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

Maternal inheritance of a mutation in the UBE3A gene causes Angelman syndrome*
If you need additional information, you can contact us at the following e-mail address: cgtspain@igenomix.com