|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| Name | Ch\_aff | gene\_aff | m\_type | inheritance | description | reference |
| Achondroplasia | 4 | FGFR3 | Gene alteration | Autosomal Dominant |  |  |
| Huntington's disease | 4 | HTT | Gene alteration | Autosomal Dominant |  |  |
| Marfan syndrome | 15 | FBN1 | Gene alteration | Autosomal Dominant |  |  |
| Polycystic kidney disease 1 | 16 | PKD1 | Gene alteration | Autosomal Dominant |  |  |
| Polycystic kidney disease 2 | 4 | PKD2 | Gene alteration | Autosomal Dominant |  |  |
| Familial hypercholesterolemia | 19 | LDLRP1 | Gene alteration | Autosomal Recessive |  |  |
| Sickle cell anaemia | 11 | HBB | Point mutation | Autosomal Recessive |  |  |
| Cystic fibrosis | 7 | CFTR | Gene alteration | Autosomal Recessive |  |  |
| Tay Sachs disease | 15 | HEXA | Gene alteration | Autosomal Recessive |  |  |
| Phenylketonuria | 12 | PAH | Gene alteration | Autosomal Recessive |  |  |
| Galactosemia | 9 | GALT | Gene alteration | Autosomal Recessive |  |  |
| Haemophilia | X | F8 | Gene alteration | X-linked Recessive |  |  |
| Duchenne muscular dystrophy | X | DMD | Gene alteration | X-linked Recessive |  |  |
| Hereditary multiple exostoses | 8  11  19 | EXT1  EXT2  EXT3 | Gene alteration | Autosomal Dominant |  |  |
| Tuberous sclerosis | 9  16 | TSC1  TSC2 | Gene alteration | Autosomal Dominant |  |  |
| Von Willebrand disease | 12 | VWF | Gene alteration | Autosomal Dominant |  |  |
| Albinism, oculocutaneous, type II | 4 | OCA2 | Gene alteration | Autosomal Recessive |  |  |
| Niemann Pick disease | 14 | SMPD1 | Gene alteration | Autosomal Recessive |  |  |
| Spinal muscular atrophy | 5 | SMN1 | Gene alteration | Autosomal Recessive |  |  |
| Roberts syndrome | 8 | ESCO2 | Gene alteration | Autosomal Recessive |  |  |
| Rett syndrome | X | MECP2 | Gene alteration | X-linked dominant |  |  |
| Down syndrome | 21 | Extrachromosome | trisomy | Not inherited |  |  |
| Turner syndrome | X | Missing X chromosome | Loss of chromosome | Not inherited |  |  |
| Aicardi syndrome | X | unknown | Gene alteration | X-linked dominan |  |  |
| Edward syndrome | 18 | Extrachromosome | trisomy | Autosomal Recessive |  |  |