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CARRIER TEST



diseases tested

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Some diseases are more common in certain ethnicities, like Ashkenazi Jewish. But these diseases can happen to anyone, regardless of ethnicity.



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*
Dihydroliipoamide 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*
Mucopolipidosis, 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.