Sema4 carrier screening guide

Choosing the best panel for your patient

sema4

Comprehensive options to help guide family planning

This guide provides information about all of the diseases and genes covered by Sema4's carrier screening panels. Sema4 offers a wide range of panels. Testing can be performed for more than 280 genes, a smaller subset of genes, or even just 1 gene. All of our carrier screening technologies are >95% accurate.

Sema4 Expanded Carrier Screen

283 genes

Sema4 Expanded Carrier Screen (ECS) is one of the most comprehensive carrier screens available. It screens for inherited disorders associated with 283 genes, including many conditions that are prevalent in people of certain ethnic backgrounds. For example, our Expanded Carrier Screen includes a comprehensive Jewish carrier screen for 101 genes.

Sema4 Expanded Carrier Screen

152 genes

The Expanded Carrier Screen panel of 152 genes includes 84 genes recommended for expanded carrier screening panels by Stevens, et al.* based on a 2013 position statement from American College of Medical Genetics and Genomics (ACMG) and American College of Obstetricians and Gynecologists (ACOG) Committee Opinion 690. It also includes an additional 53 genes from our comprehensive Jewish carrier screening panel and 15 other genes with a carrier frequency of >1 in 100 in an ethnic subgroup.

Sema4 Expanded Carrier Screen

39 genes

The Expanded Carrier Screen panel of 39 genes includes 23 genes highlighted in ACOG Committee Opinion 690: Carrier Screening in the Age of Genomic Medicine. It also includes 16 additional higher-frequency genes associated with conditions such as Duchenne muscular dystrophy, autosomal recessive polycystic kidney disease, and congenital disorder of glycosylation, type 1A.

Comprehensive Jewish carrier screen

101 genes

The comprehensive Jewish carrier screen tests for variants in 101 genes associated with genetic diseases found at an increased frequency in the Ashkenazi Jewish (Central and Eastern European), Sephardi Jewish (Southern European and Northern African), and Mizrahi Jewish (Middle Eastern/Arab) populations.

Ashkenazi Jewish carrier screen

64 genes

The Ashkenazi Jewish carrier screen tests for variants in 64 genes associated with genetic diseases found at an increased frequency in the Ashkenazi Jewish population. Because of the serious and life-threatening nature of several of these conditions and the lack of available treatment, prevention is the best strategy for combating these diseases. Carrier screening and appropriate genetic counseling can be used to dramatically reduce the incidence of these disorders in the Ashkenazi Jewish population.

*Stevens B, et al. Finding middle ground in constructing a clinically useful expanded carrier screening panel. Obstet Gynecol 2017;130(2):279-284.

Sephardi-Mizrahi Jewish carrier screen

54 genes

There are several pathogenic variants that occur at increased frequencies in the Sephardi Jewish (Southern European and Northern African) and Mizrahi Jewish (Middle Eastern/Arab) populations. Our Sephardi-Mizrahi Jewish carrier screen covers 54 genes that fall into this category.

East Asian carrier screen

95 genes

This panel includes 95 genes reported to have an increased carrier frequency in the East Asian population, such as *USH2A* (Usher syndrome, type 2A), *SLC12A3* (Gitelman syndrome), and *SLC26A4* (Pendred syndrome). Genes with known founder mutations in the East Asian population like, *SLC25A13* (citrin deficiency), *ATP7B* (Wilson disease), and *GJB2* (non-syndromic hearing loss), are also covered by this panel.

High frequency pan-ethnic panel

11 genes

The high frequency pan-ethnic panel provides carrier screening for the following genetic disorders due to the relatively elevated carrier frequencies and high detection rates in most ethnic groups with severe, early onset clinical presentation.

Standard pan-ethnic panel

4 genes

The standard pan-ethnic panel is a basic carrier screening panel that covers cystic fibrosis (CF), fragile X syndrome, Smith-Lemli-Opitz syndrome (SLOS), and spinal muscular atrophy (SMA). This panel was developed based on the recommendations from ACOG Committee Opinion 691: Carrier Screening for Genetic Conditions.

If a customized panel is desired, any subset of the 283 genes included on the Expanded Carrier Screen panel may be selected for testing.

Conditions covered by our panels

Disease	Expanded Carrier Screen (283 genes)	Expanded Carrier Screen (152 genes)	Expanded Carrier Screen (39 genes)
Abetalipoproteinemia *(MTTP)*	✓	✓	
Achromatopsia *(CNGB3)*	✓	✓	
Acrodermatitis Enteropathica *(SLC39A4)*	✓		
Acute Infantile Liver Failure *(TRMU)*	✓	✓	
Acyl-CoA Oxidase I Deficiency *(ACOX1)*	✓		
Adenosine Deaminase Deficiency *(ADA)*	✓	✓	
Adrenoleukodystrophy, X-Linked *(ABCD1)*	✓	✓	
Aicardi-Goutières Syndrome *(SAMHD1)*	✓		
Alpha-Mannosidosis *(MAN2B1)*	✓		
Alpha-Thalassemia *(HBA1 and HBA2)*	✓	✓	✓
Alpha-Thalassemia Mental Retardation Syndrome *(ATRX)*	✓		
Alport Syndrome *(COL4A3)*	✓	✓	
Alport Syndrome *(COL4A4)*	✓		
Alport Syndrome *(COL4A5)*	✓		
Alstrom Syndrome *(ALMS1)*	✓		
Andermann Syndrome *(SLC12A6)*	✓	✓	
Argininosuccinic Aciduria *(ASL)*	✓	✓	
Aromatase Deficiency *(CYP19A1)*	✓		
Arthrogryposis, Mental Retardation, and Seizures *(SLC35A3)*	✓	✓	
Asparagine Synthetase Deficiency *(ASNS)*	✓	✓	
Aspartylglycosaminuria *(AGA)*	✓	✓	
Ataxia with Isolated Vitamin E Deficiency *(TTPA)*	✓		
Ataxia-Telangiectasia *(ATM)*	✓	✓	
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay *(SACS)*	✓	✓	
Bardet-Biedl Syndrome *(BBS1)*	✓	✓	
Bardet-Biedl Syndrome *(BBS2)*	✓	✓	
Bardet-Biedl Syndrome *(BBS10)*	✓		
Bardet-Biedl Syndrome *(BBS12)*	✓		
Bare Lymphocyte Syndrome, Type II *(CIITA)*	✓		

	Comprehensive Jewish carrier screen (101 genes)	Ashkenazi Jewish carrier screen (64 genes)	Sephardi-Mizrahi Jewish carrier screen (54 genes)	East Asian carrier screen (95 genes)	High frequency pan-ethnic carrier screen (11 genes)	Standard pan-ethnic carrier screen (4 genes)
	✓	✓				
	✓		✓			
				✓		
	✓		✓	✓		
	✓	✓	✓	✓	✓	
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	✓	✓				

Conditions covered by our panels

Disease	Expanded Carrier Screen (283 genes)	Expanded Carrier Screen (152 genes)	Expanded Carrier Screen (39 genes)
Bartter Syndrome, Type 4A *(BSND)*	✓		
Bernard-Soulier Syndrome, Type A1 *(GP1BA)*	✓		
Bernard-Soulier Syndrome, Type C *(GP9)*	✓		
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency *(HSD3B2)*	✓		
Beta-Globin-Related Hemoglobinopathies *(HBB)*	✓	✓	✓
Beta-Ketothiolase Deficiency *(ACAT1)*	✓		
Bilateral Frontoparietal Polymicrogyria *(GPR56)*	✓		
Biotinidase Deficiency *(BTD)*	✓	✓	
Bloom Syndrome *(BLM)*	✓	✓	✓
Canavan Disease *(ASPA)*	✓	✓	✓
Carbamoylphosphate Synthetase I Deficiency *(CPS1)*	✓		
Carnitine Palmitoyltransferase IA Deficiency *(CPT1A)*	✓		
Carnitine Palmitoyltransferase II Deficiency *(CPT2)*	✓	✓	
Carpenter Syndrome *(RAB23)*	✓		
Cartilage-Hair Hypoplasia *(RMRP)*	✓	✓	
Cerebral Creatine Deficiency Syndrome 1 *(SLC6A8)*	✓		
Cerebral Creatine Deficiency Syndrome 2 *(GAMT)*	✓		
Cerebrotendinous Xanthomatosis *(CYP27A1)*	✓	✓	
Charcot-Marie-Tooth Disease, Type 4D *(NDRG1)*	✓		
Charcot-Marie-Tooth Disease, Type 5 / Arts Syndrome *(PRPS1)*	✓		
Charcot-Marie-Tooth Disease, X-Linked *(GJB1)*	✓		
Choreoacanthocytosis *(VPS13A)*	✓	✓	
Choroideremia *(CHM)*	✓		
Chronic Granulomatous Disease *(CYBA)*	✓	✓	
Chronic Granulomatous Disease *(CYBB)*	✓		
Citrin Deficiency *(SLC25A13)*	✓	✓	
Citrullinemia, Type 1 *(ASS1)*	✓		
Cohen Syndrome *(VPS13B)*	✓		
Combined Malonic and Methylmalonic Aciduria *(ACSF3)*	✓	✓	

	Comprehensive Jewish carrier screen (101 genes)	Ashkenazi Jewish carrier screen (64genes)	Sephardi-Mizrahi Jewish carrier screen (54 genes)	East Asian carrier screen (95 genes)	High frequency pan-ethnic carrier screen (11 genes)	Standard pan-ethnic carrier screen (4 genes)
	✓	✓	✓	✓	✓	
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				✓		

Conditions covered by our panels

Disease	Expanded Carrier Screen (283 genes)	Expanded Carrier Screen (152 genes)	Expanded Carrier Screen (39 genes)
Combined Oxidative Phosphorylation Deficiency 1 *(GFM1)*	✓		
Combined Oxidative Phosphorylation Deficiency 3 *(TSFM)*	✓	✓	
Combined Pituitary Hormone Deficiency 2 *(PROP1)*	✓	✓	
Combined Pituitary Hormone Deficiency 3 *(LHX3)*	✓		
Combined SAP Deficiency *(PSAP)*	✓		
Congenital Adrenal Hyperplasia due to17-Alpha-Hydroxylase Deficiency *(CYP17A1)*	✓		
Congenital Adrenal Hyperplasia due to 21-Alpha-Hydroxylase Deficiency *(CYP21A2)*	✓		
Congenital Amegakaryocytic Thrombocytopenia *(MPL)*	✓	✓	
Congenital Disorder of Glycosylation, Type Ia *(PMM2)*	✓	✓	✓
Congenital Disorder of Glycosylation, Type Ib *(MPI)*	✓		
Congenital Disorder of Glycosylation, Type Ic *(ALG6)*	✓		
Congenital Insensitivity to Pain with Anhidrosis *(NTRK1)*	✓	✓	
Congenital Myasthenic Syndrome *(CHRNE)*	✓		
Congenital Myasthenic Syndrome *(RAPSN)*	✓	✓	
Congenital Neutropenia *(HAX1)*	✓		
Congenital Neutropenia *(VPS45)*	✓		
Corneal Dystrophy and Perceptive Deafness *(SLC4A11)*	✓		
Corticosterone Methyloxidase Deficiency *(CYP11B2)*	✓	✓	
Cystic Fibrosis *(CFTR)*	✓	✓	✓
Cystinosis *(CTNS)*	✓	✓	
D-Bifunctional Protein Deficiency *(HSD17B4)*	✓		
Deafness, Autosomal Recessive 77 *(LOXHD1)*	✓	✓	
Duchenne Muscular Dystrophy / Becker Muscular Dystrophy *(DMD)*	✓	✓	✓
Dyskeratosis Congenita *(RTEL1)*	✓	✓	
Dystrophic Epidermolysis Bullosa *(COL7A1)*	✓	✓	
Ehlers-Danlos Syndrome, Type VIIC *(ADAMTS2)*	✓	✓	
Ellis-van Creveld Syndrome *(EVC)*	✓	✓	
Emery-Dreifuss Myopathy 1 *(EMD)*	✓		
Enhanced S-Cone Syndrome *(NR2E3)*	✓	✓	

	Comprehensive Jewish carrier screen (101 genes)	Ashkenazi Jewish carrier screen (64 genes)	Sephardi-Mizrahi Jewish carrier screen (54 genes)	East Asian carrier screen (95 genes)	High frequency pan-ethnic carrier screen (11 genes)	Standard pan-ethnic carrier screen (4 genes)
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Conditions covered by our panels

Disease	Expanded Carrier Screen (283 genes)	Expanded Carrier Screen (152 genes)	Expanded Carrier Screen (39 genes)
Ethylmalonic Encephalopathy *(ETHE1)*	✓		
Fabry Disease *(GLA)*	✓		
Factor IX Deficiency *(F9)*	✓		
Factor XI Deficiency *(F11)*	✓	✓	
Familial Autosomal Recessive Hypercholesterolemia *(LDLRAP1)*	✓		
Familial Dysautonomia *(IKBKAP)*	✓	✓	✓
Familial Hypercholesterolemia *(LDLR)*	✓	✓	
Familial Hyperinsulinism *(ABCC8)*	✓	✓	✓
Familial Hyperinsulinism *(KCNJ11)*	✓	✓	
Familial Mediterranean Fever *(MEFV)*	✓	✓	
Fanconi Anemia, Group A *(FANCA)*	✓	✓	
Fanconi Anemia, Group C *(FANCC)*	✓	✓	✓
Fanconi Anemia, Group G *(FANCG)*	✓	✓	
Fragile X Syndrome *(FMR1)*	✓	✓	✓
Fumarase Deficiency *(FH)*	✓	✓	
Galactokinase Deficiency *(GALK1)*	✓	✓	
Galactosemia *(GALT)*	✓	✓	✓
Gaucher Disease *(GBA)*	✓	✓	✓
Gitelman Syndrome *(SLC12A3)*	✓	✓	
Glutaric Acidemia, Type I *(GCDH)*	✓	✓	
Glutaric Acidemia, Type IIa *(ETFA)*	✓		
Glutaric Acidemia, Type IIc *(ETFDH)*	✓	✓	
Glycine Encephalopathy *(AMT)*	✓		
Glycine Encephalopathy *(GLDC)*	✓		
Glycogen Storage Disease, Type Ia *(G6PC)*	✓	✓	✓
Glycogen Storage Disease, Type Ib *(SLC37A4)*	✓		
Glycogen Storage Disease, Type II *(GAA)*	✓	✓	
Glycogen Storage Disease, Type III *(AGL)*	✓	✓	
Glycogen Storage Disease, Type IV / Adult Polyglucosan Body Disease *(GBE1)*	✓	✓	

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Conditions covered by our panels

Disease	Expanded Carrier Screen (283 genes)	Expanded Carrier Screen (152 genes)	Expanded Carrier Screen (39 genes)
Glycogen Storage Disease, Type V *(PYGM)*	✓	✓	
Glycogen Storage Disease, Type VII *(PFKM)*	✓	✓	
GRACILE Syndrome and Other *BCS1L*-Related Disorders *(BCS1L)*	✓	✓	
Hemochromatosis, Type 2A *(HFE2)*	✓		
Hemochromatosis, Type 3 *(TFR2)*	✓		
Hereditary Fructose Intolerance *(ALDOB)*	✓	✓	
Hereditary Spastic Paraparesis 49 *(TECPR2)*	✓	✓	
Hermansky-Pudlak Syndrome, Type 1 *(HPS1)*	✓	✓	
Hermansky-Pudlak Syndrome, Type 3 *(HPS3)*	✓	✓	
HMG-CoA Lyase Deficiency *(HMGCL)*	✓		
Holocarboxylase Synthetase Deficiency *(HLCS)*	✓	✓	
Homocystinuria *(CBS)*	✓	✓	
Homocystinuria due to MTHFR Deficiency *(MTHFR)*	✓	✓	
Homocystinuria, cblE Type *(MTRR)*	✓		
Hydrolethalus Syndrome *(HYLS1)*	✓	✓	
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome *(SLC25A15)*	✓		
Hypohidrotic Ectodermal Dysplasia 1 *(EDA)*	✓		
Hypophosphatasia *(ALPL)*	✓	✓	
Inclusion Body Myopathy 2 *(GNE)*	✓	✓	
Infantile Cerebral and Cerebellar Atrophy *(MED17)*	✓	✓	
Isovaleric Acidemia *(IVD)*	✓		✓
Joubert Syndrome 2 *(TMEM216)*	✓	✓	✓
Joubert Syndrome 7 / Meckel Syndrome 5 / COACH Syndrome *(RPGRIP1L)*	✓		
Junctional Epidermolysis Bullosa *(LAMA3)*	✓		
Junctional Epidermolysis Bullosa *(LAMB3)*	✓		
Junctional Epidermolysis Bullosa *(LAMC2)*	✓		
Krabbe Disease *(GALC)*	✓	✓	
Lamellar Ichthyosis, Type 1 *(TGM1)*	✓	✓	
Leber Congenital Amaurosis 10 and Other *CEP290*-Related Ciliopathies *(CEP290)*	✓	✓	

Comprehensive Jewish carrier screen (101 genes)	Ashkenazi Jewish carrier screen (64 genes)	Sephardi-Mizrahi Jewish carrier screen (54 genes)	East Asian carrier screen (95 genes)	High frequency pan-ethnic carrier screen (11 genes)	Standard pan-ethnic carrier screen (4 genes)
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Conditions covered by our panels

Disease	Expanded Carrier Screen (283 genes)	Expanded Carrier Screen (152 genes)	Expanded Carrier Screen (39 genes)
Leber Congenital Amaurosis 13 *(RDH12)*	✓		
Leber Congenital Amaurosis 2 / Retinitis Pigmentosa 20 *(RPE65)*	✓	✓	
Leber Congenital Amaurosis 5 *(LCA5)*	✓		
Leber Congenital Amaurosis 8 / Retinitis Pigmentosa 12 / Pigmented Paravenous Chorioretinal Atrophy *(CRB1)*	✓		
Leigh Syndrome, French-Canadian Type *(LRPPRC)*	✓	✓	
Lethal Congenital Contracture Syndrome 1 / Lethal Arthrogryposis with Anterior Horn Cell Disease *(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 2C *(SGCG)*	✓		
Limb-Girdle Muscular Dystrophy, Type 2D *(SGCA)*	✓		
Limb-Girdle Muscular Dystrophy, Type 2E *(SGCB)*	✓		
Limb-Girdle Muscular Dystrophy, Type 2I *(FKRP)*	✓		
Lipoamide Dehydrogenase Deficiency *(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 1a *(BCKDHA)*	✓	✓	✓
Maple Syrup Urine Disease, Type 1b *(BCKDHB)*	✓	✓	✓
Meckel-Gruber Syndrome 1 / Bardet-Biedl Syndrome 13 *(MKS1)*	✓	✓	
Medium Chain Acyl-CoA Dehydrogenase Deficiency *(ACADM)*	✓	✓	✓
Megalencephalic Leukoencephalopathy with Subcortical Cysts *(MLC1)*	✓	✓	
Menkes Disease *(ATP7A)*	✓		
Metachromatic Leukodystrophy *(ARSA)*	✓	✓	
3-Methylcrotonyl-CoA Carboxylase Deficiency *(MCCC1)*	✓		
3-Methylcrotonyl-CoA Carboxylase Deficiency *(MCCC2)*	✓		

	Comprehensive Jewish carrier screen (101 genes)	Ashkenazi Jewish carrier screen (64 genes)	Sephardi-Mizrahi Jewish carrier screen (54 genes)	East Asian carrier screen (95 genes)	High frequency pan-ethnic carrier screen (11 genes)	Standard pan-ethnic carrier screen (4 genes)
	✓		✓			
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				✓		

Conditions covered by our panels

Disease	Expanded Carrier Screen (283 genes)	Expanded Carrier Screen (152 genes)	Expanded Carrier Screen (39 genes)
3-Methylglutaconic Aciduria, Type III / Optic Atrophy 3, with Cataract *(OPA3)*	✓	✓	
Methylmalonic Acidemia *(MMAA)*	✓		
Methylmalonic Acidemia *(MMAB)*	✓		
Methylmalonic Acidemia *(MUT)*	✓		
Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type *(MMACHC)*	✓		✓
Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type *(MMADHC)*	✓		
Microphthalmia / Anophthalmia *(VSX2)*	✓	✓	
Mitochondrial Complex I Deficiency *(ACAD9)*	✓		
Mitochondrial Complex I Deficiency *(NDUFAF5)*	✓	✓	
Mitochondrial Complex I Deficiency *(NDUFS6)*	✓	✓	
Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy *(MPV17)*	✓		
Mitochondrial Myopathy and Sideroblastic Anemia 1 *(PUS1)*	✓	✓	
Mucolipidosis II / IIIA *(GNPTAB)*	✓	✓	
Mucolipidosis III Gamma *(GNPTG)*	✓		
Mucolipidosis IV *(MCOLN1)*	✓	✓	✓
Mucopolysaccharidosis Type I *(IDUA)*	✓		✓
Mucopolysaccharidosis Type II *(IDS)*	✓		
Mucopolysaccharidosis Type IIIA *(SGSH)*	✓		
Mucopolysaccharidosis Type IIIB *(NAGLU)*	✓		
Mucopolysaccharidosis Type IIIC *(HGSNAT)*	✓		
Mucopolysaccharidosis Type IIID *(GNS)*	✓		
Mucopolysaccharidosis Type IVb / GM1 Gangliosidosis *(GLB1)*	✓	✓	
Mucopolysaccharidosis Type VI *(ARSB)*	✓		
Mucopolysaccharidosis Type IX *(HYAL1)*	✓		
Multiple Sulfatase Deficiency *(SUMF1)*	✓	✓	
Muscle-Eye-Brain Disease and Other *POMGNT1*-Related Congenital Muscular Dystrophy-Dystroglycanopathies *(POMGNT1)*	✓	✓	

	Comprehensive Jewish carrier screen (101 genes)	Ashkenazi Jewish carrier screen (64 genes)	Sephardi-Mizrahi Jewish carrier screen (54 genes)	East Asian carrier screen (95 genes)	High frequency pan-ethnic carrier screen (11 genes)	Standard pan-ethnic carrier screen (4 genes)
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	✓	✓				

Conditions covered by our panels

Disease	Expanded Carrier Screen (283 genes)	Expanded Carrier Screen (152 genes)	Expanded Carrier Screen (39 genes)
Myoneurogastrointestinal Encephalopathy *(TYMP)*	✓	✓	
Myotubular Myopathy 1 *(MTM1)*	✓		
N-Acetylglutamate Synthase Deficiency *(NAGS)*	✓		
Nemaline Myopathy 2 *(NEB)*	✓	✓	✓
Nephrogenic Diabetes Insipidus, Type II *(AQP2)*	✓		
Nephrotic Syndrome / Congenital Finnish Nephrosis *(NPHS1)*	✓	✓	
Nephrotic Syndrome / Steroid-Resistant Nephrotic Syndrome *(NPHS2)*	✓		
Neuronal Ceroid-Lipofuscinosis *(CLN3)*	✓		✓
Neuronal Ceroid-Lipofuscinosis *(CLN5)*	✓	✓	
Neuronal Ceroid-Lipofuscinosis *(CLN6)*	✓		
Neuronal Ceroid-Lipofuscinosis *(CLN8)*	✓		
Neuronal Ceroid-Lipofuscinosis *(MFSD8)*	✓		
Neuronal Ceroid-Lipofuscinosis *(PPT1)*	✓	✓	
Neuronal Ceroid-Lipofuscinosis *(TPP1)*	✓	✓	
Niemann-Pick Disease, Type A/B *(SMPD1)*	✓	✓	✓
Niemann-Pick Disease, Type C *(NPC1)*	✓		
Niemann-Pick Disease, Type C *(NPC2)*	✓		
Nijmegen Breakage Syndrome *(NBN)*	✓		
Non-Syndromic Hearing Loss *(GJB2)*	✓	✓	
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome *(WNT10A)*	✓		
Omenn Syndrome *(RAG2)*	✓	✓	
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type *(DCLRE1C)*	✓		
Ornithine Aminotransferase Deficiency *(OAT)*	✓	✓	
Ornithine Transcarbomylase Deficiency *(OTC)*	✓		
Osteopetrosis 1 *(TCIRG1)*	✓	✓	
Pendred Syndrome *(SLC26A4)*	✓	✓	
Phenylalanine Hydroxylase Deficiency *(PAH)*	✓	✓	✓

Comprehensive Jewish carrier screen (101 genes)	Ashkenazi Jewish carrier screen (64 genes)	Sephardi-Mizrahi Jewish carrier screen (54 genes)	East Asian carrier screen (95 genes)	High frequency pan-ethnic carrier screen (11 genes)	Standard pan-ethnic carrier screen (4 genes)
✓		✓			
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✓	✓	✓	✓	✓	

Conditions covered by our panels

Disease	Expanded Carrier Screen (283 genes)	Expanded Carrier Screen (152 genes)	Expanded Carrier Screen (39 genes)
3-Phosphoglycerate Dehydrogenase Deficiency *(PHGDH)*	✓	✓	
Polycystic Kidney Disease, Autosomal Recessive *(PKHD1)*	✓	✓	✓
Polyglandular Autoimmune Syndrome, Type 1 *(AIRE)*	✓	✓	
Pontocerebellar Hypoplasia, Type 1A *(VRK1)*	✓	✓	
Pontocerebellar Hypoplasia, Type 6 *(RARS2)*	✓	✓	
Primary Carnitine Deficiency *(SLC22A5)*	✓	✓	
Primary Ciliary Dyskinesia *(DNAH5)*	✓	✓	
Primary Ciliary Dyskinesia *(DNAI1)*	✓	✓	
Primary Ciliary Dyskinesia *(DNAI2)*	✓	✓	
Primary Hyperoxaluria, Type 1 *(AGXT)*	✓		
Primary Hyperoxaluria, Type 2 *(GRHPR)*	✓		
Primary Hyperoxaluria, Type 3 *(HOGA1)*	✓	✓	
Progressive Cerebello-Cerebral Atrophy *(SEPSECS)*	✓	✓	
Progressive Familial Intrahepatic Cholestasis, Type 2 *(ABCB11)*	✓		
Propionic Acidemia *(PCCA)*	✓		
Propionic Acidemia *(PCCB)*	✓		
Pycnodysostosis *(CTSK)*	✓		
Pyruvate Dehydrogenase E1-Alpha Deficiency *(PDHA1)*	✓		
Pyruvate Dehydrogenase E1-Beta Deficiency *(PDHB)*	✓		
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency *(PTS)*	✓		
Renal Tubular Acidosis and Deafness *(ATP6V1B1)*	✓	✓	
Retinitis Pigmentosa 25 *(EYS)*	✓	✓	
Retinitis Pigmentosa 26 *(CERKL)*	✓	✓	
Retinitis Pigmentosa 28 *(FAM161A)*	✓	✓	
Retinitis Pigmentosa 59 *(DHDDS)*	✓	✓	
Rhizomelic Chondrodysplasia Punctata, Type 1 *(PEX7)*	✓		✓
Rhizomelic Chondrodysplasia Punctata, Type 3 *(AGPS)*	✓		
Roberts Syndrome *(ESCO2)*	✓		
Salla Disease *(SLC17A5)*	✓	✓	

Comprehensive Jewish carrier screen (101 genes)	Ashkenazi Jewish carrier screen (64 genes)	Sephardi-Mizrahi Jewish carrier screen (54 genes)	East Asian carrier screen (95 genes)	High frequency pan-ethnic carrier screen (11 genes)	Standard pan-ethnic carrier screen (4 genes)
✓	✓				
✓	✓				
✓		✓			
✓	✓				
✓		✓			
			✓		
✓	✓				
✓	✓				
✓	✓				
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✓	✓		✓		
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			✓		
			✓		
			✓		
✓		✓			
✓		✓			
✓		✓			
✓	✓	✓			
✓	✓				

Conditions covered by our panels

Disease	Expanded Carrier Screen (283 genes)	Expanded Carrier Screen (152 genes)	Expanded Carrier Screen (39 genes)
Sandhoff Disease *(HEXB)*	✓	✓	
Schimke Immunoosseous Dysplasia *(SMARCAL1)*	✓		
Segawa Syndrome *(TH)*	✓		
Sjogren-Larsson Syndrome *(ALDH3A2)*	✓		
Smith-Lemli-Opitz Syndrome *(DHCR7)*	✓	✓	✓
Spinal Muscular Atrophy (Includes Enhanced SMA Testing) *(SMN1)*	✓	✓	✓
Spondylothoracic Dysostosis *(MESP2)*	✓		
Steel Syndrome *(COL27A1)*	✓		
Stuve-Wiedemann Syndrome *(LIFR)*	✓		
Sulfate Transporter-Related Osteochondrodysplasia *(SLC26A2)*	✓	✓	
Tay-Sachs Disease *(HEXA)**	✓	✓	✓
Tyrosinemia, Type I *(FAH)*	✓	✓	✓
Usher Syndrome, Type IB *(MYO7A)*	✓	✓	
Usher Syndrome, Type IC *(USH1C)*	✓	✓	
Usher Syndrome, Type ID *(CDH23)*	✓	✓	
Usher Syndrome, Type IF *(PCDH15)*	✓	✓	✓
Usher Syndrome, Type IIA *(USH2A)*	✓	✓	
Usher Syndrome, Type III *(CLRN1)*	✓	✓	✓
Very Long Chain Acyl-CoA Dehydrogenase Deficiency *(ACADVL)*	✓	✓	
Walker-Warburg Syndrome and Other *FKTN*-Related Dystrophies *(FKTN)*	✓	✓	✓
Wilson Disease *(ATP7B)*	✓	✓	
Wolman Disease / Cholesteryl Ester Storage Disease *(LIPA)*	✓	✓	
X-Linked Juvenile Retinoschisis *(RS1)*	✓		
X-Linked Severe Combined Immunodeficiency *(IL2RG)*	✓		
Zellweger Syndrome Spectrum *(PEX10)*	✓		
Zellweger Syndrome Spectrum *(PEX1)*	✓		✓
Zellweger Syndrome Spectrum *(PEX2)*	✓	✓	
Zellweger Syndrome Spectrum *(PEX6)*	✓	✓	

	Comprehensive Jewish carrier screen (101 genes)	Ashkenazi Jewish carrier screen (64 genes)	Sephardi-Mizrahi Jewish carrier screen (54 genes)	East Asian carrier screen (95 genes)	High frequency pan-ethnic carrier screen (11 genes)	Standard pan-ethnic carrier screen (4 genes)
				✓		
	✓	✓	✓	✓	✓	✓
	✓	✓	✓	✓	✓	✓
	✓	✓	✓	✓		
	✓	✓				
	✓	✓				
	✓		✓	✓		
	✓	✓				
				✓		
	✓	✓				
	✓	✓	✓	✓		
	✓		✓			
				✓		
				✓		
				✓		
	✓	✓				
	✓		✓			

Questions?

For additional information about any of Sema4's carrier screening panels, please visit **sema4.com/testcatalog**.
If you need further assistance, you can also reach customer support at **800-298-6470** or at **sema4.com/contact-us**.

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