

CGT Basic

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; 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