



## GUIDELINES-BASED PANEL

Single panel for 19 genetic diseases recommended for testing by international organizations, ACMG and ACOG\*, due to their high incidence and severity.

<p><b>Alpha Thalassemia, HBA1, HBA2</b></p> <p>Carrier Frequency: 1 in 25 General Population</p> <p>Early onset, Interventions &amp; Therapy, Life quality, Life-threatening</p>	<p><b>Beta Thalassemia, HBB</b></p> <p>Carrier Frequency: 1 in 28 Mediterranean</p> <p>Early onset, Interventions &amp; Therapy, Life quality, Life-threatening</p>	<p><b>Bloom Syndrome, BLM</b></p> <p>Carrier Frequency: 1 in 100 Ashkenazi Jewish &lt;1 in 500 General Population</p> <p>Cognitive and Developmental, Interventions &amp; Therapy, Life quality, Life-threatening, Multi-organ, Predisposition to malignancy, Reduced lifespan</p>
<p><b>Canavan Disease, ASPA</b></p> <p>Carrier Frequency: 1 in 57 Ashkenazi Jewish 1 in 3392 European</p> <p>Cognitive &amp; Developmental, Interventions &amp; Therapy, Life quality, Motor development, Progressive</p>	<p><b>Cystic Fibrosis, CFTR</b></p> <p>Carrier Frequency: 1 in 20 European 1 in 45 General Population</p> <p>Early onset, Interventions &amp; Therapy, Life quality, Life-threatening, Multi-organ, Progressive</p>	<p><b>Duchenne Muscular Dystrophy, X-linked, DMD</b></p> <p>Carrier Frequency: &lt;1 in 500</p> <p>Early onset, Interventions &amp; Therapy, Life quality, Life-threatening, Motor development, Progressive, Reduced lifespan</p>
<p><b>Familial Dysautonomia, IKBKAP</b></p> <p>Carrier Frequency: 1 in 31 Ashkenazi Jewish</p> <p>Cognitive &amp; Developmental, Early onset, Failure to thrive, Interventions &amp; Therapy, Life quality, Multi-organ</p>	<p><b>Fanconi Anemia, Type C, FANCC</b></p> <p>Carrier Frequency: 1 in 89 Ashkenazi Jewish 1 in 417 General Population</p> <p>Early onset, Interventions &amp; Therapy, Life quality, Life-threatening, Progressive, Multi-Organ, Predisposition to malignancy</p>	<p><b>Fragile X Syndrome, X-Linked, FMRI</b></p> <p>Carrier Frequency: 1 in 102 Ashkenazi Jewish 1 in 201 General Population</p> <p>Cognitive &amp; Developmental, Early onset, Interventions &amp; Therapy, Motor development</p>
<p><b>Galactosemia, GALT</b></p> <p>Carrier Frequency: 1 in 100 General Population</p> <p>Cognitive &amp; Developmental, Early onset, Interventions &amp; Therapy, Life quality, Life-threatening</p>	<p><b>Gaucher Disease, GBA</b></p> <p>Carrier Frequency: 1 in 158 General Population</p> <p>Early onset, Interventions &amp; Therapy, Life quality, Life-threatening, Multi-organ</p>	<p><b>Medium Chain Acyl-CoA Dehydrogenase Deficiency, ACADM</b></p> <p>Carrier Frequency: 1 in 66 General Population</p> <p>Early onset, Interventions &amp; Therapy, Life quality, Life-threatening</p>
<p><b>Mucopolidiosis, Type IV, MCOLN1</b></p> <p>Carrier Frequency: 1 in 100 Ashkenazi Jewish &lt;1 in 500 General Population</p> <p>Cognitive &amp; Developmental, Early onset, Interventions &amp; Therapy, Life quality, Motor development, Progressive, Reduced lifespan</p>	<p><b>Niemann-Pick Disease, Types A/B, SMPD</b></p> <p>Carrier Frequency: 1 in 115 Ashkenazi Jewish 1 in 250 General Population</p> <p>Early onset, Failure to thrive, Interventions &amp; Therapy, Life quality, Life-threatening, Multi-organ, Progressive, Reduced lifespan</p>	<p><b>Phenylketonurea, PAH</b></p> <p>Carrier Frequency: 1 in 50 European 1 in 140 General Population</p> <p>Cognitive &amp; Developmental, Early onset, Interventions &amp; Therapy</p>
<p><b>Sickle-Cell Disease, HBB</b></p> <p>Birth prevalence: 112 per 100,000 Globally</p> <p>Early onset, Interventions &amp; Therapy, Life quality, Life-threatening, Multi-organ</p>	<p><b>Spinal Muscular Atrophy, SMN1, SMN2</b></p> <p>Carrier Frequency: 1 in 35 Caucasian 1 in 41 Ashkenazi Jewish</p> <p>Early Onset, Failure to thrive, Interventions &amp; Therapy, Reduced lifespan, Progressive, Motor development, Life quality, Life-threatening, Reduced lifespan</p>	<p><b>Smith-Lemli-Opitz Syndrome, DHCR7</b></p> <p>Carrier Frequency: 1 in 54 Northern European 1 in 66 Southern European, 1 in 71 General Population</p> <p>Cognitive &amp; Developmental, Early Onset, Interventions &amp; Therapy, Life quality, Life-threatening, Multi-Organ</p>
<p><b>Tay-Sachs Disease, HEXA</b></p> <p>Carrier Frequency: 1 in 25 Ashkenazi Jewish, 1 in 250 General Population</p> <p>Early onset, Life-threatening, Progressive, Reduced lifespan</p>		

DISEASE CHARACTERISTICS	
Cognitive & Developmental	Varying degrees of reduced intellectual ability, or developmental delays
Early onset	Symptoms originate from birth, in infancy or childhood
Interventions & Therapy	Medical, physical or supportive therapies are necessary and may be available to support or improve quality of life
Life quality	Impact on quality of life
Life-threatening	Disease can be fatal
Failure to thrive	Difficulty eating, swallowing resulting in poor growth
Motor development	Reduced ability to move, delayed development of movement or muscle weakness
Multi-organ	Disease affects multiple organs
Predisposition to malignancy	Increased risk of developing cancer
Progressive	Symptoms become worse over the years
Reduced lifespan	Decreased life expectancy

The above diseases are also included in the Adventia Comprehensive Panel.

\* American College of Obstetricians and Gynaecologists (ACOG), American College of Medical Genetics and Genomics (ACMG).