Table 1: GeneScreen® Focus - List of genes screened and genetic diseases investigated

Gene	OMIM Gene	Disease	OMIM Disease	Inheritance
ACADM	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of	201450	AR
AGXT	604285	Hyperoxaluria, primary, type 1	259900	AR
ARSA	607574	Metachromatic leukodystrophy	250100	AR
ATP7B	606882	Wilson disease	277900	AR
BTD	609019	Biotinidase deficiency	253260	AR
CBS	613381	Homocystinuria	236200	AR
CFTR	602421	Cystic fibrosis	219700	AR
DHCR7	602858	Smith-Lemli-Opitz syndrome	270400	AR
EMD	300384	Emery-Dreifuss muscular dystrophy 1, X-linked	310300	XLR
FMR1	309550	Fragile X syndrome	300624	XLD
GAA	606800	Glycogen storage disease II	232300	AR
GALC	606890	Krabbe disease	245200	AR
GALT	606999	Galactosemia	230400	AR
GBA	606463	Gaucher disease type I-II-III-IIIC-Perinatal Lethal	608013 230800 230900 231000 231005	AR
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked	302800	XLD
GJB2	121011	Deafness, autosomal recessive 1A	220290	AR
GJB6	604418	Deafness, autosomal recessive 1B	612645	AR
GLA	300644	Fabry disease	301500	XL
HADHA	600890	LCHAD deficiency	609016	AR
HBA1	141800	Alpha-Thalassemia	604131	AR
HBA2	141850	Alpha-Thalassemia	604131	AR
HBB	141900	Beta-Thalassemia, Sickle cell anemia	603903 613985	AR
HEXA	606869	Tay-Sachs disease	272800	AR
MEFV	608107	Familial Mediterranean fever	249100	AR
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	277400	AR
PAH	612349	Phenylketonuria	261600	AR

PMM2	601785	Congenital disorder of glycosylation, type Ia	212065	AR
SERPINA1	107400	Alpha-1-Antitrypsin Deficiency	613490	AR
SLC26A2	606718	Achondrogenesis Ib	600972	AR
SMN1	600354	Spinal muscular atrophy (SMA1)	253300	AR