000  | 1 in 100,000 |
| Finnish                         | 98                              |   |   | 1 in 277  | 1 in 14,000                                       | 1 in 56,000   |              |              |
| Non-Finnish European/Caucasian  | 99                              |   |   | 1 in 311  | 1 in 31,000                                       | 1 in 120,000  |              |              |
| South Asian                     | 98                              |   |   | 1 in 317  | 1 in 16,000                                       | 1 in 64,000   |              |              |
| Other (population not assigned) | 99                              |   |   | 1 in 237  | 1 in 24,000                                       | 1 in 96,000   |              |              |
| Mucopolysaccharidosis type VI   | ARSB                            | African/African American  | 97  | 1 in 268  | 1 in 8,900  | 1 in 36,000   |              |              |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|-----------------------------------|--------|---|---|---|---|---|
|                                   |        | Latino/Admixed American   | 99  | 1 in 4,258  | 1 in 430,000                                      | 1 in 1,700,000  |
|                                   |        | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|                                   |        | East Asian  | 99  | 1 in 1,484  | 1 in 150,000                                      | 1 in 600,000  |
|                                   |        | Finnish   | 98  | 1 in 1,356  | 1 in 68,000                                       | 1 in 270,000  |
|                                   |        | Non-Finnish European/Caucasian  | 99  | 1 in 378  | 1 in 38,000                                       | 1 in 150,000  |
|                                   |        | South Asian   | 98  | 1 in 902  | 1 in 45,000                                       | 1 in 180,000  |
|                                   |        | Other (population not assigned)   | 97  | 1 in 1,533  | 1 in 51,000                                       | 1 in 200,000  |
| Mucopolysaccharidosis type VII    | GUSB   | African/African American  | 99  | 1 in 488  | 1 in 49,000                                       | 1 in 200,000  |
|                                   |        | Latino/Admixed American   | 99  | 1 in 685  | 1 in 68,000                                       | 1 in 270,000  |
|                                   |        | Ashkenazi Jewish  | 99  | 1 in 741  | 1 in 74,000                                       | 1 in 300,000  |
|                                   |        | East Asian  | 99  | 1 in 1,760  | 1 in 180,000                                      | 1 in 720,000  |
|                                   |        | Finnish   | 99  | 1 in 1,177  | 1 in 120,000                                      | 1 in 480,000  |
|                                   |        | Non-Finnish European/Caucasian  | 99  | 1 in 552  | 1 in 55,000                                       | 1 in 220,000  |
|                                   |        | South Asian   | 99  | 1 in 914  | 1 in 91,000                                       | 1 in 360,000  |
|                                   |        | Other (population not assigned)   | 99  | 1 in 661  | 1 in 66,000                                       | 1 in 260,000  |
|                                   |        | Mucopolysaccharidosis type IX   | HYAL1   | African/African American  | 97  | 1 in 2,692  |
| Latino/Admixed American           | 98     |   |   | 1 in 1,708  | 1 in 85,000                                       | 1 in 340,000  |
| Ashkenazi Jewish                  | 97     |   |   | N/A   | N/A   | N/A   |
| East Asian                        | 98     |   |   | 1 in 2,296  | 1 in 110,000                                      | 1 in 440,000  |
| Finnish                           | 97     |   |   | N/A   | N/A   | N/A   |
| Non-Finnish European/Caucasian    | 98     |   |   | 1 in 2,811  | 1 in 140,000                                      | 1 in 560,000  |
| South Asian                       | 98     |   |   | 1 in 1,691  | 1 in 85,000                                       | 1 in 340,000  |
| Other (population not assigned)   | 97     |   |   | N/A   | N/A   | N/A   |
| Multiple pterygium syndrome       | CHRNA3 |   |   | African/African American  | 97  | 1 in 371  |
|                                   |        | Latino/Admixed American   | 97  | 1 in 546  | 1 in 18,000                                       | 1 in 72,000   |
|                                   |        | Ashkenazi Jewish  | 99  | 1 in 2,517  | 1 in 250,000                                      | 1 in 1,000,000  |
|                                   |        | East Asian  | 98  | 1 in 529  | 1 in 26,000                                       | 1 in 100,000  |
|                                   |        | Finnish   | 99  | 1 in 637  | 1 in 64,000                                       | 1 in 260,000  |
|                                   |        | Non-Finnish European/Caucasian  | 97  | 1 in 400  | 1 in 13,000                                       | 1 in 52,000   |
|                                   |        | South Asian   | 98  | 1 in 384  | 1 in 19,000                                       | 1 in 76,000   |
|                                   |        | Other (population not assigned)   | 97  | 1 in 705  | 1 in 23,000                                       | 1 in 92,000   |
|                                   |        | Multiple sulphatase deficiency  | SUMF1   | African/African American  | 98  | 1 in 148  |
| Latino/Admixed American           | 98     |   |   | 1 in 255  | 1 in 13,000                                       | 1 in 52,000   |
| Ashkenazi Jewish                  | 97     |   |   | 1 in 47   | 1 in 1,500  | 1 in 6,000  |
| East Asian                        | 99     |   |   | 1 in 97   | 1 in 9,600  | 1 in 38,000   |
| Finnish                           | 97     |   |   | 1 in 1,864  | 1 in 62,000                                       | 1 in 250,000  |
| Non-Finnish European/Caucasian    | 98     |   |   | 1 in 269  | 1 in 13,000                                       | 1 in 52,000   |
| South Asian                       | 97     |   |   | 1 in 38   | 1 in 1,200  | 1 in 4,800  |
| Other (population not assigned)   | 97     |   |   | 1 in 120  | 1 in 4,000  | 1 in 16,000   |
| Muscular dystrophy, LAMA2-related | LAMA2  |   |   | African/African American  | 99  | 1 in 114  |
|                                   |        | Latino/Admixed American   | 98  | 1 in 159  | 1 in 7,900  | 1 in 32,000   |
|                                   |        | Ashkenazi Jewish  | 99  | 1 in 312  | 1 in 31,000                                       | 1 in 120,000  |
|                                   |        | East Asian  | 99  | 1 in 122  | 1 in 12,000                                       | 1 in 48,000   |
|                                   |        | Finnish   | 98  | 1 in 915  | 1 in 46,000                                       | 1 in 180,000  |
|                                   |        | Non-Finnish European/Caucasian  | 98  | 1 in 273  | 1 in 14,000                                       | 1 in 56,000   |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

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N/A= Not Available

| Disorder                        | Gene  | Ethnicity<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |             |              |
|---------------------------------|---|---|---|---|---|---|-------------|--------------|
|                                 |   | South Asian   | 98  | 1 in 306  | 1 in 15,000                                       | 1 in 60,000   |             |              |
|                                 |   | Other (population not assigned)   | 99  | 1 in 299  | 1 in 30,000                                       | 1 in 120,000  |             |              |
| Myotubular myopathy             | MTM1  | African/African American  | 97  | N/A   | N/A   | N/A   |             |              |
|                                 |   | Latino/Admixed American   | 97  | N/A   | N/A   | N/A   |             |              |
|                                 |   | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |             |              |
|                                 |   | East Asian  | 97  | N/A   | N/A   | N/A   |             |              |
|                                 |   | Finnish   | 97  | 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)   | 97  | N/A   | N/A   | N/A   |             |              |
|                                 |   | Nemaline myopathy   | NEB   | African/African American  | 93  | 1 in 190  | 1 in 2,700  | 1 in 11,000  |
| Latino/Admixed American         | 98  |   |   | 1 in 149  | 1 in 7,400  | 1 in 30,000   |             |              |
| Ashkenazi Jewish                | 99  |   |   | 1 in 226  | 1 in 23,000                                       | 1 in 92,000   |             |              |
| East Asian                      | 97  |   |   | 1 in 87   | 1 in 2,900  | 1 in 12,000   |             |              |
| Finnish                         | 98  |   |   | 1 in 181  | 1 in 9,000  | 1 in 36,000   |             |              |
| Non-Finnish European/Caucasian  | 92  |   |   | 1 in 146  | 1 in 1,800  | 1 in 7,200  |             |              |
| South Asian                     | 98  |   |   | 1 in 159  | 1 in 7,900  | 1 in 32,000   |             |              |
| Other (population not assigned) | 99  |   |   | 1 in 94   | 1 in 9,300  | 1 in 37,000   |             |              |
| Nephrogenic diabetes insipidus  | AVPR2   |   |   | African/African American  | 98  | N/A   | N/A         | N/A          |
|                                 |   | Latino/Admixed American   | 98  | N/A   | N/A   | N/A   |             |              |
|                                 |   | Ashkenazi Jewish  | 98  | N/A   | N/A   | N/A   |             |              |
|                                 |   | East Asian  | 99  | N/A   | N/A   | N/A   |             |              |
|                                 |   | Finnish   | 98  | N/A   | N/A   | N/A   |             |              |
|                                 |   | Non-Finnish European/Caucasian  | 99  | N/A   | N/A   | N/A   |             |              |
|                                 |   | South Asian   | 98  | N/A   | N/A   | N/A   |             |              |
|                                 |   | Other (population not assigned)   | 99  | N/A   | N/A   | N/A   |             |              |
|                                 |   | Nephrotic syndrome  | NPHS1   | African/African American  | 99  | 1 in 42   | 1 in 4,100  | 1 in 16,000  |
| Latino/Admixed American         | 98  |   |   | 1 in 91   | 1 in 4,500  | 1 in 18,000   |             |              |
| Ashkenazi Jewish                | 99  |   |   | 1 in 419  | 1 in 42,000                                       | 1 in 170,000  |             |              |
| East Asian                      | 99  |   |   | 1 in 121  | 1 in 12,000                                       | 1 in 48,000   |             |              |
| Finnish                         | 98  |   |   | 1 in 27   | 1 in 1,300  | 1 in 5,200  |             |              |
| Non-Finnish European/Caucasian  | 99  |   |   | 1 in 67   | 1 in 6,600  | 1 in 26,000   |             |              |
| South Asian                     | 99  |   |   | 1 in 123  | 1 in 12,000                                       | 1 in 48,000   |             |              |
| Other (population not assigned) | 99  |   |   | 1 in 63   | 1 in 6,200  | 1 in 25,000   |             |              |
| NPHS2                           | African/African American                                |   |   | 98  | 1 in 370  | 1 in 18,000   | 1 in 72,000 |              |
|                                 | Latino/Admixed American                                 |   | 99  | 1 in 339  | 1 in 34,000                                       | 1 in 140,000  |             |              |
|                                 | Ashkenazi Jewish  |   | 98  | N/A   | N/A   | N/A   |             |              |
|                                 | East Asian  |   | 99  | 1 in 710  | 1 in 71,000                                       | 1 in 280,000  |             |              |
|                                 | Finnish   |   | 98  | 1 in 1,441  | 1 in 72,000                                       | 1 in 290,000  |             |              |
|                                 | Non-Finnish European/Caucasian                          |   | 99  | 1 in 187  | 1 in 19,000                                       | 1 in 76,000   |             |              |
|                                 | South Asian   |   | 98  | 1 in 628  | 1 in 31,000                                       | 1 in 120,000  |             |              |
|                                 | Other (population not assigned)                         |   | 99  | 1 in 201  | 1 in 20,000                                       | 1 in 80,000   |             |              |
|                                 | Neurodegeneration with brain iron accumulation disorder |   | ATP13A2   | African/African American  | 99  | 1 in 92   | 1 in 9,100  | 1 in 36,000  |
|                                 |   |   |   | Latino/Admixed American   | 98  | 1 in 751  | 1 in 38,000 | 1 in 150,000 |
| Ashkenazi Jewish                |   |   |   | 97  | 1 in 181  | 1 in 6,000  | 1 in 24,000 |              |

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

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N/A= Not Available

| Disorder | Gene                            | Ethnicity<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2) | Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2) | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |                |
|----------|---------------------------------|---|---|---|--|--|----------------|
|          |                                 | East Asian  | 99  | 1 in 449  | 1 in 45,000                                    | 1 in 180,000   |                |
|          |                                 | Finnish   | 99  | 1 in 725  | 1 in 72,000                                    | 1 in 290,000   |                |
|          |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 221  | 1 in 22,000                                    | 1 in 88,000  |                |
|          |                                 | South Asian   | 98  | 1 in 915  | 1 in 46,000                                    | 1 in 180,000   |                |
|          |                                 | Other (population not assigned)   | 99  | 1 in 263  | 1 in 26,000                                    | 1 in 100,000   |                |
|          | C19orf12                        | African/African American  | 99  | 1 in 702  | 1 in 70,000                                    | 1 in 280,000   |                |
|          |                                 | Latino/Admixed American   | 99  | 1 in 309  | 1 in 31,000                                    | 1 in 120,000   |                |
|          |                                 | Ashkenazi Jewish  | 99  | 1 in 187  | 1 in 19,000                                    | 1 in 76,000  |                |
|          |                                 | East Asian  | 99  | 1 in 4,597  | 1 in 460,000                                   | 1 in 1,800,000   |                |
|          |                                 | Finnish   | 98  | 1 in 3,680  | 1 in 180,000                                   | 1 in 720,000   |                |
|          |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 201  | 1 in 20,000                                    | 1 in 80,000  |                |
|          |                                 | South Asian   | 99  | 1 in 49   | 1 in 4,800                                     | 1 in 19,000  |                |
|          |                                 | Other (population not assigned)   | 99  | 1 in 185  | 1 in 18,000                                    | 1 in 72,000  |                |
|          |                                 | COASY   | African/African American  | 99  | 1 in 179                                       | 1 in 18,000  | 1 in 72,000    |
|          |                                 |   | Latino/Admixed American   | 98  | 1 in 906                                       | 1 in 45,000  | 1 in 180,000   |
|          | Ashkenazi Jewish                |   | 99  | 1 in 709  | 1 in 71,000                                    | 1 in 280,000   |                |
|          | East Asian                      |   | 98  | 1 in 819  | 1 in 41,000                                    | 1 in 160,000   |                |
|          | Finnish                         |   | 97  | 1 in 1,846  | 1 in 62,000                                    | 1 in 250,000   |                |
|          | Non-Finnish European/Caucasian  |   | 97  | 1 in 383  | 1 in 13,000                                    | 1 in 52,000  |                |
|          | South Asian                     |   | 99  | 1 in 437  | 1 in 44,000                                    | 1 in 180,000   |                |
|          | Other (population not assigned) |   | 97  | 1 in 234  | 1 in 7,800                                     | 1 in 31,000  |                |
|          | CP                              | African/African American  | 99  | 1 in 442  | 1 in 44,000                                    | 1 in 180,000   |                |
|          |                                 | Latino/Admixed American   | 97  | 1 in 248  | 1 in 8,200                                     | 1 in 33,000  |                |
|          |                                 | Ashkenazi Jewish  | 98  | 1 in 642  | 1 in 32,000                                    | 1 in 130,000   |                |
|          |                                 | East Asian  | 97  | 1 in 220  | 1 in 7,300                                     | 1 in 29,000  |                |
|          |                                 | Finnish   | 99  | 1 in 597  | 1 in 60,000                                    | 1 in 240,000   |                |
|          |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 497  | 1 in 50,000                                    | 1 in 200,000   |                |
|          |                                 | South Asian   | 99  | 1 in 266  | 1 in 27,000                                    | 1 in 110,000   |                |
|          |                                 | Other (population not assigned)   | 98  | 1 in 1,358  | 1 in 68,000                                    | 1 in 270,000   |                |
|          |                                 | DCAF17  | African/African American  | 97  | 1 in 8,126                                     | 1 in 270,000   | 1 in 1,100,000 |
|          |                                 |   | Latino/Admixed American   | 99  | 1 in 2,564                                     | 1 in 260,000   | 1 in 1,000,000 |
|          | Ashkenazi Jewish                |   | 99  | 1 in 199  | 1 in 20,000                                    | 1 in 80,000  |                |
|          | East Asian                      |   | 99  | 1 in 1,757  | 1 in 180,000                                   | 1 in 720,000   |                |
|          | Finnish                         |   | 99  | 1 in 1,800  | 1 in 180,000                                   | 1 in 720,000   |                |
|          | Non-Finnish European/Caucasian  |   | 99  | 1 in 1,053  | 1 in 110,000                                   | 1 in 440,000   |                |
|          | South Asian                     |   | 99  | 1 in 2,479  | 1 in 250,000                                   | 1 in 1,000,000   |                |
|          | Other (population not assigned) |   | 99  | 1 in 538  | 1 in 54,000                                    | 1 in 220,000   |                |
|          | FA2H                            |   | African/African American  | 97  | 1 in 455                                       | 1 in 15,000  | 1 in 60,000    |
|          |                                 | Latino/Admixed American   | 98  | 1 in 1,143  | 1 in 57,000                                    | 1 in 230,000   |                |
|          |                                 | Ashkenazi Jewish  | 97  | 1 in 3,630  | 1 in 120,000                                   | 1 in 480,000   |                |
|          |                                 | East Asian  | 98  | 1 in 4,124  | 1 in 210,000                                   | 1 in 840,000   |                |
|          |                                 | Finnish   | 97  | N/A   | N/A  | N/A  |                |
|          |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 1,669  | 1 in 170,000                                   | 1 in 680,000   |                |
|          |                                 | South Asian   | 98  | 1 in 2,423  | 1 in 120,000                                   | 1 in 480,000   |                |
|          |                                 | Other (population not assigned)   | 99  | 1 in 1,448  | 1 in 140,000                                   | 1 in 560,000   |                |



### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|--------------------------------|--------|---|---|---|---|---|
| Disorder                       | PLA2G6 | African/African American  | 99  | 1 in 214  | 1 in 21,000                                       | 1 in 84,000   |
|                                |        | Latino/Admixed American   | 99  | 1 in 530  | 1 in 53,000                                       | 1 in 210,000  |
|                                |        | Ashkenazi Jewish  | 99  | 1 in 254  | 1 in 25,000                                       | 1 in 100,000  |
|                                |        | East Asian  | 99  | 1 in 251  | 1 in 25,000                                       | 1 in 100,000  |
|                                |        | Finnish   | 99  | 1 in 2,725  | 1 in 270,000                                      | 1 in 1,100,000  |
|                                |        | Non-Finnish European/Caucasian  | 99  | 1 in 343  | 1 in 34,000                                       | 1 in 140,000  |
|                                |        | South Asian   | 99  | 1 in 60   | 1 in 5,900  | 1 in 24,000   |
|                                |        | Other (population not assigned)   | 99  | 1 in 242  | 1 in 24,000                                       | 1 in 96,000   |
| Neuronal ceroid-lipofuscinosis | CLN3   | African/African American  | 99  | 1 in 540  | 1 in 54,000                                       | 1 in 220,000  |
|                                |        | Latino/Admixed American   | 99  | 1 in 242  | 1 in 24,000                                       | 1 in 96,000   |
|                                |        | Ashkenazi Jewish  | 99  | 1 in 202  | 1 in 20,000                                       | 1 in 80,000   |
|                                |        | East Asian  | 99  | 1 in 373  | 1 in 37,000                                       | 1 in 150,000  |
|                                |        | Finnish   | 98  | 1 in 513  | 1 in 26,000                                       | 1 in 100,000  |
|                                |        | Non-Finnish European/Caucasian  | 99  | 1 in 411  | 1 in 41,000                                       | 1 in 160,000  |
|                                |        | South Asian   | 99  | 1 in 133  | 1 in 13,000                                       | 1 in 52,000   |
|                                |        | Other (population not assigned)   | 99  | 1 in 304  | 1 in 30,000                                       | 1 in 120,000  |
|                                | CLN5   | African/African American  | 99  | 1 in 1,021  | 1 in 100,000                                      | 1 in 400,000  |
|                                |        | Latino/Admixed American   | 98  | 1 in 644  | 1 in 32,000                                       | 1 in 130,000  |
|                                |        | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|                                |        | East Asian  | 97  | 1 in 714  | 1 in 24,000                                       | 1 in 96,000   |
|                                |        | Finnish   | 99  | 1 in 142  | 1 in 14,000                                       | 1 in 56,000   |
|                                |        | Non-Finnish European/Caucasian  | 99  | 1 in 254  | 1 in 25,000                                       | 1 in 100,000  |
|                                |        | South Asian   | 99  | 1 in 840  | 1 in 84,000                                       | 1 in 340,000  |
|                                |        | Other (population not assigned)   | 99  | 1 in 335  | 1 in 33,000                                       | 1 in 130,000  |
|                                | CLN6   | African/African American  | 99  | 1 in 196  | 1 in 20,000                                       | 1 in 80,000   |
|                                |        | Latino/Admixed American   | 99  | 1 in 358  | 1 in 36,000                                       | 1 in 140,000  |
|                                |        | Ashkenazi Jewish  | 99  | 1 in 1,256  | 1 in 130,000                                      | 1 in 520,000  |
|                                |        | East Asian  | 99  | 1 in 572  | 1 in 57,000                                       | 1 in 230,000  |
|                                |        | Finnish   | 99  | 1 in 333  | 1 in 33,000                                       | 1 in 130,000  |
|                                |        | Non-Finnish European/Caucasian  | 99  | 1 in 434  | 1 in 43,000                                       | 1 in 170,000  |
|                                |        | South Asian   | 98  | 1 in 1,000  | 1 in 50,000                                       | 1 in 200,000  |
|                                |        | Other (population not assigned)   | 99  | 1 in 435  | 1 in 43,000                                       | 1 in 170,000  |
|                                | CLN8   | African/African American  | 99  | 1 in 1,566  | 1 in 160,000                                      | 1 in 640,000  |
|                                |        | Latino/Admixed American   | 97  | 1 in 8,644  | 1 in 290,000                                      | 1 in 1,200,000  |
|                                |        | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|                                |        | East Asian  | 99  | 1 in 4,599  | 1 in 460,000                                      | 1 in 1,800,000  |
|                                |        | Finnish   | 99  | 1 in 429  | 1 in 43,000                                       | 1 in 170,000  |
|                                |        | Non-Finnish European/Caucasian  | 99  | 1 in 1,605  | 1 in 160,000                                      | 1 in 640,000  |
|                                |        | South Asian   | 99  | 1 in 1,920  | 1 in 190,000                                      | 1 in 760,000  |
|                                |        | Other (population not assigned)   | 99  | 1 in 2,744  | 1 in 270,000                                      | 1 in 1,100,000  |
|                                | CTSD   | African/African American  | 98  | 1 in 1,160  | 1 in 58,000                                       | 1 in 230,000  |
|                                |        | Latino/Admixed American   | 98  | 1 in 1,591  | 1 in 80,000                                       | 1 in 320,000  |
|                                |        | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|                                |        | East Asian  | 99  | 1 in 9,179  | 1 in 920,000                                      | 1 in 3,700,000  |
|                                |        | Finnish   | 97  | 1 in 1,801  | 1 in 60,000                                       | 1 in 240,000  |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |              |
|----------|-----------------------------|---|---|---|---|---|--------------|
|          |                             | Non-Finnish European/Caucasian  | 97  | 1 in 1,003  | 1 in 33,000                                       | 1 in 130,000  |              |
|          |                             | South Asian   | 99  | 1 in 1,912  | 1 in 190,000                                      | 1 in 760,000  |              |
|          |                             | Other (population not assigned)   | 99  | 1 in 1,153  | 1 in 120,000                                      | 1 in 480,000  |              |
|          | CTSF                        | African/African American  | 99  | 1 in 426  | 1 in 43,000                                       | 1 in 170,000  |              |
|          |                             | Latino/Admixed American   | 99  | 1 in 1,448  | 1 in 140,000                                      | 1 in 560,000  |              |
|          |                             | Ashkenazi Jewish  | 98  | N/A   | N/A   | N/A   |              |
|          |                             | East Asian  | 99  | 1 in 365  | 1 in 36,000                                       | 1 in 140,000  |              |
|          |                             | Finnish   | 99  | 1 in 10,821   | 1 in 1,100,000                                    | 1 in 4,400,000  |              |
|          |                             | Non-Finnish European/Caucasian  | 99  | 1 in 1,773  | 1 in 180,000                                      | 1 in 720,000  |              |
|          |                             | South Asian   | 98  | 1 in 881  | 1 in 44,000                                       | 1 in 180,000  |              |
|          |                             | Other (population not assigned)   | 98  | 1 in 1,449  | 1 in 72,000                                       | 1 in 290,000  |              |
|          | KCTD7                       | African/African American  | 99  | 1 in 1,971  | 1 in 200,000                                      | 1 in 800,000  |              |
|          |                             | Latino/Admixed American   | 99  | 1 in 1,022  | 1 in 100,000                                      | 1 in 400,000  |              |
|          |                             | Ashkenazi Jewish  | 98  | 1 in 2,520  | 1 in 130,000                                      | 1 in 520,000  |              |
|          |                             | East Asian  | 99  | 1 in 2,659  | 1 in 270,000                                      | 1 in 1,100,000  |              |
|          |                             | Finnish   | 99  | 1 in 5,373  | 1 in 540,000                                      | 1 in 2,200,000  |              |
|          |                             | Non-Finnish European/Caucasian  | 99  | 1 in 1,083  | 1 in 110,000                                      | 1 in 440,000  |              |
|          |                             | South Asian   | 99  | 1 in 1,116  | 1 in 110,000                                      | 1 in 440,000  |              |
|          |                             | Other (population not assigned)   | 98  | 1 in 301  | 1 in 15,000                                       | 1 in 60,000   |              |
|          | MFSD8                       | African/African American  | 99  | 1 in 1,587  | 1 in 160,000                                      | 1 in 640,000  |              |
|          |                             | Latino/Admixed American   | 99  | 1 in 1,415  | 1 in 140,000                                      | 1 in 560,000  |              |
|          |                             | Ashkenazi Jewish  | 98  | N/A   | N/A   | N/A   |              |
|          |                             | East Asian  | 99  | 1 in 992  | 1 in 99,000                                       | 1 in 400,000  |              |
|          |                             | Finnish   | 99  | 1 in 5,411  | 1 in 540,000                                      | 1 in 2,200,000  |              |
|          |                             | Non-Finnish European/Caucasian  | 99  | 1 in 1,661  | 1 in 170,000                                      | 1 in 680,000  |              |
|          |                             | South Asian   | 99  | 1 in 3,047  | 1 in 300,000                                      | 1 in 1,200,000  |              |
|          |                             | Other (population not assigned)   | 98  | 1 in 611  | 1 in 31,000                                       | 1 in 120,000  |              |
|          | PPT1                        | African/African American  | 99  | 1 in 856  | 1 in 86,000                                       | 1 in 340,000  |              |
|          |                             | Latino/Admixed American   | 99  | 1 in 4,195  | 1 in 420,000                                      | 1 in 1,700,000  |              |
|          |                             | Ashkenazi Jewish  | 99  | N/A   | N/A   | N/A   |              |
|          |                             | East Asian  | 99  | 1 in 8,621  | 1 in 860,000                                      | 1 in 3,400,000  |              |
|          |                             | Finnish   | 99  | 1 in 78   | 1 in 7,700  | 1 in 31,000   |              |
|          |                             | Non-Finnish European/Caucasian  | 99  | 1 in 350  | 1 in 35,000                                       | 1 in 140,000  |              |
|          |                             | South Asian   | 99  | 1 in 1,550  | 1 in 150,000                                      | 1 in 600,000  |              |
|          |                             | Other (population not assigned)   | 99  | 1 in 228  | 1 in 23,000                                       | 1 in 92,000   |              |
|          | TPP1                        | African/African American  | 99  | 1 in 150  | 1 in 15,000                                       | 1 in 60,000   |              |
|          |                             | Latino/Admixed American   | 99  | 1 in 426  | 1 in 43,000                                       | 1 in 170,000  |              |
|          |                             | Ashkenazi Jewish  | 99  | 1 in 1,231  | 1 in 120,000                                      | 1 in 480,000  |              |
|          |                             | East Asian  | 98  | 1 in 3,066  | 1 in 150,000                                      | 1 in 600,000  |              |
|          |                             | Finnish   | 98  | 1 in 1,573  | 1 in 79,000                                       | 1 in 320,000  |              |
|          |                             | Non-Finnish European/Caucasian  | 99  | 1 in 276  | 1 in 28,000                                       | 1 in 110,000  |              |
|          |                             | South Asian   | 99  | 1 in 1,877  | 1 in 190,000                                      | 1 in 760,000  |              |
|          |                             | Other (population not assigned)   | 99  | 1 in 474  | 1 in 47,000                                       | 1 in 190,000  |              |
|          | Niemann-Pick disease type C | NPC1  | African/African American  | 99  | 1 in 439  | 1 in 44,000   | 1 in 180,000 |
|          |                             |   | Latino/Admixed American   | 99  | 1 in 325  | 1 in 32,000   | 1 in 130,000 |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |              |              |
|---------------------------------|--------------------------------|---|---|---|---|---|--------------|--------------|
|                                 |                                | Ashkenazi Jewish  | 97  | 1 in 382  | 1 in 13,000                                       | 1 in 52,000   |              |              |
|                                 |                                | East Asian  | 99  | 1 in 444  | 1 in 44,000                                       | 1 in 180,000  |              |              |
|                                 |                                | Finnish   | 99  | 1 in 181  | 1 in 18,000                                       | 1 in 72,000   |              |              |
|                                 |                                | Non-Finnish European/Caucasian  | 99  | 1 in 150  | 1 in 15,000                                       | 1 in 60,000   |              |              |
|                                 |                                | South Asian   | 99  | 1 in 548  | 1 in 55,000                                       | 1 in 220,000  |              |              |
|                                 |                                | Other (population not assigned)   | 99  | 1 in 328  | 1 in 33,000                                       | 1 in 130,000  |              |              |
|                                 | NPC2                           | African/African American  | 99  | 1 in 66   | 1 in 6,500  | 1 in 26,000   |              |              |
|                                 |                                | Latino/Admixed American   | 99  | 1 in 306  | 1 in 31,000                                       | 1 in 120,000  |              |              |
|                                 |                                | Ashkenazi Jewish  | 99  | 1 in 297  | 1 in 30,000                                       | 1 in 120,000  |              |              |
|                                 |                                | East Asian  | 99  | 1 in 969  | 1 in 97,000                                       | 1 in 390,000  |              |              |
|                                 |                                | Finnish   | 99  | 1 in 171  | 1 in 17,000                                       | 1 in 68,000   |              |              |
|                                 |                                | Non-Finnish European/Caucasian  | 99  | 1 in 74   | 1 in 7,300  | 1 in 29,000   |              |              |
|                                 |                                | South Asian   | 99  | 1 in 214  | 1 in 21,000                                       | 1 in 84,000   |              |              |
|                                 |                                | Other (population not assigned)   | 99  | 1 in 136  | 1 in 14,000                                       | 1 in 56,000   |              |              |
|                                 |                                | Niemann-Pick disease types A and B  | SMPD1   | African/African American  | 99  | 1 in 269  | 1 in 27,000  | 1 in 110,000 |
|                                 |                                |   |   | Latino/Admixed American   | 98  | 1 in 255  | 1 in 13,000  | 1 in 52,000  |
|                                 |                                |   |   | Ashkenazi Jewish  | 99  | 1 in 92   | 1 in 9,100   | 1 in 36,000  |
|                                 |                                |   |   | East Asian  | 97  | 1 in 499  | 1 in 17,000  | 1 in 68,000  |
| Finnish                         | 97                             |   |   | 1 in 560  | 1 in 19,000                                       | 1 in 76,000   |              |              |
| Non-Finnish European/Caucasian  | 99                             |   |   | 1 in 131  | 1 in 13,000                                       | 1 in 52,000   |              |              |
| South Asian                     | 99                             |   |   | 1 in 338  | 1 in 34,000                                       | 1 in 140,000  |              |              |
| Other (population not assigned) | 98                             |   |   | 1 in 185  | 1 in 9,200  | 1 in 37,000   |              |              |
| Nijmegen breakage syndrome      | NBN                            |   |   | African/African American  | 99  | 1 in 379  | 1 in 38,000  | 1 in 150,000 |
|                                 |                                | Latino/Admixed American   | 99  | 1 in 213  | 1 in 21,000                                       | 1 in 84,000   |              |              |
|                                 |                                | Ashkenazi Jewish  | 99  | 1 in 255  | 1 in 25,000                                       | 1 in 100,000  |              |              |
|                                 |                                | East Asian  | 99  | 1 in 278  | 1 in 28,000                                       | 1 in 110,000  |              |              |
|                                 |                                | Finnish   | 99  | 1 in 103  | 1 in 10,000                                       | 1 in 40,000   |              |              |
|                                 |                                | Non-Finnish European/Caucasian  | 99  | 1 in 70   | 1 in 6,900  | 1 in 28,000   |              |              |
|                                 |                                | South Asian   | 99  | 1 in 767  | 1 in 77,000                                       | 1 in 310,000  |              |              |
|                                 |                                | Other (population not assigned)   | 99  | 1 in 124  | 1 in 12,000                                       | 1 in 48,000   |              |              |
|                                 |                                | Omenn syndrome  | DCLRE1C   | African/African American  | 99  | 1 in 100  | 1 in 9,900   | 1 in 40,000  |
| Latino/Admixed American         | 98                             |   |   | 1 in 973  | 1 in 49,000                                       | 1 in 200,000  |              |              |
| Ashkenazi Jewish                | 97                             |   |   | 1 in 202  | 1 in 6,700  | 1 in 27,000   |              |              |
| East Asian                      | 98                             |   |   | 1 in 836  | 1 in 42,000                                       | 1 in 170,000  |              |              |
| Finnish                         | 99                             |   |   | 1 in 2,703  | 1 in 270,000                                      | 1 in 1,100,000  |              |              |
| Non-Finnish European/Caucasian  | 98                             |   |   | 1 in 775  | 1 in 39,000                                       | 1 in 160,000  |              |              |
| South Asian                     | 97                             |   |   | 1 in 356  | 1 in 12,000                                       | 1 in 48,000   |              |              |
| Other (population not assigned) | 97                             |   |   | 1 in 203  | 1 in 6,700  | 1 in 27,000   |              |              |
| RAG1                            | African/African American       |   |   | 99  | 1 in 1,109  | 1 in 110,000  | 1 in 440,000 |              |
|                                 | Latino/Admixed American        |   | 99  | 1 in 441  | 1 in 44,000                                       | 1 in 180,000  |              |              |
|                                 | Ashkenazi Jewish               |   | 99  | 1 in 2,462  | 1 in 250,000                                      | 1 in 1,000,000  |              |              |
|                                 | East Asian                     |   | 99  | 1 in 523  | 1 in 52,000                                       | 1 in 210,000  |              |              |
|                                 | Finnish                        |   | 99  | 1 in 643  | 1 in 64,000                                       | 1 in 260,000  |              |              |
|                                 | Non-Finnish European/Caucasian |   | 99  | 1 in 344  | 1 in 34,000                                       | 1 in 140,000  |              |              |
|                                 | South Asian                    |   | 99  | 1 in 639  | 1 in 64,000                                       | 1 in 260,000  |              |              |

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

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

N/A= Not Available

| Disorder                                     | Gene     | Ethnicity<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|--|----------|---|---|---|---|---|
| Disorder                                     |          | Other (population not assigned)   | 99  | 1 in 253  | 1 in 25,000                                       | 1 in 100,000  |
|  | RAG2     | African/African American  | 99  | 1 in 653  | 1 in 65,000                                       | 1 in 260,000  |
|  |          | Latino/Admixed American   | 99  | 1 in 685  | 1 in 68,000                                       | 1 in 270,000  |
|  |          | Ashkenazi Jewish  | 99  | 1 in 704  | 1 in 70,000                                       | 1 in 280,000  |
|  |          | East Asian  | 99  | 1 in 1,266  | 1 in 130,000                                      | 1 in 520,000  |
|  |          | Finnish   | 99  | 1 in 1,217  | 1 in 120,000                                      | 1 in 480,000  |
|  |          | Non-Finnish European/Caucasian  | 99  | 1 in 736  | 1 in 74,000                                       | 1 in 300,000  |
|  |          | South Asian   | 99  | 1 in 696  | 1 in 70,000                                       | 1 in 280,000  |
|  |          | Other (population not assigned)   | 99  | 1 in 377  | 1 in 38,000                                       | 1 in 150,000  |
| Ornithine transcarbamylase deficiency        | OTC      | 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   |
| Ornithine translocase deficiency             | SLC25A15 | African/African American  | 99  | 1 in 3,829  | 1 in 380,000                                      | 1 in 1,500,000  |
|  |          | Latino/Admixed American   | 99  | 1 in 898  | 1 in 90,000                                       | 1 in 360,000  |
|  |          | Ashkenazi Jewish  | 99  | 1 in 110  | 1 in 11,000                                       | 1 in 44,000   |
|  |          | East Asian  | 99  | 1 in 298  | 1 in 30,000                                       | 1 in 120,000  |
|  |          | Finnish   | 99  | 1 in 3,608  | 1 in 360,000                                      | 1 in 1,400,000  |
|  |          | Non-Finnish European/Caucasian  | 99  | 1 in 1,244  | 1 in 120,000                                      | 1 in 480,000  |
|  |          | South Asian   | 99  | 1 in 1,703  | 1 in 170,000                                      | 1 in 680,000  |
|  |          | Other (population not assigned)   | 99  | 1 in 465  | 1 in 46,000                                       | 1 in 180,000  |
| Osteogenesis imperfecta, autosomal recessive | BMP1     | African/African American  | 99  | 1 in 49   | 1 in 4,800  | 1 in 19,000   |
|  |          | Latino/Admixed American   | 99  | 1 in 394  | 1 in 39,000                                       | 1 in 160,000  |
|  |          | Ashkenazi Jewish  | 99  | 1 in 207  | 1 in 21,000                                       | 1 in 84,000   |
|  |          | East Asian  | 99  | 1 in 740  | 1 in 74,000                                       | 1 in 300,000  |
|  |          | Finnish   | 99  | 1 in 2,688  | 1 in 270,000                                      | 1 in 1,100,000  |
|  |          | Non-Finnish European/Caucasian  | 99  | 1 in 643  | 1 in 64,000                                       | 1 in 260,000  |
|  |          | South Asian   | 98  | 1 in 647  | 1 in 32,000                                       | 1 in 130,000  |
|  |          | Other (population not assigned)   | 99  | 1 in 740  | 1 in 74,000                                       | 1 in 300,000  |
|  | CRTAP    | African/African American  | 99  | 1 in 1,122  | 1 in 110,000                                      | 1 in 440,000  |
|  |          | Latino/Admixed American   | 97  | 1 in 546  | 1 in 18,000                                       | 1 in 72,000   |
|  |          | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|  |          | East Asian  | 99  | 1 in 1,377  | 1 in 140,000                                      | 1 in 560,000  |
|  |          | Finnish   | 97  | N/A   | N/A   | N/A   |
|  |          | Non-Finnish European/Caucasian  | 99  | 1 in 1,416  | 1 in 140,000                                      | 1 in 560,000  |
|  |          | South Asian   | 98  | 1 in 1,059  | 1 in 53,000                                       | 1 in 210,000  |
|  |          | Other (population not assigned)   | 99  | 1 in 2,126  | 1 in 210,000                                      | 1 in 840,000  |
|  | FKBP10   | African/African American  | 97  | 1 in 641  | 1 in 21,000                                       | 1 in 84,000   |
|  |          | Latino/Admixed American   | 99  | 1 in 624  | 1 in 62,000                                       | 1 in 250,000  |
|  |          | Ashkenazi Jewish  | 99  | 1 in 839  | 1 in 84,000                                       | 1 in 340,000  |
|  |          | East Asian  | 99  | 1 in 457  | 1 in 46,000                                       | 1 in 180,000  |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|----------|----------|---|---|---|---|---|
|          |          | Finnish   | 97  | 1 in 1,591  | 1 in 53,000                                       | 1 in 210,000  |
|          |          | Non-Finnish European/Caucasian  | 98  | 1 in 1,025  | 1 in 51,000                                       | 1 in 200,000  |
|          |          | South Asian   | 98  | 1 in 803  | 1 in 40,000                                       | 1 in 160,000  |
|          |          | Other (population not assigned)   | 98  | 1 in 868  | 1 in 43,000                                       | 1 in 170,000  |
|          | P3H1     | African/African American  | 99  | 1 in 136  | 1 in 14,000                                       | 1 in 56,000   |
|          |          | Latino/Admixed American   | 97  | 1 in 278  | 1 in 9,200  | 1 in 37,000   |
|          |          | Ashkenazi Jewish  | 99  | 1 in 816  | 1 in 82,000                                       | 1 in 330,000  |
|          |          | East Asian  | 99  | 1 in 206  | 1 in 21,000                                       | 1 in 84,000   |
|          |          | Finnish   | 98  | 1 in 2,203  | 1 in 110,000                                      | 1 in 440,000  |
|          |          | Non-Finnish European/Caucasian  | 98  | 1 in 567  | 1 in 28,000                                       | 1 in 110,000  |
|          |          | South Asian   | 98  | 1 in 765  | 1 in 38,000                                       | 1 in 150,000  |
|          |          | Other (population not assigned)   | 98  | 1 in 912  | 1 in 46,000                                       | 1 in 180,000  |
|          | PLOD2    | African/African American  | 97  | 1 in 54   | 1 in 1,800  | 1 in 7,200  |
|          |          | Latino/Admixed American   | 97  | 1 in 504  | 1 in 17,000                                       | 1 in 68,000   |
|          |          | Ashkenazi Jewish  | 97  | 1 in 4,573  | 1 in 150,000                                      | 1 in 600,000  |
|          |          | East Asian  | 98  | 1 in 2,150  | 1 in 110,000                                      | 1 in 440,000  |
|          |          | Finnish   | 99  | 1 in 251  | 1 in 25,000                                       | 1 in 100,000  |
|          |          | Non-Finnish European/Caucasian  | 99  | 1 in 1,400  | 1 in 140,000                                      | 1 in 560,000  |
|          |          | South Asian   | 98  | 1 in 842  | 1 in 42,000                                       | 1 in 170,000  |
|          |          | Other (population not assigned)   | 98  | 1 in 456  | 1 in 23,000                                       | 1 in 92,000   |
|          | PIIB     | African/African American  | 97  | N/A   | N/A   | N/A   |
|          |          | Latino/Admixed American   | 98  | 1 in 1,722  | 1 in 86,000                                       | 1 in 340,000  |
|          |          | Ashkenazi Jewish  | 97  | 1 in 5,041  | 1 in 170,000                                      | 1 in 680,000  |
|          |          | East Asian  | 99  | 1 in 2,875  | 1 in 290,000                                      | 1 in 1,200,000  |
|          |          | Finnish   | 97  | N/A   | N/A   | N/A   |
|          |          | Non-Finnish European/Caucasian  | 99  | 1 in 2,688  | 1 in 270,000                                      | 1 in 1,100,000  |
|          |          | South Asian   | 99  | 1 in 3,061  | 1 in 310,000                                      | 1 in 1,200,000  |
|          |          | Other (population not assigned)   | 99  | 1 in 984  | 1 in 98,000                                       | 1 in 390,000  |
|          | SERPINF1 | African/African American  | 98  | 1 in 1,166  | 1 in 58,000                                       | 1 in 230,000  |
|          |          | Latino/Admixed American   | 98  | 1 in 3,209  | 1 in 160,000                                      | 1 in 640,000  |
|          |          | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|          |          | East Asian  | 99  | 1 in 1,403  | 1 in 140,000                                      | 1 in 560,000  |
|          |          | Finnish   | 97  | N/A   | N/A   | N/A   |
|          |          | Non-Finnish European/Caucasian  | 98  | 1 in 1,456  | 1 in 73,000                                       | 1 in 290,000  |
|          |          | South Asian   | 98  | 1 in 2,606  | 1 in 130,000                                      | 1 in 520,000  |
|          |          | Other (population not assigned)   | 97  | 1 in 3,068  | 1 in 100,000                                      | 1 in 400,000  |
|          | TMEM38B  | African/African American  | 99  | 1 in 2,703  | 1 in 270,000                                      | 1 in 1,100,000  |
|          |          | Latino/Admixed American   | 99  | 1 in 2,880  | 1 in 290,000                                      | 1 in 1,200,000  |
|          |          | Ashkenazi Jewish  | 99  | 1 in 336  | 1 in 34,000                                       | 1 in 140,000  |
|          |          | East Asian  | 99  | 1 in 9,196  | 1 in 920,000                                      | 1 in 3,700,000  |
|          |          | Finnish   | 97  | 1 in 2,706  | 1 in 90,000                                       | 1 in 360,000  |
|          |          | Non-Finnish European/Caucasian  | 99  | 1 in 2,249  | 1 in 220,000                                      | 1 in 880,000  |
|          |          | South Asian   | 99  | 1 in 1,364  | 1 in 140,000                                      | 1 in 560,000  |
|          |          | Other (population not assigned)   | 99  | 1 in 3,068  | 1 in 310,000                                      | 1 in 1,200,000  |
|          | WNT1     | African/African American  | 99  | 1 in 2,260  | 1 in 230,000                                      | 1 in 920,000  |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|--|---------|---|---|---|---|---|
|  |         | Latino/Admixed American   | 97  | 1 in 4,934  | 1 in 160,000                                      | 1 in 640,000  |
|  |         | Ashkenazi Jewish  | 97  | 1 in 2,468  | 1 in 82,000                                       | 1 in 330,000  |
|  |         | East Asian  | 99  | 1 in 1,003  | 1 in 100,000                                      | 1 in 400,000  |
|  |         | Finnish   | 97  | 1 in 6,767  | 1 in 230,000                                      | 1 in 920,000  |
|  |         | Non-Finnish European/Caucasian  | 97  | 1 in 2,536  | 1 in 85,000                                       | 1 in 340,000  |
|  |         | South Asian   | 97  | 1 in 3,758  | 1 in 130,000                                      | 1 in 520,000  |
|  |         | Other (population not assigned)   | 97  | 1 in 2,727  | 1 in 91,000                                       | 1 in 360,000  |
| Osteopetrosis, autosomal recessive               | OSTM1   | African/African American  | 97  | N/A   | N/A   | N/A   |
|  |         | Latino/Admixed American   | 97  | N/A   | N/A   | N/A   |
|  |         | Ashkenazi Jewish  | 99  | 1 in 2,097  | 1 in 210,000                                      | 1 in 840,000  |
|  |         | East Asian  | 99  | 1 in 9,197  | 1 in 920,000                                      | 1 in 3,700,000  |
|  |         | Finnish   | 97  | 1 in 7,821  | 1 in 260,000                                      | 1 in 1,000,000  |
|  |         | Non-Finnish European/Caucasian  | 99  | 1 in 6,173  | 1 in 620,000                                      | 1 in 2,500,000  |
|  |         | South Asian   | 99  | 1 in 6,619  | 1 in 660,000                                      | 1 in 2,600,000  |
|  |         | Other (population not assigned)   | 97  | N/A   | N/A   | N/A   |
|  | TCIRG1  | African/African American  | 99  | 1 in 729  | 1 in 73,000                                       | 1 in 290,000  |
|  |         | Latino/Admixed American   | 99  | 1 in 382  | 1 in 38,000                                       | 1 in 150,000  |
|  |         | Ashkenazi Jewish  | 99  | 1 in 491  | 1 in 49,000                                       | 1 in 200,000  |
|  |         | East Asian  | 99  | 1 in 540  | 1 in 54,000                                       | 1 in 220,000  |
|  |         | Finnish   | 99  | 1 in 1,482  | 1 in 150,000                                      | 1 in 600,000  |
|  |         | Non-Finnish European/Caucasian  | 99  | 1 in 589  | 1 in 59,000                                       | 1 in 240,000  |
|  |         | South Asian   | 99  | 1 in 701  | 1 in 70,000                                       | 1 in 280,000  |
|  |         | Other (population not assigned)   | 99  | 1 in 443  | 1 in 44,000                                       | 1 in 180,000  |
|  | TNFSF11 | African/African American  | 99  | 1 in 1,789  | 1 in 180,000                                      | 1 in 720,000  |
|  |         | Latino/Admixed American   | 99  | 1 in 3,300  | 1 in 330,000                                      | 1 in 1,300,000  |
|  |         | Ashkenazi Jewish  | 99  | 1 in 5,039  | 1 in 500,000                                      | 1 in 2,000,000  |
|  |         | East Asian  | 99  | 1 in 2,480  | 1 in 250,000                                      | 1 in 1,000,000  |
|  |         | Finnish   | 99  | N/A   | N/A   | N/A   |
| Non-Finnish European/Caucasian                   |         | 99  | 1 in 2,497  | 1 in 250,000  | 1 in 1,000,000                                    |   |
| South Asian                                      |         | 99  | 1 in 419  | 1 in 42,000   | 1 in 170,000                                      |   |
| Other (population not assigned)                  |         | 99  | N/A   | N/A   | N/A   |   |
| Pantothenate kinase-associated neurodegeneration | PANK2   | African/African American  | 98  | 1 in 1,027  | 1 in 51,000                                       | 1 in 200,000  |
|  |         | Latino/Admixed American   | 98  | 1 in 845  | 1 in 42,000                                       | 1 in 170,000  |
|  |         | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|  |         | East Asian  | 99  | 1 in 655  | 1 in 65,000                                       | 1 in 260,000  |
|  |         | Finnish   | 98  | 1 in 1,418  | 1 in 71,000                                       | 1 in 280,000  |
|  |         | Non-Finnish European/Caucasian  | 98  | 1 in 700  | 1 in 35,000                                       | 1 in 140,000  |
|  |         | South Asian   | 97  | 1 in 427  | 1 in 14,000                                       | 1 in 56,000   |
|  |         | Other (population not assigned)   | 98  | 1 in 510  | 1 in 25,000                                       | 1 in 100,000  |
|  |         | Pendred syndrome  | SLC26A4   | African/African American  | 99  | 1 in 84   |
| Latino/Admixed American                          | 99      |   |   | 1 in 144  | 1 in 14,000                                       | 1 in 56,000   |
| Ashkenazi Jewish                                 | 99      |   |   | 1 in 64   | 1 in 6,300  | 1 in 25,000   |
| East Asian                                       | 99      |   |   | 1 in 56   | 1 in 5,500  | 1 in 22,000   |
| Finnish  | 99      |   |   | 1 in 895  | 1 in 89,000                                       | 1 in 360,000  |
| Non-Finnish European/Caucasian                   | 99      |   |   | 1 in 79   | 1 in 7,800  | 1 in 31,000   |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|--|-------|---|---|---|---|---|
|  |       | South Asian   | 99  | 1 in 48   | 1 in 4,700  | 1 in 19,000   |
|  |       | Other (population not assigned)   | 99  | 1 in 84   | 1 in 8,300  | 1 in 33,000   |
| Peroxisomal acyl-CoA oxidase deficiency        | ACOX1 | African/African American  | 98  | 1 in 866  | 1 in 43,000                                       | 1 in 170,000  |
|  |       | Latino/Admixed American   | 98  | 1 in 1,909  | 1 in 95,000                                       | 1 in 380,000  |
|  |       | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|  |       | East Asian  | 99  | 1 in 9,197  | 1 in 920,000                                      | 1 in 3,700,000  |
|  |       | Finnish   | 99  | 1 in 2,706  | 1 in 270,000                                      | 1 in 1,100,000  |
|  |       | Non-Finnish European/Caucasian  | 99  | 1 in 504  | 1 in 50,000                                       | 1 in 200,000  |
|  |       | South Asian   | 97  | 1 in 1,177  | 1 in 39,000                                       | 1 in 160,000  |
|  |       | Other (population not assigned)   | 97  | 1 in 3,069  | 1 in 100,000                                      | 1 in 400,000  |
|  |       | Phenylalanine hydroxylase deficiency, includes<br>phenylketonuria (PKU)   | PAH   | African/African American  | 99  | 1 in 213  |
| Latino/Admixed American                        | 99    |   |   | 1 in 74   | 1 in 7,300  | 1 in 29,000   |
| Ashkenazi Jewish                               | 99    |   |   | 1 in 17   | 1 in 1,600  | 1 in 6,400  |
| East Asian                                     | 98    |   |   | 1 in 57   | 1 in 2,800  | 1 in 11,000   |
| Finnish  | 99    |   |   | 1 in 195  | 1 in 19,000                                       | 1 in 76,000   |
| Non-Finnish European/Caucasian                 | 99    |   |   | 1 in 41   | 1 in 4,000  | 1 in 16,000   |
| South Asian                                    | 99    |   |   | 1 in 76   | 1 in 7,500  | 1 in 30,000   |
| Other (population not assigned)                | 99    |   |   | 1 in 39   | 1 in 3,800  | 1 in 15,000   |
| Phosphoglycerate dehydrogenase deficiency      | PHGDH |   |   | African/African American  | 99  | 1 in 3,826  |
|  |       | Latino/Admixed American   | 99  | 1 in 4,291  | 1 in 430,000                                      | 1 in 1,700,000  |
|  |       | Ashkenazi Jewish  | 99  | 1 in 290  | 1 in 29,000                                       | 1 in 120,000  |
|  |       | East Asian  | 99  | 1 in 1,500  | 1 in 150,000                                      | 1 in 600,000  |
|  |       | Finnish   | 99  | 1 in 1,859  | 1 in 190,000                                      | 1 in 760,000  |
|  |       | Non-Finnish European/Caucasian  | 99  | 1 in 1,575  | 1 in 160,000                                      | 1 in 640,000  |
|  |       | South Asian   | 99  | 1 in 1,862  | 1 in 190,000                                      | 1 in 760,000  |
|  |       | Other (population not assigned)   | 99  | N/A   | N/A   | N/A   |
|  |       | Pitt-Hopkins-like syndrome 1  | CNTNAP2   | African/African American  | 97  | 1 in 114  |
| Latino/Admixed American                        | 98    |   |   | 1 in 355  | 1 in 18,000                                       | 1 in 72,000   |
| Ashkenazi Jewish                               | 97    |   |   | 1 in 1,874  | 1 in 62,000                                       | 1 in 250,000  |
| East Asian                                     | 99    |   |   | 1 in 571  | 1 in 57,000                                       | 1 in 230,000  |
| Finnish  | 97    |   |   | 1 in 381  | 1 in 13,000                                       | 1 in 52,000   |
| Non-Finnish European/Caucasian                 | 98    |   |   | 1 in 314  | 1 in 16,000                                       | 1 in 64,000   |
| South Asian                                    | 97    |   |   | 1 in 419  | 1 in 14,000                                       | 1 in 56,000   |
| Other (population not assigned)                | 98    |   |   | 1 in 349  | 1 in 17,000                                       | 1 in 68,000   |
| Polycystic kidney disease, autosomal recessive | PKHD1 |   |   | African/African American  | 99  | 1 in 71   |
|  |       | Latino/Admixed American   | 99  | 1 in 61   | 1 in 6,000  | 1 in 24,000   |
|  |       | Ashkenazi Jewish  | 98  | 1 in 33   | 1 in 1,600  | 1 in 6,400  |
|  |       | East Asian  | 99  | 1 in 193  | 1 in 19,000                                       | 1 in 76,000   |
|  |       | Finnish   | 99  | 1 in 25   | 1 in 2,400  | 1 in 9,600  |
|  |       | Non-Finnish European/Caucasian  | 99  | 1 in 31   | 1 in 3,000  | 1 in 12,000   |
|  |       | South Asian   | 99  | 1 in 127  | 1 in 13,000                                       | 1 in 52,000   |
|  |       | Other (population not assigned)   | 99  | 1 in 34   | 1 in 3,300  | 1 in 13,000   |
|  |       | Pompe disease   | GAA   | African/African American  | 99  | 1 in 104  |
| Latino/Admixed American                        | 99    |   |   | 1 in 111  | 1 in 11,000                                       | 1 in 44,000   |
| Ashkenazi Jewish                               | 99    |   |   | 1 in 82   | 1 in 8,100  | 1 in 32,000   |

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

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N/A= Not Available

| Disorder                   | Gene                            | Ethnicity<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|----------------------------|---------------------------------|---|---|---|---|---|
|                            |                                 | East Asian  | 99  | 1 in 51   | 1 in 5,000  | 1 in 20,000   |
|                            |                                 | Finnish   | 99  | 1 in 201  | 1 in 20,000                                       | 1 in 80,000   |
|                            |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 63   | 1 in 6,200  | 1 in 25,000   |
|                            |                                 | South Asian   | 99  | 1 in 146  | 1 in 15,000                                       | 1 in 60,000   |
|                            |                                 | Other (population not assigned)   | 99  | 1 in 104  | 1 in 10,000                                       | 1 in 40,000   |
| Pontocerebellar hypoplasia | AMPD2                           | African/African American  | 98  | 1 in 761  | 1 in 38,000                                       | 1 in 150,000  |
|                            |                                 | Latino/Admixed American   | 97  | 1 in 400  | 1 in 13,000                                       | 1 in 52,000   |
|                            |                                 | Ashkenazi Jewish  | 98  | 1 in 1,494  | 1 in 75,000                                       | 1 in 300,000  |
|                            |                                 | East Asian  | 97  | 1 in 195  | 1 in 6,500  | 1 in 26,000   |
|                            |                                 | Finnish   | 97  | 1 in 834  | 1 in 28,000                                       | 1 in 110,000  |
|                            |                                 | Non-Finnish European/Caucasian  | 98  | 1 in 488  | 1 in 24,000                                       | 1 in 96,000   |
|                            |                                 | South Asian   | 98  | 1 in 358  | 1 in 18,000                                       | 1 in 72,000   |
|                            |                                 | Other (population not assigned)   | 97  | 1 in 445  | 1 in 15,000                                       | 1 in 60,000   |
|                            |                                 | CHMP1A  | African/African American  | 99  | 1 in 1,214  | 1 in 120,000  |
|                            | Latino/Admixed American         |   | 99  | 1 in 907  | 1 in 91,000                                       | 1 in 360,000  |
|                            | Ashkenazi Jewish                |   | 99  | 1 in 620  | 1 in 62,000                                       | 1 in 250,000  |
|                            | East Asian                      |   | 99  | 1 in 331  | 1 in 33,000                                       | 1 in 130,000  |
|                            | Finnish                         |   | 99  | 1 in 1,064  | 1 in 110,000                                      | 1 in 440,000  |
|                            | Non-Finnish European/Caucasian  |   | 99  | 1 in 717  | 1 in 72,000                                       | 1 in 290,000  |
|                            | South Asian                     |   | 99  | 1 in 734  | 1 in 73,000                                       | 1 in 290,000  |
|                            | Other (population not assigned) |   | 99  | 1 in 365  | 1 in 36,000                                       | 1 in 140,000  |
|                            | CLP1                            | African/African American  | 98  | N/A   | N/A   | N/A   |
|                            |                                 | Latino/Admixed American   | 99  | 1 in 5,682  | 1 in 570,000                                      | 1 in 2,300,000  |
|                            |                                 | Ashkenazi Jewish  | 98  | N/A   | N/A   | N/A   |
|                            |                                 | East Asian  | 98  | 1 in 3,066  | 1 in 150,000                                      | 1 in 600,000  |
|                            |                                 | Finnish   | 99  | 1 in 10,821   | 1 in 1,100,000                                    | 1 in 4,400,000  |
|                            |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 2,855  | 1 in 290,000                                      | 1 in 1,200,000  |
|                            |                                 | South Asian   | 98  | 1 in 2,162  | 1 in 110,000                                      | 1 in 440,000  |
|                            |                                 | Other (population not assigned)   | 98  | N/A   | N/A   | N/A   |
|                            | EXOSC3                          | African/African American  | 98  | 1 in 507  | 1 in 25,000                                       | 1 in 100,000  |
|                            |                                 | Latino/Admixed American   | 99  | 1 in 287  | 1 in 29,000                                       | 1 in 120,000  |
|                            |                                 | Ashkenazi Jewish  | 98  | 1 in 719  | 1 in 36,000                                       | 1 in 140,000  |
|                            |                                 | East Asian  | 97  | 1 in 336  | 1 in 11,000                                       | 1 in 44,000   |
|                            |                                 | Finnish   | 99  | 1 in 43   | 1 in 4,200  | 1 in 17,000   |
|                            |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 139  | 1 in 14,000                                       | 1 in 56,000   |
|                            |                                 | South Asian   | 99  | 1 in 213  | 1 in 21,000                                       | 1 in 84,000   |
|                            |                                 | Other (population not assigned)   | 99  | 1 in 149  | 1 in 15,000                                       | 1 in 60,000   |
|                            |                                 | RARS2   | African/African American  | 99  | 1 in 158  | 1 in 16,000   |
|                            | Latino/Admixed American         |   | 99  | 1 in 109  | 1 in 11,000                                       | 1 in 44,000   |
|                            | Ashkenazi Jewish                |   | 99  | 1 in 89   | 1 in 8,800  | 1 in 35,000   |
|                            | East Asian                      |   | 99  | 1 in 70   | 1 in 6,900  | 1 in 28,000   |
|                            | Finnish                         |   | 99  | 1 in 245  | 1 in 24,000                                       | 1 in 96,000   |
|                            | Non-Finnish European/Caucasian  |   | 99  | 1 in 109  | 1 in 11,000                                       | 1 in 44,000   |
|                            | South Asian                     |   | 99  | 1 in 260  | 1 in 26,000                                       | 1 in 100,000  |
|                            | Other (population not assigned) |   | 99  | 1 in 132  | 1 in 13,000                                       | 1 in 52,000   |



### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2) | Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2) | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|---------|---|---|---|--|--|
|          | SEPSECS | African/African American  | 99  | 1 in 672  | 1 in 67,000                                    | 1 in 270,000   |
|          |         | Latino/Admixed American   | 99  | 1 in 711  | 1 in 71,000                                    | 1 in 280,000   |
|          |         | Ashkenazi Jewish  | 99  | 1 in 1,665  | 1 in 170,000                                   | 1 in 680,000   |
|          |         | East Asian  | 97  | 1 in 154  | 1 in 5,100                                     | 1 in 20,000  |
|          |         | Finnish   | 99  | 1 in 88   | 1 in 8,700                                     | 1 in 35,000  |
|          |         | Non-Finnish European/Caucasian  | 98  | 1 in 523  | 1 in 26,000                                    | 1 in 100,000   |
|          |         | South Asian   | 99  | 1 in 470  | 1 in 47,000                                    | 1 in 190,000   |
|          |         | Other (population not assigned)   | 99  | 1 in 390  | 1 in 39,000                                    | 1 in 160,000   |
|          | TSEN2   | African/African American  | 97  | 1 in 104  | 1 in 3,400                                     | 1 in 14,000  |
|          |         | Latino/Admixed American   | 99  | 1 in 230  | 1 in 23,000                                    | 1 in 92,000  |
|          |         | Ashkenazi Jewish  | 99  | 1 in 389  | 1 in 39,000                                    | 1 in 160,000   |
|          |         | East Asian  | 98  | 1 in 913  | 1 in 46,000                                    | 1 in 180,000   |
|          |         | Finnish   | 99  | 1 in 258  | 1 in 26,000                                    | 1 in 100,000   |
|          |         | Non-Finnish European/Caucasian  | 99  | 1 in 152  | 1 in 15,000                                    | 1 in 60,000  |
|          |         | South Asian   | 98  | 1 in 789  | 1 in 39,000                                    | 1 in 160,000   |
|          |         | Other (population not assigned)   | 99  | 1 in 109  | 1 in 11,000                                    | 1 in 44,000  |
|          | TSEN34  | African/African American  | 98  | N/A   | N/A  | N/A  |
|          |         | Latino/Admixed American   | 98  | 1 in 3,635  | 1 in 180,000                                   | 1 in 720,000   |
|          |         | Ashkenazi Jewish  | 99  | 1 in 1,007  | 1 in 100,000                                   | 1 in 400,000   |
|          |         | East Asian  | 99  | 1 in 8,990  | 1 in 900,000                                   | 1 in 3,600,000   |
|          |         | Finnish   | 98  | N/A   | N/A  | N/A  |
|          |         | Non-Finnish European/Caucasian  | 99  | 1 in 2,062  | 1 in 210,000                                   | 1 in 840,000   |
|          |         | South Asian   | 99  | 1 in 8,464  | 1 in 850,000                                   | 1 in 3,400,000   |
|          |         | Other (population not assigned)   | 98  | N/A   | N/A  | N/A  |
|          | TSEN54  | African/African American  | 99  | 1 in 260  | 1 in 26,000                                    | 1 in 100,000   |
|          |         | Latino/Admixed American   | 99  | 1 in 491  | 1 in 49,000                                    | 1 in 200,000   |
|          |         | Ashkenazi Jewish  | 99  | 1 in 561  | 1 in 56,000                                    | 1 in 220,000   |
|          |         | East Asian  | 97  | 1 in 186  | 1 in 6,200                                     | 1 in 25,000  |
|          |         | Finnish   | 99  | 1 in 566  | 1 in 57,000                                    | 1 in 230,000   |
|          |         | Non-Finnish European/Caucasian  | 99  | 1 in 223  | 1 in 22,000                                    | 1 in 88,000  |
|          |         | South Asian   | 98  | 1 in 686  | 1 in 34,000                                    | 1 in 140,000   |
|          |         | Other (population not assigned)   | 99  | 1 in 204  | 1 in 20,000                                    | 1 in 80,000  |
|          | VPS53   | African/African American  | 99  | 1 in 1,577  | 1 in 160,000                                   | 1 in 640,000   |
|          |         | Latino/Admixed American   | 98  | 1 in 2,317  | 1 in 120,000                                   | 1 in 480,000   |
|          |         | Ashkenazi Jewish  | 97  | 1 in 5,041  | 1 in 170,000                                   | 1 in 680,000   |
|          |         | East Asian  | 98  | 1 in 943  | 1 in 47,000                                    | 1 in 190,000   |
|          |         | Finnish   | 99  | 1 in 309  | 1 in 31,000                                    | 1 in 120,000   |
|          |         | Non-Finnish European/Caucasian  | 98  | 1 in 1,506  | 1 in 75,000                                    | 1 in 300,000   |
|          |         | South Asian   | 99  | 1 in 1,549  | 1 in 150,000                                   | 1 in 600,000   |
|          |         | Other (population not assigned)   | 97  | 1 in 3,066  | 1 in 100,000                                   | 1 in 400,000   |
|          | VRK1    | African/African American  | 97  | 1 in 8,120  | 1 in 270,000                                   | 1 in 1,100,000   |
|          |         | Latino/Admixed American   | 99  | 1 in 324  | 1 in 32,000                                    | 1 in 130,000   |
|          |         | Ashkenazi Jewish  | 99  | 1 in 329  | 1 in 33,000                                    | 1 in 130,000   |
|          |         | East Asian  | 99  | 1 in 930  | 1 in 93,000                                    | 1 in 370,000   |
|          |         | Finnish   | 97  | N/A   | N/A  | N/A  |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |              |              |
|---------------------------------|---------------------------------|---|---|---|---|---|--------------|--------------|
|                                 |                                 | Non-Finnish European/Caucasian  | 97  | 1 in 774  | 1 in 26,000                                       | 1 in 100,000  |              |              |
|                                 |                                 | South Asian   | 98  | 1 in 7,488  | 1 in 370,000                                      | 1 in 1,500,000  |              |              |
|                                 |                                 | Other (population not assigned)   | 99  | 1 in 202  | 1 in 20,000                                       | 1 in 80,000   |              |              |
| Primary carnitine deficiency    | SLC22A5                         | African/African American  | 99  | 1 in 70   | 1 in 6,900  | 1 in 28,000   |              |              |
|                                 |                                 | Latino/Admixed American   | 99  | 1 in 72   | 1 in 7,100  | 1 in 28,000   |              |              |
|                                 |                                 | Ashkenazi Jewish  | 99  | 1 in 702  | 1 in 70,000                                       | 1 in 280,000  |              |              |
|                                 |                                 | East Asian  | 99  | 1 in 84   | 1 in 8,300  | 1 in 33,000   |              |              |
|                                 |                                 | Finnish   | 99  | 1 in 390  | 1 in 39,000                                       | 1 in 160,000  |              |              |
|                                 |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 69   | 1 in 6,800  | 1 in 27,000   |              |              |
|                                 |                                 | South Asian   | 99  | 1 in 42   | 1 in 4,100  | 1 in 16,000   |              |              |
|                                 |                                 | Other (population not assigned)   | 99  | 1 in 56   | 1 in 5,500  | 1 in 22,000   |              |              |
|                                 |                                 | Primary congenital glaucoma   | CYP1B1  | African/African American  | 98  | 1 in 1,667  | 1 in 83,000  | 1 in 330,000 |
| Latino/Admixed American         | 98                              |   |   | 1 in 246  | 1 in 12,000                                       | 1 in 48,000   |              |              |
| Ashkenazi Jewish                | 99                              |   |   | 1 in 2,465  | 1 in 250,000                                      | 1 in 1,000,000  |              |              |
| East Asian                      | 99                              |   |   | 1 in 399  | 1 in 40,000                                       | 1 in 160,000  |              |              |
| Finnish                         | 97                              |   |   | 1 in 2,786  | 1 in 93,000                                       | 1 in 370,000  |              |              |
| Non-Finnish European/Caucasian  | 99                              |   |   | 1 in 196  | 1 in 20,000                                       | 1 in 80,000   |              |              |
| South Asian                     | 99                              |   |   | 1 in 325  | 1 in 32,000                                       | 1 in 130,000  |              |              |
| Other (population not assigned) | 98                              |   |   | 1 in 145  | 1 in 7,200  | 1 in 29,000   |              |              |
| Primary hyperoxaluria           | AGXT                            |   |   | African/African American  | 99  | 1 in 528  | 1 in 53,000  | 1 in 210,000 |
|                                 |                                 | Latino/Admixed American   | 99  | 1 in 451  | 1 in 45,000                                       | 1 in 180,000  |              |              |
|                                 |                                 | Ashkenazi Jewish  | 99  | 1 in 1,595  | 1 in 160,000                                      | 1 in 640,000  |              |              |
|                                 |                                 | East Asian  | 99  | 1 in 170  | 1 in 17,000                                       | 1 in 68,000   |              |              |
|                                 |                                 | Finnish   | 99  | 1 in 1,963  | 1 in 200,000                                      | 1 in 800,000  |              |              |
|                                 |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 254  | 1 in 25,000                                       | 1 in 100,000  |              |              |
|                                 |                                 | South Asian   | 99  | 1 in 235  | 1 in 23,000                                       | 1 in 92,000   |              |              |
|                                 |                                 | Other (population not assigned)   | 99  | 1 in 289  | 1 in 29,000                                       | 1 in 120,000  |              |              |
|                                 |                                 | GRHPR   | African/African American  | 99  | 1 in 1,836  | 1 in 180,000  | 1 in 720,000 |              |
|                                 |                                 |   | Latino/Admixed American   | 98  | 1 in 557  | 1 in 28,000   | 1 in 110,000 |              |
|                                 |                                 |   | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A          |              |
|                                 |                                 |   | East Asian  | 97  | 1 in 288  | 1 in 9,600  | 1 in 38,000  |              |
|                                 |                                 |   | Finnish   | 99  | 1 in 636  | 1 in 64,000   | 1 in 260,000 |              |
|                                 |                                 |   | Non-Finnish European/Caucasian  | 98  | 1 in 353  | 1 in 18,000   | 1 in 72,000  |              |
|                                 | South Asian                     |   | 99  | 1 in 222  | 1 in 22,000                                       | 1 in 88,000   |              |              |
|                                 | Other (population not assigned) |   | 99  | 1 in 573  | 1 in 57,000                                       | 1 in 230,000  |              |              |
|                                 | HOGA1                           |   | African/African American  | 99  | 1 in 287  | 1 in 29,000   | 1 in 120,000 |              |
|                                 |                                 |   | Latino/Admixed American   | 99  | 1 in 267  | 1 in 27,000   | 1 in 110,000 |              |
|                                 |                                 | Ashkenazi Jewish  | 98  | 1 in 37   | 1 in 1,800  | 1 in 7,200  |              |              |
|                                 |                                 | East Asian  | 99  | 1 in 182  | 1 in 18,000                                       | 1 in 72,000   |              |              |
|                                 |                                 | Finnish   | 99  | 1 in 507  | 1 in 51,000                                       | 1 in 200,000  |              |              |
|                                 |                                 | Non-Finnish European/Caucasian  | 99  | 1 in 167  | 1 in 17,000                                       | 1 in 68,000   |              |              |
|                                 |                                 | South Asian   | 99  | 1 in 265  | 1 in 26,000                                       | 1 in 100,000  |              |              |
|                                 |                                 | Other (population not assigned)   | 99  | 1 in 145  | 1 in 14,000                                       | 1 in 56,000   |              |              |
|                                 |                                 | Progressive familial intrahepatic cholestasis   | ABCB11  | African/African American  | 99  | 1 in 510  | 1 in 51,000  | 1 in 200,000 |
|                                 |                                 |   |   | Latino/Admixed American   | 99  | 1 in 512  | 1 in 51,000  | 1 in 200,000 |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |              |              |
|---------------------------------|--|---|---|---|---|---|--------------|--------------|
| Disorder                        |  | Ashkenazi Jewish  | 99  | N/A   | N/A   | N/A   |              |              |
|                                 |  | East Asian  | 99  | 1 in 951  | 1 in 95,000                                       | 1 in 380,000  |              |              |
|                                 |  | Finnish   | 99  | 1 in 1,180  | 1 in 120,000                                      | 1 in 480,000  |              |              |
|                                 |  | Non-Finnish European/Caucasian  | 99  | 1 in 476  | 1 in 48,000                                       | 1 in 190,000  |              |              |
|                                 |  | South Asian   | 99  | 1 in 774  | 1 in 77,000                                       | 1 in 310,000  |              |              |
|                                 |  | Other (population not assigned)   | 99  | 1 in 317  | 1 in 32,000                                       | 1 in 130,000  |              |              |
|                                 | ABCB4                                  | African/African American  | 99  | 1 in 76   | 1 in 7,500  | 1 in 30,000   |              |              |
|                                 |  | Latino/Admixed American   | 99  | 1 in 75   | 1 in 7,400  | 1 in 30,000   |              |              |
|                                 |  | Ashkenazi Jewish  | 98  | 1 in 367  | 1 in 18,000                                       | 1 in 72,000   |              |              |
|                                 |  | East Asian  | 98  | 1 in 673  | 1 in 34,000                                       | 1 in 140,000  |              |              |
|                                 |  | Finnish   | 98  | 1 in 110  | 1 in 5,500  | 1 in 22,000   |              |              |
|                                 |  | Non-Finnish European/Caucasian  | 99  | 1 in 60   | 1 in 5,900  | 1 in 24,000   |              |              |
|                                 |  | South Asian   | 98  | 1 in 113  | 1 in 5,600  | 1 in 22,000   |              |              |
|                                 |  | Other (population not assigned)   | 99  | 1 in 70   | 1 in 6,900  | 1 in 28,000   |              |              |
|                                 |  | ATP8B1  | African/African American  | 99  | 1 in 1,567  | 1 in 160,000  | 1 in 640,000 |              |
|                                 |  |   | Latino/Admixed American   | 98  | 1 in 718  | 1 in 36,000   | 1 in 140,000 |              |
|                                 | Ashkenazi Jewish                       |   | 99  | 1 in 4,925  | 1 in 490,000                                      | 1 in 2,000,000  |              |              |
|                                 | East Asian                             |   | 99  | 1 in 1,127  | 1 in 110,000                                      | 1 in 440,000  |              |              |
|                                 | Finnish                                |   | 97  | 1 in 2,724  | 1 in 91,000                                       | 1 in 360,000  |              |              |
|                                 | Non-Finnish European/Caucasian         |   | 99  | 1 in 738  | 1 in 74,000                                       | 1 in 300,000  |              |              |
|                                 | South Asian                            |   | 99  | 1 in 1,679  | 1 in 170,000                                      | 1 in 680,000  |              |              |
|                                 | Other (population not assigned)        |   | 99  | 1 in 767  | 1 in 77,000                                       | 1 in 310,000  |              |              |
|                                 | Progressive pseudorheumatoid dysplasia |   | CCN6  | African/African American  | 98  | 1 in 643  | 1 in 32,000  | 1 in 130,000 |
|                                 |  |   |   | Latino/Admixed American   | 97  | 1 in 469  | 1 in 16,000  | 1 in 64,000  |
|                                 |  | Ashkenazi Jewish  |   | 97  | 1 in 885  | 1 in 29,000   | 1 in 120,000 |              |
|                                 |  | East Asian  |   | 98  | 1 in 706  | 1 in 35,000   | 1 in 140,000 |              |
| Finnish                         |  | 98  |   | 1 in 2,593  | 1 in 130,000                                      | 1 in 520,000  |              |              |
| Non-Finnish European/Caucasian  |  | 98  |   | 1 in 674  | 1 in 34,000                                       | 1 in 140,000  |              |              |
| South Asian                     |  | 99  |   | 1 in 303  | 1 in 30,000                                       | 1 in 120,000  |              |              |
| Other (population not assigned) |  | 97  |   | 1 in 383  | 1 in 13,000                                       | 1 in 52,000   |              |              |
| Propionic acidemia              |  | PCCA  |   | African/African American  | 99  | 1 in 493  | 1 in 49,000  | 1 in 200,000 |
|                                 |  |   |   | Latino/Admixed American   | 98  | 1 in 348  | 1 in 17,000  | 1 in 68,000  |
|                                 | Ashkenazi Jewish                       |   | 97  | 1 in 548  | 1 in 18,000                                       | 1 in 72,000   |              |              |
|                                 | East Asian                             |   | 98  | 1 in 662  | 1 in 33,000                                       | 1 in 130,000  |              |              |
|                                 | Finnish                                |   | 99  | 1 in 2,722  | 1 in 270,000                                      | 1 in 1,100,000  |              |              |
|                                 | Non-Finnish European/Caucasian         |   | 99  | 1 in 756  | 1 in 76,000                                       | 1 in 300,000  |              |              |
|                                 | South Asian                            |   | 99  | 1 in 522  | 1 in 52,000                                       | 1 in 210,000  |              |              |
|                                 | Other (population not assigned)        |   | 97  | 1 in 846  | 1 in 28,000                                       | 1 in 110,000  |              |              |
|                                 | PCCB                                   |   | African/African American  | 98  | 1 in 238  | 1 in 12,000   | 1 in 48,000  |              |
|                                 |  |   | Latino/Admixed American   | 99  | 1 in 285  | 1 in 28,000   | 1 in 110,000 |              |
|                                 |  | Ashkenazi Jewish  | 97  | 1 in 4,924  | 1 in 160,000                                      | 1 in 640,000  |              |              |
|                                 |  | East Asian  | 99  | 1 in 233  | 1 in 23,000                                       | 1 in 92,000   |              |              |
|                                 |  | Finnish   | 99  | 1 in 83   | 1 in 8,200  | 1 in 33,000   |              |              |
|                                 |  | Non-Finnish European/Caucasian  | 99  | 1 in 144  | 1 in 14,000                                       | 1 in 56,000   |              |              |
|                                 |  | South Asian   | 99  | 1 in 842  | 1 in 84,000                                       | 1 in 340,000  |              |              |

### 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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD<br>and ClinVar except CYP21A2, GBA, and<br>HBA1/HBA2) | Individual Carrier Risk<br>Before Testing (Derived<br>from gnomAD and ClinVar<br>except CYP21A2 and<br>HBA1/HBA2) | Individual Residual Risk After Negative<br>Result | Risk of Affected Fetus When<br>One Partner has a Carrier<br>Result and One Partner has<br>a Negative Result |
|---|---------|---|---|---|---|---|
|   |         | Other (population not assigned)   | 99  | 1 in 251  | 1 in 25,000                                       | 1 in 100,000  |
| Pseudocholinesterase deficiency           | BCHE    | African/African American  | 99  | 1 in 72   | 1 in 7,100  | 1 in 28,000   |
|   |         | Latino/Admixed American   | 99  | 1 in 34   | 1 in 3,300  | 1 in 13,000   |
|   |         | Ashkenazi Jewish  | 99  | 1 in 15   | 1 in 1,400  | 1 in 5,600  |
|   |         | East Asian  | 99  | 1 in 67   | 1 in 6,600  | 1 in 26,000   |
|   |         | Finnish   | 99  | 1 in 36   | 1 in 3,500  | 1 in 14,000   |
|   |         | Non-Finnish European/Caucasian  | 99  | 1 in 17   | 1 in 1,600  | 1 in 6,400  |
|   |         | South Asian   | 99  | 1 in 45   | 1 in 4,400  | 1 in 18,000   |
|   |         | Other (population not assigned)   | 99  | 1 in 19   | 1 in 1,800  | 1 in 7,200  |
| Pycnodysostosis                           | CTSK    | African/African American  | 99  | 1 in 663  | 1 in 66,000                                       | 1 in 260,000  |
|   |         | Latino/Admixed American   | 98  | 1 in 897  | 1 in 45,000                                       | 1 in 180,000  |
|   |         | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|   |         | East Asian  | 97  | 1 in 1,435  | 1 in 48,000                                       | 1 in 190,000  |
|   |         | Finnish   | 99  | 1 in 2,810  | 1 in 280,000                                      | 1 in 1,100,000  |
|   |         | Non-Finnish European/Caucasian  | 99  | 1 in 1,700  | 1 in 170,000                                      | 1 in 680,000  |
|   |         | South Asian   | 99  | 1 in 1,183  | 1 in 120,000                                      | 1 in 480,000  |
|   |         | Other (population not assigned)   | 99  | 1 in 948  | 1 in 95,000                                       | 1 in 380,000  |
| Pyridoxal 5'-phosphate-dependent epilepsy | PNPO    | African/African American  | 98  | 1 in 386  | 1 in 19,000                                       | 1 in 76,000   |
|   |         | Latino/Admixed American   | 99  | 1 in 1,846  | 1 in 180,000                                      | 1 in 720,000  |
|   |         | Ashkenazi Jewish  | 99  | 1 in 5,039  | 1 in 500,000                                      | 1 in 2,000,000  |
|   |         | East Asian  | 97  | 1 in 1,484  | 1 in 49,000                                       | 1 in 200,000  |
|   |         | Finnish   | 99  | 1 in 1,193  | 1 in 120,000                                      | 1 in 480,000  |
|   |         | Non-Finnish European/Caucasian  | 99  | 1 in 1,107  | 1 in 110,000                                      | 1 in 440,000  |
|   |         | South Asian   | 99  | 1 in 1,920  | 1 in 190,000                                      | 1 in 760,000  |
|   |         | Other (population not assigned)   | 99  | 1 in 1,449  | 1 in 140,000                                      | 1 in 560,000  |
| Pyridoxine-dependent epilepsy             | ALDH7A1 | African/African American  | 98  | 1 in 245  | 1 in 12,000                                       | 1 in 48,000   |
|   |         | Latino/Admixed American   | 99  | 1 in 121  | 1 in 12,000                                       | 1 in 48,000   |
|   |         | Ashkenazi Jewish  | 99  | 1 in 309  | 1 in 31,000                                       | 1 in 120,000  |
|   |         | East Asian  | 99  | 1 in 310  | 1 in 31,000                                       | 1 in 120,000  |
|   |         | Finnish   | 99  | 1 in 1,295  | 1 in 130,000                                      | 1 in 520,000  |
|   |         | Non-Finnish European/Caucasian  | 99  | 1 in 88   | 1 in 8,700  | 1 in 35,000   |
|   |         | South Asian   | 99  | 1 in 321  | 1 in 32,000                                       | 1 in 130,000  |
|   |         | Other (population not assigned)   | 99  | 1 in 125  | 1 in 12,000                                       | 1 in 48,000   |
| Pyruvate dehydrogenase deficiency         | DLAT    | African/African American  | 99  | 1 in 1,244  | 1 in 120,000                                      | 1 in 480,000  |
|   |         | Latino/Admixed American   | 97  | 1 in 1,613  | 1 in 54,000                                       | 1 in 220,000  |
|   |         | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|   |         | East Asian  | 97  | 1 in 2,220  | 1 in 74,000                                       | 1 in 300,000  |
|   |         | Finnish   | 99  | 1 in 1,736  | 1 in 170,000                                      | 1 in 680,000  |
|   |         | Non-Finnish European/Caucasian  | 98  | 1 in 1,265  | 1 in 63,000                                       | 1 in 250,000  |
|   |         | South Asian   | 99  | 1 in 1,262  | 1 in 130,000                                      | 1 in 520,000  |
|   |         | Other (population not assigned)   | 99  | 1 in 602  | 1 in 60,000                                       | 1 in 240,000  |
|   | PDHA1   | African/African American  | 97  | N/A   | N/A   | N/A   |
|   |         | Latino/Admixed American   | 97  | N/A   | N/A   | N/A   |
|   |         | Ashkenazi Jewish  | 97  | N/A   | N/A   | N/A   |
|   |         | East Asian  | 97  | N/A   | N/A   | N/A   |

**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 and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is 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<br>(Source:<br><a href="https://gnomad.broadinstitute.org/">https://gnomad.broadinstitute.org/</a><br>except CYP21A2 and HBA1/HBA2) | Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2) | Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2) | Individual Residual Risk After Negative Result | Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result |
|----------|------|---|---|---|--|--|
|          |      |   |   |   |  |  |
| Disorder |      | Finnish   | 97  | N/A   | N/A  | N/A  |
|          |      | Non-Finnish European/Caucasian  | 97  | 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  |
|          | PDHB | African/African American  | 99  | 1 in 8,128  | 1 in 810,000                                   | 1 in 3,200,000   |
|          |      | Latino/Admixed American   | 97  | 1 in 1,352  | 1 in 45,000                                    | 1 in 180,000   |
|          |      | Ashkenazi Jewish  | 97  | 1 in 5,041  | 1 in 170,000                                   | 1 in 680,000   |
|          |      | East Asian  | 97  | 1 in 9,197  | 1 in 310,000                                   | 1 in 1,200,000   |
|          |      | Finnish   | 98  | 1 in 3,587  | 1 in 180,000                                   | 1 in 720,000   |
|          |      | Non-Finnish European/Caucasian  | 98  | 1 in 6,213  | 1 in 310,000                                   | 1 in 1,200,000   |
|          |      | South Asian   | 98  | 1 in 1,797  | 1 in 90,000                                    | 1 in 360,000   |
|          |      | Other (population not assigned)   | 97  | N/A   | N/A  | N/A  |
|          | PDHX | African/African American  | 99  | 1 in 155  | 1 in 15,000                                    | 1 in 60,000  |
|          |      | Latino/Admixed American   | 98  | 1 in 4,294  | 1 in 210,000                                   | 1 in 840,000   |
|          |      | Ashkenazi Jewish  | 97  | 1 in 4,921  | 1 in 160,000                                   | 1 in 640,000   |
|          |      | East Asian  | 99  | 1 in 461  | 1 in 46,000                                    | 1 in 180,000   |
|          |      | Finnish   | 98  | 1 in 2,164  | 1 in 110,000                                   | 1 in 440,000   |
|          |      | Non-Finnish European/Caucasian  | 99  | 1 in 1,524  | 1 in 150,000                                   | 1 in 600,000   |
|          |      | South Asian   | 99  | 1 in 372  | 1 in 37,000                                    | 1 in 150,000   |
|          |      | Other (population not assigned)   | 99  | 1 in 1,242  | 1 in 120,000                                   | 1 in 480,000   |
|          | PDP1 | African/African American  | 99  | 1 in 4,064  | 1 in 410,000                                   | 1 in 1,600,000   |
|          |      | Latino/Admixed American   | 98  | 1 in 8,647  | 1 in 430,000                                   | 1 in 1,700,000   |
|          |      | Ashkenazi Jewish  | 97  | N/A   | N/A  | N/A  |
|          |      | East Asian  | 99  | 1 in 184  | 1 in 18,000                                    | 1 in 72,000  |
|          |      | Finnish   | 97  | 1 in 5,324  | 1 in 180,000                                   | 1 in 720,000   |
|          |      | Non-Finnish European/Caucasian  | 98  | 1 in 4,651  | 1 in 230,000                                   | 1 in 920,000   |
|          |      | South Asian   | 97  | 1 in 15,296   | 1 in 510,000                                   | 1 in 2,000,000   |
|          |      | Other (population not assigned)   | 97  | N/A   | N/A  | N/A  |