

# Disorders Tested

## Standard Panel

### CLINICAL IMPACT AREAS

● Life Expectancy	Decreased life expectancy and increased childhood mortality
● Quality of Life	Severe impact on quality of life; typically no effect on life expectancy
● Treatment Benefits	Early medical intervention can increase life expectancy or reduce symptoms
● Intellectual Ability	Associated with varying degrees of intellectual disability

	Life Expectancy	Quality of Life	Treatment Benefits	Intellectual Ability
Alpha-thalassemia	●		●	
Beta-thalassemia		●	●	
Bloom syndrome	●			●
Canavan disease	●			●
Congenital amegakaryocytic thrombocytopenia	●			
Cystic fibrosis (600 mutations)	●			
Dihydroliipoamide dehydrogenase deficiency	●		●	●
Familial dysautonomia	●			
Familial hyperinsulinism, ABCC8-related		●		
Fanconi anemia, type C	●		●	●
Fragile X syndrome (X-linked)*		●		●
Gaucher disease	●		●	
Glycogen storage disease, type IA	●		●	
Hemoglobinopathy, Hb C		●	●	
Hemoglobinopathy, Hb D		●	●	
Hemoglobinopathy, Hb E		●	●	
Hemoglobinopathy, Hb O		●	●	
Joubert syndrome 2	●			●
Maple syrup urine disease, type IA	●		●	●
Maple syrup urine disease, type IB	●		●	●
Mucopolipidosis, type IV		●		●
Nemaline myopathy 2	●			
Niemann-Pick disease, type A and B	●			●
Sickle cell anemia (Hb S)	●		●	
Spinal muscular atrophy	●		●	
Tay-Sachs disease	●			●
Usher syndrome, type IF		●	●	
Usher syndrome, type III		●	●	
Walker-Warburg syndrome	●			●

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\*X-linked disorders are not tested in males.

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