CGT Essential

Gene	Disease
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
AGXT	Hyperoxaluria, primary, type 1
ARSA	Metachromatic leukodystrophy
BTD	Biotinidase deficiency
CBS	Homocystinuria due to cystathionine beta-synthase
CFTR	Cystic fibrosis
DHCR7	Smith-Lemli-Opitz syndrome
EMD	Emery-Dreifuss muscular dystrophy, type 1, X-linked
FMR1	Fragile X syndrome
GAA	Glycogen storage disease, type 2
GALT	Galactosemia
GLA	Fabry disease
HADHA	LCHAD deficiency
HBB	HBB-related hemoglobinopathy
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
PAH	Phenylketonuria
PMM2	Congenital disorder of glycosylation, type 1A
SLC26A2	Achondrogenesis, type 1B (diastrophic dysplasia)
SMN1	Spinal muscular atrophy