diseases tested
DISEASES TESTED

Alpha-Thalassemia HBA1, HBA2
Beta-Hemoglobinopathies, including Sickle Cell Anemia HBB
Bloom Syndrome BLM
Canavan Disease ASPA
Congenital Amegakaryocytic Thrombocytopenia MPL
Cystic Fibrosis CFTR
Dihydrolipoamide Dehydrogenase Deficiency DLD
Familial Dysautonomia ELP1
Familial Hyperinsulinism ABCC8
Fanconi Anemia FANCC
Fragile X Syndrome FMR1
Gaucher Disease GBA
Glycogen Storage Disease, Type Ia G6PC
Joubert Syndrome TMEM216
Maple Syrup Urine Disease BCKDHA, BCKDHB
Mucolipidosis, Type IV MCOLN1
Nemaline Myopathy NEB
Niemann-Pick Disease, Type A/B SMPD1
Spinal Muscular Atrophy SMN1
Tay-Sachs Disease HEXA
Usher Syndrome CLRN1, PCDH15
Walker-Warburg Syndrome and Other FKTN-Related Diseases FKTN

WHY TEST FOR THESE DISEASES?

❖ These diseases cause serious health problems, beginning in infancy or childhood.
❖ Children born with these diseases often benefit from early diagnosis and treatment.
❖ Most children born with a genetic disease have no affected family members.
❖ Medical guidelines recommend testing for these diseases.