

# CGT Basic v2.1

| chrom | OMIM (gene) | gene | previous | OMIM (phen) | disease name (phenotype)  | inheritance  |
|-------|-------------|------|----------|-------------|---|--|
| 7     | 602421      | CFTR |          | 219700      | Cystic fibrosis   | Autosomal recessive                                  |
| X     | 309550      | FMR1 |          | 300624      | Fragile X syndrome  | X-linked   |
| X     | 305900      | G6PD |          | 300908      | Hemolytic anemia, G6PD deficient (favism)                               | X-linked   |
| 13    | 121011      | GJB2 |          | 220290      | Deafness, autosomal recessive, type 1A;<br>Deafness, digenic, GJB2/GJB6 | Autosomal recessive; Digenic inheritance (GJB6 gene) |
| 16    | 141800      | HBA1 |          | 604131      | Thalassemia, alpha-   | Autosomal recessive                                  |
| 16    | 141850      | HBA2 |          | 604131      | Thalassemia, alpha-   | Autosomal recessive                                  |
| 11    | 141900      | HBB  |          | 603903      | HBB-related hemoglobinopathy  | Autosomal recessive                                  |
| 5     | 600354      | SMN1 |          | 253300      | Spinal muscular atrophy   | Autosomal recessive                                  |