

# CGT Bank v2.1

| chrom | OMIM (gene) | Gene   | OMIM (phen)    | DISEASE   | MOI                 |
|-------|-------------|--------|----------------|---|---------------------|
| X     | 300371      | ABCD1  | 300100         | Adrenoleukodystrophy  | X-linked            |
| X     | 313700      | AR     | 300068         | Androgen insensitivity syndrome, complete                                       | X-linked            |
| X     | 300180      | ARSE   | 302950         | Chondrodysplasia punctata, brachytelephalangic                                  | X-linked            |
| X     | 300011      | ATP7A  | 309400; 304150 | Menkes disease; Occipital horn syndrome   | X-linked            |
| X     | 300300      | BTK    | 300755         | Agammaglobulinemia X-linked, type 1   | X-linked            |
| X     | 300386      | CD40LG | 308230         | Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1) | X-linked            |
| 7     | 602421      | CFTR   | 219700         | Cystic fibrosis   | Autosomal recessive |
| X     | 300390      | CHM    | 303100         | Choroideremia   | X-linked            |
| X     | 300481      | CYBB   | 306400         | Chronic granulomatous disease, X-linked   | X-linked            |
| X     | 300451      | EDA    | 305100         | Ectodermal dysplasia, type 1, hypohidrotic, X-linked                            | X-linked            |
| X     | 300384      | EMD    | 310300         | Emery-Dreifuss muscular dystrophy, type 1, X-linked                             | X-linked            |
| X     | 300841      | F8     | 306700         | Hemophilia A  | X-linked            |
| X     | 300746      | F9     | 306900         | Hemophilia B  | X-linked            |
| 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  | Autosomal recessive |
| X     | 300644      | GLA    | 301500         | Fabry disease   | X-linked            |
| X     | 300808      | GPR143 | 300500         | Ocular albinism, type 1 (Nettleship-Falls type)                                 | X-linked            |
| 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 |
| X     | 300823      | IDS    | 309900         | Mucopolysaccharidosis, type 2   | X-linked            |

|   |        |        |                |  |                        |
|---|--------|--------|----------------|--|------------------------|
| X | 308380 | IL2RG  | 300400         | Severe combined immunodeficiency, X-linked           | X-linked               |
| X | 308840 | L1CAM  | 307000; 303350 | L1 Syndrome  | X-linked               |
| X | 300005 | MECP2  | 300673         | Encephalopathy, neonatal severe                      | X-linked               |
| X | 300415 | MTM1   | 310400         | Myotubular myopathy, X-linked                        | X-linked               |
| X | 300473 | NR0B1  | 300200         | Adrenal hypoplasia, congenital                       | X-linked               |
| X | 300535 | OCRL   | 309000; 300555 | Lowe Syndrome; Dent disease type 2                   | X-linked               |
| X | 300461 | OTC    | 311250         | Ornithine transcarbamylase deficiency                | X-linked               |
| X | 300502 | PDHA1  | 312170         | Pyruvate dehydrogenase E1-alpha deficiency           | X-linked               |
| X | 300490 | SH2D1A | 308240         | Lymphoproliferative syndrome, X-linked, type 1       | X-linked               |
| X | 300036 | SLC6A8 | 300352         | Cerebral creatine deficiency syndrome, type 1        | X-linked               |
| 5 | 600354 | SMN1   | 253300         | Spinal muscular atrophy                              | Autosomal<br>recessive |
| X | 300392 | WAS    | 301000; 313900 | Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked | X-linked               |