standard panel
diseases tested
Alpha-thalassemia
Beta-thalassemia
Bloom syndrome
Canavan disease
Congenital amegakaryocytic thrombocytopenia
Cystic fibrosis
Dihydrolipoamide dehydrogenase deficiency
Familial dysautonomia
Familial hyperinsulinism, ABCC8-related
Fanconi anemia, type C
Fragile X syndrome
Gaucher disease
Glycogen storage disease, type IA
Joubert syndrome 2
Maple syrup urine disease, type IA
Maple syrup urine disease, type IB
Mucolipidosis, type IV
Nemaline myopathy 2
Niemann-Pick disease, type A and B
Sickle cell anemia and related diseases
Spinal muscular atrophy
Tay-Sachs disease
Usher syndrome, type IF
Usher syndrome, type III
Walker-Warburg syndrome

About 1 in 8 people are found to be a carrier when tested for these diseases.

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.