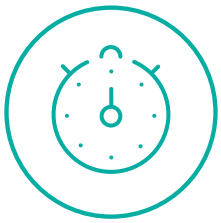
Invitae Carrier Screening

The insight your patients need to prepare for tomorrow

Invitae offers carrier screening with flexibility and customization. Select a pre-curated test, combine multiple tests, or customize your own test for each patient.

In network for 250+ million

$250 patient-pay price
$100 partner-pay price



10–21 day average turnaround time

Access to board-certified genetic counselors

Invitae's carrier screen includes:

- Severe and prevalent disorders seen across all ethnicities
- Enhanced SMA testing to help identify silent carriers
- Comprehensive Fragile X analysis, including AGG interruptions
- Full gene sequencing with deletion and duplication analysis leading to a 99% detection rate for most genes
- Actionable results; no reporting of variants of unknown significance

	Invitae CORE Carrier Screen	Invitae Broad Carrier Screen	Invitae Comprehensive Carrier Screen
Number of genes	3	46	288
Includes all ACOG & ACMG recommended disorders		●	●
Number of X-linked disorders	1*	5*	21*
Sample type	Blood or saliva	Blood or saliva	Blood or saliva

ACOG: American College of Obstetricians and Gynecologists
ACMG: American College of Medical Genetics and Genomics

*All panels are available without X-linked disorders.

About Invitae

Invitae's mission is to bring comprehensive genetic information into mainstream medical practice to improve the quality of healthcare for everyone. Specializing in genetic diagnostics in clinical areas across all stages of life, Invitae is aggregating most of the world's genetic tests into a single service with higher quality, faster turnaround time, and lower prices. For more information, please visit www.invitae.com.

Invitae Carrier Screening *(continued)*

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COMPREHENSIVE CARRIER SCREEN

CORE

Disorder	Gene
Cystic fibrosis and other CFTR-related disorders	CFTR
Fragile X syndrome*	FMR1
Spinal muscular atrophy	SMN1

BROAD

Disorder	Gene
ABCC8-related disorders	ABCC8
Alpha-thalassemia	HBA1/HBA2
Bloom syndrome	BLM
Canavan disease	ASPA
Citrullinemia type 1	ASS1
Congenital disorder of glycosylation (PMM2-related)	PMM2
Dihydrolipoamide dehydrogenase deficiency (DLD)	DLD
DMD-related dystrophinopathy* (including Duchenne/Becker muscular dystrophy and dilated cardiomyopathy)	DMD
Familial dysautonomia	ELP1
Fanconi anemia type C	FANCC
FKTN-related disorders (including Walker-Warburg syndrome)	FKTN
Galactosemia (GALT-related)	GALT
Gaucher disease	GBA
GJB2-related DFNB1 nonsyndromic hearing loss and deafness	GJB2
Glycogen storage disease type Ia	G6PC
Glycogen storage disease type II (Pompe disease)	GAA
HBB-related hemoglobinopathies (including beta-thalassemia and sickle cell disease)	HBB
Krabbe disease	GALC
Maple syrup urine disease (MSUD) type 1A	BCKDHA
Maple syrup urine disease (MSUD) type 1B	BCKDHB

Disorder	Gene
Medium chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM
Mucolipidosis type IV	MCOLN1
Mucopolysaccharidosis type I (including Hurler, Hurler-Scheie, and Scheie syndromes)	IDUA
Nemaline myopathy 2	NEB
Neuronal ceroid-lipofuscinosis (CLN3-related)	CLN3
Niemann-Pick disease type A/B	SMPD1
Ornithine transcarbamylase (OTC) deficiency*	OTC
Pendred syndrome	SLC26A4
Phenylalanine hydroxylase deficiency (including phenylketonuria (PKU))	PAH
Polycystic kidney disease (PKHD1-related)	PKHD1
Rhizomelic chondrodysplasia punctata type 1/Refsum disease (PEX7-related)	PEX7
Smith-Lemli-Opitz syndrome	DHCR7
Tay-Sachs disease/hexosaminidase A deficiency	HEXA
TMEM216-related disorders (including Joubert syndrome 2 and Meckel syndrome 2)	TMEM216
Tyrosinemia type I	FAH
Usher syndrome type IF/PCDH15-related disorders	PCDH15
Usher syndrome type IIA/USH2A-related disorders	USH2A
Usher syndrome type IIIA	CLRN1
X-linked adrenoleukodystrophy*	ABCD1
X-linked severe combined immunodeficiency (X-SCID)*	IL2RG
Zellweger spectrum disorder (PEX1-related)	PEX1
Zellweger spectrum disorder (PEX6-related)	PEX6

Disorder	Gene
3-beta-hydroxysteroid dehydrogenase type II deficiency (congenital adrenal hyperplasia)	HSD3B2
3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency	HMGCL

Disorder	Gene
3-methylglutaconic aciduria type III (Costeff optic atrophy)	OPA3
11-beta-hydroxylase-deficient congenital adrenal hyperplasia	CYP11B1

**Indicates disorder with X-linked inheritance.*

Invitae Carrier Screening *(continued)*

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Disorder	Gene
17-alpha-hydroxylase-deficient congenital adrenal hyperplasia	CYP17A1
Abetalipoproteinemia	MTTP
ACAD9 deficiency	ACAD9
Achromatopsia (CNGB3-related)	CNGB3
Acrodermatitis enteropathica	SLC39A4
Adenosine deaminase deficiency	ADA
Aicardi-Goutieres syndrome (SAMHD1-related)	SAMHD1
Aldosterone synthase deficiency	CYP11B2
Alpha-mannosidosis	MAN2B1
Alpha-thalassemia X-linked intellectual disability syndrome*	ATRX
Alport syndrome (COL4A3-related)	COL4A3
Alport syndrome (COL4A4-related)	COL4A4
Alport syndrome, X-linked (COL4A5-related)*	COL4A5
Alström syndrome	ALMS1
Andermann syndrome	SLC12A6
Arginase deficiency	ARG1
Argininosuccinic aciduria	ASL
Aromatase deficiency	CYP19A1
Asparagine synthetase deficiency	ASNS
Aspartylglucosaminuria	AGA
Ataxia with vitamin E deficiency	TTPA
Ataxia-telangiectasia	ATM
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia	AIRE
Autosomal recessive deafness 77 (DFNB77)	LOXHD1
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	SACS
Bardet-Biedl syndrome (BBS1-related)	BBS1
Bardet-Biedl syndrome (BBS2-related)	BBS2
Bardet-Biedl syndrome (BBS10-related)	BBS10
Bardet-Biedl syndrome (BBS12-related)	BBS12
Bartter syndrome type 4A	BSND

Disorder	Gene
Beta-ketothiolase deficiency	ACAT1
Carbamoylphosphate synthetase I deficiency	CPS1
Carnitine palmitoyltransferase I deficiency	CPT1A
Carnitine palmitoyltransferase II deficiency	CPT2
Carpenter syndrome (RAB23-related)	RAB23
Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders	RMRP
Cerebrotendinous xanthomatosis	CYP27A1
Charcot-Marie-Tooth disease (NDRG1-related)	NDRG1
Charcot-Marie-Tooth disease, X-linked (GJB1-related)*	GJB1
Chorea-acanthocytosis	VPS13A
Choroideremia*	CHM
Chronic granulomatous disease (CYBA-related)	CYBA
Chronic granulomatous disease (CYBB-related)*	CYBB
Citrin deficiency	SLC25A13
Cockayne syndrome type A	ERCC8
Cockayne syndrome type B	ERCC6
Cohen syndrome	VPS13B
Combined malonic and methylmalonic aciduria (ACSF3-related)	ACSF3
Combined oxidative phosphorylation deficiency (GFM1-related)	GFM1
Combined oxidative phosphorylation deficiency (TSFM-related)	TSFM
Combined pituitary hormone deficiency (LHX3-related)	LHX3
Combined pituitary hormone deficiency (PROP1-related)	PROP1
Congenital amegakaryocytic thrombocytopenia	MPL
Congenital disorder of glycosylation (ALG6-related)	ALG6
Congenital disorder of glycosylation (MPI-related)	MPI
Congenital ichthyosis (TGM1-related)	TGM1
Congenital insensitivity to pain with anhidrosis	NTRK1
Congenital myasthenic syndrome (CHRNE-related)	CHRNE
Congenital myasthenic syndrome (RAPSN-related)	RAPSN

**Indicates disorder with X-linked inheritance.*

Invitae Carrier Screening *(continued)*

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COMPREHENSIVE CARRIER SCREEN *(CONTINUED)*

Disorder	Gene
Congenital neutropenia (HAX1-related)	HAX1
Corneal dystrophy and perceptive deafness	SLC4A11
Cystinosis	CTNS
D-bifunctional protein deficiency	HSD17B4
DHDDS-related disorders	DHDDS
Dysferlinopathy (including limb-girdle muscular dystrophy type 2B)	DYSF
Dystrophic epidermolysis bullosa (COL7A1-related)	COL7A1
Ehlers-Danlos syndrome, dermatosparaxis type	ADAMTS2
Ellis-van Creveld syndrome (EVC-related)	EVC
Ellis-van Creveld syndrome (EVC2-related)	EVC2
Emery-Dreifuss muscular dystrophy (EMD-related)*	EMD
Enhanced S-cone syndrome/retinitis pigmentosa	NR2E3
Ethylmalonic encephalopathy	ETHE1
Fabry disease*	GLA
Factor IX deficiency (hemophilia B)*	F9
Familial hypercholesterolemia (LDLR-related)	LDLR
Familial hypercholesterolemia (LDLRAP1-related)	LDLRAP1
Fanconi anemia type A	FANCA
Fanconi anemia type G	FANCG
FKRP-related disorders (including Walker-Warburg syndrome)	FKRP
Fumarate hydratase deficiency	FH
Galactokinase deficiency galactosemia	GALK1
Gitelman syndrome (SLC12A3-related)	SLC12A3
Glutaric acidemia type I	GCDH
Glutaric acidemia type II (ETFA-related)	ETFA
Glutaric acidemia type II (ETFDH-related)	ETFDH
Glycine encephalopathy (AMT-related)	AMT
Glycine encephalopathy (GLDC-related)	GLDC
Glycogen storage disease type Ib	SLC37A4
Glycogen storage disease type III	AGL
Glycogen storage disease type IV/adult polyglucosan body disease	GBE1

Disorder	Gene
Glycogen storage disease type V	PYGM
Glycogen storage disease type VII	PFKM
GRACILE syndrome/BCS1L-related disorders (including mitochondrial complex III deficiency, Bjornstad syndrome, and Leigh syndrome)	BCS1L
Guanidinoacetate methyltransferase deficiency	GAMT
Hereditary fructose intolerance	ALDOB
Hereditary hemochromatosis (HJV-related)	HJV
Hereditary hemochromatosis (TFR2-related)	TFR2
Hermansky-Pudlak syndrome (HPS1-related)	HPS1
Hermansky-Pudlak syndrome (HPS3-related)	HPS3
Holocarboxylase synthetase deficiency	HLCS
Homocystinuria (CBS-related)	CBS
Homocystinuria due to MTHFR deficiency	MTHFR
Homocystinuria, cobalamin E type	MTRR
Hydrolethalus syndrome type 1	HYLS1
Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	SLC25A15
Hypohidrotic ectodermal dysplasia (EDA-related)*	EDA
Hypophosphatasia	ALPL
Inclusion body myopathy 2	GNE
Isovaleric acidemia	IVD
Junctional epidermolysis bullosa (LAMA3-related)	LAMA3
Junctional epidermolysis bullosa (LAMB3-related)	LAMB3
Junctional epidermolysis bullosa (LAMC2-related)	LAMC2
KCNJ11-related disorders	KCNJ11
LAMA2-related muscular dystrophy	LAMA2
Leber congenital amaurosis 2	RPE65
Leber congenital amaurosis 5	LCA5
Leber congenital amaurosis 8/CRB1-related disorders	CRB1
Leber congenital amaurosis 10/CEP290-related disorders	CEP290
Leber congenital amaurosis 13	RDH12

**Indicates disorder with X-linked inheritance.*

Invitae Carrier Screening *(continued)*

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COMPREHENSIVE CARRIER SCREEN *(CONTINUED)*

Disorder	Gene
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-related)	EIF2B5
Limb-girdle muscular dystrophy type 2A (calpainopathy)	CAPN3
Limb-girdle muscular dystrophy type 2C	SGCG
Limb-girdle muscular dystrophy type 2D	SGCA
Limb-girdle muscular dystrophy type 2E	SGCB
Lipoid congenital adrenal hyperplasia (STAR-related)	STAR
Lipoprotein lipase deficiency	LPL
Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	HADHA
Lysinuric protein intolerance	SLC7A7
Lysosomal acid lipase deficiency (includes Wolman disease and cholesterol ester storage disease)	LIPA
Major histocompatibility complex class II deficiency (CIITA-related)	CIITA
Maple syrup urine disease (MSUD) type 2	DBT
Megalencephalic leukoencephalopathy with subcortical cysts type 1	MLC1
Menkes disease/ATP7A-related disorders* (including occipital horn syndrome and distal hereditary motor neuropathy)	ATP7A
Metachromatic leukodystrophy (ARSA-related)	ARSA
Methylmalonic acidemia (MMAA-related)	MMAA
Methylmalonic acidemia (MMAB-related)	MMAB
Methylmalonic acidemia (MUT-related)	MUT (also known as MMUT)
Methylmalonic acidemia with homocystinuria, cobalamin C type	MMACHC
Methylmalonic acidemia with homocystinuria, cobalamin D type	MMADHC
Microphthalmia/clinical anophthalmia (VSX2-related)	VSX2
Mitochondrial complex I deficiency/Leigh syndrome (NDUFAF5-related)	NDUFAF5
Mitochondrial complex I deficiency/Leigh syndrome (NDUFS6-related)	NDUFS6
Mitochondrial DNA depletion syndrome (MPV17-related)	MPV17
Mitochondrial myopathy and sideroblastic anemia 1	PUS1
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	TYMP
MKS1-related disorders	MKS1
Mucolipidosis type II/III (GNPTAB-related)	GNPTAB
Mucolipidosis type III (GNPTG-related)	GNPTG
Mucopolysaccharidosis type II (Hunter syndrome)*	IDS
Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)	SGSH
Mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)	NAGLU
Mucopolysaccharidosis type IIIC (Sanfilippo C syndrome)/retinitis pigmentosa	HGSNAT
Mucopolysaccharidosis type IIID (Sanfilippo D syndrome)	GNS
Mucopolysaccharidosis type IVB (Morquio B syndrome)/GM1 gangliosidosis	GLB1
Mucopolysaccharidosis type IX	HYAL1
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	ARSB
Multiple sulfatase deficiency	SUMF1
N-acetylglutamate synthase deficiency	NAGS
Nephrogenic diabetes insipidus (AQP2-related)	AQP2
Nephrotic syndrome/congenital Finnish nephrosis (NPHS1-related)	NPHS1
Nephrotic syndrome/steroid-resistant nephrotic syndrome (NPHS2-related)	NPHS2
Neuronal ceroid-lipofuscinosis (MFSD8-related)	MFSD8
Neuronal ceroid-lipofuscinosis (PPT1-related)	PPT1
Neuronal ceroid-lipofuscinosis (TPP1-related)	TPP1
Neuronal ceroid-lipofuscinosis (CLN5-related)	CLN5
Neuronal ceroid-lipofuscinosis (CLN6-related)	CLN6
Neuronal ceroid-lipofuscinosis/Northern epilepsy (CLN8-related)	CLN8
Niemann-Pick disease type C (NPC1-related)	NPC1
Niemann-Pick disease type C (NPC2-related)	NPC2

**Indicates disorder with X-linked inheritance.*

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COMPREHENSIVE CARRIER SCREEN *(CONTINUED)*

Disorder	Gene
Nijmegen breakage syndrome	NBN
Ornithine aminotransferase deficiency	OAT
Osteopetrosis (TCIRG1-related)	TCIRG1
Peroxisomal acyl-CoA oxidase deficiency	ACOX1
Phosphoglycerate dehydrogenase deficiency/ Neu-Laxova syndrome	PHGDH
Polymicrogyria (ADGRG1-related)	ADGRG1
POMGNT1-related disorders (including muscle-eye-brain disease and Walker-Warburg syndrome)	POMGNT1
Pontocerebellar hypoplasia (RARS2-related)	RARS2
Pontocerebellar hypoplasia (SEPSECS-related)	SEPSECS
Pontocerebellar hypoplasia (VRK1-related)	VRK1
Postnatal progressive microcephaly with seizures and brain atrophy/infantile cerebral and cerebellar atrophy (MED17-related)	MED17
Primary carnitine deficiency	SLC22A5
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
PRPS1-related disorders	PRPS1
PSAP-related disorders	PSAP
Pycnodysostosis	CTSK
Pyruvate carboxylase deficiency	PC
Pyruvate dehydrogenase deficiency (PDHA1-related)*	PDHA1
Pyruvate dehydrogenase deficiency (PDHB-related)	PDHB
Renal tubular acidosis with deafness (ATP6V1B1-related)	ATP6V1B1
Retinitis pigmentosa 25	EYS
Retinitis pigmentosa 26	CERKL
Retinitis pigmentosa 28	FAM161A
Rhizomelic chondrodysplasia punctata type 3	AGPS
Roberts syndrome	ESCO2
RPGRIP1L-related disorders (including Joubert syndrome 7, COACH syndrome, and Meckel syndrome 5)	RPGRIP1L
RTEL-1-related disorders (including dyskeratosis congenita)	RTEL1
Sandhoff disease	HEXB
Schimke immuno-osseous dysplasia	SMARCAL1
Severe combined immune deficiency (DCLRE1C-related)	DCLRE1C
Severe combined immunodeficiency/Omenn syndrome (RAG2-related)	RAG2
Severe congenital neutropenia (VPS45-related)	VPS45
Sialic acid storage disorders	SLC17A5
Sjögren-Larsson syndrome	ALDH3A2
SLC26A2-related disorders (including diastrophic dysplasia, atelosteogenesis type 2, and achondrogenesis type 1B/multiple metaphyseal dysplasia)	SLC26A2
SLC35A3-related disorder	SLC35A3
Spastic paraplegia type 15	ZFYVE26
Spastic paraplegia type 49	TECPR2
Spondylothoracic dysostosis	MESP2
Steel syndrome	COL27A1
Stüve-Wiedemann syndrome	LIFR
Tetrahydrobiopterin deficiency (PTS-related)	PTS
Transient infantile liver failure (TRMU-related)	TRMU
Tyrosine hydroxylase deficiency	TH
Tyrosinemia type II	TAT
Usher syndrome type IB/MYO7A-related disorders	MYO7A
Usher syndrome type IC/USH1C-related disorders	USH1C
Usher syndrome type ID	CDH23
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL

**Indicates disorder with X-linked inheritance.*

Invitae Carrier Screening *(continued)*

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COMPREHENSIVE CARRIER SCREEN

Disorder	Gene
Wilson disease	ATP7B
WNT10A-related disorders (including odonto-onycho-dermal dysplasia and Schopf-Schulz-Passarge syndrome)	WNT10A
X-linked creatine transporter deficiency*	SLC6A8
X-linked juvenile retinoschisis*	RS1
X-linked myotubular myopathy*	MTM1
Xeroderma pigmentosum complementation group A	XPA
Xeroderma pigmentosum complementation group C	XPC
Zellweger spectrum disorder (PEX2-related)	PEX2
Zellweger spectrum disorder (PEX10-related)	PEX10
Zellweger spectrum disorder (PEX12-related)	PEX12

ADD-ON GENES *(CAN BE ADDED TO ANY PANEL)*

Disorder	Gene
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC1-related)	MCCC1
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC2-related)	MCCC2
Alkaptonuria	HGD
Alpha-1 antitrypsin deficiency	SERPINA1
Bernard-Soulier syndrome (GP1BA-related)	GP1BA
Bernard-Soulier syndrome (GP9-related)	GP9
Biotinidase deficiency	BTD
Factor V Leiden thrombophilia	F5
Factor XI deficiency (hemophilia C)	F11
Familial Mediterranean fever	MEFV
Glucose-6-phosphate dehydrogenase (G6PD) deficiency*	G6PD
Hereditary hemochromatosis (HFE-related)	HFE
Prothrombin-related thrombophilia	F2

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