Adventia®
carrier screening

CORE PANEL

Single panel that tests for 20 selected genetic diseases of high incidence and severity

Alpha Thalassemia, *HBA1, HBA2*

Carrier Frequency: 1 in 25 General Population

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

Beta Thalassemia, *HBB*

Carrier Frequency: 1 in 28 Mediterranean

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

Bloom Syndrome, *BLM*

Carrier Frequency: 1 in 100 Ashkenazi Jewish
<1 in 500 General Population

Cognitive and Developmental, Interventions &Therapy, Life quality, Life-threatening, Multi-organ, Predisposition to malignancy, Reduced lifespan

Canavan Disease, *ASPA*

Carrier Frequency: 1 in 57 Ashkenazi Jewish
1 in 3392 European

Cognitive & Developmental, Interventions & Therapy, Life quality, Motor development, Progressive

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

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

Familial Dysautonomia, *IKBKAP*

Carrier Frequency: 1 in 31 Ashkenazi Jewish

Cognitive & Developmental, Early onset, Failure to thrive, Interventions & Therapy, Life quality, Multi-organ

Fanconi Anemia, Type C, *FANCC*

Carrier Frequency: 1 in 89 Ashkenazi Jewish
1 in 417 General Population

Early onset, Interventions & Therapy, Life quality, Life-threatening, Progressive, Multi-Organ, Predisposition to malignancy

Fragile X Syndrome, *X-Linked, FMR1*

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

Cognitive & Developmental, Early onset, Interventions & Therapy, Motor development

Galactosemia, *GALT*

Carrier Frequency: 1 in 100 General Population

Cognitive & Developmental, Early onset, Interventions & Therapy, Life quality, Life-threatening

Gaucher Disease, *GBA*

Carrier Frequency: 1 in 158 General Population

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

Medium Chain Acyl-CoA Dehydrogenase Deficiency, *ACADM*

Carrier Frequency: 1 in 66 General Population

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

Mucolipidosis, Type IV, *MCOLN1*

Carrier Frequency: 1 in 100 Ashkenazi Jewish
<1 in 500 General Population

Cognitive & Developmental, Early onset, Interventions & Therapy, Life quality, Motor development, Progressive, Reduced lifespan

Niemann-Pick Disease, Types A/B, *SMPD1*

Carrier Frequency: 1 in 115 Ashkenazi Jewish
1 in 250 General Population

Early onset, Failure to thrive, Interventions & Therapy, Life quality, Life-threatening, Multi-organ, Progressive, Reduced lifespan

Non-Syndromic Hearing Loss GJB2-Related and GJB6-Related, *GJB2, GJB6*

Carrier Frequency: 1 in 50 General Population (GJB2)
1 in 423 General Population (GJB6)

Early onset, Interventions & Therapy

Phenylketonurea, *PAH*

Carrier Frequency: 1 in 50 European
1 in 140 General Population

Cognitive & Developmental, Early onset, Interventions & Therapy

Sickle-Cell Disease, *HBB*

Birth prevalence: 112 per 100,000 Globally

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

Spinal Muscular Atrophy, *SMN1, SMN2*

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

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

Smith-Lemli-Opitz Syndrome, *DHCR7*

Carrier Frequency: 1 in 54 Northern European
1 in 66 Southern European,
1 in 71 General Population

Cognitive & Developmental, Early Onset, Interventions & Therapy, Life quality, Life-threatening, Multi-Organ

Tay-Sachs Disease, *HEXA*

Carrier Frequency: 1 in 25 Ashkenazi Jewish, 1 in 250 General Population

Early onset, Life-threatening, 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 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

All the above diseases are included in either Tier 1, Tier 2 or Tier 3 of the carrier screening system proposed by the American College of Medical Genetics and Genomics (ACMG). All the diseases tested in the Core Panel are also included in the Adventia Comprehensive Panel.

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

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