

CONDITION	GENE	AUTOSOMAL RECESSIVE	X-LINKED	SCREENING RECOMMENDATIONS			PANEL AVAILABILITY				
				ACOG*	ACMG	VICTOR CENTER	H 4	H 14	H 27	H 106	H 274
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	HSD3B2	•									•
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	HMGCL	•									•
3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	MCCC1	•									•
3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	MCCC2	•									•
3-Phosphoglycerate Dehydrogenase Deficiency	PHGDH	•				o				•	•
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	PTS	•									•
Abetalipoproteinemia	MTTP	•				o				•	•
Achondrogenesis, Type 1B	SLC26A2	•									•
Achromatopsia, CNGB3-Related	CNGB3	•									•
Acrodermatitis Enteropathica	SLC39A4	•									•
Acute Infantile Liver Failure, TRMU-Related	TRMU	•								•	•
Acyl-CoA Oxidase I Deficiency	ACOX1	•									•
Adrenoleukodystrophy, X-Linked	ABCD1		•							•	•
Aicardi-Goutières Syndrome	SAMHD1	•									•
Alpha-Thalassemia Intellectual Disability Syndrome	ATRX		•								•
Alpha-Mannosidosis	MAN2B1	•									•
Alpha-Thalassemia	HBA1/HBA2	•			o			•	•	•	•
Alport Syndrome, COL4A3-Related	COL4A3	•				o				•	•
Alport Syndrome, COL4A4-Related	COL4A4	•									•
Alport Syndrome, X-Linked	COL4A5		•								•
Alstrom Syndrome	ALMS1	•									•
Andermann Syndrome	SLC12A6	•									•
Argininosuccinate Lyase Deficiency	ASL	•									•
Aromatase Deficiency	CYP19A1	•									•
Asparagine Synthetase Deficiency	ASNS	•								•	•
Aspartylglycosaminuria	AGA	•									•
Ataxia with Vitamin E Deficiency	TTPA	•									•
Ataxia-Telangiectasia	ATM	•									•
Autism Spectrum, Epilepsy and Arthrogyposis	SLC35A3	•				o				•	•
Autoimmune Polyglandular Syndrome, Type 1	AIRE	•								•	•
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	SACS	•									•
Bardet-Biedl Syndrome, BBS10-Related	BBS10	•									•
Bardet-Biedl Syndrome, BBS12-Related	BBS12	•									•
Bardet-Biedl Syndrome, BBS1-Related	BBS1	•									•
Bardet-Biedl Syndrome, BBS2-Related	BBS2	•				o				•	•
Bare Lymphocyte Syndrome, CIITA-Related	CIITA	•									•
Bartter Syndrome, BSND-Related	BSND	•									•
Batten Disease, CLN3-Related	CLN3	•								•	•
Beta-Hemoglobinopathies (including sickle cell disease)	HBB	•			o			•	•	•	•
Bilateral Frontoparietal Polymicrogyria	GPR56	•									•
Biotinidase Deficiency	BTBD	•									•
Bloom Syndrome	BLM	•			o	o	o		•	•	•
Canavan Disease	ASPA	•			o	o	o		•	•	•
Carbamoyl Phosphate Synthetase I Deficiency	CPS1	•									•
Carnitine Deficiency	SLC22A5	•									•
Carnitine Palmitoyltransferase IA Deficiency	CPT1A	•									•
Carnitine Palmitoyltransferase II Deficiency	CPT2	•					o			•	•
Carpenter Syndrome	RAB23	•									•
Cartilage-Hair Hypoplasia	RMRP	•									•
Cerebrotendinous Xanthomatosis	CYP27A1	•								•	•
Charcot-Marie-Tooth Disease with Deafness, X-Linked	GJB1		•								•
Charcot-Marie-Tooth Disease, Type 4D	NDRG1	•									•
Choreoacanthocytosis	VPS13A	•								•	•
Choroideremia	CHM		•								•
Chronic Granulomatous Disease, CYBA-Related	CYBA	•								•	•
Chronic Granulomatous Disease, X-Linked	CYBB		•								•
Ciliopathies, RPGRIP1L-Related	RPGRIP1L	•									•
Citrin Deficiency	SLC25A13	•									•
Citrullinemia, Type I	ASS1	•							•	•	•
Cohen Syndrome	VPS13B	•									•
Combined Malonic and Methylmalonic Aciduria	ACSF3	•									•
Combined Oxidative Phosphorylation Deficiency (Complex 4 Deficiency)	GFM1	•									•
Combined Oxidative Phosphorylation Deficiency 3	TSMF	•									•
Combined Pituitary Hormone Deficiency-2	PROP1	•									•
Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficiency	CYP17A1	•									•
Congenital Amegakaryocytic Thrombocytopenia	MPL	•				o				•	•

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Hypohidrotic Ectodermal Dysplasia, X-Linked	EDA		•								•
Hypophosphatasia, ALPL-Related	ALPL	•									•
Inclusion Body Myopathy 2	GNE	•								•	•
Infantile Cerebral and Cerebellar Atrophy	MED17	•								•	•
Isovaleric Acidemia	IVD	•							•	•	•
Joubert Syndrome 2 / Meckel Syndrome 2	TMEM216	•		o		o				•	•
Juvenile Retinoschisis, X-Linked	RS1		•								•
Ketothiolase Deficiency	ACAT1	•									•
Krabbe Disease	GALC	•									•
Lamellar Ichthyosis, Type 1	TGM1	•									•
Leber Congenital Amaurosis	LCA5	•									•
Leber Congenital Amaurosis 2	RPE65	•								•	•
Leber Congenital Amaurosis, Type CEP290	CEP290	•									•
Leber Congenital Amaurosis, Type RDH12	RDH12	•									•
Leigh Syndrome, French-Canadian Type	LRPPRC	•									•
Lethal Congenital Contracture Syndrome 1	GLE1	•									•
Leukoencephalopathy with Vanishing White Matter	EIF2B5	•									•
Limb-Girdle Muscular Dystrophy, Type 2A	CAPN3	•									•
Limb-Girdle Muscular Dystrophy, Type 2B	DYSF	•								•	•
Limb-Girdle Muscular Dystrophy, Type 2I	FKRP	•									•
Limb-Girdle Muscular Dystrophy, Type 2C	SGCG	•									•
Limb-Girdle Muscular Dystrophy, Type 2D	SGCA	•									•
Limb-Girdle Muscular Dystrophy, Type 2E	SGCB	•									•
Lipoamide Dehydrogenase Deficiency (Dihydropolipoamide Dehydrogenase Deficiency)	DLD	•				o				•	•
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 1A	BCKDHA	•									•
Maple Syrup Urine Disease, Type 1B	BCKDHB	•		o		o				•	•
Meckel-Gruber Syndrome, Type 1	MKS1	•									•
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM	•		o				•	•	•	•
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1	•								•	•
Menkes Syndrome	ATP7A		•								•
Metachromatic Leukodystrophy, PSAP-Related	PSAP	•									•
Metachromatic Leukodystrophy, ARSA-Related	ARSA	•								•	•
Methylmalonic Aciduria and Homocystinuria, Type cblC	MMACHC	•							•	•	•
Methylmalonic Aciduria and Homocystinuria, Type cblD	MMADHC	•									•
Methylmalonic Aciduria, MMAA-Related	MMAA	•									•
Methylmalonic Aciduria, MMAB-Related	MMAB	•									•
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	•								•	•
Mucopolipidosis II/IIIA	GNPTAB	•									•
Mucopolipidosis III gamma	GNPTG	•									•
Mucopolipidosis, Type IV	MCOLN1	•		o	o	o			•	•	•
Mucopolysaccharidosis, Type IIIA (Sanfilippo A)	SGSH	•									•
Mucopolysaccharidosis Type IX	HYAL1	•									•
Mucopolysaccharidosis, Type I (Hurler Syndrome)	IDUA	•							•	•	•
Mucopolysaccharidosis, Type II (Hunter Syndrome)	IDS		•								•
Mucopolysaccharidosis, Type IIIB (Sanfilippo B)	NAGLU	•									•
Mucopolysaccharidosis, Type IIIC (Sanfilippo C)	HGSNAT	•									•
Mucopolysaccharidosis, Type IIID (Sanfilippo D)	GNS	•									•
Mucopolysaccharidosis, Type IVB / GM1 Gangliosidosis	GLB1	•									•
Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)	ARSB	•									•
Multiple Sulphatase Deficiency	SUMF1	•				o				•	•
Muscle-Eye-Brain Disease, POMGNT1-Related	POMGNT1	•									•
Myoneurogastrointestinal Encephalopathy (MNGIE)	TYMP	•								•	•
Myotubular Myopathy, X-Linked	MTM1		•								•
N-acetylglutamate Synthase Deficiency	NAGS	•									•
Nemaline Myopathy, NEB-Related	NEB	•				o				•	•
Neuronal Ceroid Lipofuscinosis, CLN5-Related	CLN5	•									•
Neuronal Ceroid Lipofuscinosis, MFSD8-Related	MFSD8	•									•
Neuronal Ceroid Lipofuscinosis, PPT1-Related	PPT1	•									•
Neuronal Ceroid Lipofuscinosis, TPP1-Related	TPP1	•									•
Neuronal Ceroid-Lipofuscinosis, CLN6-Related	CLN6	•									•
Neuronal Ceroid-Lipofuscinosis, CLN8-Related	CLN8	•									•
Niemann-Pick Disease, Types C1/D	NPC1	•		o							•
Niemann-Pick Disease, Type C2	NPC2	•		o							•

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Niemann-Pick Disease, Types A/B	SMPD1	•		o	o	o			•	•	•
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		•								•
Osteopetrosis, Infantile Malignant, TCIRG1-Related	TCIRG1	•								•	•
Pendred Syndrome	SLC26A4	•									•
Phenylketonuria	PAH	•		o						•	•
Pituitary Hormone Deficiency, Combined 3	LHX3	•									•
Polycystic Kidney Disease, Autosomal Recessive	PKHD1	•				o		•	•	•	•
Pontocerebellar Hypoplasia, RARS2-Related	RARS2	•								•	•
Pontocerebellar Hypoplasia, Type 1A	VRK1	•								•	•
Pontocerebellar Hypoplasia, Type 2D	SEPSECS	•								•	•
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	•								•	•
Progressive Familial Intrahepatic Cholestasis, Type 2	ABCB11	•									•
Propionic Acidemia, PCCA-Related	PCCA	•									•
Propionic Acidemia, PCCB-Related	PCCB	•									•
Pycnodysostosis	CTSK	•									•
Pyruvate Dehydrogenase Deficiency, PDHB-Related	PDHB	•									•
Pyruvate Dehydrogenase Deficiency, X-Linked	PDHA1		•								•
Renal Tubular Acidosis and Deafness, ATP6V1B1-Related	ATP6V1B1	•								•	•
Retinitis Pigmentosa 25	EYS	•									•
Retinitis Pigmentosa 26	CERKL	•									•
Retinitis Pigmentosa 28	FAM161A	•									•
Retinitis Pigmentosa 59	DHDDS	•				o				•	•
Rhizomelic Chondrodysplasia Punctata, Type 3	AGPS	•									•
Rhizomelic Chondrodysplasia Punctata, Type 1	PEX7	•						•	•	•	•
Roberts Syndrome	ESCO2	•									•
Salla Disease	SLC17A5	•									•
Sandhoff Disease	HEXB	•									•
Schimke Immunoosseous Dysplasia	SMARCAL1	•									•
Segawa Syndrome, TH-Related	TH	•									•
Severe Combined Immunodeficiency, ADA-Related	ADA	•									•
Severe Combined Immunodeficiency, Type Athabaskan	DCLRE1C	•									•
Severe Combined Immunodeficiency, X-Linked	IL2RG		•								•
Sjogren-Larsson Syndrome	ALDH3A2	•									•
Smith-Lemli-Opitz Syndrome	DHCR7	•		o		o		•	•	•	•
Spinal Muscular Atrophy	SMN1	•		o	o	o	•	•	•	•	•
Spondylothoracic Dysostosis, MESP2-Related	MESP2	•									•
Steroid-Resistant Nephrotic Syndrome	NPHS2	•									•
Stuve-Wiedemann Syndrome	LIFR	•									•
Tay-Sachs Disease	HEXA	•		o	o	o		•	•	•	•
Tyrosinemia, Type I	FAH	•				o		•	•	•	•
Usher Syndrome, Type 1B	MYO7A	•									•
Usher Syndrome, Type 1C	USH1C	•									•
Usher Syndrome, Type 1D	CDH23	•									•
Usher Syndrome, Type 1F	PCDH15	•		o		o				•	•
Usher Syndrome, Type 2A	USH2A	•								•	•
Usher Syndrome, Type 3	CLRN1	•		o		o				•	•
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	•									•
Walker-Warburg Syndrome, FKTN-Related	FKTN	•				o				•	•
Wilson Disease	ATP7B	•				o				•	•
Wolman Disease	LIPA	•								•	•
Zellweger Spectrum Disorders, PEX10-Related	PEX10	•									•
Zellweger Spectrum Disorders, PEX1-Related	PEX1	•						•	•	•	•
Zellweger Spectrum Disorders, PEX2-Related	PEX2	•				o				•	•
Zellweger Spectrum Disorders, PEX6-Related	PEX6	•								•	•

* Note that ACOG screening recommendations listed here include diseases in ACOG Committee Opinion 690, example panel, as well as the diseases listed in ACOG Committee Opinion 691.