Adventia®
carrier screening

COMPREHENSIVE PANEL

231 genes associated with genetic diseases that have moderate to severe symptoms

3-Hydroxy-3-Methylglutaryl- Coenzyme A Lyase Deficiency, *HMGCL*	3-Methylcrotonyl-CoA Carboxylase Deficiency 1, *MCCC1*	3-Methylcrotonyl-CoA Carboxylase Deficiency 2, *MCCC2*
3-Methylglutaconic Aciduria, Type 3 [Costeff Syndrome], *OPA3*	3-Phosphoglycerate Dehydrogenase Deficiency, *PHGDH*	6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency, *PTS*
Abetalipoproteinemia, *MTTP*	Achondrogenesis, Type 1B, *SLC26A2*	Achromatopsia (CNGB3-related), *CNGB3*
Acute Infantile Liver Failure (TRMU-related), *TRMU*	Acyl-CoA Oxidase I Deficiency, *ACOX1*	Adrenoleukodystrophy, X-Linked, *ABCD1*
Aicardi-Goutières Syndrome, *SAMHD1*	Alpha Thalassemia, *HBA1, HBA2* •	Alport Syndrome (COL4A3-related), *COL4A3*
Alport Syndrome, X-Linked, *COL4A5*	Alstrom Syndrome, *ALMS1*	Andermann Syndrome, *SLC12A6*
Argininosuccinate Lyase Deficiency, *ASL*	Aromatase Deficiency, *CYP19A1*	Arthrogryposis Mental Retardation Seizures, *SLC35A3*
Asparagine Synthetase Deficiency, *ASNS*	Aspartylglycosaminuria, *AGA*	Ataxia with Vitamin E Deficiency, *TTPA*
Ataxia-Telangiectasia, *ATM*	Autoimmune Polyglandular Syndrome, Type 1, *AIRE*	Autosomal Recessive Polycystic Kidney Disease, *PKHD1*
Autosomal Recessive Spastic Ataxia of Charlevoix- Saguenay, *SACS*	Bardet-Biedl Syndrome (BBS1-related), *BBS1*	Bardet Biedl syndrome (BBS12-related), *BBS12*
Bare Lymphocyte Syndrome (CIITA-related), *CIITA*	Bartter Syndrome (BSND-related), *BSND*	Batten Disease (CLN3-related), *CLN3*
Becker Muscular Dystrophy, *DMD* •	Beta Thalassemia, *HBB* •	Biotinidase Deficiency, *BTD*
Bloom Syndrome, *BLM* ◆	Canavan Disease, *ASPA* ◆	Carnitine Palmitoyltransferase IA Deficiency, *CPT1A*
Carnitine Palmitoyltransferase II Deficiency, *CPT2*	Carpenter Syndrome, *RAB23*	Cartilage-Hair Hypoplasia, *RMRP*
Cerebrotendinous Xanthomatosis, *CYP27A1*	Choreacanthocytosis, *VPS13A*	Choroideremia, X-Linked, *CHM*
Chronic Granulomatous Disease, X-Linked, *CYBB*	Citrin Deficiency, *SLC25A13*	Citrullinemia, Type 1, *ASS1*
Combined Malonic and Methylmalonic Acidemia, *ACSF3*	Combined Oxidative Phosphorylation Deficiency 1, *GFM1*	Combined Oxidative Phosphorylation Deficiency 3, *TSFM*
Combined Pituitary Hormone Deficiency 2, *PROP1*	Congenital Disorder of Glycosylation, Type 1A (PMM2- related), *PMM2*	Congenital Disorder of Glycosylation, Type 1B, *MPI*
Congenital Disorder of Glycosylation Type 1C, *ALG6*	Congenital Finnish Nephrosis, *NPHS1*	Congenital Insensitivity to Pain with Anhidrosis, *NTRK1*
Congenital Myasthenic Syndrome (CHRNE-related), *CHRNE*	Congenital Myasthenic Syndrome (CHRNE-related), *CHRNE*	Congenital Neutropenia (HAX1-related), *HAX1*
Congenital Neutropenia (VPS45-related), *VPS45*	Corneal Dystrophy and Perceptive Deafness, *SLC4A11*	Corticosterone Methyloxidase Deficiency, *CYP11B2*
CRB1-related Retinal Dystrophies, *CRB1*	Creatine Transporter Defect [Cerebral Creatine Deficiency Syndrome 1] X-Linked, *SLC6A8*	Crigler Najjar syndrome, Type I, *UGT1A1*
Cystic Fibrosis, *CFTR* •	Cystinosis, *CTNS*	D-Bifunctional Protein Deficiency, *HSD17B4*
Deafness, Autosomal Recessive 77, *LOXHD1*	Duchenne Muscular Dystrophy, X-linked, *DMD* •	Dystrophic Epidermolysis Bullosa (COL7A1-related), *COL7A1*
Ehlers-Danlos Syndrome, Type VIIC, *ADAMTS2*	Emery-Dreifuss Muscular Dystrophy 1, X-Linked, *EMD*	Enhanced S-Cone Syndrome, *NR2E3*
Ethylmalonic Encephalopathy, *ETHE1*	Fabry Disease, X-Linked, *GLA*	Factor IX Deficiency, X-Linked, *F9*
Factor V Leiden Thrombophilia, *F5*	Factor XI Deficiency, *F11*	Familial Dysautonomia, *ELP1* ◆
Familial Hypercholesterolemia (LDLR-related), *LDLR*	Familial Mediterranean Fever, *MEFV*	Familial Nephrogenic Diabetes Insipidus (AQP2- related), *AQP2*
Fanconi Anemia, Type G, *FANCG*	Fanconi Anemia, Type C, *FANCC* ◆	Fragile X Syndrome, X-Linked, *FMR1* •
Galactokinase Deficiency [Galactosemia, Type II], *GALK1*	Galactosemia, *GALT* ◆	Gaucher Disease, *GBA* ◆

Glutaric Acidemia, Type 1, *GCDH*	Glutaric Acidemia, Type 2A, *ETFA*	Glycine Encephalopathy (GLDC-related), *GLDC*
Glycine Encephalopathy (AMT-related), *AMT*	Glycogen Storage Disease, Type 1A, *G6PC*	Glycogen Storage Disease, Type 1B, *SLC37A4*
Glycogen Storage Disease, Type 2 [Pompe Disease], *GAA*	Glycogen Storage Disease, Type 3, *AGL*	Glycogen Storage Disease, Type 4, *GBE1*
Glycogen Storage Disease, Type 5 [McArdle Disease], *PYGM*	Glycogen Storage Disease, Type 7, *PFKM*	GRACILE Syndrome, *BCS1L*
Hemochromatosis, Type 2A, *HFE2*	Hemochromatosis, Type 3 (TFR2-related), *TFR2*	Hereditary Fructose Intolerance, *ALDOB*
Hermansky-Pudlak Syndrome (HPS1-related), *HPS1*	Hermansky-Pudlak Syndrome (HPS3-related), *HPS3*	Holocarboxylase Synthetase Deficiency, *HLCS*
Homocystinuria due to cystathionine beta-synthase deficiency, *CBS*	Homocystinuria-Megaloblastic Anemia, cblE Type, *MTRR*	Hydrolethalus Syndrome, *HYLS1*
Hypohidrotic Ectodermal Dysplasia, X-Linked, *EDA*	Hypophosphatasia (ALPL-related), *ALPL*	Inclusion Body Myopathy Type 2, *GNE*
Isovaleric Acidemia, *IVD*	Joubert Syndrome, Type 2, *TMEM216*	Junctional Epidermolysis Bullosa, Herlitz type, *LAMC2*
Juvenile Retinoschisis, X-Linked, *RS1*	Krabbe Disease, *GALC*	Lamellar Ichthyosis, Type 1, *TGM1*
Leber Congenital Amaurosis (LCA5-related), *LCA5*	Leber Congenital Amaurosis, Type CEP290, *CEP290*	Leigh Syndrome, French-Canadian Type, *LRPPRC*
Leukoencephalopathy with Vanishing White Matter, *EIF2B5*	Leydig Cell Hypoplasia [Luteinizing Hormone Resistance], *LHCGR*	Limb-Girdle Muscular Dystrophy, Type 2A, *CAPN3*
Limb-Girdle Muscular Dystrophy, Type 2B, *DYSF*	Limb-Girdle Muscular Dystrophy, Type 2C, *SGCG*	Limb-Girdle Muscular Dystrophy, Type 2D, *SGCA*
Limb-Girdle Muscular Dystrophy, Type 2E, *SGCB*	Lipoamide Dehydrogenase Deficiency [Maple Syrup Urine Disease, Type 3], *DLD*	Lipoid Adrenal Hyperplasia, *STAR*
Lipoprotein Lipase Deficiency, *LPL*	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency, *HADHA*	Lysinuric Protein Intolerance, *SLC7A7*
Maple Syrup Urine Disease, Type 1B, BCKDHB	Meckel-Gruber Syndrome, Type 1, *MKS1*	Medium Chain Acyl-CoA Dehydrogenase Deficiency, *ACADM* ◆
Megalencephalic Leukoencephalopathy with Subcortical Cysts, MLC1	Metachromatic Leukodystrophy (ARSA-related), *ARSA*	Metachromatic Leukodystrophy (PSAP-related), *PSAP*
Methylmalonic Acidemia, cblA Type, *MMAA*	Methylmalonic Acidemia, cblB Type, *MMAB*	Methylmalonic Acidemia and Homocystinuria, cblC Type, *MMACHC*
Methylmalonic acidemia and homocystinuria, cblD Type, *MMADHC*	Methylmalonic acidemia due to Methylmalonyl-CoA Mutase Deficiency, *MMUT*	Microphthalmia/Anophthalmia (VSX2-related), *VSX2*
Mitochondrial Complex 1 Deficiency (ACAD9-related), *ACAD9*	Mitochondrial Complex 1 Deficiency (NDUFAF5- related), *NDUFAF5*	Mitochondrial Complex 1 Deficiency (NDUFS6-related), *NDUFS6*
Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1), *PUS1*	Mucolipidosis II/III, *GNPTAB*	Mucolipidosis III Gamma, *GNPTG*
Mucolipidosis, Type IV, *MCOLN1* ◆	Mucopolysaccharidosis, Type II [Hunter Syndrome], X-Linked, *IDS*	Mucopolysaccharidosis, Type IIIB [Sanfilippo B], *NAGLU*
Mucopolysaccharidosis, Type IIIC [Sanfilippo C], *HGSNAT*	Mucopolysaccharidosis IIID [Sanfilippo D], *GNS*	Mucopolysaccharidosis, Type IX, *HYAL1*
Multiple Sulfatase Deficiency, *SUMF1*	Myoneurogastrointestinal Encephalopathy (MNGIE), *TYMP*	Myotubular Myopathy, X-Linked, *MTM1*
N-acetylglutamate Synthase Deficiency, *NAGS*	Navajo Neurohepatopathy [MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome], *MPV17*	Neurological Ceroid Lipofuscinosis, TPP1-related, *TPP1*
Neuronal Ceroid Lipofuscinosis (MFSD8-related), *MFSD8*	Neuronal Ceroid Lipofuscinosis (CLN5-related), *CLN5*	Neuronal Ceroid Lipofuscinosis (CLN6-related), *CLN6*
Neuronal Ceroid Lipofuscinosis (CLN8-related), *CLN8*	Neuronal Ceroid Lipofuscinosis (PPT1-related), *PPT1*	Niemann-Pick Disease, Types A/B, *SMPD1* ◆
Niemann-Pick Disease, Type C1/D, *NPC1*	Niemann-Pick Disease, Type C2, *NPC2*	Nijmegen Breakage Syndrome, *NBN*
Non-Syndromic Hearing Loss (GJB2-related, GJB6-related), *GJB2, GJB6*	Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz- Passarge Syndrome, *WNT10A*	Omenn Syndrome, RAG2-related, *RAG2*
Ornithine Aminotransferase Deficiency, *OAT*	Ornithine Transcarbamylase Deficiency, *OTC*	Ornithine Translocase Deficiency [Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome], *SLC25A15*

Pendred Syndrome, *SLC26A4*	Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related), *PEX1*	Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related), *PEX2*
Phenylalanine Hydroxylase Deficiency, *PAH* ◆	Pituitary Hormone Deficiency, Combined 3, *LHX3*	Pontocerebellar Hypoplasia, RARS2-related, *RARS2*
Pontocerebellar Hypoplasia, Type 1A, *VRK1*	Pontocerebellar Hypoplasia, Type 2D, *SEPSECS*	Pontocerebellar Hypoplasia, Type 2E, *VPS53*
Primary Ciliary Dyskinesia (DNAH5-related), *DNAH5*	Primary Ciliary Dyskinesia, DNAI1-related, *DNAI1*	Primary Ciliary Dyskinesia, DNAI2-related, *DNAI2*
Primary Hyperoxaluria, Type 1, *AGXT*	Primary Hyperoxaluria, Type 2, *GRHPR*	Primary Hyperoxaluria, Type 3, *HOGA1*
Pycnodysostosis, *CTSK*	Pyruvate Dehydrogenase Deficiency (PDHB-related), *PDHB*	Pyruvate Dehydrogenase Deficiency, X-Linked, *PDHA1*
Renal Tubular Acidosis and Deafness (ATP6V1B1- related), *ATP6V1B*	Retinal Dystrophy (RLBP1-related) [Bothnia Retinal Dystrophy], *RLBP1*	Retinitis Pigmentosa 59 (DHDDS-related), *DHDDS*
Retinitis Pigmentosa 25 (EYS-related), *EYS*	Retinitis Pigmentosa 26, *CERKL*	Retinitis Pigmentosa 28, *FAM161A*
Retinitis Pigmentosa, X-linked, *RPGR*	Rhizomelic Chondrodysplasia Punctata, Type 1, *PEX7*	Rhizomelic Chondrodysplasia Punctata, Type 3, *AGPS*
Roberts Syndrome, *ESCO2*	Salla Disease, *SLC17A5*	Sandhoff Disease, *HEXB*
Schimke Immunoosseous Dysplasia, *SMARCAL1*	Segawa Syndrome, (TH-related), *TH*	Severe Combined Immunodeficiency, Type Athabaskan, *DCLRE1C*
Severe Combined Immunodeficiency, X-Linked, *IL2RG*	Sickle-Cell Disease, *HBB* •	Sjögren-Larsson Syndrome, *ALDH3A2*
Smith-Lemli-Opitz Syndrome, *DHCR7* ◆	Spinal Muscular Atrophy, *SMN1, SMN2* •	Steroid-Resistant Nephrotic Syndrome, *NPHS2*
Stuve-Wiedemann Syndrome, *LIFR*	Tay-Sachs Disease, *HEXA* ◆	Tyrosinemia, Type 1, *FAH*
Usher Syndrome, Type 1C, *USH1C*	Usher Syndrome, Type 1F, *PCDH15*	Usher Syndrome, Type 2A, *USH2A*
Usher Syndrome, Type 3, *CLRN1*	Wilson Disease, *ATP7B*	Wolman Disease, *LIPA*
Zellweger Spectrum Disorders, (PEX6-related), *PEX6*	Zellweger Spectrum Disorders (PEX10-related), PEX10	

• AVAILABLE AS FOCUS PANELS AND AS PART OF THE CORE PANEL ◆ INCLUDED IN THE CORE PANEL

MEDICOVER GENETICS
Neas Engomis 31, Nicosia, 2409 Cyprus
Phone: +357 22266888 Fax: +357 22266899
Web: www.medicover-genetics.com
Email: info.genetics@medicover.com