Picture™

PICTURE PARENTING

Genes and Conditions Tested

Gene / Condition Name

ACADM **-** Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
ASPA **-** Canavan disease
ASS1 **-** Citrullinemia
BLM **-** Bloom syndrome
CFTR **-** Cystic fibrosis
CLN3 **-** Neuronal ceroid lipofuscinosis, CLN3-related
DHCR7 **-** Smith-Lemli-Opitz syndrome
DMD* **-** Duchenne muscular dystrophy
FANCC **-** Fanconi anemia group C
FAH **-** Tyrosinemia, type 1
FMR1* **-** Fragile X syndrome
G6PC **-** Glycogen storage disease, type 1a
GAA **-** Pompe disease
GALT **-** Galactosemia
GBA **-** Gaucher disease
HBA1 / HBA2 **-** Alpha thalassemia
HBB **-** Sickle cell disease; beta thalassemia
HEXA **-** Tay-Sachs disease
IDUA **-** Mucopolysaccharidosis, type I (Hurler syndrome)
PAH **-** Phenylalanine hydroxylase deficiency (Phenylketonuria)
IVD **-** Isovaleric acidemia
PEX1 **-** Zellweger syndrome, PEX1-related
PEX7 **-** Rhizomelic chondrodysplasia punctata, type 1
PKHD1 **-** Polycystic kidney disease, PKHD1-related
PMM2 **-** Congenital disorder of glycosylation type 1a
SMN1 **-** Spinal muscular atrophy
SMPD1 **-** Niemann-Pick disease, type A/B
IKBKAP **-** Familial dysautonomia
MMACHC **-** Methylmalonic aciduria and homocystinuria, cblC type

* Male patients will not be screened for X-linked conditions. If an X-linked condition is suspected in a male patient, please contact a genetics professional about diagnostic testing for that particular disorder.

