INVITAE

ORDER ID
For Invitae internal use only

INVITAE PEDIATRIC GENETICS, METABOLIC, NEPHROLOGY, & ENDOCRINOLOGY REQUISITION FORM

PATIENT INFORMATION

First name | MI | Last name | Date of birth (MM/DD/YYYY) | If patient is deceased Date of death:

Sex assigned at birth ◯ Female ◯ Male

Gender (if differs from sex assigned at birth) ◯ Man ◯ Non-binary ◯ Woman ◯ Self-described: _____

Race/Ethnicity (select all that apply): ◯ Ashkenazi Jewish ◯ Asian ◯ Black ◯ French Canadian ◯ Hispanic ◯ Native American ◯ Pacific Islander ◯ Sephardic Jewish ◯ White ◯ Other: _____

Patient ID (MRN) | Email address (billing and report access after clinician releases) | Mobile Phone (patient consents to receive texts from Invitae)

Address | City | State/Prov | Zip/Postal code | Country

Ship a saliva kit to this patient (optional) **Ship to:** ◯ Address above ◯ Alternate address: _____

INSURANCE INFORMATION (Provide only if applicable. Attach front and back of insurance card, clinical notes and medical records. Insurance is not accepted for patients outside the US.)

Policyholder name | Primary insurance company name | Primary member ID # | Primary insurance phone | Prior-authorization #

Patient relationship to policyholder ◯ Self ◯ Spouse ◯ Child ◯ Other: _____ | Secondary insurance company name | Secondary member ID # | Secondary insurance phone | Prior-authorization #

Medicare insurance billing only (select one): ◯ Patient was treated as a hospital inpatient (more than a 24 hour stay) in the last 14 days ◯ Not a hospital patient

PROVIDER INFORMATION

Organization name | Phone | Fax

Address | City | State/Prov | Zip/Postal code | Country

Primary clinical contact name (if different from ordering provider) | NPI | Email address (for report access)

Ordering provider (Pre-populate your provider list below. For each order, indicate **one** ordering provider by marking the checkbox before the name)

Name	NPI	Email address (for report access)	Name	NPI	Email address (for report access)
◯			◯		
◯			◯		

Additional clinical or laboratory contacts (optional) ◯ Share this order with the primary clinical contact's default clinical team (manage team online at www.invitae.com/signin)

Name | Email address (for report access) | Name | Email address (for report access)

BILLING SELECTION

Billing selection (select one): ◯ Patient pay (patient email required) ◯ Institutional ◯ Insurance, **ICD-10 code(s) required:** _____

PARTNERSHIP PROGRAMS
Invitae partner code (if applicable): _____

SPECIMEN INFORMATION

Specimen type: Blood -OR- Saliva -OR- DNA - source: _____

We are unable to accept blood or saliva from patients with allogeneic bone marrow transplants or a blood transfusion <2 weeks prior to specimen collection. DNA must be extracted in a CLIA or other suitably certified laboratory.

Specimen collection date (MM/DD/YYYY): ___ ___ _____ *If not provided, the day before specimen receipt will be used. For DNA, provide date retrieved from archive.*

Specimen ID (IB on tube): _____

TEST SELECTION

OPTION 1: SELECT AN INVITAE PANEL FROM OUR TEST CATALOG

Select your desired tests(s) from the attached test catalog and discard any pages without a selection. -OR- Indicate test IDs below. If test IDs contain add-on codes, the original panel will also be included.

Test code	Add-on code (optional)	Test code	Add-on code (optional)
.		.	

AUTOMATIC REFLEX: Invitae offers one re-requisition at no additional charge for tests within the same clinical area. Preschedule below or online. Learn more at www.invitae.com/re-requisition.

Conditions for reflex:
◯ Regardless of initial results
◯ Only if negative (no pathogenic/likely pathogenic results)

Test code	Add-on code (optional)
.	

OPTION 2: FAMILY FOLLOW-UP TESTING

Invitae family follow-up testing is available at no additional charge for blood relatives of patients who receive pathogenic or likely pathogenic results (or approved VUS). Learn more at www.invitae.com/family.

Invitae proband RQ# _____

Relationship to proband _____

Gene(s) _____

Variant(s) _____

NOTE: Invitae's family follow-up testing analyzes the variant(s) indicated above.

◯ This report should include any variants of uncertain significance and be eligible for re-requisition. Billing information **must** be included above.

By signing this form, I acknowledge that the patient (or the individual authorized to make decisions for the patient) has been supplied information regarding and consented to undergo genetic testing, as set forth in Invitae's Informed Consent for Genetic Testing (www.invitae.com/forms). I acknowledge that the patient has agreed that (1) for orders originating outside the US, the patient's personal information and specimen will be transferred to and processed in the US (2) Invitae may notify the patient of clinical updates related to genetic test results (in consultation with the ordering clinician) (3) Invitae and its designees may release information concerning testing to the patient's insurer (if billing to insurance) (4) if the patient's insurer does not reimburse Invitae in full for any reason then Invitae may bill the patient for the services and the patient will remit payment to Invitae and (5) for amounts the patient receives from the insurer, patient will remit payment to Invitae for services rendered. I attest that I am authorized under applicable law to order this test. If required by patient's insurer, I attest that I offered pre-test genetic counseling to the patient or authorize Invitae to assist the patient in obtaining pre-test genetic counseling from a third party. I agree to the transfer of information from this TRF to a letter of medical necessity and/or other documentation using my name as the signature. For US ordering clinicians only: I consent and direct Invitae to share my contact information with third parties who may contact me directly in connection with patient results (opt out via online portal). If I am a delegate, I confirm I have authorization to (1) agree to all of the above and (2) sign this form and any supporting documents for Invitae on behalf of the ordering clinician.

Medical professional or delegate signature (required) | **Date (MM/DD/YYYY)**

CONTINUE TO NEXT PAGE TO COMPLETE REASONS FOR TESTING

INVITAE

Patient's first name _____ Patient's last name _____

INVITAE PEDIATRIC GENETICS, METABOLIC, NEPHROLOGY & ENDOCRINOLOGY

REASONS FOR TESTING

PERSONAL HISTORY

Is/was this patient affected or symptomatic†? ◯ Yes ◯ No If yes, describe briefly. Complete checklist below and attach clinical notes.

Age at diagnosis: _____

†Symptomatic means the patient has features or signs known or suspected to be related to the genetic testing being ordered and could include findings on physical examination, laboratory tests, or imaging.

Is there a hematological malignancy in this patient (current or history of)? ◯ Yes ◯ No

Has this patient had genetic testing before? ◯ Yes ◯ No If yes, write test results and attach the report.

FAMILY HISTORY

Is there a family history of disease for which the patient is being tested? ◯ Yes ◯ No
If yes, describe below and attach pedigree and/or clinical notes.

Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis

REQUESTED VARIANTS (attach proband's report)

To have the presence or absence of specific variants commented on in this patient's report, provide details below (optional). For Invitae family follow-up testing see the **NOTE** below that section.

This patient's relationship to proband: ◯ Sibling ◯ Child ◯ Other: _____

Gene(s)/Variant(s): _____

_____

CLINICAL INFORMATION CHECKLIST

Complete this **required** clinical information checklist and/or attach a clinic note if patient is/was affected or symptomatic.
IMPORTANT: Clinical information is essential for insurance billing and variant interpretation.

Clinical diagnosis

_____

_____

Cardiac

◯ Yes ◯ No ◯ Unknown

- ☐ Aortic root dilation
- ☐ Arrhythmia
- ☐ Atrial septal defect
- ☐ Cardiomyopathy – type: _____
- ☐ Coarctation of aorta
- ☐ Heart murmur
- ☐ Heterotaxy
- ☐ Hypertension
- ☐ Patent ductus arteriosus
- ☐ Tetralogy of Fallot
- ☐ Ventricular septal defect
- ☐ Other: _____

Craniofacial

◯ Yes ◯ No ◯ Unknown

- ☐ Abnormal facial shape
- ☐ Cleft lip
- ☐ Cleft palate
- ☐ Craniosynostosis
- ☐ Downslanted palpebral fissures
- ☐ Epicanthus
- ☐ External ear malformation
- ☐ Facial asymmetry
- ☐ Frontal bossing
- ☐ High palate
- ☐ Hypertelorism
- ☐ Low set ears
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Micrognathia
- ☐ Retrognathia
- ☐ Short neck
- ☐ Synophrys
- ☐ Wide nasal bridge
- ☐ Other: _____

Developmental/Behavioral

◯ Yes ◯ No ◯ Unknown

- ☐ Absent speech
- ☐ Attention deficit hyperactivity disorder
- ☐ Autistic behavior
- ☐ Behavioral abnormality
- ☐ Delayed fine motor development
- ☐ Delayed gross motor development
- ☐ Delayed speech & language development
- ☐ Developmental regression
- ☐ Global developmental delay
- ☐ Hyperactivity
- ☐ Intellectual disability
- ☐ Obsessive compulsive disorder
- ☐ Specific learning disability
- ☐ Stereotypy
- ☐ Other: _____

Endocrine

◯ Yes ◯ No ◯ Unknown

- ☐ Abnormality of adrenal glands
- ☐ Amenorrhea
- ☐ BMI: _____
- ☐ Delayed puberty
- ☐ Diabetes insipidus/mellitus
- ☐ Elevated hemoglobin A1c
- ☐ Goiter
- ☐ Hypercalcemia
- ☐ Hyperthyroidism
- ☐ Hypophosphatemia
- ☐ Hypothyroidism
- ☐ Low alkaline phosphatase
- ☐ MODY – age of onset: _____
- ☐ Pancreatic islet autoantibody negativity
- ☐ Rickets
- ☐ Other: _____

Eye/Vision Abnormalities

◯ Yes ◯ No ◯ Unknown

- ☐ Aniridia
- ☐ Anophthalmia
- ☐ Blue sclerae
- ☐ Cataracts
- ☐ Central vision loss
- ☐ Coloboma
- ☐ Cortical visual impairment
- ☐ Ectopia lentis
- ☐ External ophthalmoplegia
- ☐ Macular dystrophy
- ☐ Microphthalmia
- ☐ Myopia
- ☐ Night blindness
- ☐ Nystagmus
- ☐ Peripheral vision loss
- ☐ Photophobia
- ☐ Ptosis
- ☐ Retinal dystrophy
- ☐ Retinitis Pigmentosa
- ☐ Strabismus
- ☐ Other: _____

Gastrointestinal

◯ Yes ◯ No ◯ Unknown

- ☐ Cholestasis
- ☐ Congenital diaphragmatic hernia
- ☐ Constipation
- ☐ Diarrhea
- ☐ Duodenal stenosis/atresia
- ☐ Exocrine pancreatic insufficiency
- ☐ Failure to thrive
- ☐ Feeding difficulties
- ☐ Gastroesophageal reflux
- ☐ Gastroschisis
- ☐ Hepatomegaly/Splenomegaly
- ☐ Hepatic fibrosis
- ☐ Hirschsprung's disease
- ☐ Inflammatory bowel disease
- ☐ Intestinal perforation
- ☐ Intrahepatic biliary atresia
- ☐ Laryngomalacia
- ☐ Nausea/vomiting
- ☐ Pancreatitis
- ☐ Pyloric stenosis
- ☐ Tracheoesophageal fistula
- ☐ Other: _____

Genitourinary

◯ Yes ◯ No ◯ Unknown

- ☐ Abnormal renal biopsy: _____
- ☐ Abnormal urine analysis: _____
- ☐ Ambiguous genitalia
- ☐ Chronic kidney disease
- ☐ Congenital malformation of reproductive organs
- ☐ Cryptorchidism
- ☐ Hydronephrosis
- ☐ Hypospadias
- ☐ Micropenis
- ☐ Nephrocalcinosis
- ☐ Nephrotic syndrome
- ☐ Nephrolithiasis
- ☐ Polycystic kidney disease
- ☐ Renal agenesis
- ☐ Renal dysplasia
- ☐ Renal insufficiency
- ☐ Renal tubular dysfunction/acidosis
- ☐ Other: _____

(continues)

CONTINUE TO NEXT PAGE TO COMPLETE REASONS FOR TESTING

INVITAE

Patient's first name

Patient's last name

INVITAE PEDIATRIC GENETICS, METABOLIC, NEPHROLOGY & ENDOCRINOLOGY

CLINICAL INFORMATION CHECKLIST

Hearing Impairment

◯ Yes ◯ No ◯ Unknown

- ☐ Conductive hearing impairment: ◯ bilateral ◯ unilateral
- ☐ Sensorineural hearing impairment: ◯ bilateral ◯ unilateral
- ☐ Hearing impairment, mixed or unknown: ◯ bilateral ◯ unilateral
- ☐ Other:_____

Hematological or Immunological

◯ Yes ◯ No ◯ Unknown

- ☐ Anemia
- ☐ Bone marrow hypocellularity
- ☐ Immunodeficiency
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Recurrent infections
- ☐ Recurrent otitis media
- ☐ Thrombocytopenia
- ☐ Thromboembolism
- ☐ Other:_____

Musculoskeletal

◯ Yes ◯ No ◯ Unknown

- ☐ Abnormal connective tissue
- ☐ Abnormality of bone mineral density
- ☐ Abnormality of the arm – specify:_____
- ☐ Abnormality of the hand – specify:_____
- ☐ Abnormality of the ribs
- ☐ Bowing of the long bones
- ☐ Bruising susceptibility
- ☐ Recurrent fractures
- ☐ Hemihypertrophy
- ☐ Hyperostosis
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Kyphosis
- ☐ Joint contractures
- ☐ Overgrowth %ile: _____
- ☐ Pectus excavatum
- ☐ Scoliosis
- ☐ Short stature
- ☐ Skeletal dysplasia
- ☐ Tall stature
- ☐ Thoracic hypoplasia
- ☐ Vertebral abnormalities
- ☐ Other:_____

Neurological

◯ Yes ◯ No ◯ Unknown

- ☐ Anosmia, congenital
- ☐ Ataxia
- ☐ Cerebral palsy
- ☐ Dystonia
- ☐ Encephalopathy
- ☐ Epileptic encephalopathy
- ☐ Headaches
- ☐ Hemiplegic migraine
- ☐ Hyperreflexia
- ☐ Peripheral neuropathy
- ☐ Reduced tendon reflexes
- ☐ Seizures – type: _____
- ☐ Sensory neuropathy
- ☐ Spasticity
- ☐ Stroke-like episode(s)
- ☐ Other:_____

Pre/Perinatal history

◯ Yes ◯ No ◯ Unknown

- ☐ Cystic hygroma
- ☐ Increased nuchal translucency
- ☐ Intrauterine growth restriction
- ☐ Nonimmune hydrops fetalis
- ☐ Multiple prenatal fractures
- ☐ Oligohydramnios
- ☐ Omphalocele
- ☐ Polyhydramnios
- ☐ Other:_____

Respiratory

◯ Yes ◯ No ◯ Unknown

- ☐ Asthma
- ☐ Bronchiectasis
- ☐ Pneumothorax
- ☐ Pulmonary fibrosis
- ☐ Recurrent upper respiratory infections
- ☐ Respiratory distress/insufficiency
- ☐ Other:_____

Skin/Hair

◯ Yes ◯ No ◯ Unknown

- ☐ Abnormality of hair – specify:_____
- ☐ Abnormality of nail
- ☐ Alopecia
- ☐ Angiokeratoma
- ☐ Blistering of the skin
- ☐ Café-au-lait macules – count:_____
- ☐ Eczema
- ☐ Hyperextensible skin
- ☐ Hyperpigmentation of the skin
- ☐ Hypertrichosis
- ☐ Hypopigmentation of the skin
- ☐ Ichthyosis
- ☐ Recurrent skin infections
- ☐ Velvety skin (soft skin)
- ☐ Xanthomatosis
- ☐ Other:_____

Structural Brain Abnormalities

◯ Yes ◯ No ◯ Unknown

- ☐ Abnormal/delayed myelination
- ☐ Abnormality of basal ganglia
- ☐ Abnormality of brainstem
- ☐ Abnormality of the corpus callosum
- ☐ Aplasia/hypoplasia of cerebellum
- ☐ Arnold Chiari malformation
- ☐ Brain atrophy
- ☐ Cerebellar atrophy
- ☐ Cortical dysplasia
- ☐ Cortical tubers
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Hypomyelination
- ☐ Leukodystrophy
- ☐ Lissencephaly
- ☐ Molar tooth sign on MRI
- ☐ Pachygria
- ☐ Periventricular heterotopia
- ☐ Polymicrogyria
- ☐ Pontocerebellar hypoplasia
- ☐ Subcortical band heterotopia
- ☐ Ventriculomegaly
- ☐ Other:_____

Vascular System

◯ Yes ◯ No ◯ Unknown

- ☐ Aneurysm
- ☐ Arterial calcification
- ☐ Arterial dissection
- ☐ Arteriovenous malformation
- ☐ Lymphedema
- ☐ Stroke
- ☐ Other:_____

Imaging, metabolic, other lab results (attach lab reports/values)

_____

_____

_____

_____

_____

Additional clinical findings

_____

_____

_____

_____

_____

CONTINUE TO NEXT PAGE TO COMPLETE TEST CATALOG

WITHIN THE US | p: 800-436-3037 | f: 415-276-4164 | e: clientservices@invitae.com | OUTSIDE THE US | p: +1 415-930-4018 | f: +1 510 417-3922 | e: globalsupport@invitae.com
INVITAE | 1400 16th Street, San Francisco, CA 94103, USA | www.invitae.com/contact |

PEDIATRIC GENETICS TEST CATALOG

All tests on this form are organized by clinical area. If your order contains tests from multiple clinical areas, you will need to send a separate specimen for each clinical area. Each clinical area represents an individual billable event and report. Contact Client Services with any questions. For Invitae's full test menu, please visit www.invitae.com.

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Ciliopathies			
○ 04102	Invitae Ciliopathies Panel	174	ADAMTS9, AHI1, AK7, ALG8, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC4, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C11orf70, C2CD3, C8orf37, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CCNQ, CELSR2, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP55, CEP83, CFAP298, CFAP410, CFTR, CLUAP1, CPLANE1, CRB2, CSPP1, CTU2, DCDC2, DDX59, DHCR7, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB11, DNAJB13, DNAL1, DRC1, DYNC2H1, DYNC2LI1, DZIP1L, EVC, EVC2, EXOC3L2, EXOC8, FAM186B, FBN3, FGFR1, FGFR2, FGFR3, GANAB, GAS8, GLIS2, HNF1B, HYLS1, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, LRP5, LRRC56, LRRC6, LRRCC1, LZTFL1, MAPKBP1, MCIDAS, MKKS, MKS1, MRE11, NEK1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PIAS1, PIBF1, PIH1D3, PKD2, PKHD1, PMM2, PRKCSH, RBM48, RCOR1, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SCLT1, SDCCAG8, SEC63, SLC30A7, SPAG1, SUFU, TBC1D32, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TRAPPC3, TRIM32, TTC21B, TTC26, TTC8, TXNDC15, USP9X, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZMYND10, ZNF423
○ 04103	Invitae Perinatal Lethal Skeletal Dysplasia and Skeletal Ciliopathies Panel	72	AGPS, ALPL, ASCC1, CEP120, CFAP410, CHUK, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CREB3L1, CRTAP, CSPP1, DLL1, DLL3, DMRT2, DYNC2H1, DYNC2LI1, EBP, EVC, EVC2, FAM111A, FGFR2, FGFR3, GNPAT, GPX4, HES7, HSPG2, ICK, IFT122, IFT140, IFT172, IFT43, IFT52, IFT74, IFT80, IFT81, IMPAD1, INPPL1, INTU, KIAA0586, KIAA0753, LBR, LFNG, LRP5, MBTPS2, MESP2, NEK1, P3H1, PAM16, PEX5, PEX7, PPIB, RIPPLY2, SLC26A2, SLC35D1, SOX9, TBX6, TCTEX1D2, TMEM38B, TRAF3IP1, TRIP11, TRIP4, TRPV4, TTC21B, WDR19, WDR34, WDR35, WDR60, WNT1, WNT3
○ 04101	Invitae Primary Ciliary Dyskinesia Panel	42	AK7, ARMC4, C11orf70, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP164, CFAP298, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, GAS8, LRRC56, LRRC6, MCIDAS, NOTCH2, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10
○ 04112	Invitae Bardet-Biedl Syndrome Panel	28	ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CEP164, CEP19, CEP290, FBN3, IFT172, IFT27, IFT74, KIF7, LZTFL1, MKKS, MKS1, SCLT1, SDCCAG8, TRAPPC3, TRIM32, TTC8, WDPCP
○ 55012	Invitae Neonatal Respiratory Distress Panel	111	ABCA3, ACE, AFF4, AGT, AGTR1, AK7, ALB, ARL6, ARMC4, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C11orf70, C8orf37, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CD40, CD40LG, CEP164, CEP19, CEP290, CFAP298, CFTR, COPA, CSF2RA, CSF2RB, CXCR4, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, ELANE, FBN3, FLNA, FOXF1, GAS8, GATA2, HSD11B2, IFT172, IFT27, IFT74, IL1RN, INPPL1, ITGA3, KIF7, LRRC56, LRRC6, LZTFL1, MARS, MCIDAS, MKKS, MKS1, MTHFR, MTM1, NDST1, NKX2-1, NME8, NOTCH2, OFD1, PARN, PIEZO2, PIH1D3, REN, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SARS2, SCLT1, SDCCAG8, SFTPB, SFTPC, SLC27A4, SLC34A2, SLC7A7, SPAG1, TERC, TERT, TINF2, TMEM165, TMEM173, TRAPPC3, TRIM32, TTC8, WDPCP, ZMYND10
○ 04111	Invitae Joubert and Meckel-Gruber Syndromes Panel	31	AHI1, ARL13B, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, MRE11, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423

If an order is placed using an outdated test requisition form, Invitae reserves the right to upgrade ordered tests to their current versions. View current requisition forms online at www.invitae.com/forms or consider placing your order online in the Invitae portal. Please note: Test IDs containing add-on codes will include the original panel as well as the add-on.

PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Ciliopathies (continued)			
○ 04113	Invitae Nephronophthisis Panel	27	AHI1, ANKS6, CC2D2A, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PKHD1, RPGRIP1L, SDCCAG8, TCTN1, TMEM216, TMEM237, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423
Congenital Heart Defects			
○ 04201	Invitae Congenital Heart Defects and Heterotaxy Panel	82	ACTC1, ACVR2B, ALMS1, ANKS6, ARMC4, BBS10, BCOR, BRAF, CBL, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP290, CFAP53, CFAP298, CHD7, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, ELN, FOXH1, GAS8, GATA4, GDF1, GJA1, GPC3, HRAS, INVS, JAG1, KRAS, LEFTY2, LRRC6, MAP2K1, MAP2K2, MCIDAS, MED13L, MEIS2, MKS1, NEK8, NF1, NKX2-5, NKX2-6, NME8, NODAL, NOTCH1, NOTCH2, NPHP3, NR2F2, NRAS, NSD1, OFD1, PTPN11, RAF1, RIT1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SHOC2, SOS1, SPAG1, TBX1, TBX5, TTC8, ZIC3, ZMYND10, ZNF423
Connective Tissue Disorders			
○ 434340	Invitae Connective Tissue Disorders Panel	92	ABCC6, ABL1, ACTA2, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ARIH1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B3GAT3, B4GALT7, BGN, C1S, CBS, CHST14, CHST3, COG7, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, CRTAP, DCHS1, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLCN, FLNA, FLNB, FOXE3, GGCX, GORAB, HCN4, LEMD3, LOX, LOXL3, LTBP2, LTBP3, LTBP4, LZTS1, MAT2A, MED12, MFAP5, MYH11, MYLK, NOG, NOTCH1, P3H1, PKD2, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, RIN2, SKI, SLC26A2, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SPARC, TALDO1, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, UPF3B, ZNF469
Cystic Fibrosis			
○ 04714	Invitae Cystic Fibrosis Test	1	CFTR
Developmental Disorders			
○ 04215	Invitae Alagille Syndrome Panel	2	JAG1, NOTCH2
○ 04721	Invitae Alpha Thalassemia X-linked Intellectual Disability Test	1	ATRX
○ 04741	Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel	2	ACTB, ACTG1
○ 04725	Invitae Carpenter Syndrome Panel	2	MEGF8, RAB23
○ 04738	Invitae Coffin-Lowry Syndrome Test	1	RPS6KA3
○ 04737	Invitae Cohen Syndrome Test	1	VPS13B
○ 04727	Invitae Cornelia de Lange Syndrome and Related Disorders Panel	31	ADNP, AFF4, ANKRD11, ARID1A, ARID1B, BRD4, CREBBP, DPF2, EP300, ESCO2, HDAC8, KMT2A, MED13L, NIPBL, PHF6, PHIP, RAD21, SETD5, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SOX11, SRCAP, STAG1, STAG2, TAF1, TAF6, ZMYND11
○ 04303	Invitae Facial Dysostosis and Frontonasal Dysplasia Panel	28	ALX1, ALX3, ALX4, CHD7, DHODH, EDN1, EDNRA, EFNB1, EFTUD2, EVC, EVC2, GATA1, GNAI3, IRX5, PDE4D, PLCB4, POLR1A, POLR1C, POLR1D, PRKAR1A, RPL11, RPL5, RPS28, SF3B4, TCOF1, TSR2, TWIST1, ZSWIM6
○ 04736	Invitae Hypogonadotropic Hypogonadism Panel	46	ANOS1, AXL, CCDC141, CHD4, CHD7, CYP19A1, DHCR7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IGSF10, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, LMNA, NR0B1, NSMF, PCSK1, PLXNA1, POLR3B, PROK2, PROKR2, PROP1, RELN, RNF216, SEMA3A, SOX10, SOX2, SPRY4, SRA1, SRY, TAC3, TACR3, WDR11
○ 04731	Invitae Kabuki Syndrome Panel	2	KDM6A, KMT2D
○ 04747	Invitae KBG Syndrome Test	1	ANKRD11
○ 04213	Invitae Oculo-Facio-Cardio-Dental Syndrome Test	1	BCOR

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Developmental Disorders (continued)			
○ 04739	Invitae Rubinstein-Taybi Syndrome Panel	2	CREBBP, EP300
○ 01739	Invitae Simpson-Golabi-Behmel Syndrome Test	1	GPC3
○ 04735	Invitae van der Woude Syndrome Panel	2	GRHL3, IRF6
○ 01740	Invitae Weaver Syndrome Test	1	EZH2
Disorders of Sex Development/Endocrinology (if available, please provide karyotype information)			
○ 04413	Invitae Disorders of Female Sex Development Test	1	SRY
○ 04412	Invitae Androgen Insensitivity Panel	2	AR, SRD5A2
○ 04736	Invitae Hypogonadotropic Hypogonadism Panel	46	ANOS1, AXL, CCDC141, CHD4, CHD7, CYP19A1, DHCR7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IGSF10, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, LMNA, NR0B1, NSMF, PCSK1, PLXNA1, POLR3B, PROK2, PROKR2, PROP1, RELN, RNF216, SEMA3A, SOX10, SOX2, SPRY4, SRA1, SRY, TAC3, TACR3, WDR11
○ 55007	Invitae Disorders of Sex Development Panel	53	AMH, AMHR2, ANOS1, AR, ARX, ATRX, B3GLCT, CBX2, CCNQ, CHD7, CKAP2L, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, DHH, DMRT1, ESR2, FRAS1, FREM2, H6PD, HHAT, HOXA13, HSD17B3, HSD3B2, KL, LHCGR, LHX3, LHX4, MAMLD1, MAP3K1, NR0B1, NR5A1, POR, PROP1, PSMC3IP, RSPO1, SOX9, SPECC1L, SRD5A2, SRY, STAR, TNK2, TOE1, TSPYL1, TWIST2, UBR1, WNT4, WNT9B, WT1, WWOX, ZFPM2
Epilepsy Seizures and Developmental Brain Abnormalities			
○ 434349	Invitae Septo-optic Dysplasia Panel	8	GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3
○ 04741	Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel	2	ACTB, ACTG1
○ 04422	Invitae Cerebral Cavernous Malformations Panel	3	CCM2, KRIT1, PDCD10
○ 04424	Invitae Holoprosencephaly Panel	6	FGFR1, GLI2, SHH, SIX3, TGIF1, ZIC2
	○ 04424.1 Add-on preliminary-evidence genes	4	CDON, FOXH1, NODAL, PTCH1
○ 03404	Invitae Rett and Angelman Syndromes and Related Disorders Panel	40	ADSL, ALDH5A1, ARX, ATRX, CAMK2B, CDKL5, CLTC, CNTNAP2, CTNNB1, DDX3X, DYRK1A, EHMT1, FOLR1, FOXG1, GABBR2, GRIA3, GRIN2A, GRIN2B, HDAC8, IQSEC2, KANSL1, KCNA2, MBD5, MECP2, MEF2C, NGLY1, NRXN1, SATB2, SCN2A, SCN8A, SLC6A1, SLC9A6, SMC1A, STXBP1, SYNGAP1, TBL1XR1, TCF4, UBE3A, WDR45, ZEB2
○ 01721	Invitae Tuberous Sclerosis Complex Panel	2	TSC1, TSC2
○ 55002	Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel	697	AARS, AARS2, ABAT, ABCA1, ABCD1, ABCD4, ABHD5, ACADS, ACBD5, ACER3, ACO2, ACOX1, ACP5, ACSF3, ACTA2, ACTB, ACY1, ADAR, ADGRG1, ADK, ADNP, ADSL, AGA, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, ALDH18A1, ALDH3A2, ALDH5A1, ALDH6A1, ALG12, ALG13, ALG2, ALG6, ALG9, AMACR, AMPD2, AMT, ANK3, AP1S2, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOPT1, ARCN1, ARFGEF2, ARHGAP31, ARHGEF9, ARNT2, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL2, ATP13A2, ATP5A1, ATP6AP2, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRN, ATRX, AUH, B3GALNT2, B4GALNT1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCL11B, BCS1L, BICD2, BMP4, BOLA3, BPTF, BRAT1, BTD, C12orf57, C12orf65, C19orf12, C2CD3, CACNA1A, CACNA1E, CARS2, CBS, CC2D2A, CCDC88A, CDC42, CDKL5, CEP290, CHMP1A, CLCN2, CLCN4, CLCN7, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLP1, CLPP, CNNM2, CNOT1, CNTNAP1, CNTNAP2, COA7, COASY, COG7, COL18A1, COL3A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, COX8A, CPLANE1, CPLX1, CPS1, CRAT, CREBBP, CRIPT, CRLF1, CSF1R, CSPP1, CTBP1, CTC1, CTDP1, CTNS, CTSA, CTSD, CUL4B, CYB5R3, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEAF1, DEGS1, DGUOK, DHCR24, DHFR, DHX37, DLD, DLL4, DMXL2, DNM1L, DNM2 (continues)

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Epilepsy Seizures and Developmental Brain Abnormalities (continued)			
	Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel (continued)	697	(continued) DOCK6, DOCK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DPYS, DYRK1A, EARS2, EDNRB, EHMT1, EIF2AK1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5. ELOVL1, ELOVL4, ENTPD1, EOGT, EPG5, EPRS, ERCC1, ERCC2, ERCC3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, EXOSC2, EXOSC3, EXOSC8, EXOSC9, FA2H, FAM126A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FDX2, FGFRL1, FH, FIG4, FKRP, FKTN, FLVCR2, FOLR1, FOXC1, FOXG1, FOXRED1, FUCA1, FUK, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GALC, GALT, GAN, GATAD2B, GCDH, GDAP1, GFAP, GFM1, GFM2, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLRX5, GLUL, GLYCTK, GM2A, GMNN, GNAO1, GNS, GOT2, GPHN, GRIN1, GRM7, GTF2H5, GTPBP2, HACE1, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HK1, HLCS, HMGCL, HNRNPU, HSD17B10, HSD17B4, HSPD1, IARS, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, INPP5E, IREB2, ISCA1, ISCA2, ITPA, ITPR1, IVD, JAM3, KARS, KAT6B, KATNB1, KCNJ10, KCNJ2, KCNMA1, KCNT1, KCTD7, KDM1A, KIAA0556, KIAA0586, KIAA0753, KIDINS220, KIF1A, KIF1C, KLHL7, KMT2E, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB1, LARGE1, LARS2, LETM1, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM7, MAG, MAGEL2, MAN2B1, MANBA, MARS2, MAST1, MAT1A, MCCC1, MCCC2, MCOLN1, MDH2, MECP2, MED17, MED25, MEF2C, MFSD2A, MFSD8, MGP, MICU1, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MORC2, MPLKIP, MPV17, MPZ, MRPL12, MRPL44, MRPS16, MRPS22, MRPS34, MSTO1, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTR, MTRR, MUT, NACC1, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAXD, NAXE, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK1, NFE2L2, NFU1, NGLY1, NKX6-2, NOTCH1, NPC1, NPC2, NPHP1, NRXN1, NSUN3, NT5C2, NTRK2, NUBPL, NUP62, OAT, OCRL, OPA1, OPA3, OSGEP, OTC, PACS1, PAFAH1B1, PAH, PANK2, PARS2, PAX1, PC, PCCA, PCCB, PCDH12, PCGF2, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PHGDH, PHYH, PIGA, PIGB, PIGG, PIGL, PIGM, PIGN, PIGP, PIGQ, PIGT, PIGU, PIGV, PIK3C2A, PIK3R2, PLA2G6, PLAA, PLEKHG2, PLP1, PMM2, PMP22, PNKP, PNPT1, POLG, POLG2, POLR1A, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPP1R15B, PPP2R1A, PPP3CA, PPT1, PRF1, PRKDC, PRODH, PROSC, PRPS1, PRUNE1, PSAP, PSAT1, PSPH, PTEN, PTPN23, PURA, PUS3, PYCR2, QARS, QRSL1, RAB11B, RAB3GAP1, RAB3GAP2, RARS, RARS2, RBPJ, REPS1, RERE, RHOBTB2, RIN2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPGRIP1L, RPIA, RPS6KC1, RRM2B, RTTN, RXYLT1, SAMD9L, SAMHD1, SCN3A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEPSECS, SERAC1, SETBP1, SGSH, SH3TC2, SHOC2, SHPK, SLC12A2, SLC12A5, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC35A2, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A8, SLC6A9, SLC9A1, SLC9A6, SMC1A, SNAP29, SNIP1, SNORD118, SNRPB, SNX14, SOD1, SON, SOX10, SOX2, SPART, SPAST, SPATA5, SPG11, SPG7, SPTAN1, SQSTM1, SRD5A3, SSR4, ST3GAL5, STAG2, STAMBP, STAT1, STAT2, STN1, STRADA, STX11, STXBP1, STXBP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYNE1, SYNJ1, TACO1, TAF2, TANGO2, TARS2, TBC1D24, TBCD, TBCE, TBCK, TBX1, TCF4, TCTN2, TIMM50, TIMMDC1, TM4SF20, TMEM106B, TMEM126B, TMEM165, TMEM216, TMEM67, TMEM70, TMTC3, TOE1, TP53RK, TPI1, TPK1, TRAPPC11, TRAPPC9, TREX1, TRMT10A, TRMT5, TSC1, TSC2, TSEN54, TTC19, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUFM, TWNK, TXN2, TYMP, UBA5, UBE2A, UBE3A, UFM1, UNC13D, UPB1, USP7, VARS2, VPS11, VPS13D, VPS33A, WARS2, WDR45, WDR73, WHSC1, WWOX, YARS, YME1L1, ZEB2, ZFYVE26, ZIC1, ZNF335
○ 55002.1	Add-on Adult-onset Leukodystrophy and Leukoencephalopathy Panel	32	ALDH7A1, APP, CHMP2B, CP, CTSF, DNAJC5, DNMT1, FTL, GBE1, GRN, HTRA1, KIAA1161, KIF5A, LMNB1, MAPT, NOTCH3, PDGFB, PDGFRB, PDYN, PHAX, PINK1, PRNP, PSEN1, RNF216, SCP2, SLC20A2, TREM2, TYROBP, VCP, VPS13A, XK, XPR1

PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Epilepsy Seizures and Developmental Brain Abnormalities (continued)			
○ 55004	Invitae Cerebral Palsy Spectrum Disorders Panel	425	ABAT, ABCD1, ACADM, ACADVL, ACAT1, ACBD5, ACOX1, ACTB, ADAR, ADCY5, ADD3, ADNP, ADSL, AFG3L2, AGAP1, AHDC1, AHI1, AKT3, ALDH18A1, ALDH3A2, ALDH5A1, ALDH7A1, ALG13, ALG3, ALS2, AMACR, AMPD2, AMT, ANO3, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARG1, ARHGEF9, ARL6IP1, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ATAD1, ATL1, ATM, ATP13A2, ATP1A3, ATP7A, ATP7B, ATP8A2, ATRX, AUH, AUTS2, B4GALNT1, BCAP31, BCKDHA, BCKDHB, BICD2, BSCL2, BTD, C12orf65, C19orf12, CACNA1A, CACNA1G, CAMTA1, CAPN1, CASK, CBS, CCDC88C, CCT5, CDKL5, CEP290, CHD8, CHRNA1, CIZ1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLTC, COASY, COL4A1, COL4A2, COL6A3, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ9, CPS1, CPT1C, CREBBP, CTBP1, CTNNB1, CTSD, CYP27A1, CYP2U1, CYP7B1, DARS, DARS2, DBH, DBT, DCAF17, DDC, DDHD1, DDHD2, DDX3X, DGKZ, DHDDS, DHFR, DLAT, DLD, DMD, DNAJC12, DNM2, DPAGT1, DYNC1H1, DYRK1A, EEF2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, ENTPD1, EPHA4, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, FA2H, FAM126A, FARS2, FAT2, FGF12, FGF14, FH, FOLR1, FOXG1, FRRS1L, FTL, FUCA1, GABRA2, GAD1, GALC, GAMT, GATM, GBA, GBA2, GCDH, GCH1, GFAP, GJC2, GLB1, GLDC, GLRA1, GLRB, GM2A, GNAL, GNAO1, GNB1, GNS, GPHN, GPR88, GRID2, GRIN1, GRIN2B, GRM1, HACE1, HESX1, HEXA, HEXB, HGSNAT, HLCS, HMGCL, HPCA, HPRT1, HSD17B10, HSD17B4, HSPD1, IBA57, IFIH1, IQSEC2, IREB2, ITPA, ITPR1, KANK1, KAT6A, KCNA2, KCNC3, KCNJ6, KCNMA1, KCNQ2, KCNT1, KCTD17, KCTD7, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KMT2A, KMT2B, KMT2C, L1CAM, L2HGDH, LAMA2, LIAS, LMBRD1, MAG, MAOA, MAP2K1, MARS2, MAST1, MCCC1, MCCC2, MCEE, MECP2, MECR, MFSD8, MICU1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MOCS3, MPC1, MTHFR, MTOR, MTPAP, MTR, MTRR, MTTP, MUT, NAA10, NAA35, NAGLU, NAGS, NBAS, NGLY1, NIPA1, NKX2-1, NPC1, NPC2, NPHP1, NT5C2, NUS1, OTC, PAFAH1B1, PAH, PAK3, PALM, PANK2, PCBD1, PCCA, PCCB, PCDH12, PDE10A, PDE2A, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHGDH, PHIP, PIGN, PIGT, PIK3CA, PLA2G6, PLD3, PLP1, PLXNA2, PMM2, PNKD, PNP, PNPLA6, PNPO, POLG, POLR3A, PPT1, PRKRA, PROSC, PRRT2, PRUNE1, PSAT1, PSPH, PTPN11, PTS, PURA, QDPR, RAB3GAP1, RAB3GAP2, RANBP2, REEP1, REEP2, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RTN2, SACS, SAMHD1, SATB2, SCN1A, SCN2A, SCN3A, SCN8A, SETD5, SGCE, SGSH, SHH, SIL1, SIX3, SLC16A2, SLC17A5, SLC18A2, SLC19A3, SLC1A4, SLC25A15, SLC25A22, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SON, SPART, SPAST, SPATA5, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, ST3GAL5, STAMBP, STUB1, STXBP1, SUCLA2, SUCLG1, SUOX, SURF1, SYNGAP1, TAF1, TANGO2, TBC1D24, TBCK, TBL1XR1, TCF4, TECPR2, TFG, TGIF1, TGM6, TH, THAP1, TMEM240, TMEM67, TOR1A, TREX1, TRPC3, TSEN54, TTBK2, TTPA, TUBA1A, TUBB2A, TUBB2B, TUBB3, TUBB4A, UBE3A, UCHL1, VAC14, VAMP1, VPS13A, VPS13D, VPS37A, WARS2, WASHC5, WDR45, WDR62, ZBTB18, ZC4H2, ZEB2, ZFR, ZFYVE26, ZIC1, ZIC2, ZIC4
○ 55006	Invitae Brain Malformations Panel	163	ACTB, ACTG1, ADGRG1, ADNP, AHDC1, AKT3, AMPD2, APC2, ARFGEF2, ARID1A, ARID1B, ARX, ASNS, ASPM, ATP6V0A2, B3GALNT2, B4GAT1, BMP4, C19orf12, CASK, CCM2, CCND2, CDK13, CDK5, CDON, CHMP1A, CIT, CNOT1, CNOT3, COASY, COL18A1, COL3A1, COL4A1, COL4A2, CP, CRADD, CUL4B, DAG1, DCHS1, DCX, DIAPH1, DISP1, DLL1, DMXL2, DPF2, DYNC1H1, EIF2AK2, EMC1, EML1, ERMARD, EXOSC3, FA2H, FAT4, FGFR1, FIG4, FKRP, FKTN, FLNA, FOXA2, FTL, GAS1, GLI2, GMPPB, GPSM2, GRIN2B, HIVEP2, IER3IP1, IQSEC2, ISPD, KATNB1, KCNMA1, KIF11, KIF1BP, KIF2A, KIF7, KMT2E, KRIT1, L1CAM, LAMA1, LAMB1, LAMC3, LARGE1, LRP2, MACF1, MED12, MED17, MFSD2A, MRE11, NDE1, NEDD4L, NPRL3, OCLN, OPHN1, PAFAH1B1, PANK2, PDCD10, PHGDH, PIK3R2, PLA2G6, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP1R12A, PTCH1, RAB11B, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RARS2, RBM10, RELN, RERE, RTTN, RXYLT1, SEPSECS, SHH, SIN3A, SIX3, SLC25A19, SMARCA4, SMARCB1, SMARCE1, SMC1A, SNAP29, SON, SOX2, SRD5A3, STAG2, STAMBP, STIL, TBC1D20, TGIF1, TMTC3, TOE1, TRRAP, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP6, UBE3B, USP7, USP9X, VLDLR, VPS13A, VRK1, WDR45, WDR62, YWHAE, ZBTB18, ZBTB20, ZIC2, ZMIZ1

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Epilepsy Seizures and Developmental Brain Abnormalities (continued)			
○ 03401	Invitae Epilepsy Panel	302	AARS, ABAT, ADAR, ADSL, ALDH5A1, ALDH7A1, ALG1, ALG12, ALG13, ALG6, AMACR, AMT, AP2M1, AP3B2, ARG1, ARHGEF9, ARSA, ARX, ASAH1, ASNS, ATAD1, ATP1A1, ATP1A2, ATP1A3, ATP6AP2, ATP7A, ATRX, BRAT1, C12orf57, CACNA1A, CACNA1B, CACNA1E, CACNA2D2, CAD, CAMK2B, CARS2, CASK, CCDC88A, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN4, CLCN6, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLTC, CNTN2, CNTNAP2, COG5, COL18A1, CSTB, CTNNB1, CTSD, CYFIP2, CYP27A1, DDC, DDX3X, DEAF1, DENND5A, DEPDC5, DHDDS, DHFR, DIAPH1, DMXL2, DNAJC5, DNM1, DNM1L, DOCK7, DYNC1H1, DYRK1A, ECHS1, EEF1A2, EHMT1, EMC1, EPM2A, FAR1, FARS2, FBXO11, FGF12, FLNA, FOLR1, FOXG1, FOXP1, FRRS1L, GABBR2, GABRA1, GABRA2, GABRB1, GABRB2, GABRB3, GABRG2, GAMT, GATAD2B, GATM, GCH1, GLDC, GLRA1, GLRB, GNAO1, GNB1, GOSR2, GPAA1, GPHN, GRIA3, GRIN1, GRIN2A, GRIN2B, GRIN2D, GTPBP3, HCN1, HDAC8, HEXA, HNRNPU, IER3IP1, IFIH1, IQSEC2, ITPA, KANSL1, KCNA1, KCNA2, KCNB1, KCNC1, KCND2, KCNH1, KCNH2, KCNH5, KCNJ10, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD7, KIF1A, KIF2A, KIF5A, KPNA7, LAMC3, LGI1, LIAS, MBD5, MDH2, MECP2, MEF2C, MFSD8, MICAL1, MOCS1, MOCS2, MTOR, NACC1, NAGLU, NECAP1, NEDD4L, NEXMIF, NGLY1, NHLRC1, NPC1, NPC2, NPRL3, NRXN1, NTRK2, NUS1, PACS1, PACS2, PAFAH1B1, PCDH19, PCLO, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHGDH, PIGA, PIGB, PIGG, PIGN, PIGO, PIGP, PIGQ, PIGV, PIGW, PLAA, PLCB1, PNKD, PNKP, PNPO, PNPT1, POLG, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PRICKLE1, PRIMA1, PROSC, PRRT2, PSAP, PSAT1, PSPH, PTEN, PTPN23, PURA, QARS, QDPR, RAB11A, RAB11B, RAI1, RALA, RANBP2, RELN, RFT1, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNF13, ROGDI, RORB, RUSC2, SAMHD1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCP2, SERPINI1, SETBP1, SGCE, SGSH, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A5, SLC6A8, SLC9A6, SMC1A, SNAP25, SNX27, SPATA5, SPTAN1, ST3GAL3, ST3GAL5, STAG2, STRADA, STX1B, STXBP1, STXBP2, SUMF1, SUOX, SURF1, SYN1, SYNGAP1, SYNJ1, SZT2, TANGO2, TBC1D24, TBCK, TBL1XR1, TCF4, TH, TK2, TPK1, TREX1, TRIM8, TSC1, TSC2, TSFM, TUBB2A, UBA5, UBE3A, UNC80, WDR45, WWOX, YWHAG, ZDHHC9, ZEB2, ZSWIM6
	○ 03401.1 Add-on preliminary-evidence genes	18	ARHGEF15, CACNA1H, CERS1, FASN, GABRD, GUF1, IDH3A, JMJD1C, LMNB2, MOCS3, PIK3AP1, PRDM8, PRICKLE2, RBFOX1, RBFOX3, SCN5A, SNIP1, TUBA8
Eye Disorders			
○ 04305	Invitae Achromatopsia Panel	8	ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H, RGS9, RGS9BP
○ 04301	Invitae Congenital Stationary Night Blindness Panel	22	CABP4, CACNA1F, CACNA2D4, CHM, CYP4V2, FRMD7, GNAT1, GNB3, GPR179, GRM6, GUCY2D, LRIT3, NYX, PDE6B, RBP4, RDH5, RHO, RLBP1, RPE65, SAG, SLC24A1, TRPM1
○ 04302	Invitae Corneal Dystrophies Panel	33	CHRDL1, CHST6, COL17A1, COL5A1, COL8A2, CYP4V2, DCN, FOXE3, GJA8, GRHL2, GSN, KERA, KRT12, KRT3, LCAT, LOXHD1, MAF, MIR184, NLRP1, OVOL2, PAX6, PIKFYVE, PITX2, PRDM5, PXDN, SLC4A11, TACSTD2, TGFBI, UBIAD1, VSX1, ZEB1, ZNF143, ZNF469
○ 434345	Invitae Macular Dystrophy Panel	36	ABCA4, BEST1, C1QTNF5, CDH3, CERKL, CFI, CHST6, CNGB3, CRB1, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, FSCN2, GUCA1B, HMCN1, IMPG1, IMPG2, KCNV2, MFSD8, NMNAT1, PRDM13, PROM1, PRPH2, RAX2, RBP3, RBP4, RDH12, RDH5, RLBP1, RP1L1, RPGRIP1, RS1, SIX6, TIMP3
○ 434348	Invitae Oculocutaneous Albinism Panel	23	AP3B1, AP3D1, BLOC1S3, BLOC1S6, C10orf11, DTNBP1, FRMD7, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MLPH, MYO5A, OCA2, RAB27A, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1
○ 55015	Invitae Glaucoma Panel	27	ASB10, ATOH7, BMP4, COL4A1, COL8A2, CYP1B1, EXOS, FOXC1, FOXE3, LMX1B, LTBP2, MAF, MFRP, MYOC, OPTN, PAX6, PIK3R1, PITX2, PITX3, PRPF8, PRSS56, PXDN, SH3PXD2B, SIX6, SLC4A4, TEK, WDR36
○ 04112	Invitae Bardet-Biedl Syndrome Panel	28	ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CEP164, CEP19, CEP290, FBN3, IFT172, IFT27, IFT74, KIF7, LZTFL1, MKKS, MKS1, SCLT1, SDCCAG8, TRAPPC3, TRIM32, TTC8, WDPCP

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Eye Disorders (continued)			
○ 434349	Invitae Septo-optic Dysplasia Panel	8	GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3
○ 55005	Invitae Alport Syndrome Panel	6	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9
○ 55013	Invitae Stickler Syndrome Panel	9	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3, LRP2, VCAN
○ 05142	Invitae Microphthalmia, Anophthalmia, Coloboma (MAC) and Anterior Segment Dysgenesis Panel	81	ABCB6, ADAMTS18, ALDH1A3, ALX1, ASPH, BCOR, BMP4, BMP7, C12orf57, CDON, CHD7, COL4A1, CPAMD8, CRYAA, CRYBA4, CYP1B1, DCDC1, ELP4, ERCC2, ERCC5, ERCC6, FAT1, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FZD5, GDF6, GJA1, GRIP1, HCCS, HESX1, HMGB3, HMX1, ITPR1, KERA, MAB21L2, MFRP, MIR204, MITF, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PITX2, PITX3, PORCN, PRDM5, PRSS56, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, RERE, SALL2, SHH, SIX3, SIX6, SLC38A8, SMCHD1, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, UBE3B, VAX1, VSX1, VSX2, WNT2B, YAP1, ZDBF2, ZIC2
○ 04213	Invitae Oculo-Facio-Cardio-Dental Syndrome Test	1	BCOR
○ 05132	Invitae Cataracts Panel	103	ABCA3, ABCB6, ABHD12, ADAMTS18, ADAMTSL4, AGK, ALDH18A1, BCOR, BEST1, BFSP1, BFSP2, CHMP4B, CLN3, COL11A1, COL18A1, COL2A1, COL4A1, COL4A2, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, EPG5, EPHA2, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, FAM126A, FOXC1, FOXE3, FTL, FYCO1, FZD4, GALK1, GALT, GCNT2, GFER, GJA1, GJA3, GJA8, HMX1, HSF4, JAM3, LEMD2, LIM2, LONP1, LSS, MAF, MIP, MIR184, MYH9, NDP, NF2, NHS, OCRL, OPA3, P3H2, PAX6, PEX10, PEX11B, PEX16, PEX2, PEX7, PITX2, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RDH11, RECQL4, RGS6, RNLS, RRAGA, SC5D, SIL1, SIPA1L3, SIX6, SLC16A12, SLC33A1, TBC1D20, TDRD7, TFAP2A, TMEM70, UNC45B, VIM, VSX2, WDR87, WFS1, WRN, XYLT2
	○ 05132.1 Add-on preliminary-evidence genes for congenital cataracts	4	CHMP4B, CRYGB, LIM2, VIM
○ 72100	Invitae Inherited Retinal Disorders Panel	330	ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRA3, ADGRV1, ADIPOR1, AGBL5, AHI1, AHR, AIPL1, ALMS1, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARSG, ASRGL1, ATF6, ATOH7, B9D1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCT2, CDH23, CDH3, CDHR1, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CFAP410, CHM, CIB2, CISD2, CLCC1, CLN3, CLRN1, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CPLANE1, CRB1, CRX, CSPP1, CTNNA1, CWC27, CYP4V2, DHDDS, DHX38, DRAM2, DTHD1, EFEMP1, ELOVL4, EMC1, EXOSC2, EYS, FAM161A, FLVCR1, FRMD7, FSCN2, FZD4, GDF6, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRM6, GUCA1A, GUCA1B, GUCY2D, HARS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT80, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, JAG1, KCNJ13, KCNV2, KIAA0586, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MAPKAPK3, MERTK, MFN2, MFRP, MKKS, MKS1, MPDZ, MTTP, MYO7A, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OTX2, P3H2, PAX2, PAX6, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCLT1, SDCCAG8, SEMA4A, SLC24A1, SLC45A2, SLC7A14, SNRNP200, SPATA7, SPP2, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TRAF3IP1, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, UNC119, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WFS1, WHRN, ZNF408, ZNF423, ZNF513, ADAMTSL4, C12orf65, CLN5, CLN6, CLN8, CTSD, DHX32, DNAJC17, DSCAML1, ERCC6, FBLN5, GNS, GRN, HMCN1, IFT74, IFT88, ITM2B, MFSD8, MIR204, MTPAP, NAGLU, NBAS, OR2W3, POC5, PPT1, RBP1, RP9, RPGR, RPGR (ORF15), SGSH, SIX6, TMED7, WDR34, GPR45, SLC24A5, C10orf11, LYST

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Neurodevelopmental Disorders			
○ 728434	Invitae Neurodevelopmental Disorders Panel	241	ACTB, ACTG1, ADNP, ADSL, AGA, AHDC1, ALDH5A1, ALDH7A1, AMER1, ANKRD11, AP1S2, ARG1, ARID1A, ARID1B, ARSA, ARX, ASNS, ASXL1, ATP1A3, ATP7A, ATRX, AUTS2, BCAP31, BRAF, BRAT1, BRD4, BRWD3, CACNA1A, CACNA1E, CAMK2B, CASK, CBL, CC2D2A, CDK13, CDKL5, CHD2, CHD7, CHD8, CLCN4, CLN3, CLN5, CLN6, CLTC, CNTNAP2, COL4A1, CREBBP, CTNNB1, CUL3, DDC, DDX3X, DEAF1, DHCR7, DNM1L, DNMT3A, DOCK6, DPF2, DYNC1H1, DYRK1A, EEF1A2, EFTUD2, EHMT1, EP300, EZH2, FGD1, FOLR1, FOXG1, FOXP1, GABBR2, GABRB3, GABRG2, GALC, GAMT, GATAD2B, GATM, GLB1, GM2A, GNAO1, GNAS, GNS, GPC3, GRIA3, GRIN1, GRIN2A, GRIN2B, HDAC8, HEXA, HEXB, HGSNAT, HIVEP2, HNRNPK, HNRNPU, HRAS, HUWE1, IDS, IDUA, IGF1R, IL1RAPL1, IQSEC2, ITPR1, KANSL1, KAT6A, KAT6B, KCNA2, KCNB1, KCNH1, KCNQ2, KCNT1, KDM5C, KDM6A, KIF1A, KMT2A, KMT2B, KMT2D, KMT2E, KRAS, L1CAM, LZTR1, MAGEL2, MAN1B1, MAP2K1, MAP2K2, MBD5, MECP2, MED12, MED13L, MEF2C, MFSD8, MID1, MTOR, NAA10, NAA15, NAGLU, NALCN, NEXMIF, NF1, NFIA, NFIX, NGLY1, NHS, NIPBL, NONO, NPC1, NR2F1, NRAS, NRXN1, NSD1, NSUN2, OCRL, OPHN1, OTC, PACS1, PACS2, PAH, PCBD1, PCDH19, PDHA1, PGAP3, PHF21A, PHF6, PHIP, PLA2G6, PMM2, POLG, PPM1D, PPP1CB, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PTEN, PTPN11, PTS, PURA, QDPR, RAD21, RAF1, RAI1, RBM10, RIT1, RPS6KA3, SATB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SETBP1, SETD5, SGSH, SHOC2, SIN3A, SLC13A5, SLC16A2, SLC2A1, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SON, SOS1, SOS2, SOX11, SPAST, SPATA5, SPTAN1, STAG1, STXBP1, SURF1, SYNGAP1, TAF1, TBCK, TBL1XR1, TCF20, TCF4, TELO2, TPP1, TRAPPC9, TRRAP, TSC1, TSC2, TUBA1A, UBE3A, UNC80, USP9X, VPS13B, WDR45, WWOX, ZBTB18, ZBTB20, ZC4H2, ZDHHC9, ZEB2, ZIC2, ZMIZ1, ZMYND11
Overgrowth Syndromes			
○ 01739	Invitae Simpson-Golabi-Behmel Syndrome Test	1	GPC3
○ 01740	Invitae Weaver Syndrome Test	1	EZH2
○ 04501	Invitae Overgrowth and Macrocephaly Syndromes Panel	21	AKT2, AKT3, CDKN1C, CUL4B, DIS3L2, DNMT3A, EZH2, GLI3, GPC3, KPTN, MED12, MTOR, NF1, NFIX, NPR2, NSD1, PHF6, PIK3R2, PTEN, SETD2, SPRED1
	○ 04501.1 Add-on preliminary-evidence genes	5	EED, DICER1, PDGFRB, RNF125, UPF3B
○ 55011	Invitae Overgrowth Syndromes Panel	56	ABCC9, AKT1, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD4, CHD8, CUL4B, DICER1, DIS3L2, DNMT3A, EED, EZH2, GFAP, GLI3, GNAQ, GNAS, GPC3, HEPACAM, HERC1, KPTN, L1CAM, MED12, MLC1, MPDZ, MTOR, NFIA, NFIX, NONO, NPR2, NSD1, OFD1, PDGFRB, PHF21A, PIK3CA, PIK3R2, PPP2R5B, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RASA1, RIN2, RNF125, SETD2, STRADA, SYN1, TBC1D7, TCF20, UPF3B, ZBTB20
Pulmonary Disorders			
○ 55012	Invitae Neonatal Respiratory Distress Panel	111	ABCA3, ACE, AFF4, AGT, AGTR1, AK7, ALB, ARL6, ARMC4, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C11orf70, C8orf37, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CD40, CD40LG, CEP164, CEP19, CEP290, CFAP298, CFTR, COPA, CSF2RA, CSF2RB, CXCR4, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, ELANE, FBN3, FLNA, FOXF1, GAS8, GATA2, HSD11B2, IFT172, IFT27, IFT74, IL1RN, INPPL1, ITGA3, KIF7, LRRC56, LRRC6, LZTFL1, MARS, MCIDAS, MKKS, MKS1, MTHFR, MTM1, NDST1, NKX2-1, NME8, NOTCH2, OFD1, PARN, PIEZO2, PIH1D3, REN, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SARS2, SCLT1, SDCCAG8, SFTPB, SFTPC, SLC27A4, SLC34A2, SLC7A7, SPAG1, TERC, TERT, TINF2, TMEM165, TMEM173, TRAPPC3, TRIM32, TTC8, WDPCP, ZMYND10
○ 04306	Invitae Surfactant Metabolism Panel	21	ABCA3, COPA, CSF2RA, CSF2RB, DKC1, FLNA, FOXF1, GATA2, ITGA3, MARS, NKX2-1, NOTCH2, PARN, RTEL1, SFTPB, SFTPC, SLC7A7, TERC, TERT, TINF2, TMEM173MOCS1, MOCS2, MOCS3, PNP, PRPS1, SUOX, UMOD, XDH
RASopathies (Noonan Spectrum Disorders)			
○ 04163	Invitae Cardio-Facio-Cutaneous Syndrome Panel	6	BRAF, KRAS, MAP2K1, MAP2K2, SHOC2, SOS1

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
RASopathies (Noonan Spectrum Disorders) (continued)			
○ 04151	Invitae RASopathies and Noonan Spectrum Disorders Panel	28	A2ML1, ACTB, ACTG1, BRAF, CBL, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1, YWHAZ
○ 04165	Invitae Legius Syndrome Test	1	SPRED1
	○ 04165.1 Add-on neurofibromatosis type 1 gene	1	NF1
○ 01708	Invitae Neurofibromatosis Type 1 Test	1	NF1
	○ 01708.1 Add-on Legius syndrome gene	1	SPRED1
Skeletal Disorders			
○ 434342	Invitae Adams-Oliver Syndrome Panel	8	ARHGAP31, DLL4, DOCK6, EOGT, KCTD1, NOTCH1, RBPJ, UBR1
○ 04612	Invitae Antley-Bixler Syndrome Test	1	POR
	○ 04612.1 Add-on craniosynostosis gene	1	FGFR2
○ 04726	Invitae ARSE-Related Chondrodysplasia Punctata Test	1	ARSE
	○ 04726.1 Add-on NSDHL-related disorders gene	1	NSDHL
○ 04712	Invitae Campomelic Dysplasia Test	1	SOX9
○ 04423	Invitae Craniosynostosis Panel	65	ALPL, ASXL1, B3GAT3, CD96, CDC45, CDT1, COLEC11, CYP26B1, EFNA4, EFNB1, ERF, ESCO2, FBN1, FGF9, FGFR1, FGFR2, FGFR3, FREM1, GLI3, GPC3, IFT122, IFT140, IFT43, IGF1R, IL11RA, KAT6A, KAT6B, MASP1, MEGF8, MSX2, NFIA, ORC1, ORC4, ORC6, P4HB, PHEX, POR, PPP3CA, RAB23, RECQL4, RSPRY1, RUNX2, SCARF2, SEC24D, SIX2, SKI, SLC25A24, SMAD2, SMAD3, SMAD6, SOX6, SPECC1L, STAT3, TCF12, TCOF1, TGFB2, TGFB3, TGFBR1, TGFBR2, TMCO1, TWIST1, WDR19, WDR35, ZEB2, ZIC1
	○ 04423.1 Add-on 3MC and Treacher Collins syndromes genes	2	MASP1, TCOF1
○ 04613	Invitae Ellis-van Creveld and Weyers Acrofacial Dysostosis Panel	2	EVC, EVC2
○ 04614	Invitae Hereditary Multiple Osteochondromas Panel	2	EXT1, EXT2
	○ 04614.1 Add-on Langer-Giedion Syndrome Gene	1	TRPS1
○ 72039	Invitae Hypophosphatemia Panel	17	ALPL, CLCN5, CTNS, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, GNAS, OCRL, PHEX, SLC34A1, SLC34A3, VDR
○ 72038	Invitae X-Linked Hypophosphatemia Test	1	PHEX
○ 55010	Invitae Limb and Digital Malformations Panel	178	ACVR1, ADAMTS10, ADAMTS17, AFF4, AHI1, ANKRD11, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, B3GLCT, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP2, BMP4, BMPR1B, BTRC, C2CD3, CACNA1C, CC2D2A, CCNQ, CDH3, CEP104, CEP120, CEP290, CEP41, CHSY1, CHUK, CKAP2L, CPLANE1, CREBBP, CSPP1, DDX59, DHCR7, DHODH, DLL4, DLX5, DLX6, DOCK6, DPF2, DVL1, DVL3, DYNC1I1, EOGT, EP300, ESCO2, EVC, EVC2, FAT1, FBLN1, FBN1, FBXW4, FGF10, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FLNA, FMN1, FRAS1, FREM2, FZD2, GDF5, GDF6, GJA1, GLI2, GLI3, GNAS, GSC, HDAC4, HDAC8, HOXA13, HOXD13, IFT57, IHH, INPP5E, KDM6A, KIAA0586, KIF7, KMT2A, KMT2D, LMBR1, LRP4, LTBP2, LTBP3, MAP3K20, MEGF8, MGP, MKKS, MKS1, MRE11, MYCN, NECTIN1, NECTIN4, NIPBL, NOG, NOTCH1, NPHP1, NPHP3, NSDHL, NXN, OFD1, PDE3A, PDE4D, PDE6D, PGM3, PHF6, PIGV, PIK3CA, PITX1, POLR1A, PORCN, PRKAR1A, PRMT7, PTDSS1, PTHLH, RAB23, RAD21, RBM8A, RBPJ, RECQL4, ROR2, RPGRIP1L, SALL1, SALL4, SC5D, SDCCAG8, SF3B4, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMOC1, SOST, SOX11, SOX9, TBX15, TBX3, TBX5, TCTN1, TCTN2, TCTN3, TGDS, THPO, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TP63, TRIM32, TRPS1, TRPV4, TTC21B, TTC8, VAC14, WDPCP, WNT10B, WNT3, WNT5A, WNT7A, ZNF423, ZSWIM6

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Skeletal Disorders (continued)			
○ 04307	Invitae Microcephalic Primordial Dwarfism and Seckel Syndrome Panel	38	ATR, ATRIP, CDC45, CDC6, CDK5RAP2, CDT1, CENPJ, CEP135, CEP152, CEP63, CEP97, CRIPT, DNA2, DNMT3A, DONSON, GMNN, LARP7, LIG4, MCM5, MCPH1, NIN, NSMCE2, ORC1, ORC4, ORC6, PCNT, PLK4, POC1A, RBBP8, RNU4ATAC, RTTN, SRCAP, TRAIP, TUBGCP4, TUBGCP6, UBE3B, WDR4, XRCC4
○ 04732	Invitae Osteogenesis Imperfecta and Bone Fragility Panel	67	ALPL, ANO5, ASCC1, B3GAT3, B4GALT7, BMP1, CA2, CLCN5, CLCN7, COL1A1, COL1A2, CREB3L1, CRTAP, CTNS, CTSK, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FAM46A, FGF23, FGFR1, FKBP10, GNAS, GORAB, IFITM5, LRP5, LRRK1, MBTPS2, MESDC2, NBAS, NOTCH2, NTRK1, OCRL, OSTM1, P3H1, P4HB, PHEX, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SFRP4, SGMS2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SNX10, SP7, SPARC, SUCO, TAPT1, TCIRG1, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TRIP4, VDR, WNT1, WNT3A, XYLT2
○ 04103	Invitae Perinatal Lethal Skeletal Dysplasia and Skeletal Ciliopathies Panel	73	AGPS, ALPL, ASCC1, CEP120, CFAP410, CHUK, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CREB3L1, CRTAP, CSPP1, DLL1, DLL3, DMRT2, DYNC2H1, DYNC2LI1, EBP, EVC, EVC2, FAM111A, FGFR2, FGFR3, GNPAT, GPX4, HES7, HSPG2, ICK, IFT122, IFT140, IFT172, IFT43, IFT52, IFT74, IFT80, IFT81, IMPAD1, INPPL1, INTU, KIAA0586, KIAA0753, LBR, LFNG, LRP5, MBTPS2, MESP2, NEK1, P3H1, PAM16, PEX5, PEX7, PPIB, RIPPLY2, SLC26A2, SLC35D1, SOX9, TBX6, TCTEX1D2, TMEM38B, TRAF3IP1, TRIP11, TRIP4, TRPV4, TTC21B, WDR19, WDR34, WDR35, WDR60, WNT1, WNT3
○ 89100	Invitae Skeletal Disorders Panel	358	ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, AFF4, AGA, AGPS, AIFM1, ALPL, AMER1, ANKH, ANO5, ARCN1, ARSB, ARSE, ASCC1, ASPM, ATR, ATRIP, B3GALT6, B3GAT3, B4GALT7, BGN, BMP1, BMP2, BMPER, BMPR1B, C2CD3, CA2, CANT1, CASR, CCDC8, CDC45, CDC6, CDK5RAP2, CDKN1C, CDT1, CENPJ, CEP120, CEP135, CEP152, CEP63, CEP97, CFAP410, CHST14, CHST3, CHUK, CLCN5, CLCN7, COG1, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREB3L1, CRIPT, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTNS, CTSA, CTSK, CUL7, CWC27, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DIP2C, DLL1, DLL3, DLX3, DMP1, DMRT2, DNA2, DNMT3A, DONSON, DVL1, DVL3, DYM, DYNC2H1, DYNC2LI1, EBP, EIF2AK3, ENPP1, ESCO2, EVC, EVC2, EXOC6B, EXOSC2, EXT1, EXT2, EXTL3, FAH, FAM111A, FAM20C, FAM46A, FAR1, FAT4, FBN1, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FTO, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GHRHR, GHSR, GJA1, GLB1, GMNN, GNAS, GNE, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GSC, GUSB, GZF1, HES7, HGSNAT, HPGD, HSPG2, HYAL1, IARS2, ICK, IDS, IDUA, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IGF1, IGF2, IHH, IMPAD1, INPPL1, INTU, JAG1, KAT6B, KIAA0586, KIAA0753, KIF22, KL, KMT2A, LARP7, LBR, LEMD3, LFNG, LIFR, LIG4, LMNA, LMX1B, LONP1, LOXL3, LRP4, LRP5, LRRK1, LTBP2, LTBP3, MAFB, MAN2B1, MANBA, MAP3K7, MATN3, MBTPS1, MBTPS2, MCM5, MCPH1, MEOX1, MESDC2, MESP2, MGP, MMP13, MMP14, MMP2, MMP9, MNX1, MSX2, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NIN, NKX3-2, NOG, NOTCH2, NPPC, NPR2, NPR3, NSDHL, NSMCE2, NTRK1, NXN, OBSL1, OCRL, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PCGF2, PCNT, PCYT1A, PDE4D, PEX5, PEX7, PGM3, PHEX, PIK3C2A, PISD, PKDCC, PLK4, PLOD2, PLS3, POC1A, POLR1A, POP1, POR, PPIB, PPP3CA, PRKAR1A, PTDSS1, PTH1R, PTHLH, PTPN11, PYCR1, RAB33B, RBBP8, RECQL4, RIPPLY2, RMRP, RNU4ATAC, ROR2, RSPO2, RSPRY1, RTTN, RUNX2, SC5D, SEC24D, SERPINF1, SERPINH1, SETBP1, SFRP4, SGMS2, SGSH, SH3PXD2B, SLC17A5, SLC26A2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC39A13, SLCO2A1, SLCO5A1, SMAD4, SMARCAL1, SNRPB, SNX10, SOX9, SP7, SPARC, SQSTM1, SRCAP, SUCO, SULF1, SUMF1, TAB2, TAPT1, TBCE, TBX15, TBX3, TBX5, TBX6, TBXAS1, TCIRG1, TCTEX1D2, TCTN3, TGFB1, TMEM165, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TRAF3IP1, TRAIP, TRAPPC2, TREM2, TRIM37, TRIP11, TRIP4, TRMT10A, TRPS1, TRPV4, TTC21B, TUBGCP4, TUBGCP6, TYROBP, UBE3B, VAC14, VDR, VPS33A, WDR19, WDR34, WDR35, WDR4, WDR60, WISP3, WNT1, WNT3, WNT3A, WNT5A, XRCC4, XYLT1, XYLT2, ZMPSTE24, ZNF687
○ 434350	Invitae Spondylocostal Dysostosis Panel	8	DLL1, DLL3, DMRT2, HES7, LFNG, MESP2, RIPPLY2, TBX6
○ 55013	Invitae Stickler Syndrome Panel	9	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3, LRP2, VCAN

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Skeletal Disorders (continued)			
○ 04617	Invitae Trichorhinophalangeal Syndrome Panel	2	EXT1, TRPS1
Skin Disorders			
○ 434342	Invitae Adams-Oliver Syndrome Panel	8	ARHGAP31, DLL4, DOCK6, EOGT, KCTD1, NOTCH1, RBPJ, UBR1
○ 434344	Invitae Epidermolysis Bullosa and Palmoplantar Keratoderma Panel	46	AAGAB, AQP5, ATP2C1, CAST, CD151, CDSN, COL17A1, COL7A1, CTSC, DSG1, DSP, DST, ENPP1, EXPH5, FERMT1, GJB6, ITGA3, ITGA6, ITGB4, JUP, KANK2, KLHL24, KRT1, KRT10, KRT14, KRT16, KRT17, KRT5, KRT6A, KRT6B, KRT6C, KRT9, LAMA3, LAMB3, LAMC2, LOR, PKP1, PLEC, POMP, RHBDF2, RSPO1, SERPINB7, SERPINB8, SLURP1, TAT, TRPV3
○ 434346	Invitae Congenital Ichthyosis Panel	46	ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AQP5, CAST, CDSN, CERS3, CLDN1, CYP4F22, EBP, ELOVL1, ELOVL4, GJA1, GJB2, GJB3, GJB4, GJB6, KDSR, KRT1, KRT10, KRT2, KRT9, LIPN, LOR, MBTPS2, NIPAL4, PEX7, PHYH, PNPLA1, POMP, SDR9C7, SERPINB7, SERPINB8, SLC27A4, SNAP29, SPINK5, ST14, STS, SULT2B1, SUMF1, TGM1, VPS33B, ZMPSTE24
○ 04304	Invitae Hypopigmentation Panel	47	ACD, AP3B1, AP3D1, BLOC1S3, BLOC1S6, C10orf11, CLCN7, CTC1, DKC1, DTNBP1, EDN3, EDNRB, EPG5, FRMD7, GNAI3, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LYST, MITF, MLPH, MYO5A, NHP2, NOP10, OCA2, PARN, PAX3, RAB27A, RET, RTEL1, SLC24A5, SLC38A8, SLC45A2, SNAI2, SOX10, TERC, TERT, TINF2, TYR, TYRP1, USB1, WRAP53
○ 434348	Invitae Oculocutaneous Albinism Panel	23	AP3B1, AP3D1, BLOC1S3, BLOC1S6, C10orf11, DTNBP1, FRMD7, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MLPH, MYO5A, OCA2, RAB27A, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1
○ 434351	Invitae Xeroderma Pigmentosum Panel	9	DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC
○ 04163	Invitae Cardio-Facio-Cutaneous Syndrome Panel	6	BRAF, KRAS, MAP2K1, MAP2K2, SHOC2, SOS1
○ 05021	Invitae Ectodermal Dysplasia and Related Disorders Panel	73	ANTXR1, APCDD1, ATP7A, AXIN2, BCS1L, BMP4, CDH3, CDSN, CLDN1, CTSC, DSG4, DSP, EDA, EDAR, EDARADD, EGFR, ERCC2, ERCC3, ERCC8, GJA1, GJB2, GJB6, GRHL2, GTF2E2, GTF2H5, HOXC13, HR, IRF6, JUP, KANK2, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT25, KRT6A, KRT6B, KRT6C, KRT71, KRT74, KRT83, KRT85, LIPH, LPAR6, LRP6, LSS, LTBP3, MBTPS2, MPLKIP, MSX1, NECTIN1, NECTIN4, NFKBIA, PAX9, PKP1, POC1A, PORCN, PTH1R, RIN2, RNF113A, RPL21, SMOC2, SNRPE, SOX18, SPINK5, TFAP2B, TP63, TRPS1, TRPV3, TSPEAR, WNT10A, WNT10B
○ 04165	Invitae Legius Syndrome Test	1	SPRED1
	○ 04165.1 Add-on neurofibromatosis type 1 gene	1	NF1
○ 04735	Invitae van der Woude Syndrome Panel	2	GRHL3, IRF6
Deafness			
○ 55005	Invitae Alport Syndrome Panel	6	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9
○ 55014	Invitae Usher Syndrome Panel	17	ABHD12, ADGRV1, ARSG, CDH23, CEP250, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, PEX1, PEX6, USH1C, USH1G, USH2A, WHRN
○ 55013	Invitae Stickler Syndrome Panel	9	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3, LRP2, VCAN
○ 55009	Invitae Comprehensive Deafness Panel	224	ABHD12, ABHD5, ACOX1, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ANLN, ARSB, ARSG, ATP1A3, ATP2B2, ATP6V1B1, BCAP31, BCS1L, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CEACAM16, CEP250, CEP78, CHD7, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DBH, DCAF17, DCDC2, DFNA5, DFNB59, DIABLO, DIAPH1, DIAPH3, DMXL2, DNMT1, DSPP, EDN3, EDNRA, EDNRB, EFTUD2, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FAM65B, FGF3, FGFR3, FOXC1, FOXI1, GALNS, GATA3, GDF6, GIPC3, GJA1, GJB1, GJB2, GJB3, GJB6, GLB1, GNS, GPSM2, GRHL2, GRXCR1, GRXCR2, GUSB, HARS, HARS2, HGF, HGSNAT, HOMER2, HSD17B4, HYAL1, IDS (continues)

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Deafness (continued)			
	Invitae Comprehensive Deafness Panel (continued)	224	(continued) IDUA, ILDR1, JAG1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LHX3, LOXHD1, LOXL3, LRP2, LRTOMT, MAN2B1, MARVELD2, MCM2, MEOX1, MET, MIR96, MITF, MPZ, MSRB3, MYH14, MYH7B, MYH9, MYO15A, MYO18B, MYO3A, MYO6, MYO7A, NAGLU, NARS2, NDRG1, NF2, NLRP3, NOG, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PCGF2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, RAI1, RDX, RMND1, ROR1, RPS6KA3, S1PR2, SCP2, SERAC1, SERPINB6, SGSH, SH3TC2, SIX1, SIX5, SLC12A2, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLC29A3, SLC44A4, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMPX, SNAI2, SOX10, SYNE4, TBC1D24, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMEM126A, TMEM132E, TMIE, TMPRSS3, TPRN, TRIOBP, TRRAP, TSPEAR, TUBB4B, TWNK, UBR1, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, WHRN, ZNF469
Kidney Disorders			
○ 55005	Invitae Alport Syndrome Panel	6	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9
○ 434341	Invitae Congenital Anomalies of Kidney and Urinary Tract (CAKUT) Panel	41	ACE, AGT, AGTR1, BICC1, BNC2, CRKL, DCHS1, DSTYK, EYA1, FAT4, FGF20, FOXC1, FRAS1, FREM1, FREM2, GATA3, GLI3, GREB1L, HNF1B, HOXA13, HPSE2, ITGA8, LRP4, NRIP1, PAX2, PBX1, REN, RET, ROBO2, SALL1, SIX1, SIX2, SIX5, SLIT2, SOX17, TBX18, UMOD, VIPAS39, VPS33B, WNT4, WT1
○ 55008	Invitae Cystic Kidney Disease Panel	44	ALG8, ANKS6, BICC1, CEP164, CEP290, CEP83, CEP89, COL4A1, CRB2, DCDC2, DICER1, DNAJB11, DZIP1L, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, JAG1, LRP5, MAPKBP1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD2, PKHD1, PRKCSH, RPGRIP1L, SDCCAG8, SEC61A1, SEC63, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, ZNF423
○ 633100	Invitae Expanded Renal Disease Panel	401	ABCC6, ACE, ACTB, ACTN4, ADA2, ADAMTS13, ADCY10, AGPAT2, AGT, AGTR1, AGXT, AHI1, ALG1, ALG8, ALG9, ALMS1, ALPL, AMN, ANKS6, ANLN, ANOS1, AP2S1, APOA1, APOC2, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL13B, ARL6, ATP6V0A4, ATP6V1B1, ATP7B, AVP, AVPR2, B2M, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP4, BNC2, BRAF, BSND, C8orf37, CA2, CACNA1D, CACNA1H, CASR, CBL, CC2D2A, CD151, CD2AP, CDC73, CDKN1C, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP83, CEP89, CFH, CFHR5, CFI, CHD7, CHRM3, CHRNA3, CISD2, CLCN2, CLCN5, CLCNKB, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, COX10, COX14, CPLANE1, CPT2, CRB2, CREBBP, CRKL, CSPP1, CTNS, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, CYP27B1, CYP2R1, DCDC2, DCHS1, DGKE, DHCR7, DHTKD1, DICER1, DLC1, DMP1, DNAJB11, DNASE1L3, DSTYK, DZIP1L, EBP, EGF, EIF2AK3, ELP1, EMP2, ENPP1, EYA1, FAM20A, FAN1, FANCA, FAT1, FAT4, FBN3, FGF10, FGF20, FGF23, FGFR1, FGFR2, FN1, FOXC1, FOXC2, FOXI1, FOXP3, FRAS1, FREM1, FREM2, FXYD2, G6PC, GALNT3, GANAB, GATA3, GATM, GCM2, GLA, GLI3, GLIS2, GLIS3, GNA11, GNAS, GPC3, GPHN, GREB1L, GRHPR, GRIP1, GSN, HGD, HNF1A, HNF1B, HOGA1, HOXA13, HPRT1, HPS1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43, IFT74, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA6, ITGA8, ITGB4, ITSN2, JAG1, KANK1, KANK2, KANK4, KANSL1, KAT6B, KCNA1, KCNJ1, KCNJ10, KCNJ5, KCTD1, KIAA0586, KIF7, KLHL3, KRAS, LAGE3, LAMA5, LAMB2, LCAT, LDHA, LMNA, LMX1B, LPIN1, LRP2, LRP4, LRP5, LYZ, LZTFL1, LZTR1, MAFB, MAGED2, MAGI2, MAP2K1, MAP2K2, MAPKBP1, MEFV, MKKS, MKS1, MMACHC, MNX1, MOCOS, MOCS1, MOCS2, MRE11, MUT, MVK, MYCN, MYH9, MYO1E, NEK8, NF1, NLRP3, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR0B1, NR3C2, NRAS, NRIP1, NSD1, NSDHL, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OFD1, OPLAH, OSGEP, PAX2, PBX1, PCBD1, PDE6D, PDSS1, PDSS2, PEX6, PHEX, PKD2, PKHD1, PLCE1, PLG, PMM2, PODXL, PREPL, PRKCSH, PRODH, PROKR2, PRPS1, PTPN11, PTPRO, RAF1, REN, RET, RIT1, RMND1, ROBO1, ROBO2, RPGRIP1L, RPL11, RPL26, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS7, RRM2B, SALL1, SALL4, SARS2, SCARB2, SCLT1, SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SEMA3E, SGPL1, SHOC2, SI, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC22A12 (continues)

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Kidney Disorders (continued)			
	Invitae Expanded Renal Disease Panel (continued)	401	(continued) SLC26A1, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC37A4, SLC3A1, SLC41A1, SLC4A1, SLC4A4, SLC5A1, SLC6A19, SLC7A7, SLC7A9, SLC9A3R1, SLIT2, SLX4, SMARCAL1, SMC1A, SOS1, SOS2, SOX17, SOX18, SRCAP, STX16, SYNPO, TBC1D8B, TBX18, TCTN1, TCTN2, TCTN3, TFAP2A, THBD, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNS2, TP53RK, TP63, TPRKB, TRAPPC3, TRIM32, TRPC6, TRPM6, TSC1, TSC2, TTC21B, TTC8, TTR, UMOD, UQCC2, VDR, VHL, VIPAS39, VPS33B, WAS, WDPCP, WDR19, WDR73, WFS1, WNK1, WNK4, WNT4, WNT5A, WT1, XDH, XPNPEP3, XPO5, YRDC, ZNF423
○ 434347	Invitae Nephrotic Syndrome and Focal Segmental Glomerulosclerosis (FSGS) Panel	57	ACTN4, AMN, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, CFH, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DHTKD1, EMP2, FN1, INF2, ITGA3, ITGB4, KANK1, KANK2, KANK4, LAGE3, LAMA5, LAMB2, LMX1B, MAGI2, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OSGEP, PAX2, PDSS2, PLCE1, PODXL, PTPRO, SGPL1, SMARCAL1, TP53RK, TPRKB, TRPC6, WDR73, WT1, XPO5
○ 75000	Invitae Progressive Renal Disease Panel	195	ACE, ACTB, ACTN4, ADAMTS13, AGT, AGTR1, AHI1, ALG8, ALG9, ALMS1, AMN, ANKS6, ANLN, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL6, ATP6V0A4, ATP6V1B1, AVP, AVPR2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BSND, CA2, CACNA1D, CACNA1H, CASR, CD151, CD2AP, CDKN1C, CEP164, CEP19, CEP290, CEP83, CEP89, CFHR5, CHRM3, CLCN5, CLCNKB, CLDN16, CLDN19, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, CPLANE1, CRB2, CTNS, CUBN, CYP24A1, DCDC2, DGKE, DHCR7, DICER1, DLC1, DNAJB11, DZIP1L, EGF, EMP2, EYA1, FAM20A, FAN1, FAT1, FN1, FOXI1, FOXP3, FREM1, GANAB, GATA3, GATM, GLA, GLIS2, GRHPR, GSN, HNF1A, HNF1B, HPRT1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43, INF2, INVS, IQCB1, ITGA3, ITGA6, ITGB4, JAG1, KANK2, KANK4, KAT6B, KCNJ1, KCNJ10, LAMA5, LAMB2, LCAT, LDHA, LMX1B, LPIN1, LRP4, LRP5, LYZ, LZTFL1, MAGED2, MAGI2, MAPKBP1, MEFV, MKKS, MYH9, MYO1E, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NSDHL, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OFD1, PAX2, PBX1, PDSS2, PHEX, PKD2, PKHD1, PLCE1, PODXL, PREPL, PTPRO, REN, RMND1, RPGRIP1L, SALL1, SALL4, SARS2, SDCCAG8, SEC61A1, SEMA3E, SGPL1, SIX1, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC37A4, SLC41A1, SLC4A1, SLC4A4, SLC7A7, SMARCAL1, SYNPO, TBC1D8B, TMEM67, TNS2, TP63, TRIM32, TRPC6, TSC1, TSC2, TTC21B, TTC8, UMOD, WDPCP, WDR19, WDR73, WT1, XPNPEP3, XPO5, ZNF423
○ 04308	Invitae Renal Tubular Disorders Panel	39	ACE, AGT, AGTR1, AQP2, ATP6V0A4, ATP6V1B1, AVPR2, BSND, CA2, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CUL3, EGF, FOXI1, FXYD2, GATM, GNA11, HSD11B2, KCNJ1, KCNJ10, KLHL3, MAGED2, NR3C2, OCRL, REN, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3, SLC34A1, SLC4A1, SLC4A4, TRPM6, WNK1, WNK4

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES

○ A2ML1	○ ADAMTS10	○ AHR	○ ANOS1	○ ASRGL1	○ BBS2	○ BMPR1B	○ CANT1	○ CCT2
○ ABCA4	○ ADAMTS18	○ AIPL1	○ AR	○ ATF6	○ BBS4	○ BRAF	○ CAPN5	○ CDC45
○ ABCC6	○ ADAMTSL4	○ AKT1	○ ARHGEF18	○ ATOH7	○ BBS5	○ C10orf11	○ CBL	○ CDC6
○ ABHD12	○ ADGRA3	○ AKT2	○ ARL13B	○ ATP13A2	○ BBS7	○ C12orf65	○ CC2D2A	○ CDH23
○ ACBD5	○ ADGRV1	○ AKT3	○ ARL2BP	○ B3GALT6	○ BBS9	○ C19orf12	○ CCDC103	○ CDH3
○ ACO2	○ ADIPOR1	○ ALDH1A3	○ ARL3	○ B3GAT3	○ BCOR	○ C1QTNF5	○ CCDC114	○ CDHR1
○ ACP5	○ AFF4	○ ALDH7A1	○ ARL6	○ B9D1	○ BEST1	○ C8orf37	○ CCDC151	○ CDKN1C
○ ACTB	○ AGBL5	○ ALMS1	○ ARMC4	○ B9D2	○ BFSP1	○ C9orf72	○ CCDC39	○ CDON
○ ACTC1	○ AGK	○ ALPL	○ ARMC9	○ BBIP1	○ BFSP2	○ CA4	○ CCDC40	○ CDT1
○ ACTG1	○ AGPAT2	○ ANKH	○ ARSB	○ BBS1	○ BGN	○ CABP4	○ CCDC65	○ CEP104
○ ACVR2B	○ AGPS	○ ANKRD11	○ ARSE	○ BBS10	○ BMP2	○ CACNA1F	○ CCM2	○ CEP120
○ ADAM9	○ AHI1	○ ANKS6	○ ARSG	○ BBS12	○ BMP4	○ CACNA2D4	○ CCNO	○ CEP164

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES (continued)

○ CEP19	○ COL9A2	○ DIS3L2	○ EXT1	○ GLB1	○ IDH3B	○ LCA5	○ MMP9	○ NSDHL
○ CEP250	○ COL9A3	○ DLL3	○ EXT2	○ GLI2	○ IDS	○ LEFTY2	○ MPDZ	○ NUS1
○ CEP290	○ COMP	○ DNAAF1	○ EYA1	○ GLI3	○ IDUA	○ LEMD3	○ MRE11	○ NYX
○ CEP41	○ COQ4	○ DNAAF2	○ EYS	○ GLIS2	○ IFT122	○ LFNG	○ MSX1	○ OAT
○ CEP78	○ CP	○ DNAAF3	○ EZH2	○ GMNN	○ IFT140	○ LIFR	○ MSX2	○ OBSL1
○ CEP83	○ CPLANE1	○ DNAAF4	○ FA2H	○ GNAT1	○ IFT172	○ LIM2	○ MTPAP	○ OCA2
○ CERKL	○ CRB1	○ DNAAF5	○ FAM126A	○ GNAT2	○ IFT27	○ LMX1B	○ MTTP	○ OCRL
○ CFAP298	○ CREBBP	○ DNAH1	○ FAM161A	○ GNB3	○ IFT43	○ LRAT	○ MYH6	○ OFD1
○ CFAP410	○ CRELD1	○ DNAH11	○ FBLN5	○ GNPAT	○ IFT74	○ LRIT3	○ MYO7A	○ OPA1
○ CFAP52	○ CRTAP	○ DNAH5	○ FBN1	○ GNPTAB	○ IFT80	○ LRP2	○ NAGLU	○ OPA3
○ CFAP53	○ CRX	○ DNAH8	○ FGFR1	○ GNPTG	○ IFT81	○ LRP5	○ NBAS	○ OPN1SW
○ CFTR	○ CRYAA	○ DNAI1	○ FGFR2	○ GNS	○ IFT88	○ LRRC6	○ NDP	○ OR2W3
○ CHD7	○ CRYAB	○ DNAI2	○ FGFR3	○ GORAB	○ IHH	○ LTBP3	○ NEK1	○ ORC1
○ CHM	○ CRYBA1	○ DNAJC17	○ FLNB	○ GPC3	○ IMPAD1	○ LYST	○ NEK2	○ ORC4
○ CHMP4B	○ CRYBA4	○ DNAL1	○ FLVCR1	○ GPC6	○ IMPDH1	○ LZTFL1	○ NEK8	○ ORC6
○ CHRNA3	○ CRYBB1	○ DNMT3A	○ FN1	○ GPR143	○ IMPG1	○ MAB21L2	○ NEUROD1	○ OTX2
○ CHST14	○ CRYBB2	○ DRAM2	○ FOXC1	○ GPR179	○ IMPG2	○ MAF	○ NF1	○ P3H1
○ CHST3	○ CRYBB3	○ DRC1	○ FOXC2	○ GPR45	○ INPP5E	○ MAK	○ NF2	○ P3H2
○ CIB2	○ CRYGB	○ DSCAML1	○ FOXE3	○ GRHL3	○ INPPL1	○ MAP2K1	○ NFIX	○ PANK2
○ CISD2	○ CRYGC	○ DTHD1	○ FOXH1	○ GRM6	○ INVS	○ MAP2K2	○ NFKBIA	○ PAPSS2
○ CLCC1	○ CRYGD	○ DVL1	○ FOXL2	○ GRN	○ IQCB1	○ MAP3K1	○ NHS	○ PAX2
○ CLN2 (TPP1)	○ CRYGS	○ DVL3	○ FRMD7	○ GUCA1A	○ IRF6	○ MAP3K7	○ NIPBL	○ PAX6
○ CLN3	○ CSPP1	○ DYM	○ FSCN2	○ GUCA1B	○ ITM2B	○ MAPKAPK3	○ NKX2-5	○ PAX9
○ CLN5	○ CTDP1	○ DYNC1H1	○ FTL	○ GUCY2D	○ ITSN2	○ MASP1	○ NKX2-6	○ PCARE
○ CLN6	○ CTNNA1	○ DYNC2H1	○ FUCA1	○ GUSB	○ JAG1	○ MATN3	○ NKX3-2	○ PCDH15
○ CLN8	○ CTRC	○ EBP	○ FYCO1	○ HAND1	○ KAT6B	○ MCIDAS	○ NME8	○ PCYT1A
○ CLRN1	○ CTSD	○ EDA	○ FZD4	○ HARS	○ KCNJ13	○ MED12	○ NMNAT1	○ PDCD10
○ CLUAP1	○ CUL4B	○ EDAR	○ GABRA6	○ HDAC8	○ KCNV2	○ MED13L	○ NODAL	○ PDE6A
○ CNGA1	○ CUL7	○ EDARADD	○ GALK1	○ HES7	○ KDM6A	○ MEGF8	○ NOTCH1	○ PDE6B
○ CNGA3	○ CWC27	○ EED	○ GALNS	○ HESX1	○ KIAA0586	○ MEIS2	○ NOTCH2	○ PDE6C
○ CNGB1	○ CYP11B2	○ EFEMP1	○ GAS8	○ HGSNAT	○ KIAA1549	○ MERTK	○ NPHP1	○ PDE6D
○ CNGB3	○ CYP1B1	○ EIF2AK3	○ GATA4	○ HK1	○ KIF11	○ MESP2	○ NPHP3	○ PDE6G
○ CNNM4	○ CYP4V2	○ ELN	○ GATA6	○ HMCN1	○ KIF1A	○ MFN2	○ NPHP4	○ PDE6H
○ COASY	○ DCAF17	○ ELOVL4	○ GCNT2	○ HMX1	○ KIF22	○ MFRP	○ NPR2	○ PDGFRB
○ COL10A1	○ DCDC2	○ EMC1	○ GDF1	○ HPRT1	○ KIF7	○ MFSD8	○ NR0B1	○ PDZD7
○ COL11A1	○ DDR2	○ EP300	○ GDF3	○ HRAS	○ KIZ	○ MGP	○ NR2E3	○ PEX1
○ COL11A2	○ DHCR7	○ EPHA2	○ GDF5	○ HSF4	○ KLHL7	○ MIP	○ NR2F1	○ PEX10
○ COL18A1	○ DHDDS	○ ERCC6	○ GDF6	○ HSPG2	○ KMT2D	○ MIR204	○ NR2F2	○ PEX11B
○ COL1A1	○ DHH	○ ERF	○ GJA1	○ HTT	○ KPTN	○ MKKS	○ NR5A1	○ PEX12
○ COL1A2	○ DHX32	○ EVC	○ GJA3	○ HYAL1	○ KRAS	○ MKS1	○ NRAS	○ PEX13
○ COL2A1	○ DHX38	○ EVC2	○ GJA8	○ ICK	○ KRIT1	○ MMP13	○ NRL	○ PEX14
○ COL9A1	○ DICER1	○ EXOSC2	○ GJB6	○ IDH3A	○ LBR	○ MMP2	○ NSD1	○ PEX16

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES (continued)

○ PEX19	○ PPT1	○ RASA1	○ ROM1	○ SAMD11	○ SNRNP200	○ TEAD1	○ TTLL5	○ WDR60
○ PEX2	○ PQBP1	○ RAX	○ ROR2	○ SCLT1	○ SOS1	○ TGIF1	○ TTPA	○ WFS1
○ PEX26	○ PRCD	○ RAX2	○ RP1	○ SDCCAG8	○ SOS2	○ TIMM8A	○ TUB	○ WHRN
○ PEX3	○ PRDM13	○ RB1	○ RP1L1	○ SEMA3E	○ SOX2	○ TIMP3	○ TUBGCP4	○ WISP3
○ PEX5	○ PROKR2	○ RBM8A	○ RP2	○ SEMA4A	○ SOX9	○ TMED7	○ TUBGCP6	○ WNT10A
○ PEX6	○ PROM1	○ RBP1	○ RP9	○ SGSH	○ SPAG1	○ TMEM107	○ TULP1	○ WNT5A
○ PEX7	○ PRPF3	○ RBP3	○ RPE65	○ SH3PXD2B	○ SPATA7	○ TMEM126A	○ TWIST1	○ WT1
○ PHF6	○ PRPF31	○ RBP4	○ RPGR	○ SHH	○ SPINK1	○ TMEM138	○ TYR	○ XPNPEP3
○ PHYH	○ PRPF4	○ RCBTB1	○ RPGR (ORF15)	○ SHOC2	○ SPP2	○ TMEM216	○ TYRP1	○ XYLT1
○ PIK3CA	○ PRPF6	○ RD3	○ RPGRIP1	○ SIL1	○ SPRED1	○ TMEM231	○ UNC119	○ YRDC
○ PIK3R2	○ PRPF8	○ RDH11	○ RPGRIP1L	○ SIX1	○ SQSTM1	○ TMEM237	○ UPF3B	○ ZFPM2
○ PITPNM3	○ PRPH2	○ RDH12	○ RPS6KA3	○ SIX3	○ SRD5A2	○ TMEM67	○ USH1C	○ ZIC2
○ PITX2	○ PRPS1	○ RDH5	○ RRAS	○ SIX6	○ SRY	○ TOPORS	○ USH1G	○ ZIC3
○ PITX3	○ PRSS1	○ REEP6	○ RS1	○ SLC24A1	○ STRA6	○ TP63	○ USH2A	○ ZMYND10
○ PKD2	○ PRSS56	○ RGR	○ RSPH1	○ SLC24A5	○ TBCE	○ TRAF3IP1	○ VAX1	○ ZNF408
○ PKHD1	○ PTCH1	○ RGS9	○ RSPH3	○ SLC26A2	○ TBX1	○ TREX1	○ VCAN	○ ZNF423
○ PLA2G5	○ PTH1R	○ RGS9BP	○ RSPH4A	○ SLC36A2	○ TBX3	○ TRIM32	○ VHL	○ ZNF513
○ PLA2G6	○ PTPN11	○ RHO	○ RSPH9	○ SLC39A13	○ TBX5	○ TRNT1	○ VIM	
○ PLK4	○ PXDN	○ RIMS1	○ RSPRY1	○ SLC45A2	○ TBX6	○ TRPM1	○ VPS13B	
○ PNPLA6	○ RAB23	○ RIPPLY2	○ RTN4IP1	○ SLC7A14	○ TCOF1	○ TRPS1	○ VSX2	
○ POC1B	○ RAB28	○ RIT1	○ RUNX2	○ SMAD4	○ TCTN1	○ TRPV4	○ WDPCP	
○ POC5	○ RAD21	○ RLBP1	○ SAG	○ SMAD6	○ TCTN2	○ TSPAN12	○ WDR19	
○ POMGNT1	○ RAF1	○ RMRP	○ SALL1	○ SMARCAL1	○ TCTN3	○ TTC21B	○ WDR34	
○ POR	○ RARB	○ RNF125	○ SALL4	○ SMC3	○ TDRD7	○ TTC8	○ WDR35	

CLINICAL AREA: EPILEPSY

EPILEPSY INDIVIDUAL GENES

○ AARS	○ AP2M1	○ ATP6AP2	○ CCDC88A	○ CLN6	○ DDC	○ DYRK1A	○ FOLR1	○ GATM
○ ABAT	○ AP3B2	○ ATRX	○ CDKL5	○ CLN8	○ DDX3X	○ ECHS1	○ FOXG1	○ GCH1
○ ADAR	○ ARG1	○ BRAT1	○ CERS1	○ CLTC	○ DEAF1	○ EEF1A2	○ FRRS1L	○ GLDC
○ ADSL	○ ARHGEF15	○ C12orf57	○ CHD2	○ CNTN2	○ DEPDC5	○ EHMT1	○ GABBR2	○ GLRA1
○ ALDH5A1	○ ARHGEF9	○ CACNA1A	○ CHRNA2	○ CNTNAP2	○ DHDDS	○ EMC1	○ GABRA1	○ GLRB
○ ALDH7A1	○ ARSA	○ CACNA1E	○ CHRNA4	○ COG5	○ DHFR	○ EPM2A	○ GABRB1	○ GNAO1
○ ALG1	○ ARX	○ CACNA1H	○ CHRNB2	○ COL18A1	○ DIAPH1	○ FAR1	○ GABRB2	○ GNB1
○ ALG12	○ ASAH1	○ CACNA2D2	○ CLCN4	○ CSTB	○ DNAJC5	○ FARS2	○ GABRB3	○ GOSR2
○ ALG13	○ ASNS	○ CAD	○ CLCN6	○ CTNNB1	○ DNM1	○ FASN	○ GABRD	○ GPAA1
○ ALG6	○ ATAD1	○ CAMK2B	○ CLN2 (TPP1)	○ CTSD	○ DNM1L	○ FBXO11	○ GABRG2	○ GPHN
○ AMACR	○ ATP1A2	○ CARS2	○ CLN3	○ CYFIP2	○ DOCK7	○ FGF12	○ GAMT	○ GRIA3
○ AMT	○ ATP1A3	○ CASK	○ CLN5	○ CYP27A1	○ DYNC1H1	○ FLNA	○ GATAD2B	○ GRIN1

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: EPILEPSY

EPILEPSY INDIVIDUAL GENES (continued)

○ GRIN2A	○ KCNH5	○ MOCS2	○ PEX13	○ PNPO	○ RAI1	○ SCN8A	○ SNX27	○ TPK1
○ GRIN2B	○ KCNJ10	○ MOCS3	○ PEX14	○ PNPT1	○ RALA	○ SCN9A	○ SPATA5	○ TREX1
○ GRIN2D	○ KCNK4	○ MTOR	○ PEX16	○ POLG	○ RANBP2	○ SCP2	○ SPTAN1	○ TSC1
○ GTPBP3	○ KCNMA1	○ NACC1	○ PEX19	○ PPP2CA	○ RBFOX1	○ SERPINI1	○ ST3GAL3	○ TSC2
○ GUF1	○ KCNQ2	○ NAGLU	○ PEX2	○ PPP2R1A	○ RBFOX3	○ SETBP1	○ ST3GAL5	○ TSFM
○ HCN1	○ KCNQ3	○ NECAP1	○ PEX26	○ PPP2R5D	○ RELN	○ SGCE	○ STAG2	○ TUBA8
○ HDAC8	○ KCNQ5	○ NEDD4L	○ PEX3	○ PPP3CA	○ RFT1	○ SGSH	○ STRADA	○ TUBB2A
○ HEXA	○ KCNT1	○ NEXMIF	○ PEX5	○ PPT1	○ RHOBTB2	○ SIK1	○ STX1B	○ UBA5
○ HNRNPU	○ KCTD7	○ NGLY1	○ PEX6	○ PRDM8	○ RNASEH2A	○ SLC12A5	○ STXBP1	○ UBE3A
○ IDH3A	○ KIF1A	○ NHLRC1	○ PHGDH	○ PRICKLE1	○ RNASEH2B	○ SLC13A5	○ STXBP2	○ UNC80
○ IER3IP1	○ KIF2A	○ NPC1	○ PIGA	○ PRICKLE2	○ RNASEH2C	○ SLC19A3	○ SUMF1	○ WDR45
○ IFIH1	○ KIF5A	○ NPC2	○ PIGG	○ PRIMA1	○ RNF13	○ SLC1A2	○ SUOX	○ WWOX
○ IQSEC2	○ KPNA7	○ NPRL3	○ PIGN	○ PRRT2	○ ROGDI	○ SLC25A12	○ SYN1	○ YWHAG
○ ITPA	○ LAMC3	○ NRXN1	○ PIGO	○ PSAP	○ RORB	○ SLC25A22	○ SYNGAP1	○ ZDHHC9
○ JMJD1C	○ LGI1	○ NTRK2	○ PIGP	○ PSAT1	○ RUSC2	○ SLC2A1	○ SYNJ1	○ ZEB2
○ KANSL1	○ LIAS	○ NUS1	○ PIGQ	○ PSPH	○ SAMHD1	○ SLC35A2	○ SZT2	○ ZSWIM6
○ KCNA1	○ LMNB2	○ PACS1	○ PIGV	○ PTEN	○ SATB2	○ SLC6A1	○ TANGO2	
○ KCNA2	○ MBD5	○ PACS2	○ PIGW	○ PTPN23	○ SCARB2	○ SLC6A5	○ TBC1D24	
○ KCNB1	○ MDH2	○ PAFAH1B1	○ PIK3AP1	○ PURA	○ SCN1A	○ SLC6A8	○ TBCK	
○ KCNC1	○ MECP2	○ PCDH19	○ PLAA	○ QARS	○ SCN1B	○ SLC9A6	○ TBL1XR1	
○ KCND2	○ MEF2C	○ PCLO	○ PLCB1	○ QDPR	○ SCN2A	○ SMC1A	○ TCF4	
○ KCNH1	○ MFSD8	○ PEX10	○ PNKD	○ RAB11A	○ SCN3A	○ SNAP25	○ TH	
○ KCNH2	○ MOCS1	○ PEX12	○ PNKP	○ RAB11B	○ SCN5A	○ SNIP1	○ TK2	

CLINICAL AREA: HEREDITARY CANCER

Test code	Test name	# gene(s)	Gene list
Chronic Pancreatitis			
○ 01745	Invitae Chronic Pancreatitis Panel	6	CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1
Pediatric Oncology			
○ 01104	Invitae Pediatric Solid Tumors Panel	54	AIP, ALK, APC, AXIN2, BAP1, BLM, BMPR1A, CDC73, CDKN1C, DICER1, DIS3L2, EPCAM, EXT1, EXT2, FH, GPC3, HRAS, LZTR1, MAX, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PHOX2B, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, REST, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1

HEREDITARY CANCER INDIVIDUAL GENES

○ AKT1	○ ATR	○ BLM	○ BRIP1	○ CDH1	○ CDKN2A	○ CHEK2	○ DIS3L2	○ ELANE
○ ALK	○ AXIN2	○ BMPR1A	○ BUB1B	○ CDK4	○ CEBPA	○ CTNNA1	○ DKC1	○ ENG
○ APC	○ BAP1	○ BRCA1	○ CASR	○ CDKN1B	○ CEP57	○ CTRC	○ EGFR	○ EPCAM
○ ATM	○ BARD1	○ BRCA2	○ CDC73	○ CDKN1C	○ CFTR	○ DICER1	○ EGLN1	○ ERCC4

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: HEREDITARY CANCER

HEREDITARY CANCER INDIVIDUAL GENES (continued)								
○ EZH2	○ GALNT12	○ MITF	○ PALLD	○ RAD51D	○ RUNX1	○ SUFU	○ LZTR1	○ CTR9
○ ABRAXAS1	○ GATA1	○ MLH1	○ PDGFRA	○ RB1	○ SDHA	○ TERC	○ POT1	○ ERBB2
○ FANCA	○ GATA2	○ MLH3	○ PHOX2B	○ RECQL4	○ SDHAF2	○ TERT	○ EXT1	○ GEN1
○ FANCB	○ GPC3	○ MPL	○ PIK3CA	○ RET	○ SDHB	○ TINF2	○ EXT2	○ GNA11
○ FANCC	○ GREM1	○ MRE11	○ PMS2	○ RINT1	○ SDHC	○ TMEM127	○ ACD	○ RECQL
○ FANCD2	○ HOXB13	○ MSH2	○ POLD1	○ RPL11	○ SDHD	○ TP53	○ PARN	○ REST
○ FANCE	○ HRAS	○ MSH6	○ POLE	○ RPL26	○ SLX4	○ TSC1	○ RTEL1	○ RNF43
○ FANCF	○ KIF1B	○ MUTYH	○ PRKAR1A	○ RPL35A	○ SMAD4	○ TSC2	○ USB1	○ RPL15
○ FANCG	○ KIT	○ NBN	○ PRSS1	○ RPL5	○ SMARCA4	○ VHL	○ MSH3	○ RPL19
○ FANCI	○ MAX	○ NF1	○ PTCH1	○ RPS10	○ SMARCB1	○ WAS	○ NTHL1	○ RPS29
○ FANCL	○ MC1R	○ NF2	○ PTCH2	○ RPS19	○ SMARCE1	○ WRN	○ RPS20	○ WRAP53
○ FANCM	○ MDM2	○ NHP2	○ PTEN	○ RPS24	○ SPINK1	○ WT1	○ AIP	
○ FH	○ MEN1	○ NOP10	○ RAD50	○ RPS26	○ SPRED1	○ XRCC2	○ AP2S1	
○ FLCN	○ MET	○ PALB2	○ RAD51C	○ RPS7	○ STK11	○ CTC1	○ CPA1	

METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Metabolic Disorders Newborn Screening Confirmation			
○ 06102	Invitae Metabolic Newborn Screening Confirmation Panel	160	AAAS, ABCD1, ABCD3, ABCD4, ACAD8, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD5, ACOX1, ACOX2, ACSF3, ADAR, ADK, AHCY, ALDH4A1, ALDH6A1, ALDH7A1, AMACR, AMT, ARG1, ARSA, ASL, ASPA, ASS1, AUH, BCAP31, BCAT2, BCKDHA, BCKDHB, BTD, CBS, CD320, CFTR, CLPB, CPS1, CPT1A, CPT2, DBT, DLD, DNAJC12, DNAJC19, DNM1L, ECHS1, ETFA, ETFB, ETFDH, ETHE1, FAH, FLAD1, FTCD, G6PD, GAA, GALC, GALE, GALK1, GALM, GALT, GAMT, GATM, GCDH, GCGR, GCH1, GLA, GLDC, GNMT, GSS, HADH, HADHA, HADHB, HCFC1, HIBCH, HLCS, HMGCL, HPD, HSD17B10, HSD17B4, HTRA2, IDS, IDUA, IFIH1, IVD, LMBRD1, MAT1A, MCCC1, MCCC2, MCEE, MFF, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, NADK2, NAGS, NR0B1, OAT, OPA3, OTC, PAH, PC, PCBD1, PCCA, PCCB, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGM1, PNPO, PPM1K, PRDX1, PRODH, PROSC, PTS, QDPR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCP2, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A32, SLC2A1, SLC52A1, SLC52A2, SLC52A3, SLC6A8, SMPD1, SPR, SUCLA2, SUCLG1, TAT, TAZ, TCN2, THAP11, TIMM50, TMEM70, TREX1, ZNF143
○ 98003	Invitae Supplemental Metabolic Newborn Screening Panel	192	A4GALT, ACAD9, ALDH18A1, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ARCN1, ATP13A2, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V1A, ATP6V1E1, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, BOLA3, C1GALT1C1, CA5A, CAD, CANT1, CCDC115, CHST14, CHST3, CHST6, CHSY1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COPA, COPB2, CSGALNACT1, CTSD, CYP27A1, D2HGDH, DDOST, DHCR7, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, DSE, EOGT, EXT1, EXT2, EXTL3, FKRP, FKTN, FUK, FUT8, G6PC3, GALNT3, GANAB, GFPT1, GLRX5, GLUL, GM2A, GMPPA, GMPPB, GNE, GNPTAB, GNS, GORAB, GOSR2, GPAA1, HEXA, HEXB, HGSNAT, HMGCS2, IBA57, ISCA2, ISPD, JAGN1, KCTD7, LARGE1, LFNG, LIAS, LIPT1, LIPT2, MAGT1, MAN1B1, MFSD8, MGAT2, MOGS, MPDU1, MPI, NAGLU, NANS, NFU1, NGLY1, NPC1, NPC2, NUS1, OGT, OXCT1, PAPSS2, PGAP1, PGAP2, PGAP3, PGM3, PIGA, PIGB, PIGC, PIGG, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGT, PIGU, PIGV, PIGW, PIGY, PMM2, POFUT1, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPT1, PRKCSH, RFT1, RPN2, RXYLT1, SAR1B, SEC23A, SEC23B, SEC24D, SEC63, SGSH, SLC10A7, SLC16A1, SLC26A2, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A8, SLC6A9, SLC7A7, SLC9A7, SRD5A3, SSR3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, TGDS, TMEM165, TMEM199, TRAK1, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC6B, TRAPPC9, TRIP11, TUSC3, VMA21, VPS13B, XYLT1, XYLT2
○ 06171	Invitae Lysosomal Storage Disorders Newborn Screening Panel	10	CHIT1, GAA, GALC, GBA, GLA, IDS, IDUA, NPC1, NPC2, SMPD1
○ 06210	Invitae Elevated Very Long Chain Fatty Acids Panel (including X-ALD)	40	AAAS, ABCD1, ABCD3, ACBD5, ACOX1, ACOX2, ADAR, AMACR, ARSA, BCAP31, CLN2 (TPP1), CLN3, DNM1L, GALC, HSD17B4, IFIH1, MFF, NR0B1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PPT1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCP2, TREX1
Panels by Analyte			
○ 06145	Invitae Elevated Phenylalanine (Hyperphenylalaninemia) Panel	8	DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR, SLC25A13, SPR
○ 06146	Invitae Elevated Proline (Hyperprolinemia) Panel	2	ALDH4A1, PRODH
○ 06166	Invitae Elevated C6, C8 and C10 (MCAD deficiency) Test	1	ACADM
○ 06195	Invitae Elevated C5-DC (Glutaric Aciduria Type I) Test	1	GCDH
○ 06197	Invitae Elevated C4 and C5 (Multiple Acyl-CoA Dehydrogenase deficiency) Panel	9	ETFA, ETFB, ETFDH, ETHE1, FLAD1, SLC25A32, SLC52A1, SLC52A2, SLC52A3
○ 06210	Invitae Elevated Very Long Chain Fatty Acids Panel (including X-ALD)	40	AAAS, ABCD1, ABCD3, ACBD5, ACOX1, ACOX2, ADAR, AMACR, ARSA, BCAP31, CLN2 (TPP1), CLN3, DNM1L, GALC, HSD17B4, IFIH1, MFF, NR0B1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PPT1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCP2, TREX1

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METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Panels by Analyte (continued)			
○ 06103	Invitae Low C0 Test	1	SLC22A5
○ 06104	Invitae Elevated C0/(C16+C18) Test	1	CPT1A
○ 06105	Invitae Elevated C3 Panel	20	ABCD4, ACSF3, ALDH6A1, BTD, CD320, HCFC1, HLCS, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MUT, PCCA, PCCB, PRDX1, TCN2, THAP11, ZNF143
○ 06106	Invitae Elevated C3-DC Panel	2	ACSF3, MLYCD
○ 06107	Invitae Elevated C4 Panel	4	ACAD8, ACADS, ETHE1, FTCD
○ 06127	Invitae Elevated C4-DC Panel	2	SUCLA2, SUCLG1
○ 06108	Invitae Elevated C4-OH Panel	2	HADH, HIBCH
○ 06110	Invitae Elevated C5 Panel	2	ACADSB, IVD
○ 06112	Invitae Elevated C5-OH Panel	16	ACAT1, AUH, BTD, CLPB, DNAJC19, HLCS, HMGCL, HSD17B10, HTRA2, MCCC1, MCCC2, OPA3, SERAC1, TAZ, TIMM50, TMEM70
○ 06114	Invitae Elevated C14 and C14:1 (VLCAD deficiency) Test	1	ACADVL
○ 06115	Invitae Elevated C16-OH, C16:1-OH, C18-OH and C18:1-OH Panel	2	HADHA, HADHB
○ 06116	Invitae Elevated C16, C16:1, C18, and C18:1 Panel	2	CPT2, SLC25A20
○ 06117	Invitae Elevated Arginine (Arginase deficiency) Panel	2	ARG1, GCGR
○ 06118	Invitae Elevated Citrulline (Citrullinemia) Panel	5	ASL, ASS1, DLD, PC, SLC25A13
○ 06123	Invitae Low Citrulline Panel	4	ALDH18A1, CPS1, NAGS, OTC
○ 06124	Invitae Elevated Glycine Panel (including Glycine Encephalopathy)	12	AMT, BOLA3, GLDC, GLRX5, IBA57, ISCA2, LIAS, LIPT1, LIPT2, NFU1, PNPO, SLC6A9
○ 06119	Invitae Elevated Leucine (MSUD) Panel	6	BCAT2, BCKDHA, BCKDHB, DBT, DLD, PPM1K
○ 06125	Invitae Elevated Methionine Panel	7	ADK, AHCY, CBS, FAH, GNMT, MAT1A, SLC25A13
○ 06122	Invitae Elevated Succinylacetone Test	1	FAH
○ 06126	Invitae Elevated Tyrosine (Tyrosinemia) Panel	3	FAH, HPD, TAT
Aminoacidopathies			
○ 06117	Invitae Elevated Arginine (Arginase deficiency) Panel	2	ARG1, GCGR
○ 06118	Invitae Elevated Citrulline (Citrullinemia) Panel	5	ASL, ASS1, DLD, PC, SLC25A13
○ 06119	Invitae Elevated Leucine (MSUD) Panel	6	BCAT2, BCKDHA, BCKDHB, DBT, DLD, PPM1K
○ 06124	Invitae Elevated Glycine Panel (including Glycine Encephalopathy)	12	AMT, BOLA3, GLDC, GLRX5, IBA57, ISCA2, LIAS, LIPT1, LIPT2, NFU1, PNPO, SLC6A9
○ 06126	Invitae Elevated Tyrosine (Tyrosinemia) Panel	3	FAH, HPD, TAT
○ 06140	Invitae Alkaptonuria Test	1	HGD
○ 06141	Invitae Methylmalonic Acidemia and Homocystinuria Panel	31	ABCD4, ACSF3, ADK, AHCY, ALDH6A1, AMN, CBS, CD320, CUBN, GIF, GNMT, HCFC1, LMBRD1, MAT1A, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTHFD1, MTHFR, MTR, MTRR, MUT, PRDX1, SUCLA2, SUCLG1, TCN1, TCN2, THAP11, ZNF143
○ 06142	Invitae Cystinuria Panel	3	PREPL, SLC3A1, SLC7A9
○ 06145	Invitae Elevated Phenylalanine (Hyperphenylalaninemia) Panel	8	DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR, SLC25A13, SPR
○ 06146	Invitae Elevated Proline (Hyperprolinemia) Panel	2	ALDH4A1, PRODH

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METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Carbohydrate Disorders			
○ 98006	Invitae Hypoglycemia Panel	120	AAAS, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACSF3, ADK, AGL, AKT2, ALDOA, ALDOB, ALG3, ALG6, BCKDHA, BCKDHB, CA5A, CACNA1C, CACNA1D, CDKN1C, COG7, CPT1A, CPT2, CYP7B1, DBH, DBT, DDC, DGUOK, DLD, DMXL2, DOLK, ENO3, ETFA, ETFB, ETFDH, FAH, FBP1, FLAD1, G6PC, GAA, GALE, GALK1, GALT, GBE1, GCK, GH1, GHR, GLUD1, GPC3, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HESX1, HK1, HMGCL, HMGCS2, HNF1A, HNF4A, HRAS, HSD3B7, INSR, KCNJ11, KDM6A, KMT2D, LAMP2, LDHA, LHX3, MLYCD, MPI, MPV17, NADK2, NNT, NR0B1, NR3C1, NSD1, OPLAH, OTX2, OXCT1, PC, PCK1, PCK2, PCSK1, PDX1, PFKM, PGAM2, PGM1, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PMM2, POMC, PROP1, PTF1A, PYGL, PYGM, RBCK1, SERAC1, SLC16A1, SLC22A5, SLC25A20, SLC25A32, SLC2A2, SLC37A4, SLC52A1, SLC52A2, SLC52A3, SOX2, SOX3, TAZ, TBX19, TRMT10A, UCP2
○ 06152	Invitae Galactosemia Panel	7	GALE, GALK1, GALM, GALT, PGM1, SLC25A13, SLC2A2
○ 06153	Invitae Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency Test	1	G6PD
○ 06156	Invitae Comprehensive Glycogen Storage Disease Panel	29	AGL, ALDOA, ENO3, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, LPIN1, PCK1, PCK2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, POLG, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4
○ 06159	Invitae Hereditary Fructose Intolerance Test	1	ALDOB
○ 06160	Invitae Rare Carbohydrate Disorders Panel	2	FBP1, SLC5A1
Cerebrotendinous Xanthomatosis			
○ 06161	Invitae Cerebrotendinous Xanthomatosis Test	1	CYP27A1
	○ 06161.1 Add-on sitosterolemia genes	2	ABCG5, ABCG8
Congenital Disorders of Glycosylation			
○ 06155	Invitae Congenital Disorders of Glycosylation Panel	154	A4GALT, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ARCN1, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V1A, ATP6V1E1, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, C1GALT1C1, CAD, CANT1, CCDC115, CHST14, CHST3, CHST6, CHSY1, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COPA, COPB2, CSGALNACT1, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, DSE, EOGT, EXT1, EXT2, EXTL3, FKRP, FKTN, FUK, FUT8, G6PC3, GALNT3, GANAB, GFPT1, GMPPA, GMPPB, GNE, GNPTAB, GORAB, GOSR2, GPAA1, ISPD, JAGN1, LARGE1, LFNG, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NANS, NGLY1, NUS1, OGT, PAPSS2, PGAP1, PGAP2, PGAP3, PGM1, PGM3, PIGA, PIGB, PIGC, PIGG, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGT, PIGU, PIGV, PIGW, PIGY, PMM2, POFUT1, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRKCSH, RFT1, RPN2, RXYLT1, SAR1B, SEC23A, SEC23B, SEC24D, SEC63, SLC10A7, SLC26A2, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A8, SLC9A7, SRD5A3, SSR3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, TGDS, TMEM165, TMEM199, TRAK1, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC6B, TRAPPC9, TRIP11, TUSC3, VMA21, VPS13B, XYLT1, XYLT2
Creatine Biosynthesis Disorders			
○ 06162	Invitae Cerebral Creatine Deficiency Panel	3	GAMT, GATM, SLC6A8
Cystic Fibrosis			
○ 04714	Invitae Cystic Fibrosis Test	1	CFTR
Fatty Acid Oxidation Defects			
○ 06114	Invitae Elevated C14 and C14:1 (VLCAD deficiency) Test	1	ACADVL
○ 06165	Invitae Fatty Acid Oxidation Defects Panel	25	ACAD9, ACADM, ACADS, ACADSB, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, FLAD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, NADK2, SLC22A5, SLC25A20, SLC25A32, SLC52A1, SLC52A2, SLC52A3, TANGO2

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CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Fatty Acid Oxidation Defects (continued)			
○ 06169	Invitae Ketolysis Disorders Panel	3	ACAT1, OXCT1, SLC16A1
○ 06166	Invitae Elevated C6, C8 and C10 (MCAD deficiency) Test	1	ACADM
○ 06197	Invitae Elevated C4 and C5 (Multiple Acyl-CoA Dehydrogenase deficiency) Panel	9	ETFA, ETFB, ETFDH, ETHE1, FLAD1, SLC25A32, SLC52A1, SLC52A2, SLC52A3
Lysosomal Storage Disorders			
○ 06170	Invitae Comprehensive Lysosomal Storage Disorders Panel	58	AGA, ARSA, ARSB, ASAH1, ASPA, ATP13A2, CHIT1, CLCN7, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSD, CTSK, FUCA1, GAA, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNE, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, KCTD7, LAMP2, LIPA, LYST, MAN2B1, MANBA, MCOLN1, MFSD8, MMP14, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SCARB2, SGSH, SLC17A5, SMPD1, SUMF1, VPS33A
	○ 06170.3 Add-on Adult-onset Neuronal Ceroid Lipofuscinosis Genes	3	CTSF, DNAJC5, GRN
○ 06172	Invitae Cystinosis Test	1	CTNS
○ 06179	Invitae Farber Lipogranulomatosis Test	1	ASAH1
○ 02266	Invitae Fabry Disease Test	1	GLA
○ 06000	Invitae Gaucher Common Variants Test	1	GBA
○ 06180	Invitae GM2 Gangliosidosis Panel	3	GM2A, HEXA, HEXB
○ 06173	Invitae Krabbe Disease Test	1	GALC
	○ 06173.1 Add-on prosaposin deficiency gene	1	PSAP
○ 06181	Invitae Lysosomal Acid Lipase Deficiency Test	1	LIPA
○ 06174	Invitae Metachromatic Leukodystrophy Panel	3	ARSA, PSAP, SUMF1
○ 06185	Invitae Mucopolysaccharidoses Plus (MPS+) Panel	27	AGA, ARSB, CTSA, CTSK, FUCA1, GALNS, GLB1, GNE, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MAN2B1, MANBA, MBTPS1, MCOLN1, NAGA, NAGLU, NEU1, SGSH, SLC17A5, SUMF1, VPS33A
○ 03405	Invitae Neuronal Ceroid Lipofuscinoses Panel	10	ATP13A2, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1
	○ 03405.1 Add-on adult-onset neuronal ceroid lipofuscinoses genes	3	CTSF, DNAJC5, GRN
○ 06190	Invitae Niemann-Pick Disease Types A and B Test	1	SMPD1
	○ 06190.1 Add-on chitotriosidase deficiency gene	1	CHIT1
○ 06176	Invitae Niemann-Pick Type C Panel	2	NPC1, NPC2
	○ 06176.1 Add-on lysosomal acid lipase deficiency gene	1	LIPA
	○ 06176.2 Add-on chitotriosidase deficiency gene	1	CHIT1
○ 06177	Invitae Pompe Disease Test	1	GAA
	○ 06177.1 Add-on Danon disease gene	1	LAMP2
	○ 06177.2 Add-on primary carnitine deficiency gene	1	SLC22A5
○ 06178	Invitae Sandhoff Disease Test	1	HEXB
	○ 06178.1 Add-on Tay-Sachs disease gene	1	HEXA
○ 04719	Invitae Tay-Sachs Disease Test	1	HEXA

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METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Metal Transport Disorders			
○ 06202	Invitae Copper Metabolism Disorders Panel	10	AP1S1, ATP6AP1, ATP6AP2, ATP7A, ATP7B, CCDC115, CP, PGM1, SLC33A1, TMEM199
○ 06183	Invitae Wilson Disease Test	1	ATP7B
○ 03406	Invitae Neurodegeneration with Brain Iron Accumulation Panel	20	AP4M1, ATP13A2, C19orf12, COASY, CP, CRAT, DCAF17, FA2H, FTL, FUCA1, GJA1, GTPBP2, KIF1A, PANK2, PLA2G6, REPS1, SCP2, SLC25A42, SQSTM1, WDR45
Neurotransmitter and Neurometabolic Disorders			
○ 06124	Invitae Elevated Glycine Panel (including Glycine Encephalopathy)	12	AMT, BOLA3, GLDC, GLRX5, IBA57, ISCA2, LIAS, LIPT1, LIPT2, NFU1, PNPO, SLC6A9
○ 06222	Invitae Treatable Neurometabolic Disorders Panel	195	ABCD1, ABCD4, ACAT1, AGA, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, AMN, AMT, APTX, ARG1, ARHGEF9, ARSA, ASAH1, ASL, ASNS, ASPA, ASS1, ATAD1, ATP7A, ATP7B, AUH, BCKDHA, BCKDHB, BSCL2, BSND, BTD, CA5A, CAD, CASR, CBS, CD320, CLCNKB, CLDN16, CLDN19, CLN2 (TPP1), CLN3, CLN5, CLN6, CNNM2, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, CP, CPOX, CPS1, CUBN, CYP27A1, DBT, DDC, DHCR7, DHFR, DLAT, DLD, DNAJC12, EGF, ETFA, ETFB, ETFDH, ETHE1, FAM111A, FOLR1, FXYD2, GALC, GAMT, GATM, GCDH, GCH1, GCLC, GIF, GLA, GLB1, GLDC, GLRA1, GLRB, GLUD1, GNS, GOT2, GPHN, GSS, GUSB, HCFC1, HEXA, HEXB, HGSNAT, HLCS, HMBS, HMGCL, HMGCS2, HNF1B, HSD17B10, IDS, IDUA, IVD, KCNA1, KCNJ10, LIPA, LMBRD1, MAN2B1, MCCC1, MCCC2, MCEE, MFSD8, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MSMO1, MTHFR, MTR, MTRR, MUT, NAGLU, NAGS, NPC1, NPC2, NT5C3A, OAT, OTC, OXCT1, PAH, PANK2, PCBD1, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PGM3, PHGDH, PHYH, PNPO, PPM1K, PRDX1, PROSC, PRPS1, PSAT1, PSPH, PTS, QDPR, RAPSN, SCN4A, SGSH, SLC12A1, SLC12A3, SLC13A5, SLC18A2, SLC19A1, SLC19A2, SLC19A3, SLC1A3, SLC25A13, SLC25A15, SLC25A19, SLC2A1, SLC30A10, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SPR, TAT, TCN1, TCN2, TH, TPK1, TRPM6, TTPA
○ 98005	Invitae Comprehensive Neurometabolic Disorders Panel	253	ABAT, ABCD1, ABCD4, ACAT1, ACO2, ADSL, AGA, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, AMN, AMT, AP1S1, AP4M1, APTX, ARG1, ARHGEF9, ARSA, ASAH1, ASL, ASNS, ASPA, ASS1, ATAD1, ATP13A2, ATP6AP1, ATP6AP2, ATP7A, ATP7B, AUH, BCKDHA, BCKDHB, BCKDK, BSCL2, BSND, BTD, C19orf12, CA5A, CAD, CASR, CBS, CD320, CLCNKB, CLDN16, CLDN19, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLPB, CNNM2, COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, CP, CPOX, CPS1, CRAT, CTSD, CUBN, CYP27A1, D2HGDH, DBH, DBT, DCAF17, DDC, DHCR7, DHFR, DLAT, DLD, DNAJC12, EGF, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAM111A, FBXL4, FH, FOLR1, FTL, FUCA1, FXYD2, GAD1, GALC, GAMT, GATM, GCDH, GCH1, GCLC, GIF, GJA1, GLA, GLB1, GLDC, GLRA1, GLRB, GLUD1, GNS, GOT2, GPHN, GSS, GTPBP2, GUSB, HCFC1, HEXA, HEXB, HGSNAT, HLCS, HMBS, HMGCL, HMGCS2, HNF1B, HPRT1, HSD17B10, IDH2, IDS, IDUA, IVD, KCNA1, KCNJ10, KCTD7, KIF1A, L2HGDH, LIPA, LMBRD1, MAN2B1, MAOA, MAT1A, MCCC1, MCCC2, MCEE, MFSD8, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCOS, MOCS1, MOCS2, MPV17, MSMO1, MTHFR, MTR, MTRR, MUT, NAGLU, NAGS, NAXE, NGLY1, NPC1, NPC2, NT5C3A, OAT, OTC, OXCT1, PAH, PANK2, PC, PCBD1, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PGM3, PHGDH, PHYH, PLA2G6, PNP, PNPO, POLG, PPM1K, PPT1, PRDX1, PROSC, PRPS1, PSAT1, PSPH, PTS, QDPR, RAPSN, REPS1, SCN4A, SCP2, SGSH, SLC12A1, SLC12A3, SLC13A5, SLC18A2, SLC19A1, SLC19A2, SLC19A3, SLC1A3, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A42, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SPR, SQSTM1, SUCLA2, SUCLG1, SUOX, TAT, TCN1, TCN2, TH, TPI1, TPK1, TRPM6, TTPA, TWNK, WDR45, XDH
○ 06203	Invitae Neurotransmitter Disorders Panel	45	ABAT, ALDH5A1, ALDH7A1, AMT, ARHGEF9, ASNS, ATAD1, DBH, DDC, DHFR, DNAJC12, GABBR2, GABRA1, GABRA2, GABRB1, GABRB3, GABRG2, GAD1, GCH1, GLDC, GLRA1, GLRB, GOT2, GPHN, GRIN2B, GRIN2D, MAOA, PCBD1, PHGDH, PNPO, PROSC, PSAT1, PSPH, PTS, QDPR, SLC18A2, SLC1A2, SLC1A4, SLC25A22, SLC6A1, SLC6A3, SLC6A5, SLC6A9, SPR, TH

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CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Neurotransmitter and Neurometabolic Disorders (continued)			
○ 06204	Invitae Hereditary Hyperekplexia Panel	13	ARHGEF9, ASNS, ATAD1, CLPB, CTNNB1, GLRA1, GLRB, GPHN, RPS6KA3, SCN8A, SLC6A5, SLC6A9, TRAK1
Organic Acidemias			
○ 06169	Invitae Ketolysis Disorders Panel	3	ACAT1, OXCT1, SLC16A1
○ 06191	Invitae Organic Acidemias Panel	101	ABCD4, ACAD8, ACADSB, ACAT1, ACSF3, ADK, AGK, AHCY, ALDH6A1, AMN, ASPA, ATP5D, AUH, BCKDHA, BCKDHB, BOLA3, BTD, C19orf70, CBS, CD320, CLPB, CPS1, CUBN, D2HGDH, DBT, DHTKD1, DLD, DNAJC19, ECHS1, ETFA, ETFB, ETFDH, ETHE1, FBP1, FH, FLAD1, FTCD, GCDH, GIF, GLRX5, GLYCTK, GNMT, GSS, HCFC1, HIBCH, HLCS, HMGCL, HSD17B10, HTRA2, IBA57, IDH2, ISCA2, IVD, L2HGDH, LIAS, LIPT1, LIPT2, LMBRD1, MAT1A, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, NFU1, OGDH, OPA3, OPLAH, OXCT1, PCCA, PCCB, PCK1, POLG, PPM1K, PRDX1, SERAC1, SLC13A3, SLC13A5, SLC25A1, SLC25A19, SLC25A32, SLC52A1, SLC52A2, SLC52A3, SUCLA2, SUCLG1, SUGCT, TAZ, TCN1, TCN2, THAP11, TIMM50, TMEM70, ZNF143
○ 06192	Invitae 3-Methylcrotonyl CoA Carboxylase Panel	2	MCCC1, MCCC2
○ 06194	Invitae Biotinidase Deficiency Test	1	BTD
○ 06195	Invitae Elevated C5-DC (Glutaric Aciduria Type I) Test	1	GCDH
○ 06141	Invitae Methylmalonic Acidemia and Homocystinuria Panel	31	ABCD4, ACSF3, ADK, AHCY, ALDH6A1, AMN, CBS, CD320, CUBN, GIF, GNMT, HCFC1, LMBRD1, MAT1A, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTHFD1, MTHFR, MTR, MTRR, MUT, PRDX1, SUCLA2, SUCLG1, TCN1, TCN2, THAP11, ZNF143
○ 06197	Invitae Elevated C4 and C5 (Multiple Acyl-CoA Dehydrogenase deficiency) Panel	9	ETFA, ETFB, ETFDH, ETHE1, FLAD1, SLC25A32, SLC52A1, SLC52A2, SLC52A3
○ 06199	Invitae Propionic Acidemia Panel	2	PCCA, PCCB
Peroxisomal Disorders			
○ 06207	Invitae Adult Refsum Disease Panel	2	PEX7, PHYH
○ 06211	Invitae Zellweger Spectrum Disorder Panel	18	ACBD5, ACOX1, AMACR, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7
○ 06210	Invitae Elevated Very Long Chain Fatty Acids Panel (including X-ALD)	40	AAAS, ABCD1, ABCD3, ACBD5, ACOX1, ACOX2, ADAR, AMACR, ARSA, BCAP31, CLN2 (TPP1), CLN3, DNM1L, GALC, HSD17B4, IFIH1, MFF, NR0B1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PPT1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCP2, TREX1
Porphyrias			
○ 06226	Invitae Acute Hepatic Porphyrias Panel	4	ALAD, CPOX, HMBS, PPOX
○ 55003	Invitae Comprehensive Porphyrias Panel	10	ALAD, ALAS2, CLPX, CPOX, FECH, GATA1, HMBS, PPOX, UROD, UROS
Purine Metabolism Disorders			
○ 06213	Invitae Purine Metabolism Disorders Panel	18	ADA, ADSL, AMPD1, APRT, ATIC, DGUOK, GPHN, HPRT1, IMPDH1, MOCOS, MOCS1, MOCS2, MOCS3, PNP, PRPS1, SUOX, UMOD, XDH
Pyruvate Metabolism and Tricarboxylic Acid Cycle Defects			
○ 98004	Invitae Pyruvate Metabolism and Related Disorders Panel	38	BOLA3, DLAT, DLD, ECHS1, FBXL4, FDX2, GLRX5, GOT2, HIBCH, IBA57, ISCA1, ISCA2, ISCU, LIAS, LIPT1, LIPT2, LONP1, LYRM4, MPC1, NFS1, NFU1, NUBPL, PC, PCK1, PDHA1, PDHB, PDHX, PDK3, PDP1, SLC19A2, SLC19A3, SLC25A1, SLC25A19, SUCLA2, SUCLG1, SUCLG2, TAZ, TPK1

METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Symptom-based Metabolic Panels			
○ 98001	Invitae Monogenic Obesity Panel	68	ADCY3, AFF4, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BDNF, C8orf37, CEP164, CEP19, CEP290, CPE, CREBBP, CUL4B, DYRK1B, EP300, FBN3, GNAS, GPR101, IFT172, IFT27, IFT74, KDM6A, KIDINS220, KIF7, KMT2D, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, NLGN2, NPY, NR0B2, NTRK2, PCSK1, PHF6, POMC, PPARG, PRMT7, RAB23, RAI1, RPS6KA3, SCLT1, SDCCAG8, SETD2, SH2B1, SIM1, TRAPPC3, TRAPPC9, TRIM32, TTC8, UCP3, VPS13B, WDPCP
○ 06001	Invitae Cholestasis Panel	136	ABCB11, ABCB4, ABCC2, ABCD3, ABCG5, ABCG8, ACOX1, ACOX2, ADK, AKR1C4, AKR1D1, ALAS2, ALDOB, ALG1, ALG8, AMACR, ANKS6, AP1S1, ARG1, ASS1, ATP6AP1, ATP7B, ATP8B1, B4GALT1, BAAT, BCS1L, BLVRA, C19orf70, CC2D2A, CCDC115, CDAN1, CFTR, CLDN1, CLPX, COG6, COG7, CTNS, CYP27A1, CYP7A1, CYP7B1, DCDC2, DGUOK, DHCR7, FAH, FECH, FH, G6PD, GALE, GALT, GLI2, GLIS3, GNAS, HADHA, HADHB, HNF1A, HNF1B, HSD17B4, HSD3B7, IARS, INVS, ITCH, JAG1, KIF12, KMT2D, LIPA, LSR, MKS1, MMACHC, MPV17, MVK, MYO5B, NEK8, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, OTC, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PKD1L1, PKHD1, POLG, POMC, PPM1F, PTF1A, RFX6, SC5D, SCP2, SCYL1, SERAC1, SERPINA1, SHPK, SLC10A2, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC27A5, SLC30A10, SLC51B, SMPD1, SMS, STXBP2, TALDO1, TBX19, TFAM, TFR2, TJP2, TMEM216, TRAPPC11, TRMU, TSFM, TTC26, TWNK, UGT1A1, USP53, UTP4, VIPAS39, VMA21, VPS33B, WDR83OS
○ 98006	Invitae Hypoglycemia Panel	120	AAAS, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACSF3, ADK, AGL, AKT2, ALDOA, ALDOB, ALG3, ALG6, BCKDHA, BCKDHB, CA5A, CACNA1C, CACNA1D, CDKN1C, COG7, CPT1A, CPT2, CYP7B1, DBH, DBT, DDC, DGUOK, DLD, DMXL2, DOLK, ENO3, ETFA, ETFB, ETFDH, FAH, FBP1, FLAD1, G6PC, GAA, GALE, GALK1, GALT, GBE1, GCK, GH1, GHR, GLUD1, GPC3, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HESX1, HK1, HMGCL, HMGCS2, HNF1A, HNF4A, HRAS, HSD3B7, INSR, KCNJ11, KDM6A, KMT2D, LAMP2, LDHA, LHX3, MLYCD, MPI, MPV17, NADK2, NNT, NR0B1, NR3C1, NSD1, OPLAH, OTX2, OXCT1, PC, PCK1, PCK2, PCSK1, PDX1, PFKM, PGAM2, PGM1, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PMM2, POMC, PROP1, PTF1A, PYGL, PYGM, RBCK1, SERAC1, SLC16A1, SLC22A5, SLC25A20, SLC25A32, SLC2A2, SLC37A4, SLC52A1, SLC52A2, SLC52A3, SOX2, SOX3, TAZ, TBX19, TRMT10A, UCP2
○ 06228	Invitae Mendelian Disorders with Psychiatric Symptoms Panel	167	ABCB4, ABCD1, ADSL, ALAD, ALDH5A1, AMACR, AMT, ANK3, AP1S1, ARG1, ARSA, ARX, ASL, ASS1, ATP13A2, ATP1A3, ATP2A2, ATP7B, B4GALNT1, BCKDHA, BCKDHB, BCKDK, C19orf12, CA5A, CACNA1A, CBS, CHCHD10, CLCN2, CLCN4, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLPX, CNTNAP2, COASY, COL4A1, CP, CPOX, CPS1, CSTB, CTSD, CYP27A1, DARS, DBH, DBT, DCAF17, DDC, DEPDC5, DLD, DNAJC12, DNAJC6, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPM2A, FA2H, FAH, FECH, FIG4, FOLR1, FUCA1, GALC, GAMT, GATM, GCH1, GFAP, GLA, GLB1, GLDC, GM2A, GNS, GRIA3, GSS, HARS, HEXA, HEXB, HGSNAT, HMBS, HMGCL, HPRT1, HTRA1, IQSEC2, KCNT1, KCTD17, MAN2B1, MANBA, MAOA, MECP2, MED12, MFSD8, MMACHC, MMADHC, MSTO1, MTHFR, MTR, MUT, NAGLU, NAGS, NDP, NHLRC1, NPC1, NPC2, OTC, PAH, PAK3, PANK2, PCBD1, PCCA, PCCB, PCDH19, PLA2G6, PLP1, POLG, PPOX, PPT1, PRDX1, PRKAR1A, PRODH, PSAP, PTS, QDPR, REPS1, RPS6KA3, SETX, SGCE, SGSH, SLC12A6, SLC20A2, SLC25A13, SLC25A15, SLC30A10, SLC39A14, SLC52A1, SLC52A2, SLC52A3, SLC6A19, SLC6A3, SLC6A8, SLC7A7, SPART, SPG11, SPR, SUMF1, TBC1D7, TBX1, TH, TIMM8A, TOR1A, TREX1, TRRAP, TTC19, TWNK, TYMP, TYROBP, UBQLN2, UROD, UROS, VPS13A, WDR45, WFS1, YWHAG, ZFYVE26
	○ 06228.1 Add-on adult onset Mendelian Disorders with Psychiatric Symptoms Panel	40	APP, CHCHD2, CHMP2B, CSF1R, CTSF, DCTN1, DNAJC5, DNMT1, FBXO7, FTL, GRN, ITM2B, ITPR1, KIAA1161, LMNB1, LRRK2, MAPT, MATR3, NOTCH3, OPTN, PARK7, PDGFB, PFN1, PINK1, PRKN, PRNP, PSEN1, PSEN2, SNCA, SOD1, SQSTM1, TARDBP, TBK1, TREM2, TTR, VAPB, VCP, VPS35, XK, XPR1

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METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Symptom-based Metabolic Panels (continued)			
○ 06230	Invitae Hyperammonemia Panel	76	ABCD4, ACADM, ACADVL, ALDH18A1, AMT, ARG1, ASL, ASS1, ATP5A1, ATP5D, ATP5E, ATPAF2, BCKDHA, BCKDHB, BTD, CA5A, CPS1, CPT1A, CPT2, CYC1, DBT, DLAT, DLD, ETFA, ETFB, ETFDH, FBXL4, GLDC, GLUD1, GLUL, HADHA, HADHB, HCFC1, HLCS, HMGCL, IVD, LMBRD1, LYRM7, MCCC1, MCCC2, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, NAGS, NBAS, NR1H4, OAT, OTC, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PRDX1, RINT1, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A42, SLC7A7, TANGO2, TAZ, TMEM70, TUFM, UMPS, UQCRC2, YARS2
○ 55004	Invitae Cerebral Palsy Spectrum Disorders Panel	425	ABAT, ABCD1, ACADM, ACADVL, ACAT1, ACBD5, ACOX1, ACTB, ADAR, ADCY5, ADD3, ADNP, ADSL, AFG3L2, AGAP1, AHDC1, AHI1, AKT3, ALDH18A1, ALDH3A2, ALDH5A1, ALDH7A1, ALG13, ALG3, ALS2, AMACR, AMPD2, AMT, ANO3, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARG1, ARHGEF9, ARL6IP1, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ATAD1, ATL1, ATM, ATP13A2, ATP1A3, ATP7A, ATP7B, ATP8A2, ATRX, AUH, AUTS2, B4GALNT1, BCAP31, BCKDHA, BCKDHB, BICD2, BSCL2, BTD, C12orf65, C19orf12, CACNA1A, CACNA1G, CAMTA1, CAPN1, CASK, CBS, CCDC88C, CCT5, CDKL5, CEP290, CHD8, CHRNA1, CIZ1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLTC, COASY, COL4A1, COL4A2, COL6A3, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ9, CPS1, CPT1C, CREBBP, CTBP1, CTNNB1, CTSD, CYP27A1, CYP2U1, CYP7B1, DARS, DARS2, DBH, DBT, DCAF17, DDC, DDHD1, DDHD2, DDX3X, DGKZ, DHDDS, DHFR, DLAT, DLD, DMD, DNAJC12, DNM2, DPAGT1, DYNC1H1, DYRK1A, EEF2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, ENTPD1, EPHA4, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, FA2H, FAM126A, FARS2, FAT2, FGF12, FGF14, FH, FOLR1, FOXG1, FRRS1L, FTL, FUCA1, GABRA2, GAD1, GALC, GAMT, GATM, GBA, GBA2, GCDH, GCH1, GFAP, GJC2, GLB1, GLDC, GLRA1, GLRB, GM2A, GNAL, GNAO1, GNB1, GNS, GPHN, GPR88, GRID2, GRIN1, GRIN2B, GRM1, HACE1, HESX1, HEXA, HEXB, HGSNAT, HLCS, HMGCL, HPCA, HPRT1, HSD17B10, HSD17B4, HSPD1, IBA57, IFIH1, IQSEC2, IREB2, ITPA, ITPR1, KANK1, KAT6A, KCNA2, KCNC3, KCNJ6, KCNMA1, KCNQ2, KCNT1, KCTD17, KCTD7, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KMT2A, KMT2B, KMT2C, L1CAM, L2HGDH, LAMA2, LIAS, LMBRD1, MAG, MAOA, MAP2K1, MARS2, MAST1, MCCC1, MCCC2, MCEE, MECP2, MECR, MFSD8, MICU1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MOCS3, MPC1, MTHFR, MTOR, MTPAP, MTR, MTRR, MTTP, MUT, NAA10, NAA35, NAGLU, NAGS, NBAS, NGLY1, NIPA1, NKX2-1, NPC1, NPC2, NPHP1, NT5C2, NUS1, OTC, PAFAH1B1, PAH, PAK3, PALM, PANK2, PCBD1, PCCA, PCCB, PCDH12, PDE10A, PDE2A, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHGDH, PHIP, PIGN, PIGT, PIK3CA, PLA2G6, PLD3, PLP1, PLXNA2, PMM2, PNKD, PNP, PNPLA6, PNPO, POLG, POLR3A, PPT1, PRKRA, PROSC, PRRT2, PRUNE1, PSAT1, PSPH, PTPN11, PTS, PURA, QDPR, RAB3GAP1, RAB3GAP2, RANBP2, REEP1, REEP2, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RTN2, SACS, SAMHD1, SATB2, SCN1A, SCN2A, SCN3A, SCN8A, SETD5, SGCE, SGSH, SHH, SIL1, SIX3, SLC16A2, SLC17A5, SLC18A2, SLC19A3, SLC1A4, SLC25A15, SLC25A22, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SON, SPART, SPAST, SPATA5, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, ST3GAL5, STAMBP, STUB1, STXBP1, SUCLA2, SUCLG1, SUOX, SURF1, SYNGAP1, TAF1, TANGO2, TBC1D24, TBCK, TBL1XR1, TCF4, TECPR2, TFG, TGIF1, TGM6, TH, THAP1, TMEM240, TMEM67, TOR1A, TREX1, TRPC3, TSEN54, TTBK2, TTPA, TUBA1A, TUBB2A, TUBB2B, TUBB3, TUBB4A, UBE3A, UCHL1, VAC14, VAMP1, VPS13A, VPS13D, VPS37A, WARS2, WASHC5, WDR45, WDR62, ZBTB18, ZC4H2, ZEB2, ZFR, ZFYVE26, ZIC1, ZIC2, ZIC4
○ 06229	Invitae Metabolic Non-Immune Fetal Hydrops Panel	51	AHCY, ALG1, ALG12, ALG8, ALG9, ARSB, ASAH1, CTSA, DHCR7, G6PD, GAA, GALC, GALNS, GBE1, GLB1, GLUL, GNPTAB, GUSB, HADH, HADHA, HADHB, IDUA, LIPA, MVK, NEU1, NPC1, NPC2, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIGA, PMM2, PSAT1, SLC17A5, SLC22A5, SLC26A2, SMPD1, SUMF1, TAZ, TRIP11
	○ 06229.1 Add-on limited evidence gene	1	SEC23B

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METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Symptom-based Metabolic Panels (continued)			
○ 55002	Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel	697	AARS, AARS2, ABAT, ABCA1, ABCD1, ABCD4, ABHD5, ACADS, ACBD5, ACER3, ACO2, ACOX1, ACP5, ACSF3, ACTA2, ACTB, ACY1, ADAR, ADGRG1, ADK, ADNP, ADSL, AGA, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, ALDH18A1, ALDH3A2, ALDH5A1, ALDH6A1, ALG12, ALG13, ALG2, ALG6, ALG9, AMACR, AMPD2, AMT, ANK3, AP1S2, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOPT1, ARCN1, ARFGEF2, ARHGAP31, ARHGEF9, ARNT2, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL2, ATP13A2, ATP5A1, ATP6AP2, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRN, ATRX, AUH, B3GALNT2, B4GALNT1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCL11B, BCS1L, BICD2, BMP4, BOLA3, BPTF, BRAT1, BTD, C12orf57, C12orf65, C19orf12, C2CD3, CACNA1A, CACNA1E, CARS2, CBS, CC2D2A, CCDC88A, CDC42, CDKL5, CEP290, CHMP1A, CLCN2, CLCN4, CLCN7, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLP1, CLPP, CNNM2, CNOT1, CNTNAP1, CNTNAP2, COA7, COASY, COG7, COL18A1, COL3A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, COX8A, CPLANE1, CPLX1, CPS1, CRAT, CREBBP, CRIPT, CRLF1, CSF1R, CSPP1, CTBP1, CTC1, CTDP1, CTNS, CTSA, CTSD, CUL4B, CYB5R3, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEAF1, DEGS1, DGUOK, DHCR24, DHFR, DHX37, DLD, DLL4, DMXL2, DNM1L, DNM2, DOCK6, DOCK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DPYS, DYRK1A, EARS2, EDNRB, EHMT1, EIF2AK1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL1, ELOVL4, ENTPD1, EOGT, EPG5, EPRS, ERCC1, ERCC2, ERCC3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, EXOSC2, EXOSC3, EXOSC8, EXOSC9, FA2H, FAM126A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FDX2, FGFRL1, FH, FIG4, FKRP, FKTN, FLVCR2, FOLR1, FOXC1, FOXG1, FOXRED1, FUCA1, FUK, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GALC, GALT, GAN, GATAD2B, GCDH, GDAP1, GFAP, GFM1, GFM2, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLRX5, GLUL, GLYCTK, GM2A, GMNN, GNAO1, GNS, GOT2, GPHN, GRIN1, GRM7, GTF2H5, GTPBP2, HACE1, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HK1, HLCS, HMGCL, HNRNPU, HSD17B10, HSD17B4, HSPD1, IARS, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, INPP5E, IREB2, ISCA1, ISCA2, ITPA, ITPR1, IVD, JAM3, KARS, KAT6B, KATNB1, KCNJ10, KCNJ2, KCNMA1, KCNT1, KCTD7, KDM1A, KIAA0556, KIAA0586, KIAA0753, KIDINS220, KIF1A, KIF1C, KLHL7, KMT2E, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB1, LARGE1, LARS2, LETM1, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM7, MAG, MAGEL2, MAN2B1, MANBA, MARS2, MAST1, MAT1A, MCCC1, MCCC2, MCOLN1, MDH2, MECP2, MED17, MED25, MEF2C, MFSD2A, MFSD8, MGP, MICU1, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MORC2, MPLKIP, MPV17, MPZ, MRPL12, MRPL44, MRPS16, MRPS22, MRPS34, MSTO1, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTR, MTRR, MUT, NACC1, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAXD, NAXE, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK1, NFE2L2, NFU1, NGLY1, NKX6-2, NOTCH1, NPC1, NPC2, NPHP1, NRXN1, NSUN3, NT5C2, NTRK2, NUBPL, NUP62, OAT, OCRL, OPA1, OPA3, OSGEP, OTC, PACS1, PAFAH1B1, PAH, PANK2, PARS2, PAX1, PC, PCCA, PCCB, PCDH12, PCGF2, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PHGDH, PHYH, PIGA, PIGB, PIGG, PIGL, PIGM, PIGN, PIGP, PIGQ, PIGT, PIGU, PIGV, PIK3C2A, PIK3R2, PLA2G6, PLAA, PLEKHG2, PLP1, PMM2, PMP22, PNKP, PNPT1, POLG, POLG2, POLR1A, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPP1R15B, PPP2R1A, PPP3CA, PPT1, PRF1, PRKDC, PRODH, PROSC, PRPS1, PRUNE1, PSAP, PSAT1, PSPH, PTEN, PTPN23, PURA, PUS3, PYCR2, QARS, QRSL1, RAB11B, RAB3GAP1, RAB3GAP2, RARS, RARS2, RBPJ, REPS1, RERE, RHOBTB2, RIN2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPGRIP1L, RPIA, RPS6KC1, RRM2B, RTTN, RXYLT1, SAMD9L, SAMHD1, SCN3A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEPSECS, SERAC1, SETBP1, SGSH, SH3TC2, SHOC2, SHPK, SLC12A2, SLC12A5, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC35A2, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A8, SLC6A9, SLC9A1, SLC9A6, SMC1A, SNAP29, SNIP1, SNORD118, SNRPB, SNX14 (continues)

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Test code	Test name	# gene(s)	Gene list
Symptom-based Metabolic Panels (continued)			
	Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel (continued)	697	(continued) SOD1, SON, SOX10, SOX2, SPART, SPAST, SPATA5, SPG11, SPG7, SPTAN1, SQSTM1, SRD5A3, SSR4, ST3GAL5, STAG2, STAMBP, STAT1, STAT2, STN1, STRADA, STX11, STXBP1, STXBP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYNE1, SYNJ1, TACO1, TAF2, TANGO2, TARS2, TBC1D24, TBCD, TBCE, TBCK, TBX1, TCF4, TCTN2, TIMM50, TIMMDC1, TM4SF20, TMEM106B, TMEM126B, TMEM165, TMEM216, TMEM67, TMEM70, TMTC3, TOE1, TP53RK, TPI1, TPK1, TRAPPC11, TRAPPC9, TREX1, TRMT10A, TRMT5, TSC1, TSC2, TSEN54, TTC19, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUFM, TWNK, TXN2, TYMP, UBA5, UBE2A, UBE3A, UFM1, UNC13D, UPB1, USP7, VARS2, VPS11, VPS13D, VPS33A, WARS2, WDR45, WDR73, WHSC1, WWOX, YARS, YME1L1, ZEB2, ZFYVE26, ZIC1, ZNF335
	○ 55002.1 Add-on Adult-onset Leukodystrophy and Leukoencephalopathy Panel	32	ALDH7A1, APP, CHMP2B, CP, CTSF, DNAJC5, DNMT1, FTL, GBE1, GRN, HTRA1, KIAA1161, KIF5A, LMNB1, MAPT, NOTCH3, PDGFB, PDGFRB, PDYN, PHAX, PINK1, PRNP, PSEN1, RNF216, SCP2, SLC20A2, TREM2, TYROBP, VCP, VPS13A, XK, XPR1
Mitochondial Disorders			
○ 98002	Invitae Nuclear Mitochondrial Disorders Panel	333	AARS2, AASS, ABAT, ABCB7, ACACA, ACAD9, ACADM, ACADS, ACADVL, ACAT1, ACO2, ADAR, AFG3L2, AGK, AIFM1, AK2, ALAS2, ALDH3A2, AMPD1, AMT, APOPT1, APTX, ATP5A1, ATP5D, ATP5E, ATP7B, ATPAF2, AUH, BAG3, BCS1L, BOLA3, BTD, C12orf65, C19orf12, C19orf70, C1QBP, CA5A, CARS2, CEP89, CHAT, CHCHD10, CLPB, CLPP, COA3, COA5, COA6, COA7, COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, COX8A, CPS1, CPT1A, CPT2, CYC1, CYCS, D2HGDH, DARS2, DES, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNM1L, EARS2, ECHS1, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FH, FLAD1, FOXRED1, GAMT, GARS, GATM, GCDH, GDAP1, GFER, GFM1, GFM2, GLDC, GLRX5, GTPBP3, GYG2, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HLCS, HMGCL, HMGCS2, HSD17B10, HSPD1, HTRA2, IARS2, IBA57, IDH2, IDH3B, IFIH1, ISCA1, ISCA2, ISCU, KARS, L2HGDH, LAMP2, LARS, LARS2, LIAS, LIPT1, LIPT2, LMBRD1, LONP1, LRPPRC, LYRM4, LYRM7, MARS2, MECR, MFF, MFN2, MGME1, MICU1, MIPEP, MPC1, MPV17, MRPL12, MRPL3, MRPL40, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS34, MRPS7, MSTO1, MTFMT, MTHFD1, MTO1, MTPAP, NADK2, NARS2, NAXE, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NGLY1, NNT, NR2F1, NSUN3, NUBPL, NUP62, OGDH, OPA1, OPA3, OTC, OXCT1, PANK2, PARS2, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PET100, PINK1, PITRM1, PMPCA, PMPCB, PNKD, PNPLA8, PNPT1, POLG, POLG2, POP1, PPA2, PPOX, PSAP, PUS1, QARS, QRSL1, RANBP2, RARS, RARS2, REEP1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RRM2B, SACS, SAMHD1, SARS2, SCO1, SCO2, SCN1A, SDHA, SDHAF1, SDHB, SDHC, SDHD, SERAC1, SFXN4, SIRT1, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC39A8, SLC52A2, SLC52A3, SLC6A8, SLC7A13, SPAST, SPG7, STAT2, STXBP1, SUCLA2, SUCLG1, SUCLG2, SUGCT, SURF1, TACO1, TANGO2, TARS2, TAZ, TFAM, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TOP1MT, TOP3A, TPK1, TRAP1, TREX1, TRIT1, TRMT10C, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, VARS2, WARS2, WDR45, WFS1, XPNPEP3, YARS2, YME1L1
Treatable Disorders			
○ 55000	Invitae Alpha-1 Antitrypsin Deficiency Test	1	SERPINA1
○ 06003	Invitae Crigler Najjar and Gilbert Syndrome Test	1	UGT1A1
○ 55001	Invitae Monogenic Diabetes Panel	28	ABCC8, APPL1, BLK, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, INS, KCNJ11, KLF11, MNX1, NEUROD1, NEUROG3, NKX2-2, PAX4, PDX1, PPARG, PTF1A, RFX6, SLC19A2, WFS1, ZFP57

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METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Treatable Disorders (continued)			
○ 06222	Invitae Treatable Neurometabolic Disorders Panel	195	ABCD1, ABCD4, ACAT1, AGA, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, AMN, AMT, APTX, ARG1, ARHGEF9, ARSA, ASAH1, ASL, ASNS, ASPA, ASS1, ATAD1, ATP7A, ATP7B, AUH, BCKDHA, BCKDHB, BSCL2, BSND, BTD, CA5A, CAD, CASR, CBS, CD320, CLCNKB, CLDN16, CLDN19, CLN2 (TPP1), CLN3, CLN5, CLN6, CNNM2, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, CP, CPOX, CPS1, CUBN, CYP27A1, DBT, DDC, DHCR7, DHFR, DLAT, DLD, DNAJC12, EGF, ETFA, ETFB, ETFDH, ETHE1, FAM111A, FOLR1, FXYD2, GALC, GAMT, GATM, GCDH, GCH1, GCLC, GIF, GLA, GLB1, GLDC, GLRA1, GLRB, GLUD1, GNS, GOT2, GPHN, GSS, GUSB, HCFC1, HEXA, HEXB, HGSNAT, HLCS, HMBS, HMGCL, HMGCS2, HNF1B, HSD17B10, IDS, IDUA, IVD, KCNA1, KCNJ10, LIPA, LMBRD1, MAN2B1, MCCC1, MCCC2, MCEE, MFSD8, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MSMO1, MTHFR, MTR, MTRR, MUT, NAGLU, NAGS, NPC1, NPC2, NT5C3A, OAT, OTC, OXCT1, PAH, PANK2, PCBD1, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PGM3, PHGDH, PHYH, PNPO, PPM1K, PRDX1, PROSC, PRPS1, PSAT1, PSPH, PTS, QDPR, RAPSN, SCN4A, SGSH, SLC12A1, SLC12A3, SLC13A5, SLC18A2, SLC19A1, SLC19A2, SLC19A3, SLC1A3, SLC25A13, SLC25A15, SLC25A19, SLC2A1, SLC30A10, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SPR, TAT, TCN1, TCN2, TH, TPK1, TRPM6, TTPA
Urea Cycle Disorders			
○ 06212	Invitae Urea Cycle Disorders Panel	10	ALDH18A1, ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC25A13, SLC25A15
○ 06212.1	Add-on hyperammonemia genes	4	CA5A, GLUD1, GLUL, SLC7A7
○ 06212.2	Add-on hereditary orotic aciduria gene	1	UMPS
○ 06225	Invitae Ornithine Transcarbamylase (OTC) Deficiency Test	1	OTC
○ 06225.1	Add-on hereditary orotic aciduria gene	1	UMPS
○ 06225.2	Add-on low citrulline genes	2	CPS1, NAGS
○ 06117	Invitae Elevated Arginine (Arginase deficiency) Panel	2	ARG1, GCGR
○ 06118	Invitae Elevated Citrulline (Citrullinemia) Panel	5	ASL, ASS1, DLD, PC, SLC25A13

NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY INDIVIDUAL GENES

○ ABAT	○ ACTB	○ ALDH4A1	○ AMACR	○ ATP13A2	○ BCKDHB	○ CBS	○ CHST14	○ COG5
○ ABCD1	○ ADA	○ ALDH5A1	○ AMN	○ ATP6V0A2	○ BCKDK	○ CD247	○ CHST3	○ COG6
○ ABCD4	○ ADA2	○ ALDH7A1	○ AMPD1	○ ATP7A	○ BCL10	○ CD27	○ CHST6	○ COG7
○ ABCG5	○ ADAM17	○ ALDOA	○ AMT	○ ATP7B	○ BLNK	○ CD320	○ CHSY1	○ COG8
○ ABCG8	○ ADAR	○ ALDOB	○ AP1S1	○ AUH	○ BLOC1S6	○ CD3D	○ CIITA	○ COPA
○ ACAD8	○ ADSL	○ ALG1	○ AP3B1	○ B2M	○ BTD	○ CD3E	○ CLN2 (TPP1)	○ CORO1A
○ ACADM	○ AGA	○ ALG11	○ ARG1	○ B3GALNT2	○ BTK	○ CD3G	○ CLN3	○ CP
○ ACADS	○ AGL	○ ALG12	○ ARHGEF9	○ B3GALT6	○ C19orf12	○ CD40LG	○ CLN5	○ CPOX
○ ACADSB	○ AGPS	○ ALG13	○ ARSA	○ B3GAT3	○ C1GALT1C1	○ CD79A	○ CLN6	○ CPS1
○ ACADVL	○ AHCY	○ ALG14	○ ARSB	○ B3GLCT	○ CA5A	○ CD79B	○ CLN8	○ CPT1A
○ ACAT1	○ AICDA	○ ALG2	○ ASAH1	○ B4GALNT1	○ CARD11	○ CD8A	○ CLPB	○ CPT2
○ ACD	○ AIRE	○ ALG3	○ ASL	○ B4GALT1	○ CARD14	○ CEBPE	○ COASY	○ CR2
○ ACOX1	○ AK2	○ ALG6	○ ASPA	○ B4GALT7	○ CARD9	○ CFTR	○ COG1	○ CSF2RA
○ ACP5	○ ALAD	○ ALG8	○ ASS1	○ B4GAT1	○ CASP10	○ CHD7	○ COG2	○ CSF3R
○ ACSF3	○ ALDH18A1	○ ALG9	○ ATM	○ BCKDHA	○ CASP8	○ CHIT1	○ COG4	○ CTC1

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METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY INDIVIDUAL GENES (continued)

○ CTLA4	○ EOGT	○ GCSH	○ HPD	○ L2HGDH	○ MTR	○ PCCA	○ PLCG2	○ RHOH
○ CTNS	○ EPG5	○ GFI1	○ HPRT1	○ LAMP2	○ MTRR	○ PCCB	○ PMM2	○ RMRP
○ CTPS1	○ ETFA	○ GFPT1	○ HSD17B10	○ LAMTOR2	○ MUT	○ PDHA1	○ PMS2	○ RNASEH2A
○ CTSA	○ ETFB	○ GIF	○ HSD17B4	○ LARGE1	○ MVK	○ PDHB	○ PNP	○ RNASEH2B
○ CTSC	○ ETFDH	○ GLA	○ HYAL1	○ LCK	○ MYD88	○ PDHX	○ PNPO	○ RNASEH2C
○ CTSD	○ ETHE1	○ GLB1	○ ICOS	○ LDHA	○ NADK2	○ PDP1	○ POFUT1	○ RORC
○ CTSF	○ EXT1	○ GLDC	○ IDH2	○ LFNG	○ NAGA	○ PEX1	○ POGLUT1	○ RPN2
○ CTSK	○ EXT2	○ GLRA1	○ IDS	○ LIAS	○ NAGLU	○ PEX10	○ POLE	○ RTEL1
○ CUBN	○ FA2H	○ GLRB	○ IDUA	○ LIG4	○ NAGS	○ PEX12	○ POLG	○ RXYLT1
○ CXCR4	○ FADD	○ GLUD1	○ IFIH1	○ LIPA	○ NBN	○ PEX13	○ POMGNT1	○ SAMHD1
○ CYBA	○ FAH	○ GLUL	○ IFNGR1	○ LMBRD1	○ NCF2	○ PEX14	○ POMGNT2	○ SEC23A
○ CYBB	○ FAS	○ GM2A	○ IFNGR2	○ LPIN2	○ NCF4	○ PEX16	○ POMK	○ SEC23B
○ CYP27A1	○ FASLG	○ GMPPA	○ IGLL1	○ LRBA	○ NEU1	○ PEX19	○ POMT1	○ SEMA3E
○ D2HGDH	○ FBP1	○ GMPPB	○ IKBKB	○ LYST	○ NFAT5	○ PEX2	○ POMT2	○ SERAC1
○ DBH	○ FERMT3	○ GNE	○ IL10	○ MAGT1	○ NFKB2	○ PEX26	○ PPM1K	○ SGSH
○ DBT	○ FH	○ GNMT	○ IL10RA	○ MALT1	○ NFKBIA	○ PEX3	○ PPOX	○ SH2D1A
○ DCAF17	○ FKRP	○ GNPAT	○ IL10RB	○ MAN1B1	○ NFU1	○ PEX5	○ PPT1	○ SH3BP2
○ DCLRE1B	○ FKTN	○ GNPTAB	○ IL12B	○ MAN2B1	○ NGLY1	○ PEX6	○ PREPL	○ SLC13A5
○ DCLRE1C	○ FOXN1	○ GNPTG	○ IL12RB1	○ MANBA	○ NHEJ1	○ PEX7	○ PRF1	○ SLC17A5
○ DDC	○ FOXP3	○ GNS	○ IL17F	○ MAOA	○ NHP2	○ PFKM	○ PRKCD	○ SLC19A3
○ DDOST	○ FPR1	○ GPHN	○ IL17RA	○ MAP3K14	○ NLRC4	○ PGAM2	○ PRKDC	○ SLC22A5
○ DECR1	○ FTCD	○ GRN	○ IL17RC	○ MAT1A	○ NLRP12	○ PGM1	○ PRODH	○ SLC25A1
○ DHCR7	○ FTL	○ GSS	○ IL1RN	○ MCCC1	○ NLRP3	○ PGM3	○ PSAP	○ SLC25A13
○ DHDDS	○ FUCA1	○ GUSB	○ IL21	○ MCCC2	○ NOD2	○ PHGDH	○ PSAT1	○ SLC25A15
○ DHTKD1	○ G6PC	○ GYG1	○ IL21R	○ MCEE	○ NOP10	○ PHKA1	○ PSMB8	○ SLC25A19
○ DKC1	○ G6PC3	○ GYS1	○ IL2RA	○ MCOLN1	○ NPC1	○ PHKA2	○ PSPH	○ SLC25A20
○ DLAT	○ G6PD	○ GYS2	○ IL2RG	○ MEFV	○ NPC2	○ PHKB	○ PSTPIP1	○ SLC25A22
○ DLD	○ GAA	○ HADH	○ IL36RN	○ MFSD8	○ NUS1	○ PHKG2	○ PTPRC	○ SLC26A2
○ DNAJC19	○ GAD1	○ HADHA	○ IL7R	○ MGAT2	○ OAT	○ PHYH	○ PTS	○ SLC29A3
○ DNAJC5	○ GALC	○ HADHB	○ IRAK4	○ MLYCD	○ OGDH	○ PIGA	○ PYGL	○ SLC2A1
○ DNMT3B	○ GALE	○ HAX1	○ IRF7	○ MMAA	○ OPA3	○ PIGL	○ PYGM	○ SLC2A2
○ DOCK2	○ GALK1	○ HCFC1	○ IRF8	○ MMAB	○ OPLAH	○ PIGM	○ QDPR	○ SLC33A1
○ DOCK8	○ GALNS	○ HEXA	○ ISG15	○ MMACHC	○ ORAI1	○ PIGN	○ RAB27A	○ SLC35A1
○ DOLK	○ GALNT3	○ HEXB	○ ISPD	○ MMADHC	○ OTC	○ PIGO	○ RAC2	○ SLC35A2
○ DPAGT1	○ GALT	○ HGD	○ ITCH	○ MOCOS	○ OXCT1	○ PIGQ	○ RAG1	○ SLC35A3
○ DPM1	○ GAMT	○ HGSNAT	○ ITGB2	○ MOCS1	○ PAH	○ PIGT	○ RAG2	○ SLC35C1
○ DPM2	○ GATA2	○ HIBCH	○ ITK	○ MOGS	○ PANK2	○ PIGV	○ RBCK1	○ SLC35D1
○ DPM3	○ GATM	○ HLCS	○ IVD	○ MPC1	○ PAPSS2	○ PIGW	○ RFT1	○ SLC37A4
○ DSE	○ GBE1	○ HMBS	○ JAGN1	○ MPDU1	○ PARN	○ PIK3CD	○ RFX5	○ SLC3A1
○ ELANE	○ GCDH	○ HMGCL	○ JAK3	○ MPI	○ PC	○ PIK3R1	○ RFXANK	○ SLC52A1
○ ENO3	○ GCH1	○ HMGCS2	○ KCTD7	○ MTHFR	○ PCBD1	○ PLA2G6	○ RFXAP	○ SLC52A2

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METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY INDIVIDUAL GENES (continued)								
○ SLC52A3	○ SMPD1	○ STAT2	○ SUCLA2	○ TBK1	○ TLR3	○ TNFRSF4	○ TUSC3	○ WAS
○ SLC5A1	○ SP110	○ STAT3	○ SUCLG1	○ TBX1	○ TMC6	○ TNFSF12	○ TYK2	○ WDR45
○ SLC6A3	○ SPINK5	○ STAT5B	○ SUMF1	○ TCN1	○ TMC8	○ TPP2	○ UMPS	○ WIPF1
○ SLC6A5	○ SPR	○ STIM1	○ SUOX	○ TCN2	○ TMEM165	○ TRAF3	○ UNC13D	○ XDH
○ SLC6A8	○ SRD5A3	○ STK4	○ TAP1	○ TERC	○ TMEM173	○ TRAF3IP2	○ UNC93B1	○ XIAP
○ SLC6A9	○ SSR4	○ STT3A	○ TAP2	○ TERT	○ TMEM70	○ TREX1	○ UNG	○ XYLT1
○ SLC7A7	○ ST3GAL3	○ STT3B	○ TAPBP	○ TH	○ TNFRSF13B	○ TRIP11	○ USB1	○ ZAP70
○ SLC7A9	○ ST3GAL5	○ STX11	○ TAT	○ TICAM1	○ TNFRSF13C	○ TRNT1	○ VPS13B	○ ZBTB24
○ SMARCAL1	○ STAT1	○ STXBP2	○ TAZ	○ TINF2	○ TNFRSF1A	○ TTC7A	○ VPS45	

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Skeletal Disorders			
○ 434342	Invitae Adams-Oliver Syndrome Panel	8	ARHGAP31, DLL4, DOCK6, EOGT, KCTD1, NOTCH1, RBPJ, UBR1
○ 04612	Invitae Antley-Bixler Syndrome Test	1	POR
	○ 04612.1 Add-on craniosynostosis gene	1	FGFR2
○ 04726	Invitae ARSE-Related Chondrodysplasia Punctata Test	1	ARSE
	○ 04726.1 Add-on NSDHL-related disorders gene	1	NSDHL
○ 04712	Invitae Campomelic Dysplasia Test	1	SOX9
○ 04423	Invitae Craniosynostosis Panel	65	ALPL, ASXL1, B3GAT3, CD96, CDC45, CDT1, COLEC11, CYP26B1, EFNA4, EFNB1, ERF, ESCO2, FBN1, FGF9, FGFR1, FGFR2, FGFR3, FREM1, GLI3, GPC3, IFT122, IFT140, IFT43, IGF1R, IL11RA, KAT6A, KAT6B, MASP1, MEGF8, MSX2, NFIA, ORC1, ORC4, ORC6, P4HB, PHEX, POR, PPP3CA, RAB23, RECQL4, RSPRY1, RUNX2, SCARF2, SEC24D, SIX2, SKI, SLC25A24, SMAD2, SMAD3, SMAD6, SOX6, SPECC1L, STAT3, TCF12, TCOF1, TGFB2, TGFB3, TGFBR1, TGFBR2, TMCO1, TWIST1, WDR19, WDR35, ZEB2, ZIC1
	○ 04423.1 Add-on 3MC and Treacher Collins syndromes genes	2	MASP1, TCOF1
○ 04613	Invitae Ellis-van Creveld and Weyers Acrofacial Dysostosis Panel	2	EVC, EVC2
○ 04614	Invitae Hereditary Multiple Osteochondromas Panel	2	EXT1, EXT2
	○ 04614.1 Add-on Langer-Giedion Syndrome Gene	1	TRPS1
○ 72039	Invitae Hypophosphatemia Panel	17	ALPL, CLCN5, CTNS, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, GNAS, OCRL, PHEX, SLC34A1, SLC34A3, VDR
○ 55010	Invitae Limb and Digital Malformations Panel	178	ACVR1, ADAMTS10, ADAMTS17, AFF4, AHI1, ANKRD11, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, B3GLCT, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP2, BMP4, BMPR1B, BTRC, C2CD3, CACNA1C, CC2D2A, CCNQ, CDH3, CEP104, CEP120, CEP290, CEP41, CHSY1, CHUK, CKAP2L, CPLANE1, CREBBP, CSPP1, DDX59, DHCR7, DHODH, DLL4, DLX5, DLX6, DOCK6, DPF2, DVL1, DVL3, DYNC1I1, EOGT, EP300, ESCO2, EVC, EVC2, FAT1, FBLN1, FBN1, FBXW4, FGF10, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FLNA, FMN1, FRAS1, FREM2, FZD2, GDF5, GDF6, GJA1, GLI2, GLI3, GNAS, GSC, HDAC4, HDAC8, HOXA13, HOXD13, IFT57, IHH, INPP5E, KDM6A, KIAA0586, KIF7, KMT2A (continues)

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METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Skeletal Disorders (continued)			
	Invitae Limb and Digital Malformations Panel (continued)	178	(continued) KMT2D, LMBR1, LRP4, LTBP2, LTBP3, MAP3K20, MEGF8, MGP, MKKS, MKS1, MRE11, MYCN, NECTIN1, NECTIN4, NIPBL, NOG, NOTCH1, NPHP1, NPHP3, NSDHL, NXN, OFD1, PDE3A, PDE4D, PDE6D, PGM3, PHF6, PIGV, PIK3CA, PITX1, POLR1A, PORCN, PRKAR1A, PRMT7, PTDSS1, PTHLH, RAB23, RAD21, RBM8A, RBPJ, RECQL4, ROR2, RPGRIP1L, SALL1, SALL4, SC5D, SDCCAG8, SF3B4, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMOC1, SOST, SOX11, SOX9, TBX15, TBX3, TBX5, TCTN1, TCTN2, TCTN3, TGDS, THPO, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TP63, TRIM32, TRPS1, TRPV4, TTC21B, TTC8, VAC14, WDPCP, WNT10B, WNT3, WNT5A, WNT7A, ZNF423, ZSWIM6
○ 04307	Invitae Microcephalic Primordial Dwarfism and Seckel Syndrome Panel	38	ATR, ATRIP, CDC45, CDC6, CDK5RAP2, CDT1, CENPJ, CEP135, CEP152, CEP63, CEP97, CRIPT, DNA2, DNMT3A, DONSON, GMNN, LARP7, LIG4, MCM5, MCPH1, NIN, NSMCE2, ORC1, ORC4, ORC6, PCNT, PLK4, POC1A, RBBP8, RNU4ATAC, RTTN, SRCAP, TRAIP, TUBGCP4, TUBGCP6, UBE3B, WDR4, XRCC4
○ 04732	Invitae Osteogenesis Imperfecta and Bone Fragility Panel	67	ALPL, ANO5, ASCC1, B3GAT3, B4GALT7, BMP1, CA2, CLCN5, CLCN7, COL1A1, COL1A2, CREB3L1, CRTAP, CTNS, CTSK, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FAM46A, FGF23, FGFR1, FKBP10, GNAS, GORAB, IFITM5, LRP5, LRRK1, MBTPS2, MESDC2, NBAS, NOTCH2, NTRK1, OCRL, OSTM1, P3H1, P4HB, PHEX, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SFRP4, SGMS2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SNX10, SP7, SPARC, SUCO, TAPT1, TCIRG1, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TRIP4, VDR, WNT1, WNT3A, XYLT2
○ 04103	Invitae Perinatal Lethal Skeletal Dysplasia and Skeletal Ciliopathies Panel	73	AGPS, ALPL, ASCC1, CEP120, CFAP410, CHUK, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CREB3L1, CRTAP, CSPP1, DLL1, DLL3, DMRT2, DYNC2H1, DYNC2LI1, EBP, EVC, EVC2, FAM111A, FGFR2, FGFR3, GNPAT, GPX4, HES7, HSPG2, ICK, IFT122, IFT140, IFT172, IFT43, IFT52, IFT74, IFT80, IFT81, IMPAD1, INPPL1, INTU, KIAA0586, KIAA0753, LBR, LFNG, LRP5, MBTPS2, MESP2, NEK1, P3H1, PAM16, PEX5, PEX7, PPIB, RIPPLY2, SLC26A2, SLC35D1, SOX9, TBX6, TCTEX1D2, TMEM38B, TRAF3IP1, TRIP11, TRIP4, TRPV4, TTC21B, WDR19, WDR34, WDR35, WDR60, WNT1, WNT3
○ 89100	Invitae Skeletal Disorders Panel	358	ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, AFF4, AGA, AGPS, AIFM1, ALPL, AMER1, ANKH, ANO5, ARCN1, ARSB, ARSE, ASCC1, ASPM, ATR, ATRIP, B3GALT6, B3GAT3, B4GALT7, BGN, BMP1, BMP2, BMPER, BMPR1B, C2CD3, CA2, CANT1, CASR, CCDC8, CDC45, CDC6, CDK5RAP2, CDKN1C, CDT1, CENPJ, CEP120, CEP135, CEP152, CEP63, CEP97, CFAP410, CHST14, CHST3, CHUK, CLCN5, CLCN7, COG1, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREB3L1, CRIPT, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTNS, CTSA, CTSK, CUL7, CWC27, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DIP2C, DLL1, DLL3, DLX3, DMP1, DMRT2, DNA2, DNMT3A, DONSON, DVL1, DVL3, DYM, DYNC2H1, DYNC2LI1, EBP, EIF2AK3, ENPP1, ESCO2, EVC, EVC2, EXOC6B, EXOSC2, EXT1, EXT2, EXTL3, FAH, FAM111A, FAM20C, FAM46A, FAR1, FAT4, FBN1, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FTO, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GHRHR, GHSR, GJA1, GLB1, GMNN, GNAS, GNE, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GSC, GUSB, GZF1, HES7, HGSNAT, HPGD, HSPG2, HYAL1, IARS2, ICK, IDS, IDUA, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IGF1, IGF2, IHH, IMPAD1, INPPL1, INTU, JAG1, KAT6B, KIAA0586, KIAA0753, KIF22, KL, KMT2A, LARP7, LBR, LEMD3, LFNG, LIFR, LIG4, LMNA, LMX1B, LONP1, LOXL3, LRP4, LRP5, LRRK1, LTBP2, LTBP3, MAFB, MAN2B1, MANBA, MAP3K7, MATN3, MBTPS1, MBTPS2, MCM5, MCPH1, MEOX1, MESDC2, MESP2, MGP, MMP13, MMP14, MMP2, MMP9, MNX1, MSX2, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NIN, NKX3-2, NOG, NOTCH2, NPPC, NPR2, NPR3, NSDHL, NSMCE2, NTRK1, NXN, OBSL1, OCRL, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PCGF2, PCNT, PCYT1A, PDE4D, PEX5, PEX7, PGM3, PHEX, PIK3C2A, PISD, PKDCC, PLK4, PLOD2, PLS3, POC1A, POLR1A, POP1, POR, PPIB, PPP3CA, PRKAR1A, PTDSS1, PTH1R, PTHLH, PTPN11, PYCR1, RAB33B, RBBP8, RECQL4, RIPPLY2, RMRP, RNU4ATAC, ROR2, RSPO2, RSPRY1, RTTN, RUNX2, SC5D, SEC24D, SERPINF1, SERPINH1, SETBP1, SFRP4, SGMS2, SGSH, SH3PXD2B, SLC17A5, SLC26A2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC39A13 (continues)

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METABOLIC DISORDERS AND NEWBORN SCREENING TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Skeletal Disorders (continued)			
	Invitae Skeletal Disorders Panel (continued)	358	(continued) SLCO2A1, SLCO5A1, SMAD4, SMARCAL1, SNRPB, SNX10, SOX9, SP7, SPARC, SQSTM1, SRCAP, SUCO, SULF1, SUMF1, TAB2, TAPT1, TBCE, TBX15, TBX3, TBX5, TBX6, TBXAS1, TCIRG1, TCTEX1D2, TCTN3, TGFB1, TMEM165, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TRAF3IP1, TRAIP, TRAPPC2, TREM2, TRIM37, TRIP11, TRIP4, TRMT10A, TRPS1, TRPV4, TTC21B, TUBGCP4, TUBGCP6, TYROBP, UBE3B, VAC14, VDR, VPS33A, WDR19, WDR34, WDR35, WDR4, WDR60, WISP3, WNT1, WNT3, WNT3A, WNT5A, XRCC4, XYLT1, XYLT2, ZMPSTE24, ZNF687
○ 434350	Invitae Spondylocostal Dysostosis Panel	8	DLL1, DLL3, DMRT2, HES7, LFNG, MESP2, RIPPLY2, TBX6
○ 55013	Invitae Stickler Syndrome Panel	9	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3, LRP2, VCAN
○ 04617	Invitae Trichorhinophalangeal Syndrome Panel	2	EXT1, TRPS1
○ 72038	Invitae X-Linked Hypophosphatemia Test	1	PHEX

CLINICAL AREA: PRIMARY HYPEROXALURIAS

Test code	Test name	# gene(s)	Gene list
Primary Hyperoxalurias			
○ 06227	Invitae Primary Hyperoxaluria Panel	3	AGXT, GRHPR, HOGA1

PRIMARY HYPEROXALURIAS INDIVIDUAL GENES

○ AGXT	○ GRHPR	○ HOGA1

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NEPHROLOGY TEST CATALOG

All tests on this form are organized by clinical area. If your order contains tests from multiple clinical areas, you will need to send a separate specimen for each clinical area. Each clinical area represents an individual billable event and report. Contact Client Services with any questions. For Invitae's full test menu, please visit www.invitae.com.

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Ciliopathies			
○ 04112	Invitae Bardet-Biedl Syndrome Panel	28	ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CEP164, CEP19, CEP290, FBN3, IFT172, IFT27, IFT74, KIF7, LZTFL1, MKKS, MKS1, SCLT1, SDCCAG8, TRAPPC3, TRIM32, TTC8, WDPCP
○ 04102	Invitae Ciliopathies Panel	174	ADAMTS9, AHI1, AK7, ALG8, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC4, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C11orf70, C2CD3, C8orf37, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CCNQ, CELSR2, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP55, CEP83, CFAP298, CFAP410, CFTR, CLUAP1, CPLANE1, CRB2, CSPP1, CTU2, DCDC2, DDX59, DHCR7, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB11, DNAJB13, DNAL1, DRC1, DYNC2H1, DYNC2LI1, DZIP1L, EVC, EVC2, EXOC3L2, EXOC8, FAM186B, FBN3, FGFR1, FGFR2, FGFR3, GANAB, GAS8, GLIS2, HNF1B, HYLS1, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, LRP5, LRRC56, LRRC6, LRRCC1, LZTFL1, MAPKBP1, MCIDAS, MKKS, MKS1, MRE11, NEK1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PIAS1, PIBF1, PIH1D3, PKD2, PKHD1, PMM2, PRKCSH, RBM48, RCOR1, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SCLT1, SDCCAG8, SEC63, SLC30A7, SPAG1, SUFU, TBC1D32, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TRAPPC3, TRIM32, TTC21B, TTC26, TTC8, TXNDC15, USP9X, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZMYND10, ZNF423
○ 04111	Invitae Joubert and Meckel-Gruber Syndromes Panel	31	AHI1, ARL13B, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, MRE11, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423
○ 55012	Invitae Neonatal Respiratory Distress Panel	111	ABCA3, ACE, AFF4, AGT, AGTR1, AK7, ALB, ARL6, ARMC4, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C11orf70, C8orf37, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CD40, CD40LG, CEP164, CEP19, CEP290, CFAP298, CFTR, COPA, CSF2RA, CSF2RB, CXCR4, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, ELANE, FBN3, FLNA, FOXF1, GAS8, GATA2, HSD11B2, IFT172, IFT27, IFT74, IL1RN, INPPL1, ITGA3, KIF7, LRRC56, LRRC6, LZTFL1, MARS, MCIDAS, MKKS, MKS1, MTHFR, MTM1, NDST1, NKX2-1, NME8, NOTCH2, OFD1, PARN, PIEZO2, PIH1D3, REN, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SARS2, SCLT1, SDCCAG8, SFTPB, SFTPC, SLC27A4, SLC34A2, SLC7A7, SPAG1, TERC, TERT, TINF2, TMEM165, TMEM173, TRAPPC3, TRIM32, TTC8, WDPCP, ZMYND10
○ 04113	Invitae Nephronophthisis Panel	27	AHI1, ANKS6, CC2D2A, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PKHD1, RPGRIP1L, SDCCAG8, TCTN1, TMEM216, TMEM237, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423
○ 04103	Invitae Perinatal Lethal Skeletal Dysplasia and Skeletal Ciliopathies Panel	73	AGPS, ALPL, ASCC1, CEP120, CFAP410, CHUK, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CREB3L1, CRTAP, CSPP1, DLL1, DLL3, DMRT2, DYNC2H1, DYNC2LI1, EBP, EVC, EVC2, FAM111A, FGFR2, FGFR3, GNPAT, GPX4, HES7, HSPG2, ICK, IFT122, IFT140, IFT172, IFT43, IFT52, IFT74, IFT80, IFT81, IMPAD1, INPPL1, INTU, KIAA0586, KIAA0753, LBR, LFNG, LRP5, MBTPS2, MESP2, NEK1, P3H1, PAM16, PEX5, PEX7, PPIB, RIPPLY2, SLC26A2, SLC35D1, SOX9, TBX6, TCTEX1D2, TMEM38B, TRAF3IP1, TRIP11, TRIP4, TRPV4, TTC21B, WDR19, WDR34, WDR35, WDR60, WNT1, WNT3
○ 04101	Invitae Primary Ciliary Dyskinesia Panel	42	AK7, ARMC4, C11orf70, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP164, CFAP298, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, GAS8, LRRC56, LRRC6, MCIDAS, NOTCH2, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10

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NEPHROLOGY TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Kidney Disorders			
○ 55005	Invitae Alport Syndrome Panel	6	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9
○ 434341	Invitae Congenital Anomalies of Kidney and Urinary Tract (CAKUT) Panel	41	ACE, AGT, AGTR1, BICC1, BNC2, CRKL, DCHS1, DSTYK, EYA1, FAT4, FGF20, FOXC1, FRAS1, FREM1, FREM2, GATA3, GLI3, GREB1L, HNF1B, HOXA13, HPSE2, ITGA8, LRP4, NRIP1, PAX2, PBX1, REN, RET, ROBO2, SALL1, SIX1, SIX2, SIX5, SLIT2, SOX17, TBX18, UMOD, VIPAS39, VPS33B, WNT4, WT1
○ 55008	Invitae Cystic Kidney Disease Panel	44	ALG8, ANKS6, BICC1, CEP164, CEP290, CEP83, CEP89, COL4A1, CRB2, DCDC2, DICER1, DNAJB11, DZIP1L, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, JAG1, LRP5, MAPKBP1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD2, PKHD1, PRKCSH, RPGRIP1L, SDCCAG8, SEC61A1, SEC63, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, ZNF423
○ 633100	Invitae Expanded Renal Disease Panel	401	ABCC6, ACE, ACTB, ACTN4, ADA2, ADAMTS13, ADCY10, AGPAT2, AGT, AGTR1, AGXT, AHI1, ALG1, ALG8, ALG9, ALMS1, ALPL, AMN, ANKS6, ANLN, ANOS1, AP2S1, APOA1, APOC2, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL13B, ARL6, ATP6V0A4, ATP6V1B1, ATP7B, AVP, AVPR2, B2M, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP4, BNC2, BRAF, BSND, C8orf37, CA2, CACNA1D, CACNA1H, CASR, CBL, CC2D2A, CD151, CD2AP, CDC73, CDKN1C, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP83, CEP89, CFH, CFHR5, CFI, CHD7, CHRM3, CHRNA3, CISD2, CLCN2, CLCN5, CLCNKB, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, COX10, COX14, CPLANE1, CPT2, CRB2, CREBBP, CRKL, CSPP1, CTNS, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, CYP27B1, CYP2R1, DCDC2, DCHS1, DGKE, DHCR7, DHTKD1, DICER1, DLC1, DMP1, DNAJB11, DNASE1L3, DSTYK, DZIP1L, EBP, EGF, EIF2AK3, ELP1, EMP2, ENPP1, EYA1, FAM20A, FAN1, FANCA, FAT1, FAT4, FBN3, FGF10, FGF20, FGF23, FGFR1, FGFR2, FN1, FOXC1, FOXC2, FOXI1, FOXP3, FRAS1, FREM1, FREM2, FXYD2, G6PC, GALNT3, GANAB, GATA3, GATM, GCM2, GLA, GLI3, GLIS2, GLIS3, GNA11, GNAS, GPC3, GPHN, GREB1L, GRHPR, GRIP1, GSN, HGD, HNF1A, HNF1B, HOGA1, HOXA13, HPRT1, HPS1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43, IFT74, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA6, ITGA8, ITGB4, ITSN2, JAG1, KANK1, KANK2, KANK4, KANSL1, KAT6B, KCNA1, KCNJ1, KCNJ10, KCNJ5, KCTD1, KIAA0586, KIF7, KLHL3, KRAS, LAGE3, LAMA5, LAMB2, LCAT, LDHA, LMNA, LMX1B, LPIN1, LRP2, LRP4, LRP5, LYZ, LZTFL1, LZTR1, MAFB, MAGED2, MAGI2, MAP2K1, MAP2K2, MAPKBP1, MEFV, MKKS, MKS1, MMACHC, MNX1, MOCOS, MOCS1, MOCS2, MRE11, MUT, MVK, MYCN, MYH9, MYO1E, NEK8, NF1, NLRP3, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR0B1, NR3C2, NRAS, NRIP1, NSD1, NSDHL, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OFD1, OPLAH, OSGEP, PAX2, PBX1, PCBD1, PDE6D, PDSS1, PDSS2, PEX6, PHEX, PKD2, PKHD1, PLCE1, PLG, PMM2, PODXL, PREPL, PRKCSH, PRODH, PROKR2, PRPS1, PTPN11, PTPRO, RAF1, REN, RET, RIT1, RMND1, ROBO1, ROBO2, RPGRIP1L, RPL11, RPL26, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS7, RRM2B, SALL1, SALL4, SARS2, SCARB2, SCLT1, SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SEMA3E, SGPL1, SHOC2, SI, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC26A1, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC37A4, SLC3A1, SLC41A1, SLC4A1, SLC4A4, SLC5A1, SLC6A19, SLC7A7, SLC7A9, SLC9A3R1, SLIT2, SLX4, SMARCAL1, SMC1A, SOS1, SOS2, SOX17, SOX18, SRCAP, STX16, SYNPO, TBC1D8B, TBX18, TCTN1, TCTN2, TCTN3, TFAP2A, THBD, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNS2, TP53RK, TP63, TPRKB, TRAPPC3, TRIM32, TRPC6, TRPM6, TSC1, TSC2, TTC21B, TTC8, TTR, UMOD, UQCC2, VDR, VHL, VIPAS39, VPS33B, WAS, WDPCP, WDR19, WDR73, WFS1, WNK1, WNK4, WNT4, WNT5A, WT1, XDH, XPNPEP3, XPO5, YRDC, ZNF423
○ 434347	Invitae Nephrotic Syndrome and Focal Segmental Glomerulosclerosis (FSGS) Panel	57	ACTN4, AMN, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, CFH, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DHTKD1, EMP2, FN1, INF2, ITGA3, ITGB4, KANK1, KANK2, KANK4, LAGE3, LAMA5, LAMB2, LMX1B, MAGI2, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OSGEP, PAX2, PDSS2, PLCE1, PODXL, PTPRO, SGPL1, SMARCAL1, TP53RK, TPRKB, TRPC6, WDR73, WT1, XPO5

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NEPHROLOGY TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Kidney Disorders			
○ 75000	Invitae Progressive Renal Disease Panel	195	ACE, ACTB, ACTN4, ADAMTS13, AGT, AGTR1, AHI1, ALG8, ALG9, ALMS1, AMN, ANKS6, ANLN, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL6, ATP6V0A4, ATP6V1B1, AVP, AVPR2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BSND, CA2, CACNA1D, CACNA1H, CASR, CD151, CD2AP, CDKN1C, CEP164, CEP19, CEP290, CEP83, CEP89, CFHR5, CHRM3, CLCN5, CLCNKB, CLDN16, CLDN19, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, CPLANE1, CRB2, CTNS, CUBN, CYP24A1, DCDC2, DGKE, DHCR7, DICER1, DLC1, DNAJB11, DZIP1L, EGF, EMP2, EYA1, FAM20A, FAN1, FAT1, FN1, FOXI1, FOXP3, FREM1, GANAB, GATA3, GATM, GLA, GLIS2, GRHPR, GSN, HNF1A, HNF1B, HPRT1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43, INF2, INVS, IQCB1, ITGA3, ITGA6, ITGB4, JAG1, KANK2, KANK4, KAT6B, KCNJ1, KCNJ10, LAMA5, LAMB2, LCAT, LDHA, LMX1B, LPIN1, LRP4, LRP5, LYZ, LZTFL1, MAGED2, MAGI2, MAPKBP1, MEFV, MKKS, MYH9, MYO1E, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NSDHL, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OFD1, PAX2, PBX1, PDSS2, PHEX, PKD2, PKHD1, PLCE1, PODXL, PREPL, PTPRO, REN, RMND1, RPGRIP1L, SALL1, SALL4, SARS2, SDCCAG8, SEC61A1, SEMA3E, SGPL1, SIX1, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC37A4, SLC41A1, SLC4A1, SLC4A4, SLC7A7, SMARCAL1, SYNPO, TBC1D8B, TMEM67, TNS2, TP63, TRIM32, TRPC6, TSC1, TSC2, TTC21B, TTC8, UMOD, WDPCP, WDR19, WDR73, WT1, XPNPEP3, XPO5, ZNF423
○ 04308	Invitae Renal Tubular Disorders Panel	39	ACE, AGT, AGTR1, AQP2, ATP6V0A4, ATP6V1B1, AVPR2, BSND, CA2, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CUL3, EGF, FOXI1, FXYD2, GATM, GNA11, HSD11B2, KCNJ1, KCNJ10, KLHL3, MAGED2, NR3C2, OCRL, REN, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3, SLC34A1, SLC4A1, SLC4A4, TRPM6, WNK1, WNK4

CLINICAL AREA: NEPHROLITHIASIS

Test code	Test name	# gene(s)	Gene list
Nephrolithiasis			
○ 72037	Invitae Nephrolithiasis Panel	40	ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, ATP7B, CA2, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CYP24A1, FAM20A, FOXI1, GPHN, GRHPR, HOGA1, HPRT1, KCNJ1, MOCOS, MOCS1, MOCS2, OCRL, PEX6, PREPL, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1, UMOD, VDR, XDH

NEPHROLITHIASIS INDIVIDUAL GENES

○ ADCY10	○ ATP6V0A4	○ CLCN5	○ FAM20A	○ HPRT1	○ OCRL	○ SLC26A1	○ SLC3A1	○ UMOD
○ AGXT	○ ATP6V1B1	○ CLDN16	○ GPHN	○ KCNJ1	○ PREPL	○ SLC2A9	○ SLC4A1	○ VDR
○ ALPL	○ CA2	○ CLDN19	○ GRHPR	○ MOCOS	○ SLC12A1	○ SLC34A1	○ SLC7A9	○ XDH
○ APRT	○ CASR	○ CYP24A1	○ HOGA1	○ MOCS1	○ SLC22A12	○ SLC34A3	○ SLC9A3R1	

CLINICAL AREA: NON-MALIGNANT HEMATOLOGY

Test code	Test name	# gene(s)	Gene list
Atypical Hemolytic Uremic Syndrome and Thrombotic Microangiopathies			
○ 55682	Invitae Atypical Hemolytic Uremic Syndrome and Thrombotic Microangiopathies Panel	13	ADAMTS13, C3, CD46, CD55, CD59, CFB, CFH, CFI, DGKE, INF2, MMACHC, PLG, THBD

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NEPHROLOGY TEST CATALOG

CLINICAL AREA: NEPHROLITHIASIS

NON-MALIGNANT HEMATOLOGY INDIVIDUAL GENES LIST

○ F2	○ F9	○ HFE	○ ITGA2B	○ MPL	○ PROC	○ SERPINC1	○ TFR2	○ VHL
○ F5	○ HAMP	○ HJV	○ ITGB3	○ MTHFR	○ PROS1	○ SLC40A1		

CLINICAL AREA: PROGRESSIVE RENAL DISEASE

Test code	Test name	# gene(s)	Gene list
Progressive Renal Disease			
○ 633100	Invitae Expanded Renal Disease Panel	401	ABCC6, ACE, ACTB, ACTN4, ADA2, ADAMTS13, ADCY10, AGPAT2, AGT, AGTR1, AGXT, AHI1, ALG1, ALG8, ALG9, ALMS1, ALPL, AMN, ANKS6, ANLN, ANOS1, AP2S1, APOA1, APOC2, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL13B, ARL6, ATP6V0A4, ATP6V1B1, ATP7B, AVP, AVPR2, B2M, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP4, BNC2, BRAF, BSND, C8orf37, CA2, CACNA1D, CACNA1H, CASR, CBL, CC2D2A, CD151, CD2AP, CDC73, CDKN1C, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP83, CEP89, CFH, CFHR5, CFI, CHD7, CHRM3, CHRNA3, CISD2, CLCN2, CLCN5, CLCNKB, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, COX10, COX14, CPLANE1, CPT2, CRB2, CREBBP, CRKL, CSPP1, CTNS, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, CYP27B1, CYP2R1, DCDC2, DCHS1, DGKE, DHCR7, DHTKD1, DICER1, DLC1, DMP1, DNAJB11, DNASE1L3, DSTYK, DZIP1L, EBP, EGF, EIF2AK3, ELP1, EMP2, ENPP1, EYA1, FAM20A, FAN1, FANCA, FAT1, FAT4, FBN3, FGF10, FGF20, FGF23, FGFR1, FGFR2, FN1, FOXC1, FOXC2, FOXI1, FOXP3, FRAS1, FREM1, FREM2, FXYD2, G6PC, GALNT3, GANAB, GATA3, GATM, GCM2, GLA, GLI3, GLIS2, GLIS3, GNA11, GNAS, GPC3, GPHN, GREB1L, GRHPR, GRIP1, GSN, HGD, HNF1A, HNF1B, HOGA1, HOXA13, HPRT1, HPS1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43, IFT74, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA6, ITGA8, ITGB4, ITSN2, JAG1, KANK1, KANK2, KANK4, KANSL1, KAT6B, KCNA1, KCNJ1, KCNJ10, KCNJ5, KCTD1, KIAA0586, KIF7, KLHL3, KRAS, LAGE3, LAMA5, LAMB2, LCAT, LDHA, LMNA, LMX1B, LPIN1, LRP2, LRP4, LRP5, LYZ, LZTFL1, LZTR1, MAFB, MAGED2, MAGI2, MAP2K1, MAP2K2, MAPKBP1, MEFV, MKKS, MKS1, MMACHC, MNX1, MOCOS, MOCS1, MOCS2, MRE11, MUT, MVK, MYCN, MYH9, MYO1E, NEK8, NF1, NLRP3, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR0B1, NR3C2, NRAS, NRIP1, NSD1, NSDHL, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OFD1, OPLAH, OSGEP, PAX2, PBX1, PCBD1, PDE6D, PDSS1, PDSS2, PEX6, PHEX, PKD2, PKHD1, PLCE1, PLG, PMM2, PODXL, PREPL, PRKCSH, PRODH, PROKR2, PRPS1, PTPN11, PTPRO, RAF1, REN, RET, RIT1, RMND1, ROBO1, ROBO2, RPGRIP1L, RPL11, RPL26, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS7, RRM2B, SALL1, SALL4, SARS2, SCARB2, SCLT1, SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SEMA3E, SGPL1, SHOC2, SI, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC26A1, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC37A4, SLC3A1, SLC41A1, SLC4A1, SLC4A4, SLC5A1, SLC6A19, SLC7A7, SLC7A9, SLC9A3R1, SLIT2, SLX4, SMARCAL1, SMC1A, SOS1, SOS2, SOX17, SOX18, SRCAP, STX16, SYNPO, TBC1D8B, TBX18, TCTN1, TCTN2, TCTN3, TFAP2A, THBD, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNS2, TP53RK, TP63, TPRKB, TRAPPC3, TRIM32, TRPC6, TRPM6, TSC1, TSC2, TTC21B, TTC8, TTR, UMOD, UQCC2, VDR, VHL, VIPAS39, VPS33B, WAS, WDPCP, WDR19, WDR73, WFS1, WNK1, WNK4, WNT4, WNT5A, WT1, XDH, XPNPEP3, XPO5, YRDC, ZNF423
○ 75000	Invitae Progressive Renal Disease Panel	195	ACE, ACTB, ACTN4, ADAMTS13, AGT, AGTR1, AHI1, ALG8, ALG9, ALMS1, AMN, ANKS6, ANLN, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL6, ATP6V0A4, ATP6V1B1, AVP, AVPR2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BSND, CA2, CACNA1D, CACNA1H, CASR, CD151, CD2AP, CDKN1C, CEP164, CEP19, CEP290, CEP83, CEP89, CFHR5, CHRM3, CLCN5, CLCNKB, CLDN16, CLDN19, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, CPLANE1, CRB2, CTNS, CUBN, CYP24A1, DCDC2, DGKE, DHCR7 (continues)

NEPHROLOGY TEST CATALOG

CLINICAL AREA: PROGRESSIVE RENAL DISEASE

Test code	Test name	# gene(s)	Gene list
Progressive Renal Disease (continued)			
	Invitae Progressive Renal Disease Panel (continued)	195	(continued) DICER1, DLC1, DNAJB11, DZIP1L, EGF, EMP2, EYA1, FAM20A, FAN1, FAT1, FN1, FOXI1, FOXP3, FREM1, GANAB, GATA3, GATM, GLA, GLIS2, GRHPR, GSN, HNF1A, HNF1B, HPRT1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43, INF2, INVS, IQCB1, ITGA3, ITGA6, ITGB4, JAG1, KANK2, KANK4, KAT6B, KCNJ1, KCNJ10, LAMA5, LAMB2, LCAT, LDHA, LMX1B, LPIN1, LRP4, LRP5, LYZ, LZTFL1, MAGED2, MAGI2, MAPKBP1, MEFV, MKKS, MYH9, MYO1E, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NSDHL, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OFD1, PAX2, PBX1, PDSS2, PHEX, PKD2, PKHD1, PLCE1, PODXL, PREPL, PTPRO, REN, RMND1, RPGRIP1L, SALL1, SALL4, SARS2, SDCCAG8, SEC61A1, SEMA3E, SGPL1, SIX1, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC37A4, SLC41A1, SLC4A1, SLC4A4, SLC7A7, SMARCAL1, SYNPO, TBC1D8B, TMEM67, TNS2, TP63, TRIM32, TRPC6, TSC1, TSC2, TTC21B, TTC8, UMOD, WDPCP, WDR19, WDR73, WT1, XPNPEP3, XPO5, ZNF423

PROGRESSIVE RENAL DISEASE INDIVIDUAL GENES

○ ACTN4	○ C9orf72	○ COL4A3	○ CYP11B2	○ INF2	○ MYO1E	○ PAX2	○ PKHD1	○ TRPC6
○ AGPAT2	○ CD2AP	○ COL4A4	○ FOXC2	○ ITSN2	○ NPHS1	○ PKD2	○ SLC36A2	○ YRDC
○ APOL1	○ CHRNA3	○ COL4A5	○ HNF1A	○ LMX1B	○ NPHS2			

ENDOCRINOLOGY TEST CATALOG

All tests on this form are organized by clinical area. If your order contains tests from multiple clinical areas, you will need to send a separate specimen for each clinical area. Each clinical area represents an individual billable event and report. Contact Client Services with any questions. For Invitae's full test menu, please visit www.invitae.com.

CLINICAL AREA: NEWBORN SCREENING AND METABOLISM AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Bone Mineralization Disorders			
○ 72038	Invitae X-Linked Hypophosphatemia Test	1	PHEX
○ 72039	Invitae Hypophosphatemia Panel	17	ALPL, CLCN5, CTNS, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, GNAS, OCRL, PHEX, SLC34A1, SLC34A3, VDR
Glucose Metabolism			
○ 98006	Invitae Hypoglycemia Panel	120	AAAS, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACSF3, ADK, AGL, AKT2, ALDOA, ALDOB, ALG3, ALG6, BCKDHA, BCKDHB, CA5A, CACNA1C, CACNA1D, CDKN1C, COG7, CPT1A, CPT2, CYP7B1, DBH, DBT, DDC, DGUOK, DLD, DMXL2, DOLK, ENO3, ETFA, ETFB, ETFDH, FAH, FBP1, FLAD1, G6PC, GAA, GALE, GALK1, GALT, GBE1, GCK, GH1, GHR, GLUD1, GPC3, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HESX1, HK1, HMGCL, HMGCS2, HNF1A, HNF4A, HRAS, HSD3B7, INSR, KCNJ11, KDM6A, KMT2D, LAMP2, LDHA, LHX3, MLYCD, MPI, MPV17, NADK2, NNT, NR0B1, NR3C1, NSD1, OPLAH, OTX2, OXCT1, PC, PCK1, PCK2, PCSK1, PDX1, PFKM, PGAM2, PGM1, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PMM2, POMC, PROP1, PTF1A, PYGL, PYGM, RBCK1, SERAC1, SLC16A1, SLC22A5, SLC25A20, SLC25A32, SLC2A2, SLC37A4, SLC52A1, SLC52A2, SLC52A3, SOX2, SOX3, TAZ, TBX19, TRMT10A, UCP2
○ 55001	Invitae Monogenic Diabetes Panel	28	ABCC8, APPL1, BLK, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, INS, KCNJ11, KLF11, MNX1, NEUROD1, NEUROG3, NKX2-2, PAX4, PDX1, PPARG, PTF1, ARFX6, SLC19A2, WFS1, ZFP57
Obesity			
○ 98001	Invitae Monogenic Obesity Panel	68	ADCY3, AFF4, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BDNF, C8orf37, CEP164, CEP19, CEP290, CPE, CREBBP, CUL4B, DYRK1B, EP300, FBN3, GNAS, GPR101, IFT172, IFT27, IFT74, KDM6A, KIDINS220, KIF7, KMT2D, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, NLGN2, NPY, NR0B2, NTRK2, PCSK1, PHF6, POMC, PPARG, PRMT7, RAB23, RAI1, RPS6KA3, SCLT1, SDCCAG8, SETD2, SH2B1, SIM1, TRAPPC3, TRAPPC9, TRIM32, TTC8, UCP3, VPS13B, WDPCP
Thyroid and Parathyroid Conditions			
○ 01303	Invitae Hyperparathyroidism Panel	7	AP2S1, CASR, CDC73, CDKN1B, GNA11, MEN1, RET
○ 06002	Invitae Hypoparathyroidism Panel	18	AIRE, CASR, CHD7, CYP24A1, FAM111A, GATA3, GCM2, GNA11, GNAS, HADHA, HADHB, PDE4D, PTH, PTH1R, SLC34A1, STX16, TBCE, TBX1

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Disorders of Sex Development/Endocrinology (if available, please provide karyotype information)			
○ 04413	Invitae Disorders of Female Sex Development Test	1	SRY
○ 04412	Invitae Androgen Insensitivity Panel	2	AR, SRD5A2
○ 04736	Invitae Hypogonadotropic Hypogonadism Panel	46	ANOS1, AXL, CCDC141, CHD4, CHD7, CYP19A1, DHCR7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IGSF10, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, LMNA, NR0B1, NSMF, PCSK1, PLXNA1, POLR3B, PROK2, PROKR2, PROP1, RELN, RNF216, SEMA3A, SOX10, SOX2, SPRY4, SRA1, SRY, TAC3, TACR3, WDR11

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INVITAE

Patient's first name

Patient's last name

PEDIATRIC GENETICS, METABOLIC, NEPHROLOGY & ENDOCRINOLOGY

ENDOCRINOLOGY TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Disorders of Sex Development/Endocrinology (if available, please provide karyotype information) (continued)			
○ 55007	Invitae Disorders of Sex Development Panel	53	AMH, AMHR2, ANOS1, AR, ARX, ATRX, B3GLCT, CBX2, CCNQ, CHD7, CKAP2L, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, DHH, DMRT1, ESR2, FRAS1, FREM2, H6PD, HHAT, HOXA13, HSD17B3, HSD3B2, KL, LHCGR, LHX3, LHX4, MAMLD1, MAP3K1, NR0B1, NR5A1, POR, PROP1, PSMC3IP, RSPO1, SOX9, SPECC1L, SRD5A2, SRY, STAR, TNK2, TOE1, TSPYL1, TWIST2, UBR1, WNT4, WNT9B, WT1, WWOX, ZFPM2

ENDOCRINOLOGY INDIVIDUAL GENES

○ AAAS	○ BBS10	○ CTNS	○ FBN3	○ HADHA	○ LEP	○ OPLAH	○ RAI1	○ SRY
○ ABCC8	○ BBS12	○ CUL4B	○ FBP1	○ HADHB	○ LEPR	○ OTX2	○ RBCK1	○ STAR
○ ABCD1	○ BBS2	○ CYB5A	○ FEZF1	○ HESX1	○ LHB	○ OXCT1	○ RELN	○ TAC3
○ ACAD9	○ BBS4	○ CYP11A1	○ FGF17	○ HHAT	○ LHCGR	○ PC	○ RET	○ TACR3
○ ACADM	○ BBS5	○ CYP11B1	○ FGF23	○ HK1	○ LHX3	○ PCK1	○ RNF216	○ TAZ
○ ACADS	○ BBS7	○ CYP17A1	○ FGF8	○ HMGCL	○ LHX4	○ PCK2	○ RPS6KA3	○ TBX19
○ ACADSB	○ BBS9	○ CYP19A1	○ FGFR1	○ HMGCS2	○ LMNA	○ PCSK1	○ RSPO1	○ TNK2
○ ACADVL	○ BCKDHA	○ CYP27B1	○ FLAD1	○ HNF1A	○ LZTFL1	○ PDX1	○ SCLT1	○ TOE1
○ ACAT1	○ BCKDHB	○ CYP2R1	○ FLRT3	○ HNF4A	○ MAGEL2	○ PFKM	○ SDCCAG8	○ TRAPPC3
○ ACSF3	○ BDNF	○ CYP7B1	○ FRAS1	○ HOXA13	○ MAMLD1	○ PGAM2	○ SEMA3A	○ TRAPPC9
○ ADCY3	○ C8orf37	○ DBH	○ FREM2	○ HRAS	○ MAP3K1	○ PGM1	○ SERAC1	○ TRIM32
○ ADK	○ CA5A	○ DBT	○ FSHB	○ HS6ST1	○ MC3R	○ PHEX	○ SETD2	○ TRMT10A
○ AFF4	○ CACNA1C	○ DDC	○ G6PC	○ HSD17B3	○ MC4R	○ PHF6	○ SH2B1	○ TSPYL1
○ AGL	○ CACNA1D	○ DGUOK	○ GAA	○ HSD3B2	○ MEGF8	○ PHGDH	○ SIM1	○ TTC8
○ AKT2	○ CASR	○ DHCR7	○ GALE	○ HSD3B7	○ MEN1	○ PHKA1	○ SLC16A1	○ TWIST2
○ ALDOA	○ CBX2	○ DHH	○ GALK1	○ IFT172	○ MKKS	○ PHKA2	○ SLC22A5	○ UBR1
○ ALDOB	○ CCDC141	○ DLD	○ GALT	○ IFT27	○ MKS1	○ PHKB	○ SLC25A20	○ UCP2
○ ALG3	○ CCNQ	○ DMP1	○ GBE1	○ IFT74	○ MLYCD	○ PHKG2	○ SLC25A32	○ UCP3
○ ALG6	○ CDC73	○ DMRT1	○ GCK	○ IGSF10	○ MPI	○ PLXNA1	○ SLC2A2	○ VDR
○ ALMS1	○ CDKN1B	○ DMXL2	○ GH1	○ IL17RD	○ MPV17	○ PMM2	○ SLC34A1	○ VPS13B
○ ALPL	○ CDKN1C	○ DOLK	○ GHR	○ INSR	○ MRAP2	○ POLR3B	○ SLC34A3	○ WDPCP
○ AMH	○ CEP164	○ DUSP6	○ GLUD1	○ KCNJ11	○ NADK2	○ POMC	○ SLC37A4	○ WDR11
○ AMHR2	○ CEP19	○ DYRK1B	○ GNA11	○ KDM6A	○ NLGN2	○ POR	○ SLC52A1	○ WNT4
○ ANOS1	○ CEP290	○ ENO3	○ GNAS	○ KIDINS220	○ NNT	○ PPARG	○ SLC52A2	○ WNT9B
○ AP2S1	○ CHD4	○ ENPP1	○ GNRH1	○ KIF7	○ NPY	○ PRMT7	○ SLC52A3	○ WT1
○ AR	○ CHD7	○ EP300	○ GNRHR	○ KISS1	○ NR0B1	○ PROK2	○ SOX10	○ WWOX
○ ARL6	○ CKAP2L	○ ERBB4	○ GPC3	○ KISS1R	○ NR0B2	○ PROKR2	○ SOX2	○ ZFPM2
○ ARX	○ CLCN5	○ ESR2	○ GPR101	○ KL	○ NR3C1	○ PROP1	○ SOX3	
○ ATRX	○ COG7	○ ETFA	○ GYG1	○ KLB	○ NR5A1	○ PSMC3IP	○ SOX9	
○ AXL	○ CPE	○ ETFB	○ GYS1	○ KMT2D	○ NSD1	○ PTF1A	○ SPECC1L	
○ B3GLCT	○ CPT1A	○ ETFDH	○ GYS2	○ KSR2	○ NSMF	○ PYGL	○ SPRY4	
○ BBIP1	○ CPT2	○ FAH	○ H6PD	○ LAMP2	○ NTRK2	○ PYGM	○ SRA1	
○ BBS1	○ CREBBP	○ FAM20C	○ HADH	○ LDHA	○ OCRL	○ RAB23	○ SRD5A2	

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WITHIN THE US | p: 800-436-3037 | f: 415-276-4164 | e: clientservices@invitae.com | OUTSIDE THE US | p: +1 415-930-4018 | f: +1 510 417-3922 | e: globalsupport@invitae.com
INVITAE | 1400 16th Street, San Francisco, CA 94103, USA | www.invitae.com/contact |