



FOCUS PANELS

Six individual panels for high frequency and severe genetic diseases.

<p>Alpha Thalassemia, HBA1, HBA2</p> <p>Carrier Frequency: 1 in 25 General Population</p> <ul style="list-style-type: none"> • Early onset • Interventions & Therapy • Life quality • Life-threatening 	<p>B-Haemoglobinopathies, HBB</p>	
<p>Cystic Fibrosis, CFTR</p> <p>Carrier Frequency: 1 in 20 European 1 in 45 General Population</p> <ul style="list-style-type: none"> • Early onset • Interventions & Therapy • Life quality • Life-threatening • Multi-organ • Progressive 	<p>B-Thalassemia</p> <p>Carrier Frequency: 1 in 28 Mediterranean</p> <ul style="list-style-type: none"> • Early onset • Interventions & Therapy • Life quality • Life-threatening 	<p>Sickle-Cell Disease</p> <p>Birth prevalence: 112 per 100,000 Globally</p> <ul style="list-style-type: none"> • Early onset • Interventions & Therapy • Life quality • Life-threatening • Multi-organ
<p>Spinal Muscular Atrophy, SMN1, SMN2</p> <p>Carrier Frequency: 1 in 35 Caucasian 1 in 41 Ashkenazi Jewish</p> <ul style="list-style-type: none"> • Early Onset • Failure to thrive • Interventions & Therapy 	<p>Duchenne Muscular Dystrophy, X-linked, DMD</p> <p>Carrier Frequency: <1 in 500 General Population</p> <ul style="list-style-type: none"> • Early onset • Interventions & Therapy • Life quality • Life-threatening • Motor development • Progressive • Reduced lifespan 	<p>Fragile X Syndrome, X-Linked, FMR1</p> <p>Carrier Frequency: 1 in 102 Ashkenazi Jewish 1 in 201 General Population</p> <ul style="list-style-type: none"> • Early onset • Cognitive & Developmental • Interventions & Therapy • Motor development • Progressive • Reduced lifespan

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 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
Progressive	Symptoms become worse over the years
Reduced lifespan	Decreased life expectancy

The above diseases are also included in the Adventia Guidelines-Based Panel and Comprehensive Panel.