# Horizon™
## BY NeoGenetic

**¿Dónde se realiza en Paraguay y qué condiciones detecta?**

El análisis internacional que te permite planificar
la concepción y conocer lo que tu hijo podría heredar.

NeoGenetic

# ¿Dónde realizamos el Test de portadores Horizon™ ?

- **Asunción**
- **Ciudad del Este**
- **Encarnación**
- **P.J. Caballero**

Consultanos por tu localidad

**consultar por servicio a domicilio*

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# Conocé todos los paneles disponibles

- Horizon™ 27 Genes
- Horizon™ 106Genes
- Horizon™ 274 Genes
- Horizon™ 421 Genes
- Horizon™ 445 a 835 Genes
- Horizon™ 4 Genes

Diseñados para detectar enfermedades graves y con posibilidad de acción médica, según guías internacionales

# Horizon Single-Gene PC# List

Horizon™
Advanced carrier screening

| CONDITION | GENE | AUTOSOMAL RECESSIVE | X-LINKED | SCREENING RECOMMENDATIONS | | PANEL AVAILABILITY | | | | PC# |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | ACOG* | ACMG | H445 | H569 † | H574 † | H613 | |
| 17-Beta Hydroxysteroid Dehydrogenase 3 Deficiency | HSD17B3 | • | | | | • | • | • | • | PC2.1 |
| 3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency | HSD3B2 | • | | | | • | • | • | • | PC3.1 |
| 3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency | HMGCL | • | | | | • | • | • | • | PC190.1 |
| 3-Hydroxyacyl-CoA Dehydrogenase Deficiency | HADH | • | | | | • | • | • | • | PC4.1 |
| 3-Methylcrotonyl-CoA Carboxylase 1 Deficiency | MCCC1 | • | | | | • | • | • | • | PC5.1 |
| 3-Methylcrotonyl-CoA Carboxylase 2 Deficiency | MCCC2 | • | | | o | • | • | • | • | PC6.1 |
| 3-Phosphoglycerate Dehydrogenase Deficiency | PHGDH | • | | | | • | • | • | • | PC325.1 |
| 5-Alpha-Reductase Deficiency | SRD5A2 | • | | | | | • | • | • | PC95654.1 |
| 6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency | PTS | • | | | | • | • | • | • | PC379.1 |
| ABCA4 -Related Conditions | ABCA4 | • | | | | | • | • | • | PC52817.1 |
| Abetalipoproteinemia | MTTP | • | | | | • | • | • | • | PC7.1 |
| Achondrogenesis, Type 1B | SLC26A2 | • | | | o | • | • | • | • | PC8.1 |
| Achromatopsia, CNGB3-Related | CNGB3 | • | | | o | • | • | • | • | PC9.1 |
| Acrodermatitis Enteropathica | SLC39A4 | • | | | | • | • | • | • | PC10.1 |
| Action Myoclonus–Renal Failure (AMRF) Syndrome | SCARB2 | • | | | | • | | | • | PC11.1 |
| Acute Infantile Liver Failure, TRMU-Related | TRMU | • | | | | • | • | • | • | PC12.1 |
| Acyl-CoA Oxidase I Deficiency | ACOX1 | • | | | | • | • | • | • | PC13.1 |
| Adrenal Hypoplasia Congenita, X-Linked | NR0B1 | | • | | o | • | • | • | • | PC412.1 |
| Adrenoleukodystrophy, X-Linked | ABCD1 | | • | | o | • | • | • | • | PC14.1 |
| Agammaglobulinemia, X-Linked | BTK | | • | | | • | • | • | • | PC15.1 |
| Aicardi-Goutières Syndrome | SAMHD1 | • | | | | • | • | • | • | PC19.1 |
| Aicardi-Goutières Syndrome, RNASEH2A-Related | RNASEH2A | • | | | | • | • | • | • | PC16.1 |
| Aicardi-Goutières Syndrome, RNASEH2B-Related | RNASEH2B | • | | | o | • | • | • | • | PC17.1 |
| Aicardi-Goutières Syndrome, RNASEH2C-Related | RNASEH2C | • | | | | • | • | • | • | PC18.1 |
| Aicardi-Goutières Syndrome, TREX1 -Related | TREX1 | • | | | | | • | • | • | PC99630.1 |
| Alkaptonuria | HGD | • | | | | | • | • | • | PC59465.1 |
| Alpha-1 Antitrypsin Deficiency | SERPINA1 | • | | | | • | • | • | • | PC21.1 |
| Alpha-Mannosidosis | MAN2B1 | • | | | | • | • | • | • | PC22.1 |
| Alpha-Thalassemia | HBA1/HBA2 | • | | o | o | • | • | • | • | PC23.1 |
| Alpha-Thalassemia Intellectual Disability Syndrome | ATRX | | • | | | • | • | • | • | PC20.1 |
| Alport Syndrome, COL4A3-Related | COL4A3 | • | | | | • | • | • | • | PC24.1 |
| Alport Syndrome, COL4A4-Related | COL4A4 | • | | | | • | • | • | • | PC25.1 |
| Alport Syndrome, X-Linked | COL4A5 | | • | | | • | • | • | • | PC26.1 |
| Alstrom Syndrome | ALMS1 | • | | | | • | • | • | • | PC27.1 |
| Amish Infantile Epilepsy Syndrome | ST3GAL5 | • | | | | • | • | • | • | PC28.1 |
| Andermann Syndrome | SLC12A6 | • | | | | • | • | • | • | PC183.1 |
| Androgen Insensitivity Syndrome | AR | | • | | | | • | • | • | PC43715.1 |
| Arginine:Glycine Amidinotransferase Deficiency (Agat Deficiency) | GATM | • | | | | | • | • | • | PC25716.1 |
| Argininemia | ARG1 | • | | | | • | • | • | • | PC29.1 |
| Argininosuccinate Lyase Deficiency | ASL | • | | | o | • | • | • | • | PC30.1 |
| Aromatase Deficiency | CYP19A1 | • | | | | • | • | • | • | PC31.1 |
| Arts Syndrome | PRPS1 | | • | | | • | • | • | • | PC34.1 |
| Asparagine Synthetase Deficiency | ASNS | • | | | | • | • | • | • | PC35.1 |
| Aspartylglycosaminuria | AGA | • | | | o | • | • | • | • | PC36.1 |
| Ataxia with Vitamin E Deficiency | TTPA | • | | | | • | • | • | • | PC37.1 |
| Ataxia-Telangiectasia | ATM | • | | | | • | • | • | • | PC38.1 |
| Ataxia-Telangiectasia-Like Disorder 1 | MRE11 | • | | | | • | • | • | • | PC39.1 |
| Atransferrinemia | TF | | | | o | • | • | • | • | PC465.1 |
| Autism Spectrum, Epilepsy and Arthrogryposis | SLC35A3 | • | | | | • | • | • | • | PC33.1 |
| Autoimmune Polyglandular Syndrome, Type 1 | AIRE | • | | | o | • | • | • | • | PC328.1 |
| Autosomal Recessive Congenital Ichthyosis (ARCI), SLC27A4 -Related | SLC27A4 | • | | | | | • | • | • | PC80717.1 |
| Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay | SACS | • | | | | • | • | • | • | PC32.1 |
| Bardet-Biedl Syndrome, ARL6 -Related | ARL6 | • | | | | | • | • | • | PC71221.1 |
| Bardet-Biedl Syndrome, BBS10-Related | BBS10 | • | | | | • | • | • | • | PC48.1 |
| Bardet-Biedl Syndrome, BBS12-Related | BBS12 | • | | | | • | • | • | • | PC49.1 |
| Bardet-Biedl Syndrome, BBS1-Related | BBS1 | • | | | o | • | • | • | • | PC42.1 |
| Bardet-Biedl Syndrome, BBS2-Related | BBS2 | • | | | o | • | • | • | • | PC50.1 |
| Bardet-Biedl Syndrome, BBS4-Related | BBS4 | • | | | | • | • | • | • | PC43.1 |
| Bardet-Biedl Syndrome, Bbs5-Related | BBS5 | • | | | | | • | • | • | PC49347.1 |
| Bardet-Biedl Syndrome, BBS7-Related | BBS7 | • | | | | • | • | • | • | PC45.1 |
| Bardet-Biedl Syndrome, BBS9-Related | BBS9 | • | | | | • | • | • | • | PC47.1 |
| Bardet-Biedl Syndrome, TTC8-Related | TTC8 | • | | | | • | | | • | PC46.1 |
| Bare Lymphocyte Syndrome, CIITA-Related | CIITA | • | | | | • | • | • | • | PC51.1 |
| Barth Syndrome | TAZ | | • | | | • | • | • | • | PC52.1 |
| Bartter Syndrome, BSND-Related | BSND | • | | | | • | • | • | • | PC53.1 |
| Bartter Syndrome, KCNJ1 -Related | KCNJ1 | • | | | | | • | • | • | PC92223.1 |

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|---|---|---|---|---|---|---|---|---|---|---|
| | | | | ACOG* | ACMG | H445 | H569 † | H574 † | H613 | |
| Bartter Syndrome, SLC12A1 -Related | SLC12A1 | • | | | | | • | • | • | PC14512.1 |
| Batten Disease, CLN3-Related | CLN3 | • | | | | • | • | • | • | PC54.1 |
| Bernard-Soulier Syndrome, Type A1/A2 | GP1BA | • | | | | • | • | • | • | PC55.1 |
| Bernard-Soulier Syndrome, Type C | GP9 | • | | | | • | • | • | • | PC56.1 |
| Beta-Hemoglobinopathies | HBB | • | | o | o | • | • | • | • | PC378.1 |
| Beta-Mannosidosis | MANBA | • | | | | | • | • | • | PC73957.1 |
| Beta-Ureidopropionase Deficiency | UPB1 | • | | | | • | | | • | PC57.1 |
| Bilateral Frontoparietal Polymicrogyria | GPR56 | • | | | | • | • | • | • | PC329.1 |
| Biotinidase Deficiency | BTD | • | | | o | • | • | • | • | PC58.1 |
| Biotin-Thiamine-Responsive Basal Ganglia Disease (BTBGD) | SLC19A3 | | | | o | • | • | • | • | PC464.1 |
| Bloom Syndrome | BLM | • | | o | o | • | • | • | • | PC59.1 |
| Brittle Cornea Syndrome 1 | ZNF469 | • | | | | | • | • | • | PC37592.1 |
| Brittle Cornea Syndrome 2 | PRDM5 | • | | | | | • | • | • | PC20637.1 |
| Canavan Disease | ASPA | • | | o | o | • | • | • | • | PC60.1 |
| Carbamoyl Phosphate Synthetase I Deficiency | CPS1 | • | | | | • | • | • | • | PC61.1 |
| Carnitine Deficiency | SLC22A5 | • | | | | • | • | • | • | PC63.1 |
| Carnitine Palmitoyltransferase IA Deficiency | CPT1A | • | | | | • | • | • | • | PC65.1 |
| Carnitine Palmitoyltransferase II Deficiency | CPT2 | • | | | o | • | • | • | • | PC64.1 |
| Carnitine-Acylcarnitine Translocase Deficiency | SLC25A20 | • | | | | • | • | • | • | PC66.1 |
| Carpenter Syndrome | RAB23 | • | | | | • | • | • | • | PC67.1 |
| Cartilage-Hair Hypoplasia | RMRP | • | | | | • | • | • | • | PC68.1 |
| Catecholaminergic Polymorphic Ventricular Tachycardia | CASQ2 | • | | | | | • | • | • | PC34286.1 |
| CD59 -Mediated Hemolytic Anemia | CD59 | • | | | | | • | • | • | PC83320.1 |
| CEP152 -Related Microcephaly | CEP152 | • | | | | | • | • | • | PC16422.1 |
| Cerebral Dysgenesis, Neuropathy, Ichthyosis, And Palmoplantar Keratoderma (CEDNIK) Syndrome | SNAP29 | • | | | | | • | • | • | PC55961.1 |
| Cerebrotendinous Xanthomatosis | CYP27A1 | • | | | o | • | • | • | • | PC69.1 |
| Charcot-Marie-Tooth Disease with Deafness, X-Linked | GJB1 | | • | | | • | • | • | • | PC71.1 |
| Charcot-Marie-Tooth Disease, Recessive Intermediate C | PLEKHG5 | • | | | | | • | • | • | PC63351.1 |
| Charcot-Marie-Tooth Disease, Type 4D | NDRG1 | • | | | | • | • | • | • | PC72.1 |
| Chediak-Higashi Syndrome | LYST | • | | | | • | • | • | • | PC73.1 |
| Choreoacanthocytosis | VPS13A | • | | | | • | • | • | • | PC76.1 |
| Choroideremia | CHM | | • | | | • | • | • | • | PC77.1 |
| Chronic Granulomatous Disease, CYBA-Related | CYBA | • | | | | • | • | • | • | PC79.1 |
| Chronic Granulomatous Disease, NCF2 -Related | NCF2 | • | | | | | • | • | • | PC47241.1 |
| Chronic Granulomatous Disease, X-Linked | CYBB | | • | | | • | • | • | • | PC78.1 |
| Ciliopathies, RPGRIP1L-Related | RPGRIP1L | • | | | | • | • | • | • | PC80.1 |
| Citrin Deficiency | SLC25A13 | • | | | | • | • | • | • | PC81.1 |
| Citrullinemia, Type 1 | ASS1 | • | | | | • | • | • | • | PC82.1 |
| CLN10 Disease | CTSD | • | | | | • | • | • | • | PC70.1 |
| Cohen Syndrome | VPS13B | • | | | | • | • | • | • | PC85.1 |
| COL11A2 -Related Conditions | COL11A2 | • | | | | | • | • | • | PC23804.1 |
| Combined Malonic and Methylmalonic Aciduria | ACSF3 | • | | | | • | • | • | • | PC86.1 |
| Combined Oxidative Phosphorylation Deficiency 1 | GFM1 | • | | | | • | • | • | • | PC87.1 |
| Combined Oxidative Phosphorylation Deficiency 3 | TSFM | • | | | | • | • | • | • | PC88.1 |
| Combined Pituitary Hormone Deficiency 1 | POU1F1 | • | | | | | • | • | • | PC89464.1 |
| Combined Pituitary Hormone Deficiency-2 | PROP1 | • | | | | • | • | • | • | PC345.1 |
| Congenital Adrenal Hyperplasia, 11-Beta-Hydroxylase Deficiency | CYP11B1 | • | | | | • | • | • | • | PC1.1 |
| Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficiency | CYP17A1 | • | | | | • | • | • | • | PC90.1 |
| Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency | CYP21A2 | • | | | o | • | • | • | • | PC91.1 |
| Congenital Adrenal Insufficiency, CYP11A1-Related | CYP11A1 | | | | o | • | • | • | • | PC450.1 |
| Congenital Amegakaryocytic Thrombocytopenia | MPL | • | | | | • | • | • | • | PC93.1 |
| Congenital Chronic Diarrhea | DGAT1 | • | | | | | • | • | • | PC96513.1 |
| Congenital Disorder Of Glycosylation Type 1, ALG1 -Related | ALG1 | • | | | | | • | • | • | PC35340.1 |
| Congenital Disorder of Glycosylation, Type 1A, PMM2-Related | PMM2 | • | | | o | • | • | • | • | PC95.1 |
| Congenital Disorder of Glycosylation, Type 1B | MPI | • | | | | • | • | • | • | PC96.1 |
| Congenital Disorder of Glycosylation, Type 1C | ALG6 | • | | | | • | • | • | • | PC94.1 |
| Congenital Dyserythropoietic Anemia Type 2 | SEC23B | • | | | | | • | • | • | PC20191.1 |
| Congenital Finnish Nephrosis | NPHS1 | • | | | o | • | • | • | • | PC299.1 |
| Congenital Hydrocephalus 1 | CCDC88C | | | | o | • | • | • | • | PC448.1 |
| Congenital Hyperinsulinism, KCNJ11-Related | KCNJ11 | • | | | | • | • | • | • | PC199.1 |
| Congenital Insensitivity to Pain with Anhidrosis (CIPA) | NTRK1 | • | | | | • | • | • | • | PC98.1 |
| Congenital Myasthenic Syndrome, CHAT-Related | CHAT | • | | | | • | • | • | • | PC99.1 |
| Congenital Myasthenic Syndrome, CHRNE-Related | CHRNE | • | | | o | • | • | • | • | PC100.1 |
| Congenital Myasthenic Syndrome, COLQ-Related | COLQ | • | | | | • | | | • | PC293.1 |
| Congenital Myasthenic Syndrome, DOK7-Related | DOK7 | • | | | | • | • | • | • | PC101.1 |
| Congenital Myasthenic Syndrome, RAPSN-Related | RAPSN | • | | | | • | • | • | • | PC102.1 |
| Congenital Nephrotic Syndrome, PLCE1-Related | PLCE1 | • | | | | • | | | • | PC298.1 |
| Congenital Neutropenia, G6PC3 -Related | G6PC3 | • | | | | | • | • | • | PC61295.1 |
| Congenital Neutropenia, HAX1-Related | HAX1 | • | | | | • | • | • | • | PC103.1 |
| Congenital Neutropenia, VPS45-Related | VPS45 | • | | | | • | • | • | • | PC104.1 |

| CONDITION | GENE | AUTOSOMAL RECESSIVE | X-LINKED | SCREENING RECOMMENDATIONS | | PANEL AVAILABILITY | | | | PC# |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | ACOG* | ACMG | H445 | H569 † | H574 † | H613 | |
| Congenital Secretory Chloride Diarrhea 1 | SLC26A3 | • | | | | | • | • | • | PC78928.1 |
| Corneal Dystrophy and Perceptive Deafness | SLC4A11 | • | | | | • | • | • | • | PC105.1 |
| Corticosterone Methyloxidase Deficiency | CYP11B2 | • | | | | • | • | • | • | PC106.1 |
| Costeff Syndrome (3-Methylglutaconic Aciduria, Type 3) | OPA3 | • | | | | • | • | • | • | PC107.1 |
| Cowchock Syndrome | AIFM1 | | • | | | • | | | • | PC89.1 |
| CRB1-Related Retinal Dystrophies | CRB1 | • | | | | • | • | • | • | PC108.1 |
| Creatine Transporter Defect (Cerebral Creatine Deficiency Syndrome 1, X-Linked) | SLC6A8 | | • | | o | • | • | • | • | PC109.1 |
| Cystic Fibrosis | CFTR | • | | o | o | • | • | • | • | PC110.1 |
| Cystinosis | CTNS | • | | | | • | • | • | • | PC111.1 |
| Cytochrome C Oxidase Deficiency, PET100-Related | PET100 | • | | | | • | • | • | • | PC268.1 |
| Cytochrome P450 Oxioreductase Deficiency | POR | • | | | | | • | • | • | PC6948.1 |
| D-Bifunctional Protein Deficiency | HSD17B4 | • | | | | • | • | • | • | PC112.1 |
| Deafness, Autosomal Recessive 77 | LOXHD1 | • | | | | • | • | • | • | PC113.1 |
| Dent Disease, Type 1 | CLCN5 | | • | | | • | | | • | PC114.1 |
| Dent Disease, Type 2 / Lowe Syndrome | OCRL | | • | | | • | • | • | • | PC245.1 |
| Developmental And Eplileptic Encephalopathy 36 | ALG13 | | • | | | | • | • | • | PC96040.1 |
| Dihydropteridine Reductase (DHPR) Deficiency | QDPR | • | | | | | • | • | • | PC18676.1 |
| Dihydropyrimidine Dehydrogenase Deficiency | DPYD | • | | | | • | | | • | PC118.1 |
| Donnai-Barrow Syndrome | LRP2 | | | | o | • | • | • | • | PC455.1 |
| Dubin-Johnson Syndrome | ABCC2 | • | | | | | • | • | • | PC98051.1 |
| Duchenne/Becker Muscular Dystrophy | DMD | | • | | o | • | • | • | • | PC62.1 |
| Dyskeratosis Congenita Spectrum Disorders | TERT | • | | | | | • | • | • | PC63259.1 |
| Dyskeratosis Congenita, DKC1-Related | DKC1 | | • | | | • | • | • | • | PC120.1 |
| Dyskeratosis Congenita, RTEL1-Related | RTEL1 | • | | | | • | • | • | • | PC119.1 |
| Dystrophic Epidermolysis Bullosa, COL7A1-Related | COL7A1 | • | | | o | • | • | • | • | PC121.1 |
| Early Infantile Epileptic Encephalopathy, CAD -Related | CAD | • | | | | | • | • | • | PC89446.1 |
| Ehlers-Danlos Syndrome Type VI | PLOD1 | • | | | | | • | • | • | PC30511.1 |
| Ehlers-Danlos Syndrome, Classic-Like, TNXB-Related | TNXB | | | | o | • | | • | • | PC466.1 |
| Ehlers-Danlos Syndrome, Type VIIC | ADAMTS2 | • | | | | • | • | • | • | PC122.1 |
| Ellis-van Creveld Syndrome, EVC2-Related | EVC2 | • | | | o | • | • | • | • | PC124.1 |
| Ellis-van Creveld Syndrome, EVC-Related | EVC | • | | | | • | • | • | • | PC123.1 |
| Emery-Dreifuss Muscular Dystrophy 1, X-Linked | EMD | | • | | | • | • | • | • | PC125.1 |
| Emery-Dreifuss Muscular Dystrophy 6, X Linked | FHL1 | | • | | | | • | • | • | PC35435.1 |
| Enhanced S-Cone Syndrome | NR2E3 | • | | | | • | • | • | • | PC126.1 |
| Epimerase Deficiency (Galactosemia Type III) | GALE | • | | | | | • | • | • | PC33923.1 |
| Epiphyseal Dysplasia, Multiple, 7 / Desbuquois Dysplasia 1 | CANT1 | • | | | | • | • | • | • | PC115.1 |
| ERCC6-Related Disorders | ERCC6 | • | | | | • | • | • | • | PC84.1 |
| ERCC8-Related Disorders | ERCC8 | • | | | | • | • | • | • | PC83.1 |
| Ethylmalonic Encephalopathy | ETHE1 | • | | | | • | • | • | • | PC131.1 |
| Fabry Disease | GLA | | • | | o | • | • | • | • | PC132.1 |
| Factor IX Deficiency | F9 | | • | | o | • | • | • | • | PC133.1 |
| Factor V Deficiency | F5 | • | | | | | • | • | • | PC49069.1 |
| Factor XI Deficiency | F11 | • | | | | • | • | • | • | PC134.1 |
| Familial Dysautonomia | IKBKAP | • | | o | o | • | • | • | • | PC135.1 |
| Familial Hemophagocytic Lymphohistiocytosis, PRF1-Related | PRF1 | • | | | o | • | • | • | • | PC136.1 |
| Familial Hemophagocytic Lymphohistiocytosis, STX11-Related | STX11 | • | | | | • | • | • | • | PC162.1 |
| Familial Hemophagocytic Lymphohistiocytosis, STXBP2-Related | STXBP2 | • | | | | • | • | • | • | PC137.1 |
| Familial Hemophagocytic Lymphohistiocytosis, UNC13D -Related | UNC13D | • | | | | | • | • | • | PC46264.1 |
| Familial Hypercholesterolemia, LDLRAP1-Related | LDLRAP1 | • | | | | • | • | • | • | PC139.1 |
| Familial Hypercholesterolemia, LDLR-Related | LDLR | • | | | | • | • | • | • | PC138.1 |
| Familial Hyperinsulinism, ABCC8-Related | ABCC8 | • | | o | o | • | • | • | • | PC140.1 |
| Familial Mediterranean Fever | MEFV | • | | | | • | • | • | • | PC141.1 |
| Familial Nephrogenic Diabetes Insipidus, AQP2-Related | AQP2 | • | | | | • | • | • | • | PC142.1 |
| Fanconi Anemia Group J | BRIP1 | • | | | | | • | • | • | PC31801.1 |
| Fanconi Anemia, Group A | FANCA | • | | o | | • | • | • | • | PC149.1 |
| Fanconi Anemia, Group B | FANCB | | • | | | • | • | • | • | PC143.1 |
| Fanconi Anemia, Group C | FANCC | • | | o | o | • | • | • | • | PC151.1 |
| Fanconi Anemia, Group D2 | FANCD2 | • | | | | • | • | • | • | PC144.1 |
| Fanconi Anemia, Group E | FANCE | • | | | | • | • | • | • | PC145.1 |
| Fanconi Anemia, Group F | FANCF | • | | | | • | | | • | PC146.1 |
| Fanconi Anemia, Group G | FANCG | • | | o | | • | • | • | • | PC150.1 |
| Fanconi Anemia, Group I | FANCI | • | | | | • | • | • | • | PC147.1 |
| Fanconi Anemia, Group L | FANCL | • | | | | • | • | • | • | PC148.1 |
| Farber Lipogranulomatosis | ASAH1 | • | | | | • | | | • | PC152.1 |
| Foveal Hypoplasia | SLC38A8 | • | | | | | • | • | • | PC1407.1 |
| Fragile X Syndrome | FMR1 | | • | o | o | • | • | • | • | PC153.1 |
| Fragile XE Syndrome | AFF2 | | • | | o | • | | • | • | PC446.1 |
| Fraser Syndrome 3, GRIP1-Related | GRIP1 | | | | o | • | • | • | • | PC454.1 |
| Fraser Syndrome, FRAS1 -Related | FRAS1 | • | | | | | • | • | • | PC818.1 |
| Fraser Syndrome, FREM2 -Related | FREM2 | • | | | | | • | • | • | PC5827.1 |

| CONDITION | GENE | AUTOSOMAL RECESSIVE | X-LINKED | SCREENING RECOMMENDATIONS | | PANEL AVAILABILITY | | | | PC# |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | ACOG* | ACMG | H445 | H569 † | H574 † | H613 | |
| Friedreich Ataxia | FXN | | | | o | • | | • | • | PC473.1 |
| Fructose-1,6-Bisphosphatase Deficiency | FBP1 | • | | | | | • | • | • | PC56680.1 |
| Fucosidosis | FUCA1 | • | | | | | • | • | • | PC78729.1 |
| Fumarase Deficiency | FH | • | | | | • | • | • | • | PC155.1 |
| GABA-Transaminase Deficiency | ABAT | • | | | | • | | | • | PC156.1 |
| Galactokinase Deficiency (Galactosemia, Type II) | GALK1 | • | | | | • | • | • | • | PC157.1 |
| Galactosemia | GALT | • | | | o | • | • | • | • | PC158.1 |
| Galactosialidosis | CTSA | • | | | | • | • | • | • | PC159.1 |
| Gaucher Disease | GBA | • | | | o | • | • | • | • | PC160.1 |
| GCH1 -Related Conditions | GCH1 | • | | | | | • | • | • | PC32392.1 |
| GDF5 -Related Conditions | GDF5 | • | | | | | • | • | • | PC9131.1 |
| Geroderma Osteodysplastica | GORAB | • | | | | | • | • | • | PC43013.1 |
| Gitelman Syndrome | SLC12A3 | • | | | | • | • | • | • | PC161.1 |
| Glanzmann Thrombasthenia | ITGB3 | • | | | | | • | • | • | PC67275.1 |
| Glucose-6-Phosphate Dehydrogenase Deficiency | G6PD | | • | | | • | • | • | • | PC163.1 |
| Glutaric Acidemia, Type 1 | GCDH | • | | | | • | • | • | • | PC164.1 |
| Glutaric Acidemia, Type 2A | ETFA | • | | | | • | • | • | • | PC165.1 |
| Glutaric Acidemia, Type 2B | ETFB | • | | | | • | • | • | • | PC167.1 |
| Glutaric Acidemia, Type 2C | ETFDH | • | | | | • | • | • | • | PC166.1 |
| Glutathione Synthetase Deficiency | GSS | • | | | | | • | • | • | PC17255.1 |
| Glycine Encephalopathy, AMT-Related | AMT | • | | | | • | • | • | • | PC169.1 |
| Glycine Encephalopathy, GLDC-Related | GLDC | • | | | | • | • | • | • | PC168.1 |
| Glycogen Storage Disease Type IXb | PHKB | • | | | | | • | • | • | PC52945.1 |
| Glycogen Storage Disease Type IXc | PHKG2 | • | | | | | • | • | • | PC73585.1 |
| Glycogen Storage Disease, Type 1A | G6PC | • | | o | o | • | • | • | • | PC171.1 |
| Glycogen Storage Disease, Type 1B | SLC37A4 | • | | o | o | • | • | • | • | PC172.1 |
| Glycogen Storage Disease, Type 2 (Pompe Disease) | GAA | • | | | o | • | • | • | • | PC170.1 |
| Glycogen Storage Disease, Type 3 | AGL | • | | | | • | • | • | • | PC173.1 |
| Glycogen Storage Disease, Type 4 | GBE1 | • | | | o | • | • | • | • | PC174.1 |
| Glycogen Storage Disease, Type 5 (McArdle Disease) | PYGM | • | | | | • | • | • | • | PC175.1 |
| Glycogen Storage Disease, Type 7 | PFKM | • | | | | • | • | • | • | PC176.1 |
| GRACILE Syndrome | BCS1L | • | | | | • | • | • | • | PC178.1 |
| Guanidinoacetate Methyltransferase Deficiency | GAMT | • | | | | • | • | • | • | PC179.1 |
| Harlequin Ichthyosis | ABCA12 | • | | | | • | • | • | • | PC97.1 |
| Heme Oxygenase 1 Deficiency | HMOX1 | • | | | | | • | • | • | PC34178.1 |
| Hemochromatosis, Type 2A | HFE2 | • | | | | • | • | • | • | PC180.1 |
| Hemochromatosis, Type 3, TFR2-Related | TFR2 | • | | | | • | • | • | • | PC181.1 |
| Hemophilia A | F8 | | • | | o | • | | • | • | PC452.1 |
| Hepatocerebral Mitochondrial DNA Depletion Syndrome, MPV17-Related | MPV17 | • | | | | • | • | • | • | PC269.1 |
| Hereditary Fructose Intolerance | ALDOB | • | | | o | • | • | • | • | PC182.1 |
| Hereditary Hemochromatosis Type 1 | HFE | • | | | | | • | • | • | PC63537.1 |
| Hereditary Hemochromatosis Type 2B | HAMP | • | | | | | • | • | • | PC2430.1 |
| Hereditary Spastic Paraparesis, Type 49 | TECPR2 | • | | | | • | • | • | • | PC184.1 |
| Hereditary Spastic Paraplegia, CYP7B1 -Related | CYP7B1 | • | | | | | • | • | • | PC36512.1 |
| Hermansky-Pudlak Syndrome, AP3B1-Related | AP3B1 | • | | | | • | | | • | PC185.1 |
| Hermansky-Pudlak Syndrome, BLOC1S3 -Related | BLOC1S3 | • | | | | | • | • | • | PC56794.1 |
| Hermansky-Pudlak Syndrome, BLOC1S6 -Related | BLOC1S6 | • | | | | | • | • | • | PC20653.1 |
| Hermansky-Pudlak Syndrome, HPS1-Related | HPS1 | • | | | o | • | • | • | • | PC186.1 |
| Hermansky-Pudlak Syndrome, HPS3-Related | HPS3 | • | | | o | • | • | • | • | PC187.1 |
| Hermansky-Pudlak Syndrome, HPS4-Related | HPS4 | • | | | | • | • | • | • | PC188.1 |
| Hermansky-Pudlak Syndrome, HPS5 -Related | HPS5 | • | | | | | • | • | • | PC82722.1 |
| Hermansky-Pudlak Syndrome, HPS6 -Related | HPS6 | • | | | | | • | • | • | PC12623.1 |
| Heterotaxy Syndrome, ZIC3-Related | ZIC3 | | • | | | • | | | • | PC189.1 |
| Holocarboxylase Synthetase Deficiency | HLCS | • | | | | • | • | • | • | PC191.1 |
| Homocystinuria And Megaloblastic Anemia Type Cblg | MTR | • | | | | | • | • | • | PC2862.1 |
| Homocystinuria due to Deficiency of MTHFR | MTHFR | • | | | | • | • | • | • | PC193.1 |
| Homocystinuria, CBS-Related | CBS | • | | | o | • | • | • | • | PC192.1 |
| Homocystinuria, Type cblE | MTRR | • | | | | • | • | • | • | PC194.1 |
| HSD10 Disease | HSD17B10 | | • | | | | • | • | • | PC36238.1 |
| Hydrolethalus Syndrome | HYLS1 | • | | | | • | • | • | • | PC198.1 |
| Hyper IgM Syndrome, X-Linked | CD40LG | | • | | | • | • | • | • | PC414.1 |
| Hyper-Igm Immunodeficiency | CD40 | • | | | | | • | • | • | PC52631.1 |
| Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH Syndrome) | SLC25A15 | • | | | | • | • | • | • | PC200.1 |
| Hyperphosphatemic Familial Tumoral Calcinosis, GALNT3-Related | GALNT3 | • | | | | • | • | • | • | PC201.1 |
| Hypohidrotic Ectodermal Dysplasia, X-Linked | EDA | | • | | | • | • | • | • | PC202.1 |
| Hypomyelinating Leukodystrophy 12 | VPS11 | • | | | | | • | • | • | PC17537.1 |
| Hypophosphatasia, ALPL-Related | ALPL | • | | | o | • | • | • | • | PC203.1 |
| Imerslund-Gräsbeck Syndrome 2 | AMN | • | | | | | • | • | • | PC35659.1 |
| Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-Linked (IPEX) Syndrome | FOXP3 | | • | | | • | | | • | PC204.1 |
| Immunodeficiency-Centromeric Instability-Facial Anomalies (ICF) Syndrome, DNMT3B -Related | DNMT3B | • | | | | | • | • | • | PC62096.1 |

| CONDITION | GENE | AUTOSOMAL RECESSIVE | X-LINKED | SCREENING RECOMMENDATIONS | | PANEL AVAILABILITY | | | | PC# |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | ACOG* | ACMG | H445 | H569 † | H574 † | H613 | |
| Immunodeficiency-Centromeric Instability-Facial Anomalies (ICF) Syndrome, ZBTB24 -Related | ZBTB24 | • | | | | | • | • | • | PC62195.1 |
| Inclusion Body Myopathy 2 | GNE | • | | | | • | • | • | • | PC205.1 |
| Infantile Cerebral and Cerebellar Atrophy | MED17 | • | | | | • | • | • | • | PC206.1 |
| Infantile Nephronophthisis | INVS | • | | | | • | • | • | • | PC222.1 |
| Infantile Neuroaxonal Dystrophy | PLA2G6 | • | | | | • | • | • | • | PC207.1 |
| Infantile Spinal Muscular Atrophy, X-Linked | UBA1 | | • | | | • | | | • | PC383.1 |
| Isolated Ectopia Lentis | ADAMTSL4 | • | | | | | • | • | • | PC83170.1 |
| Isolated Lissencephaly Sequence / Subcortical Band Heterotopia | DCX | | • | | | • | | | • | PC264.1 |
| Isolated Sulfite Oxidase Deficiency | SUOX | • | | | | | • | • | • | PC6377.1 |
| Isolated Thyroid-Stimulating Hormone Deficiency | TSHB | • | | | | | • | • | • | PC39517.1 |
| Isovaleric Acidemia | IVD | • | | | | • | • | • | • | PC208.1 |
| Johanson-Blizzard Syndrome | UBR1 | • | | | | • | • | • | • | PC209.1 |
| Joubert Syndrome 2 / Meckel Syndrome 2 | TMEM216 | • | | o | o | • | • | • | • | PC221.1 |
| Joubert Syndrome And Related Disorders (JSRD), TMEM67 -Related | TMEM67 | • | | | | | • | • | • | PC48967.1 |
| Joubert Syndrome, AHI1-Related | AHI1 | • | | o | o | • | • | • | • | PC216.1 |
| Joubert Syndrome, ARL13B-Related | ARL13B | • | | o | | • | | | • | PC219.1 |
| Joubert Syndrome, B9D1-Related | B9D1 | • | | o | | • | | | • | PC215.1 |
| Joubert Syndrome, B9D2-Related | B9D2 | • | | o | | • | | | • | PC218.1 |
| Joubert Syndrome, C2CD3-Related / Orofaciodigital Syndrome 14 | C2CD3 | • | | o | | • | | | • | PC318.1 |
| Joubert Syndrome, CC2D2A-Related / COACH Syndrome | CC2D2A | • | | o | o | • | • | • | • | PC220.1 |
| Joubert Syndrome, CEP104-Related | CEP104 | • | | o | | • | | | • | PC214.1 |
| Joubert Syndrome, CEP120-Related / Short-Rib Thoracic Dysplasia 13 with or without Polydactyly | CEP120 | • | | o | | • | | | • | PC217.1 |
| Joubert Syndrome, CEP41-Related | CEP41 | • | | o | | • | | | • | PC211.1 |
| Joubert Syndrome, CPLANE1-Related / Orofaciodigital Syndrome 6 | CPLANE1 | • | | o | | • | | | • | PC212.1 |
| Joubert Syndrome, CSPP1-Related | CSPP1 | • | | o | | • | | | • | PC213.1 |
| Joubert Syndrome, INPP5E-Related | INPP5E | • | | o | | • | | | • | PC210.1 |
| Junctional Epidermolysis Bullosa, COL17A1 -Related | COL17A1 | • | | | | | • | • | • | PC64093.1 |
| Junctional Epidermolysis Bullosa, ITGA6 -Related | ITGA6 | • | | | | | • | • | • | PC38788.1 |
| Junctional Epidermolysis Bullosa, ITGB4 -Related | ITGB4 | • | | | | | • | • | • | PC51277.1 |
| Junctional Epidermolysis Bullosa, LAMA3-Related | LAMA3 | • | | | | • | • | • | • | PC127.1 |
| Junctional Epidermolysis Bullosa, LAMB3-Related | LAMB3 | • | | | | • | • | • | • | PC128.1 |
| Junctional Epidermolysis Bullosa, LAMC2-Related | LAMC2 | • | | | | • | • | • | • | PC129.1 |
| Juvenile Retinoschisis, X-Linked | RS1 | | • | | o | • | • | • | • | PC415.1 |
| Ketothiolase Deficiency | ACAT1 | • | | | o | • | • | • | • | PC225.1 |
| Krabbe Disease | GALC | • | | | | • | • | • | • | PC226.1 |
| L1 Syndrome | L1CAM | | • | | o | • | • | • | • | PC197.1 |
| Lamellar Ichthyosis, Type 1 | TGM1 | • | | | | • | • | • | • | PC40.1 |
| Laron Syndrome | GHR | • | | | | | • | • | • | PC49604.1 |
| Leber Congenital Amaurosis 2 | RPE65 | • | | | | • | • | • | • | PC229.1 |
| Leber Congenital Amaurosis Type AIPL1 | AIPL1 | • | | | | | • | • | • | PC93865.1 |
| Leber Congenital Amaurosis Type LRAT | LRAT | • | | | | | • | • | • | PC54638.1 |
| Leber Congenital Amaurosis Type TULP1 | TULP1 | • | | | | | • | • | • | PC74977.1 |
| Leber Congenital Amaurosis, IQCB1-Related / Senior-Loken Syndrome 5 | IQCB1 | • | | | | • | | | • | PC372.1 |
| Leber Congenital Amaurosis, Type CEP290 | CEP290 | • | | | o | • | • | • | • | PC230.1 |
| Leber Congenital Amaurosis, Type LCA5 | LCA5 | • | | | | • | • | • | • | PC232.1 |
| Leber Congenital Amaurosis, Type RDH12 | RDH12 | • | | | | • | • | • | • | PC231.1 |
| Leber Conghenital Amaurosis Type GUCY2D | GUCY2D | • | | | | | • | • | • | PC58804.1 |
| Leigh Syndrome, French-Canadian Type | LRPPRC | • | | | | • | • | • | • | PC233.1 |
| Lesch-Nyhan Syndrome | HPRT1 | | • | | | • | • | • | • | PC195.1 |
| Lethal Congenital Contracture Syndrome 1 | GLE1 | • | | | | • | • | • | • | PC234.1 |
| Leukoencephalopathy with Vanishing White Matter | EIF2B5 | • | | | | • | • | • | • | PC235.1 |
| Leukoencephalopathy With Vanishing White Matter, EIF2B1 -Related | EIF2B1 | • | | | | | • | • | • | PC55097.1 |
| Leukoencephalopathy With Vanishing White Matter, EIF2B2 -Related | EIF2B2 | • | | | | | • | • | • | PC11836.1 |
| Leukoencephalopathy With Vanishing White Matter, EIF2B3 -Related | EIF2B3 | • | | | | | • | • | • | PC17418.1 |
| Leukoencephalopathy With Vanishing White Matter, EIF2B4 -Related | EIF2B4 | • | | | | | • | • | • | PC37159.1 |
| LIG4 Syndrome | LIG4 | • | | | | | • | • | • | PC72187.1 |
| Limb-Girdle Muscular Dystrophy Type 8 | TRIM32 | • | | | | | • | • | • | PC86939.1 |
| Limb-Girdle Muscular Dystrophy, Type 2A | CAPN3 | • | | | | • | • | • | • | PC238.1 |
| Limb-Girdle Muscular Dystrophy, Type 2B | DYSF | • | | | | • | • | • | • | PC236.1 |
| Limb-Girdle Muscular Dystrophy, Type 2C | SGCG | • | | | | • | • | • | • | PC239.1 |
| Limb-Girdle Muscular Dystrophy, Type 2D | SGCA | • | | | | • | • | • | • | PC240.1 |
| Limb-Girdle Muscular Dystrophy, Type 2E | SGCB | • | | | | • | • | • | • | PC241.1 |
| Limb-Girdle Muscular Dystrophy, Type 2F | SGCD | • | | | | • | • | • | • | PC242.1 |
| Limb-Girdle Muscular Dystrophy, Type 2I | FKRP | • | | | o | • | • | • | • | PC237.1 |
| Lipoamide Dehydrogenase Deficiency (Dihydrolipoamide Dehydrogenase Deficiency) | DLD | • | | | o | • | • | • | • | PC117.1 |
| Lipoid Adrenal Hyperplasia | STAR | • | | | | • | • | • | • | PC243.1 |
| Lipoprotein Lipase Deficiency | LPL | • | | | | • | • | • | • | PC244.1 |
| Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency | HADHA | • | | | | • | • | • | • | PC228.1 |
| Lung Disease, Immunodeficiency, And Chromosome Breakage Syndrome (LICS) | NSMCE3 | • | | | | | • | • | • | PC45178.1 |
| Lysinuric Protein Intolerance | SLC7A7 | • | | | | • | • | • | • | PC246.1 |

| CONDITION | GENE | AUTOSOMAL RECESSIVE | X-LINKED | SCREENING RECOMMENDATIONS | | PANEL AVAILABILITY | | | | PC# |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | ACOG* | ACMG | H445 | H569 † | H574 † | H613 | |
| Malonyl-CoA Decarboxylase Deficiency | MLYCD | • | | | | • | • | • | • | PC247.1 |
| Maple Syrup Urine Disease, Type 1A | BCKDHA | • | | o | | • | • | • | • | PC249.1 |
| Maple Syrup Urine Disease, Type 1B | BCKDHB | • | | o | o | • | • | • | • | PC250.1 |
| Maple Syrup Urine Disease, Type 2 | DBT | • | | o | | • | • | • | • | PC248.1 |
| McKusick-Kaufman Syndrome | MKKS | • | | | | • | • | • | • | PC44.1 |
| Meckel Syndrome 7 / Nephronophthisis 3 | NPHP3 | • | | | | • | | | • | PC224.1 |
| Meckel-Gruber Syndrome, Type 1 | MKS1 | • | | | | • | • | • | • | PC251.1 |
| MECP2 -Related Conditions | MECP2 | | • | | | | • | • | • | PC24288.1 |
| MECR -Related Neurologic Disorder | MECR | • | | | | | • | • | • | PC11767.1 |
| Medium Chain Acyl-CoA Dehydrogenase Deficiency | ACADM | • | | o | o | • | • | • | • | PC252.1 |
| MEDNIK Syndrome | AP1S1 | • | | | | • | • | • | • | PC253.1 |
| Megalencephalic Leukoencephalopathy with Subcortical Cysts | MLC1 | • | | | o | • | • | • | • | PC254.1 |
| Menkes Syndrome | ATP7A | | • | | | • | • | • | • | PC313.1 |
| Merosin-Deficient Muscular Dystrophy | LAMA2 | • | | | | • | • | • | • | PC227.1 |
| Metabolic Encephalopathy and Arrhythmias, TANGO2-Related | TANGO2 | • | | | | • | • | • | • | PC255.1 |
| Metachromatic Leukodystrophy, ARSA-Related | ARSA | • | | | o | • | • | • | • | PC257.1 |
| Metachromatic Leukodystrophy, PSAP-Related | PSAP | • | | | | • | • | • | • | PC256.1 |
| Methylmalonic Acidemia And Homocystinuria Type Cblf | LMBRD1 | • | | | | | • | • | • | PC17909.1 |
| Methylmalonic Acidemia And Homocystinuria Type Cblx | HCFC1 | | • | | | | • | • | • | PC19564.1 |
| Methylmalonic Aciduria and Homocystinuria, Type cblC | MMACHC | • | | | o | • | • | • | • | PC258.1 |
| Methylmalonic Aciduria and Homocystinuria, Type cblD | MMADHC | • | | | | • | • | • | • | PC259.1 |
| Methylmalonic Aciduria, MCEE -Related | MCEE | • | | | | | • | • | • | PC17775.1 |
| Methylmalonic Aciduria, MMAA-Related | MMAA | • | | | | • | • | • | • | PC260.1 |
| Methylmalonic Aciduria, MMAB-Related | MMAB | • | | | | • | • | • | • | PC261.1 |
| Methylmalonic Aciduria, Type mut(0) | MUT | • | | | o | • | • | • | • | PC262.1 |
| Mevalonic Kinase Deficiency | MVK | | | | o | • | • | • | • | PC458.1 |
| Microcephalic Osteodysplastic Primordial Dwarfism Type II | PCNT | • | | | | | • | • | • | PC71594.1 |
| Microphthalmia/Anophthalmia, VSX2-Related | VSX2 | • | | | | • | • | • | • | PC263.1 |
| Mitochondrial Complex 1 Deficiency, ACAD9-Related | ACAD9 | • | | | | • | • | • | • | PC365.1 |
| Mitochondrial Complex 1 Deficiency, NDUFAF5-Related | NDUFAF5 | • | | | | • | • | • | • | PC265.1 |
| Mitochondrial Complex 1 Deficiency, NDUFS6-Related | NDUFS6 | • | | | | • | • | • | • | PC266.1 |
| Mitochondrial complex I Deficiency, Nuclear Type 1 | NDUFS4 | • | | | | • | • | • | • | PC267.1 |
| Mitochondrial Complex I Deficiency, Nuclear Type 10 | NDUFAF2 | • | | | | | • | • | • | PC53435.1 |
| Mitochondrial Complex I Deficiency, Nuclear Type 17 | NDUFAF6 | • | | | | • | | | • | PC273.1 |
| Mitochondrial Complex I Deficiency, Nuclear Type 19 | FOXRED1 | • | | | | | • | • | • | PC56929.1 |
| Mitochondrial Complex I Deficiency, Nuclear Type 3 | NDUFS7 | • | | | | | • | • | • | PC76142.1 |
| Mitochondrial Complex I Deficiency, Nuclear Type 4 | NDUFV1 | • | | | | | • | • | • | PC9791.1 |
| Mitochondrial Complex IV Deficiency, Nuclear Type 2, SCO2-Related | SCO2 | | | | o | • | • | • | • | PC463.1 |
| Mitochondrial Complex IV Deficiency, Nuclear Type 6 | COX15 | • | | | | | • | • | • | PC13535.1 |
| Mitochondrial Dna Depletion Syndrome 2 | TK2 | • | | | | | • | • | • | PC85401.1 |
| Mitochondrial Dna Depletion Syndrome 3 | DGUOK | • | | | | | • | • | • | PC48922.1 |
| Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1) | PUS1 | • | | | | • | • | • | • | PC270.1 |
| Mitochondrial Trifunctional Protein Deficiency, HADHB-Related | HADHB | • | | | | • | • | • | • | PC271.1 |
| Molybdenum Cofactor Deficiency Type B | MOCS2 | • | | | | | • | • | • | PC75079.1 |
| Molybdenum Cofactor Deficiency, Type A | MOCS1 | • | | | | • | • | • | • | PC272.1 |
| Mucolipidosis II/IIIA | GNPTAB | • | | | o | • | • | • | • | PC275.1 |
| Mucolipidosis III gamma | GNPTG | • | | | | • | • | • | • | PC276.1 |
| Mucolipidosis, Type IV | MCOLN1 | • | | o | o | • | • | • | • | PC277.1 |
| Mucopolysaccharidosis, Type I (Hurler Syndrome) | IDUA | • | | | o | • | • | • | • | PC196.1 |
| Mucopolysaccharidosis, Type II (Hunter Syndrome) | IDS | | • | | | • | • | • | • | PC280.1 |
| Mucopolysaccharidosis, Type IIIA (Sanfilippo A) | SGSH | • | | | | • | • | • | • | PC281.1 |
| Mucopolysaccharidosis, Type IIIB (Sanfilippo B) | NAGLU | • | | | | • | • | • | • | PC282.1 |
| Mucopolysaccharidosis, Type IIIC (Sanfilippo C) | HGSNAT | • | | | | • | • | • | • | PC278.1 |
| Mucopolysaccharidosis, Type IIID (Sanfilippo D) | GNS | • | | | | • | • | • | • | PC283.1 |
| Mucopolysaccharidosis, Type IVA (Morquio Syndrome) | GALNS | • | | | | • | • | • | • | PC274.1 |
| Mucopolysaccharidosis, Type IVB / GM1 Gangliosidosis | GLB1 | • | | | | • | • | • | • | PC177.1 |
| Mucopolysaccharidosis, Type IX | HYAL1 | • | | | | • | • | • | • | PC279.1 |
| Mucopolysaccharidosis, Type VI (Maroteaux-Lamy) | ARSB | • | | | | • | • | • | • | PC284.1 |
| Mucopolysaccharidosis, Type VII | GUSB | • | | | | • | • | • | • | PC285.1 |
| Mulibrey Nanism | TRIM37 | • | | | | • | • | • | • | PC286.1 |
| Multiple Pterygium Syndrome, CHRNG-Related / Escobar Syndrome | CHRNG | • | | | | • | • | • | • | PC287.1 |
| Multiple Sulfatase Deficiency | SUMF1 | • | | | | • | • | • | • | PC288.1 |
| Muscle-Eye-Brain Disease, POMGNT1-Related | POMGNT1 | • | | | | • | • | • | • | PC289.1 |
| Muscular Dystrophy-Dystroglycanopathy | RXYLT1 | • | | | | | • | • | • | PC78515.1 |
| MUSK -Related Congenital Myasthenic Syndrome | MUSK | • | | | | | • | • | • | PC13167.1 |
| Myoneurogastrointestinal Encephalopathy (MNGIE) | TYMP | • | | | | • | • | • | • | PC294.1 |
| Myotonia Congenita | CLCN1 | | | | o | • | • | • | • | PC449.1 |
| Myotubular Myopathy, X-Linked | MTM1 | | • | | | • | • | • | • | PC295.1 |
| N-acetylglutamate Synthase Deficiency | NAGS | • | | | | • | • | • | • | PC296.1 |
| Nemaline Myopathy, NEB-Related | NEB | • | | | o | • | • | • | • | PC297.1 |

| CONDITION | GENE | AUTOSOMAL RECESSIVE | X-LINKED | SCREENING RECOMMENDATIONS | | PANEL AVAILABILITY | | | | PC# |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | ACOG* | ACMG | H445 | H569 † | H574 † | H613 | |
| Nephrogenic Diabetes Insipidus, AVPR2 -Related | AVPR2 | | • | | | | • | • | • | PC76349.1 |
| Nephronophthisis 1 | NPHP1 | • | | | | • | • | • | • | PC223.1 |
| Neuronal Ceroid Lipofuscinosis, CLN5-Related | CLN5 | • | | | | • | • | • | • | PC301.1 |
| Neuronal Ceroid Lipofuscinosis, CLN6-Related | CLN6 | • | | | | • | • | • | • | PC303.1 |
| Neuronal Ceroid Lipofuscinosis, CLN8-Related | CLN8 | • | | | | • | • | • | • | PC304.1 |
| Neuronal Ceroid Lipofuscinosis, MFSD8-Related | MFSD8 | • | | | | • | • | • | • | PC302.1 |
| Neuronal Ceroid Lipofuscinosis, PPT1-Related | PPT1 | • | | | | • | • | • | • | PC305.1 |
| Neuronal Ceroid Lipofuscinosis, TPP1-Related | TPP1 | • | | | | • | • | • | • | PC306.1 |
| Ngly1-Congenital Disorder Of Glycosylation | NGLY1 | • | | | | | • | • | • | PC63791.1 |
| Niemann-Pick Disease, Type C1/D | NPC1 | • | | o | | • | • | • | • | PC309.1 |
| Niemann-Pick Disease, Type C2 | NPC2 | • | | o | | • | • | • | • | PC307.1 |
| Niemann-Pick Disease, Types A/B | SMPD1 | • | | o | o | • | • | • | • | PC308.1 |
| Nijmegen Breakage Syndrome | NBN | • | | | | • | • | • | • | PC310.1 |
| Non-Syndromic Hearing Loss, GJB2-Related | GJB2 | • | | | o | • | • | • | • | PC311.1 |
| Nonsyndromic Hearing Loss, MYO15A-Related | MYO15A | • | | | | • | • | • | • | PC312.1 |
| Nonsyndromic Hearing Loss, OTOA-Related | OTOA | • | | | | | • | • | • | PC8675.1 |
| Nonsyndromic Hearing Loss, OTOF-Related | OTOF | • | | | | | • | • | • | PC51719.1 |
| Nonsyndromic Hearing Loss, PJVK-Related | PJVK | • | | | | | • | • | • | PC39556.1 |
| Nonsyndromic Hearing Loss, SYNE4-Related | SYNE4 | • | | | | | • | • | • | PC50286.1 |
| Nonsyndromic Hearing Loss, TMC1-Related | TMC1 | • | | | | | • | • | • | PC99571.1 |
| Nonsyndromic Hearing Loss, TMPRSS3 -Related | TMPRSS3 | • | | | | | • | • | • | PC78123.1 |
| Nonsyndromic Intellectual Disability | CC2D1A | • | | | | | • | • | • | PC14806.1 |
| Normophosphatemic Tumoral Calcinosis | SAMD9 | • | | | | | • | • | • | PC53991.1 |
| Oculocutaneous Albinism Type III | TYRP1 | • | | | | | • | • | • | PC46909.1 |
| Oculocutaneous Albinism Type IV | SLC45A2 | • | | | | | • | • | • | PC36690.1 |
| Oculocutaneous Albinism, OCA2-Related | OCA2 | | | | o | • | • | • | • | PC460.1 |
| Oculocutaneous Albinism, Type 1A and 1B | TYR | | | | o | • | • | • | • | PC467.1 |
| Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome | WNT10A | • | | | | • | • | • | • | PC314.1 |
| Omenn Syndrome, RAG2-Related | RAG2 | • | | | | • | • | • | • | PC315.1 |
| Opitz G/BBB Syndrome, X-Linked | MID1 | | • | | o | • | • | • | • | PC457.1 |
| Ornithine Aminotransferase Deficiency | OAT | • | | | | • | • | • | • | PC316.1 |
| Ornithine Transcarbamylase Deficiency | OTC | | • | | o | • | • | • | • | PC317.1 |
| Osteogenesis Imperfecta Type VII | CRTAP | • | | | | | • | • | • | PC84894.1 |
| Osteogenesis Imperfecta Type VIII | P3H1 | • | | | | | • | • | • | PC10407.1 |
| Osteogenesis Imperfecta Type XI | FKBP10 | • | | | | | • | • | • | PC1998.1 |
| Osteogenesis Imperfecta Type XIII | BMP1 | • | | | | | • | • | • | PC50252.1 |
| Osteopetrosis, Infantile Malignant, TCIRG1-Related | TCIRG1 | • | | | | • | • | • | • | PC319.1 |
| Osteopetrosis, OSTM1 -Related | OSTM1 | • | | | | | • | • | • | PC831.1 |
| Pantothenate Kinase-Associated Neurodegeneration | PANK2 | • | | | | | • | • | • | PC65400.1 |
| Papillon Lefèvre Syndrome | CTSC | • | | | | | • | • | • | PC2419.1 |
| Parkinson Disease 15 | FBXO7 | • | | | | | • | • | • | PC21026.1 |
| Pendred Syndrome | SLC26A4 | • | | | o | • | • | • | • | PC320.1 |
| Perlman Syndrome | DIS3L2 | • | | | | • | | | • | PC321.1 |
| Pgm3-Congenital Disorder Of Glycosylation | PGM3 | • | | | | | • | • | • | PC45785.1 |
| Phenylketonuria | PAH | • | | | o | • | • | • | • | PC324.1 |
| Pign-Congenital Disorder Of Glycosylation | PIGN | • | | | | | • | • | • | PC47990.1 |
| Pituitary Hormone Deficiency, Combined 3 | LHX3 | • | | | | • | • | • | • | PC326.1 |
| PLP1 Disorders | PLP1 | | • | | o | • | • | • | • | PC461.1 |
| POLG-Related Disorders | POLG | • | | | o | • | • | • | • | PC327.1 |
| Polycystic Kidney Disease, Autosomal Recessive | PKHD1 | • | | | o | • | • | • | • | PC41.1 |
| Pontocerebellar Hypoplasia, EXOSC3-Related | EXOSC3 | • | | | | • | • | • | • | PC330.1 |
| Pontocerebellar Hypoplasia, RARS2-Related | RARS2 | • | | | o | • | • | • | • | PC334.1 |
| Pontocerebellar Hypoplasia, TSEN2-Related | TSEN2 | • | | | | • | | | • | PC331.1 |
| Pontocerebellar Hypoplasia, TSEN54-Related | TSEN54 | • | | | | • | • | • | • | PC333.1 |
| Pontocerebellar Hypoplasia, Type 1A | VRK1 | • | | | | • | • | • | • | PC332.1 |
| Pontocerebellar Hypoplasia, Type 2D | SEPSECS | • | | | | • | • | • | • | PC342.1 |
| Pontocerebellar Hypoplasia, VPS53-Related | VPS53 | • | | | | • | • | • | • | PC92.1 |
| Primary Ciliary Dyskinesia, CCDC103 -Related | CCDC103 | • | | | | | • | • | • | PC40290.1 |
| Primary Ciliary Dyskinesia, CCDC39 -Related | CCDC39 | • | | | | | • | • | • | PC3011.1 |
| Primary Ciliary Dyskinesia, DNAH11 -Related | DNAH11 | • | | | | | • | • | • | PC95610.1 |
| Primary Ciliary Dyskinesia, DNAH5-Related | DNAH5 | • | | | | • | • | • | • | PC335.1 |
| Primary Ciliary Dyskinesia, DNAI1-Related | DNAI1 | • | | | | • | • | • | • | PC336.1 |
| Primary Ciliary Dyskinesia, DNAI2-Related | DNAI2 | • | | | | • | • | • | • | PC337.1 |
| Primary Congenital Glaucoma / Peters Anomaly | CYP1B1 | • | | | | • | • | • | • | PC338.1 |
| Primary Hyperoxaluria, Type 1 | AGXT | • | | | o | • | • | • | • | PC341.1 |
| Primary Hyperoxaluria, Type 2 | GRHPR | • | | | | • | • | • | • | PC339.1 |
| Primary Hyperoxaluria, Type 3 | HOGA1 | • | | | | • | • | • | • | PC340.1 |
| Primary Microcephaly 1, Autosomal Recessive | MCPH1 | | | | o | • | • | • | • | PC456.1 |
| Progressive Early-Onset Encepahlopathy With Brain Atrophy And Thin Corpus Callosum | TBCD | • | | | | | • | • | • | PC7411.1 |
| Progressive Familial Intrahepatic Cholestasis, ABCB4 -Related | ABCB4 | • | | | | | • | • | • | PC78083.1 |

| CONDITION | GENE | AUTOSOMAL RECESSIVE | X-LINKED | SCREENING RECOMMENDATIONS | | PANEL AVAILABILITY | | | | PC# |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | ACOG* | ACMG | H445 | H569 † | H574 † | H613 | |
| Progressive Familial Intrahepatic Cholestasis, Type 1 (PFIC1) | ATP8B1 | • | | | | • | • | • | • | PC74.1 |
| Progressive Familial Intrahepatic Cholestasis, Type 2 | ABCB11 | • | | | | • | • | • | • | PC343.1 |
| Progressive Familial Intrahepatic Cholestasis, Type 4 (PFIC4) | TJP2 | • | | | | • | | | • | PC75.1 |
| Progressive Pseudorheumatoid Dysplasia | CCN6 | • | | | | | • | • | • | PC74432.1 |
| Prolidase Deficiency | PEPD | • | | | | • | • | • | • | PC344.1 |
| Propionic Acidemia, PCCA-Related | PCCA | • | | | | • | • | • | • | PC346.1 |
| Propionic Acidemia, PCCB-Related | PCCB | • | | | | • | • | • | • | PC347.1 |
| Prothrombin-Related Thrombophilia | F2 | • | | | | | • | • | • | PC99249.1 |
| Pseudocholinesterase Deficiency | BCHE | • | | | | • | | | • | PC348.1 |
| Pseudoxanthoma Elasticum | ABCC6 | • | | | | • | | | • | PC349.1 |
| Pterin-4 Alpha-Carbinolamine Dehydratase (PCD) Deficiency | PCBD1 | • | | | | | • | • | • | PC6996.1 |
| Pycnodysostosis | CTSK | • | | | | • | • | • | • | PC350.1 |
| Pyridoxal 5'-Phosphate-Dependent Epilepsy | PNPO | • | | | | | • | • | • | PC13105.1 |
| Pyridoxine-Dependent Epilepsy | ALDH7A1 | • | | | | • | • | • | • | PC351.1 |
| Pyruvate Carboxylase Deficiency | PC | • | | | | • | • | • | • | PC352.1 |
| Pyruvate Dehydrogenase Deficiency, PDHB-Related | PDHB | • | | | | • | • | • | • | PC353.1 |
| Pyruvate Dehydrogenase Deficiency, X-Linked | PDHA1 | | • | | | • | • | • | • | PC354.1 |
| Refsum Disease, PHYH-Related | PHYH | • | | | | • | • | • | • | PC355.1 |
| Renal Tubular Acidosis and Deafness, ATP6V1B1-Related | ATP6V1B1 | • | | | | • | • | • | • | PC357.1 |
| Renal Tubular Acidosis, Proximal, with Ocular Abnormalities and Mental Retardation | SLC4A4 | • | | | | • | | | • | PC356.1 |
| Retinitis Pigmentosa 2 | RP2 | | • | | | | • | • | • | PC19284.1 |
| Retinitis Pigmentosa 25 | EYS | • | | | | • | • | • | • | PC358.1 |
| Retinitis Pigmentosa 26 | CERKL | • | | | | • | • | • | • | PC359.1 |
| Retinitis Pigmentosa 28 | FAM161A | • | | | | • | • | • | • | PC360.1 |
| Retinitis Pigmentosa 36 | PRCD | • | | | | | • | • | • | PC88696.1 |
| Retinitis Pigmentosa 59 | DHDDS | • | | | o | • | • | • | • | PC361.1 |
| Retinitis Pigmentosa 62 | MAK | • | | | | | • | • | • | PC4512.1 |
| Retinitis Pigmentosa, X-Linked, RPGR-Related | RPGR | | • | | o | • | | • | • | PC462.1 |
| Rhizomelic Chondrodysplasia Punctata, Type 1 | PEX7 | • | | | | • | • | • | • | PC362.1 |
| Rhizomelic Chondrodysplasia Punctata, Type 2 | GNPAT | • | | | | • | • | • | • | PC363.1 |
| Rhizomelic Chondrodysplasia Punctata, Type 3 | AGPS | • | | | | • | • | • | • | PC364.1 |
| RLBP1 -Related Retinopathy | RLBP1 | • | | | | | • | • | • | PC58824.1 |
| Roberts Syndrome | ESCO2 | • | | | | • | • | • | • | PC366.1 |
| RYR1 -Related Conditions | RYR1 | • | | | | | • | • | • | PC2100.1 |
| Salla Disease | SLC17A5 | • | | | | • | • | • | • | PC367.1 |
| Sandhoff Disease | HEXB | • | | | | • | • | • | • | PC368.1 |
| Schimke Immunoosseous Dysplasia | SMARCAL1 | • | | | | • | • | • | • | PC369.1 |
| Schindler Disease | NAGA | | | | o | • | • | • | • | PC459.1 |
| Segawa Syndrome, TH-Related | TH | • | | | | • | • | • | • | PC393.1 |
| Senior-Loken Syndrome 4 / Nephronophthisis 4 | NPHP4 | • | | | | • | | | • | PC371.1 |
| Sepiapterin Reductase Deficiency | SPR | • | | | | | • | • | • | PC3062.1 |
| Severe Combined Immunodeficiency (SCID ), CD3D -Related | CD3D | • | | | | | • | • | • | PC62873.1 |
| Severe Combined Immunodeficiency (SCID ), CD3E -Related | CD3E | • | | | | | • | • | • | PC32580.1 |
| Severe Combined Immunodeficiency (SCID ), FOXN1 -Related | FOXN1 | • | | | | | • | • | • | PC71077.1 |
| Severe Combined Immunodeficiency (SCID ), IKBKB -Related | IKBKB | • | | | | | • | • | • | PC6785.1 |
| Severe Combined Immunodeficiency (SCID ), Il7R-Related | IL7R | • | | | | | • | • | • | PC834.1 |
| Severe Combined Immunodeficiency (SCID ), JAK3 -Related | JAK3 | • | | | | | • | • | • | PC79789.1 |
| Severe Combined Immunodeficiency (SCID ), PTPRC -Related | PTPRC | • | | | | | • | • | • | PC95704.1 |
| Severe Combined Immunodeficiency, ADA-Related | ADA | • | | | | • | • | • | • | PC373.1 |
| Severe Combined Immunodeficiency, RAG1-Related | RAG1 | • | | | | • | • | • | • | PC375.1 |
| Severe Combined Immunodeficiency, Type Athabaskan | DCLRE1C | • | | | | • | • | • | • | PC374.1 |
| Severe Combined Immunodeficiency, X-Linked | IL2RG | | • | | | • | • | • | • | PC370.1 |
| Short-Rib Thoracic Dysplasia 3 with or without Polydactyly | DYNC2H1 | | | | o | • | • | • | • | PC451.1 |
| Shwachman-Diamond Syndrome, SBDS-Related | SBDS | • | | | | • | | | • | PC376.1 |
| Sialidosis | NEU1 | • | | | | • | • | • | • | PC377.1 |
| Sjögren-Larsson Syndrome | ALDH3A2 | • | | | | • | • | • | • | PC380.1 |
| Smith-Lemli-Opitz Syndrome | DHCR7 | • | | o | o | • | • | • | • | PC381.1 |
| Spastic Paraplegia, Type 15 | ZFYVE26 | • | | | | • | • | • | • | PC382.1 |
| Spastic Tetraplegia, Thin Corpus Callosum, and Progressive Microcephaly (SPATCCM) | SLC1A4 | • | | | | • | • | • | • | PC116.1 |
| SPG11 -Related Conditions | SPG11 | • | | | | | • | • | • | PC44490.1 |
| Spinal Muscular Atrophy | SMN1 | • | | o | o | • | • | • | • | PC384.1 |
| Spinal Muscular Atrophy With Respiratory Distress Type 1 | IGHMBP2 | • | | | | | • | • | • | PC35651.1 |
| Spinocerebellar Ataxia, Autosomal Recessive 10 | ANO10 | | | | o | • | • | • | • | PC447.1 |
| Spinocerebellar Ataxia, Autosomal Recessive 12 | WWOX | • | | | | • | | | • | PC385.1 |
| Spondylocostal Dysostosis 1 | DLL3 | • | | | | | • | • | • | PC35050.1 |
| Spondylothoracic Dysostosis, MESP2-Related | MESP2 | • | | | | • | • | • | • | PC386.1 |
| Steel Syndrome | COL27A1 | • | | | | • | • | • | • | PC387.1 |
| Steroid-Resistant Nephrotic Syndrome | NPHS2 | • | | | | • | • | • | • | PC300.1 |
| Stuve-Wiedemann Syndrome | LIFR | • | | | | • | • | • | • | PC388.1 |
| SURF1 -Related Conditions | SURF1 | • | | | | | • | • | • | PC5533.1 |

| CONDITION | GENE | AUTOSOMAL RECESSIVE | X-LINKED | SCREENING RECOMMENDATIONS | | PANEL AVAILABILITY | | | | PC# |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | ACOG* | ACMG | H445 | H569 † | H574 † | H613 | |
| Surfactant Dysfunction, ABCA3-Related | ABCA3 | | | | o | • | • | • | • | PC445.1 |
| Tay-Sachs Disease | HEXA | • | | o | o | • | • | • | • | PC389.1 |
| TBCE -Related Conditions | TBCE | • | | | | | • | • | • | PC93469.1 |
| Thiamine-Responsive Megaloblastic Anemia Syndrome | SLC19A2 | • | | | | | • | • | • | PC85692.1 |
| Thyroid Dyshormonogenesis 1 | SLC5A5 | • | | | | | • | • | • | PC12709.1 |
| Thyroid Dyshormonogenesis 2A | TPO | • | | | | | • | • | • | PC78490.1 |
| Thyroid Dyshormonogenesis 3 | TG | • | | | | | • | • | • | PC88592.1 |
| Thyroid Dyshormonogenesis 6 | DUOX2 | • | | | | | • | • | • | PC44584.1 |
| Transcobalamin II Deficiency | TCN2 | • | | | | | • | • | • | PC24244.1 |
| Trichohepatoenteric Syndrome, SKIV2L -Related | SKIC2 | • | | | | | • | • | • | PC84765.1 |
| Trichohepatoenteric Syndrome, TTC37-Related | TTC37 | • | | | | • | • | • | • | PC390.1 |
| Trichothiodystrophy 1 / Xeroderma Pigmentosum, Group D | ERCC2 | • | | | o | • | • | • | • | PC391.1 |
| Trimethylaminuria | FMO3 | | | | o | • | • | • | • | PC453.1 |
| Triple A Syndrome | AAAS | • | | | | • | • | • | • | PC392.1 |
| TSHR -Related Conditions | TSHR | • | | | | | • | • | • | PC96846.1 |
| Tyrosinemia Type III | HPD | • | | | | | • | • | • | PC72043.1 |
| Tyrosinemia, Type 1 | FAH | • | | | o | • | • | • | • | PC394.1 |
| Tyrosinemia, Type 2 | TAT | • | | | | • | • | • | • | PC395.1 |
| Usher Syndrome, Type 1B | MYO7A | • | | | | • | • | • | • | PC396.1 |
| Usher Syndrome, Type 1C | USH1C | • | | | | • | • | • | • | PC397.1 |
| Usher Syndrome, Type 1D | CDH23 | • | | | | • | • | • | • | PC398.1 |
| Usher Syndrome, Type 1F | PCDH15 | • | | o | o | • | • | • | • | PC399.1 |
| Usher Syndrome, Type 1J / Deafness, Autosomal Recessive, 48 | CIB2 | • | | | | • | | | • | PC401.1 |
| Usher Syndrome, Type 2A | USH2A | • | | | o | • | • | • | • | PC402.1 |
| Usher Syndrome, Type 2C | ADGRV1 | • | | | | • | • | • | • | PC400.1 |
| Usher Syndrome, Type 3 | CLRN1 | • | | o | o | • | • | • | • | PC403.1 |
| Very Long-Chain Acyl-CoA Dehydrogenase Deficiency | ACADVL | • | | | o | • | • | • | • | PC405.1 |
| Viatmin D-Dependent Rickets Type 2A | VDR | • | | | | | • | • | • | PC1753.1 |
| Vici Syndrome | EPG5 | • | | | | | • | • | • | PC87787.1 |
| Vitamin D Dependent Rickets, Type 1A | CYP27B1 | • | | | o | • | • | • | • | PC404.1 |
| Vldlr-Associated Cerebellar Hypoplasia | VLDLR | • | | | | | • | • | • | PC34059.1 |
| Walker-Warburg Syndrome, FKTN-Related | FKTN | • | | | o | • | • | • | • | PC154.1 |
| Walker-Warburg Syndrome, ISPD-Related | ISPD | • | | | | • | | | • | PC292.1 |
| Walker-Warburg Syndrome, LARGE1-Related | LARGE1 | • | | | | • | • | • | • | PC291.1 |
| Walker-Warburg Syndrome, POMT1-Related | POMT1 | • | | | | • | • | • | • | PC406.1 |
| Walker-Warburg Syndrome, POMT2-Related | POMT2 | • | | | | • | • | • | • | PC290.1 |
| Warsaw Breakage Syndrome | DDX11 | • | | | | | • | • | • | PC24438.1 |
| Werner Syndrome | WRN | • | | | | • | • | • | • | PC407.1 |
| Wilson Disease | ATP7B | • | | | o | • | • | • | • | PC408.1 |
| Wiskott-Aldrich Syndrome | WAS | | • | | | • | • | • | • | PC409.1 |
| Wolcott-Rallison Syndrome | EIF2AK3 | • | | | | • | • | • | • | PC410.1 |
| Wolman Disease | LIPA | • | | | | • | • | • | • | PC411.1 |
| Woodhouse-Sakati Syndrome | DCAF17 | • | | | | | • | • | • | PC75733.1 |
| Xeroderma Pigmentosum Variant Type | POLH | • | | | | | • | • | • | PC17865.1 |
| Xeroderma Pigmentosum, Group A | XPA | • | | | | • | • | • | • | PC416.1 |
| Xeroderma Pigmentosum, Group C | XPC | • | | | o | • | • | • | • | PC417.1 |
| X-Linked Chondrodysplasia Punctata 1 | ARSL | | • | | | • | • | • | • | PC413.1 |
| X-Linked Lissencephaly with Abnormal Genitalia | ARX | | • | | o | • | • | • | • | PC130.1 |
| Zellweger Spectrum Disorder, PEX13 -Related | PEX13 | • | | | | | • | • | • | PC61396.1 |
| Zellweger Spectrum Disorder, PEX16 -Related | PEX16 | • | | | | | • | • | • | PC62859.1 |
| Zellweger Spectrum Disorder, PEX5 -Related | PEX5 | • | | | | | • | • | • | PC8343.1 |
| Zellweger Spectrum Disorders, PEX10-Related | PEX10 | • | | | | • | • | • | • | PC418.1 |
| Zellweger Spectrum Disorders, PEX12-Related | PEX12 | • | | | | • | • | • | • | PC322.1 |
| Zellweger Spectrum Disorders, PEX1-Related | PEX1 | • | | | | • | • | • | • | PC419.1 |
| Zellweger Spectrum Disorders, PEX26-Related | PEX26 | • | | | | • | • | • | • | PC421.1 |
| Zellweger Spectrum Disorders, PEX2-Related | PEX2 | • | | | | • | • | • | • | PC420.1 |
| Zellweger Spectrum Disorders, PEX6-Related | PEX6 | • | | | | • | • | • | • | PC323.1 |

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