Adventia®
carrier screening

FOCUS PANELS

Six individual panels for highly frequent and severe genetic diseases.

Alpha Thalassemia, *HBA1, HBA2*

Carrier Frequency: 1 in 25 General Population

- *Early onset*
- *Interventions & Therapy*
- *Life quality*
- *Life-threatening*

Detection Rate: >90% of affected cases

B-Haemoglobinopathies, *HBB*

B-Thalassemia

Carrier Frequency: 1 in 28 Mediterranean

- *Early onset*
- *Interventions & Therapy*
- *Life quality*
- *Life-threatening*

Detection Rate: >95% of affected cases

Sickle-Cell Disease

Birth prevalence: 112 per 100,000 Globally

- *Early onset*
- *Interventions & Therapy*
- *Life quality*
- *Life-threatening*
- *Multi-organ*

Detection Rate: >95% of affected cases

Cystic Fibrosis, *CFTR*

Carrier Frequency: 1 in 20 European
1 in 45 General Population

- *Early onset*
- *Interventions & Therapy*
- *Life quality*
- *Life-threatening*
- *Multi-organ*
- *Progressive*

Detection Rate: >75% of affected cases

Duchenne Muscular Dystrophy, *X-linked, DMD*

Carrier Frequency: <1 in 500 General Population

- *Early onset*
- *Interventions & Therapy*
- *Life quality*
- *Life-threatening*
- *Motor development*
- *Progressive*
- *Reduced lifespan*

Detection Rate: >75% of affected cases

Fragile X Syndrome, *X-Linked, FMR1*

Carrier Frequency: 1 in 102 Ashkenazi Jewish
1 in 201 General Population

- *Early onset*
- *Cognitive & Developmental*
- *Interventions & Therapy*
- *Motor development*

Detection Rate: >99% of affected cases

Spinal Muscular Atrophy, *SMN1, SMN2*

Carrier Frequency: 1 in 35 Caucasian
1 in 41 Ashkenazi Jewish

- *Early Onset*
- *Failure to thrive*
- *Interventions & Therapy*
- *Life quality*
- *Life-threatening*
- *Motor development*
- *Progressive*
- *Reduced lifespan*

Detection Rate: >75% of affected cases

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 Core Panel** and **Comprehensive Panel**, which screen for all coding regions on the genes of interest, offering increased detection rates. *Exceptions include Alpha-Thalassemia, Spinal Muscular Atrophy and Fragile X Syndrome, which have the same detection rates in all panels offered.*

NIPD GENETICS
A MEDICOVER COMPANY

Neas Engomis 31, Nicosia, 2409 Cyprus
Phone: +357 22266888
Fax: +357 22266899

Web: http://www.nipd.com
Email: info@nipd.com

MKT-REP-ADV-FCS-DR-EN-V3.3