

| CONDITION NAME                     | GENE (and Refseq)                    | CARRIER RATES                       |
|------------------------------------|--------------------------------------|-------------------------------------|
| FRAGILE X SYNDROME                 | FMR1 (NM_001185076.1)                | 1 in 178                            |
| ALPHA THALASSEMIA                  | HBA1/HBA2 (NM_000553.4; NM_000517.4) | 1 in 25                             |
| CYSTIC FIBROSIS                    | CFTR (NM_000492.3)                   | 1 in 25                             |
| SPINAL MUSCULAR ATROPHY            | SMN1 (NM_000344.3)                   | 1 IN 35                             |
| NON SYNDROMIC HEARING LOSS         | GJB2 (NM_004004.5)                   | 1 in 42                             |
| DUCHENNE/BECKER MUSCULAR DYSTROPHY | DMD (NM_004006.2)                    | 1 in 500                            |
| BETA-HEMOGLOBINOPATHIES            | HBB (NM_000518.4)                    | 2-3%                                |
| PHENYLKETONURIA                    | PAH (NM_000277.1)                    | Caucasian: 1 in 50; Sicily: 1 in 26 |