NIPD GENETICS

Adventia®

carrier screening

COMPREHENSIVE PANEL

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 12, *BBS12*
Bare Lymphocyte Syndrome (CIITA-related), *CIITA*	Bartter Syndrome (BSND-related), *BSND*	Batten Disease (CLN3-related), *CLN3*
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 Aciduria, *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 (RAPSN-related), *RAPSN*	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, *ADAMTS*
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, *IKBKAP* ◆	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 (CBS-related), *CBS*
Homocystinuria, Type cblE, *MTRR*	Hydrolethalus Syndrome, *HYLS1*	Hypohidrotic Ectodermal Dysplasia, *X-Linked*, *EDA*
Hypophosphatasia (ALPL-related), *ALPL*	Inclusion Body Myopathy Type 2, *GNE*	Isovaleric Acidemia, *IVD*

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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 2C, *SGCG*
Limb-Girdle Muscular Dystrophy, Type 2B, *DYSF*	Limb-Girdle Muscular Dystrophy, Type 2C, *SGCG*	Limb-Girdle Muscular Dystrophy, Type 2D, *SGCA*
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 Aciduria (MMAA-related), *MMAA*
Methylmalonic Aciduria (MMAB-related), *MMAB*	Methylmalonic Aciduria and Homocystinuria, Type cblC, *MMACHC*	Methylmalonic Aciduria and Homocystinuria, Type cblD, *MMADHC*
Methylmalonic Aciduria, Type mut(0), *MUT*	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), *GJB2*
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*	Phenylketonurea, *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], *RLBP*	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 (PEX10-related), *PEX10*
Zellweger Spectrum Disorders, PEX6-related, *PEX6*		

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