INVITAE

ORDER ONLINE at www.invitae.com/signin.

ORDER ID
For Invitae internal use only

INVITAE REQUISITION FORM

PATIENT INFORMATION

First name | MI | Last name | Date of birth (MM/DD/YYYY)

Biological sex: ○ Male ○ Female

MRN (medical record number)

Ancestry: ○ Asian ○ Black/African American ○ White/Caucasian ○ Ashkenazi Jewish ○ Hispanic ○ Native American ○ Pacific Islander ○ French Canadian ○ Sephardic Jewish ○ Mediterranean ○ Other: _____

Email address (for billing contact and report access after clinician releases)

Mobile phone (for billing contact)

Address

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

Ship a saliva kit to this patient (to submit this request, fax this completed requisition form to Invitae Client Services at 415-276-4164)

○ Ship kit to address above ○ Ship kit to alternate address: _____

CLINICAL INFORMATION

Organization name	Phone	Fax

Address	City	State/Prov	ZIP/Postal Code	Country

CLINICAL TEAM

Primary clinical contact (contact for general inquires)

Name	NPI	Email address (for report access)

Ordering provider ○ Same as primary clinical contact

For your convenience, we have provided multiple fields below to pre-populate your organization's provider list. For each order, indicate one ordering provider.

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

Additional clinical or laboratory contacts (optional; share online access to this order with the contacts below)

○ Share this order with the primary clinical contact's default clinical team (establish and manage team online at www.invitae.com/signin)

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

○ INSURANCE BILLING (attach front and back of insurance card)

Attach clinical notes, medical records, and/or letter of medical necessity (LMN) to prevent delays. We do not accept insurance for certain tests *or* patients outside the US. www.invitae.com/billing

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

○ PATIENT PAY BILLING

Invitae will send an electronic invoice to the patient email listed above. Insurance will not be billed.

○ INSTITUTIONAL BILLING

Invitae will send an invoice to the organization address above. Please contact Invitae if this order should be billed to a different location.

○ PARTNERSHIP PROGRAMS

Invitae partner code:

INVITAE

Patient's first name

Patient's last name

INVITAE DIAGNOSTIC REQUISITION FORM

SPECIMEN INFORMATION

Label each tube with the patient's full name, date of birth, and specimen collection date. A requisition form MUST accompany each specimen. www.invitae.com/specimen-requirements

Collection date (MM/DD/YYYY)

If not provided, date will be 1 day prior to our receipt of specimen. For DNA, provide date retrieved from archive.

Specimen type

◯ Blood ◯ Saliva ◯ DNA - source: _____

DNA must be extracted in a CLIA or other suitably certified laboratory. 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.

Specimen ID (IB # on tube):

Is this patient deceased? ◯ Yes ◯ No

Deceased date (MM/DD/YYYY)

REASON FOR TESTING

Primary indication:

ONCOLOGY

◯ Hereditary breast and ovarian cancer (HBOC) syndrome
◯ Lynch syndrome
◯ Polyposis (FAP)
◯ Other: _____

CARDIOLOGY

◯ Aortopathy
◯ Cardiomyopathy
◯ Arrhythmia
◯ Other: _____

OTHER

◯ Neurology
◯ Other: _____

ICD-10 codes (required for insurance billing)

PERSONAL HISTORY

Is/was this patient affected or symptomatic†? ◯ Yes ◯ No If yes, describe 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.

Relationship to patient	Maternal or paternal	Diagnosed condition	Age at diagnosis

TEST SELECTION

OPTION 1: SELECT AN INVITAE PANEL FROM OUR TEST CATALOG

Select your desired test(s) from the attached test catalog and discard any pages without a selection.

OPTION 2: INVITAE TEST CODE

Indicate test IDs here (reference www.invitae.com/tests or our test catalog). Test IDs containing add-on codes will include the original panel as well as the add-on.

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

OR

Invitae supports customization of your test. To create a custom panel, log in to your Invitae portal account or contact Client Services. Then indicate the ID associated with that panel here.

Custom panel ID

OPTION 3: 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) _____

Invitae's family follow-up testing analyzes the variant(s) indicated above. If you would like this report to include any variants of uncertain significance and be eligible for re-requisition, please include billing information on this requisition form and check here: ☐

AUTOMATIC REFLEX: Invitae offers one re-requisition at no additional charge for tests within the same clinical area (www.invitae.com/re-requisition). Preschedule it here or in your Invitae portal.

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

Reflex test: **Test code** . **Add-on code (optional)**

By signing this form, the medical professional acknowledges that the individual/ family member authorized to make decisions for the individual (collectively, the "Patient") has been supplied information regarding and consented to undergo genetic testing, substantially as set forth in Invitae's Informed Consent for Genetic Testing (www.invitae.com/forms). For orders originating outside the US, the Patient has been informed their personal information and specimen will be transferred to and processed in the US. The Patient has been informed that Invitae may notify them of clinical updates related to genetic test results (in consultation with the ordering medical professional). If insurance billing is selected, the Patient has further been informed and authorizes Invitae Corporation ("Invitae") and its designees to release information concerning testing to their insurer in order to process and/ or appeal claims. The medical professional agrees to allow Invitae to transfer the information from this requisition to a letter of medical necessity and/or other documentation using the medical professional's name as the signature. For amounts received directly, the Patient has agreed to remit payment to Invitae for testing services rendered. I acknowledge that the Patient has agreed that if the Patient's insurer does not reimburse Invitae in full for any reason, including if the insurer considers the genetic test ordered to be a non-covered service or not medically necessary, then Invitae may bill the Patient directly for the services and the Patient will remit payment directly to Invitae. I acknowledge that I offered pre-test genetic counseling to the Patient, if required by their insurer. I attest that I am authorized under applicable law to order this test.

Medical professional signature (required)	Date (MM/DD/YYYY)

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 specimen tube for each clinical area. Each clinical area represents an individual billable event and report. Please 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	102	AHI1, ANKS6, ARL13B, ARL6, ARMC4, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP298, CPLANE1, CSPP1, DCDC2, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, DYNC2H1, EVC, EVC2, GAS8, GLIS2, IFT122, IFT140, IFT172, IFT80, INPP5E, INVS, IQCB1, KIAA0586, KIF7, LRRC6, MCIDAS, MKKS, MKS1, MRE11A, NEK1, NEK8, NME8, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PKD2, PKHD1, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SDCCAG8, SPAG1, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRIM32, TTC21B, TTC8, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZMYND10, ZNF423
○ 04103	Invitae Skeletal Ciliopathies Panel	17	CEP120, CSPP1, DYNC2H1, EVC, EVC2, IFT80, IFT122, IFT140, IFT172, KIAA0586, NEK1, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60
	○ 04103.1 Add-on FGFR3-related thanatophoric dysplasia gene	3	FGFR1, FGFR2, FGFR3
○ 04101	Invitae Primary Ciliary Dyskinesia Panel	34	ARMC4, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFAP298, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, GAS8, LRRC6, MCIDAS, NME8, OFD1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10
	○ 04101.1 Add-on preliminary-evidence gene	1	INVS
	○ 04101.2 Add-on clinically overlapping gene	1	CFTR
○ 04112	Invitae Bardet-Biedl Syndrome Panel	16	ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP
○ 04111	Invitae Joubert and Meckel-Gruber Syndromes Panel	31	AHI1, ARL13B, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, MRE11A, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423
○ 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
○ 04117	Invitae Oral-Facial-Digital Syndrome Type 1 Test	1	OFD1
○ 04115	Invitae Polycystic Kidney Disease Type 2 Panel	2	PKD2, PKHD1
○ 04114	Invitae Senior-Loken Syndrome Panel	8	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, WDR19
Congenital Heart Disease			
○ 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
	○ 04201.1 Add-on preliminary-evidence genes	7	CFAP52, CRELD1, GATA6, HAND1, MYH6, SMAD6, ZFPM2

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
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
○ 04724	Invitae Branchiootorenal Spectrum Disorders Panel	2	EYA1, SIX1
	○ 04724.1 Add-on Townes-Brocks syndrome gene	1	SALL1
○ 04725	Invitae Carpenter Syndrome Panel	2	MEGF8, RAB23
○ 01732	Invitae CASR-Related Conditions Test	1	CASR
○ 04211	Invitae CHARGE Syndrome Test	1	CHD7
○ 04743	Invitae CHOPS Syndrome Test	1	AFF4
○ 04738	Invitae Coffin-Lowry Syndrome Test	1	RPS6KA3
○ 04737	Invitae Cohen Syndrome Test	1	VPS13B
○ 04727	Invitae Cornelia de Lange Syndrome Panel	6	ANKRD11, HDAC8, NIPBL, RAD21, SMC1A, SMC3
	○ 04727.1 Add-on preliminary-evidence gene	1	EP300
○ 04744	Invitae Glass Syndrome Test	1	SATB2
○ 04730	Invitae GLI3-Related Disorders Test	1	GLI3
○ 04745	Invitae HPRT1-Related Disorders test	1	HPRT1
○ 04736	Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Panel	3	ANOS1, CHD7, FGFR1
○ 04731	Invitae Kabuki Syndrome Panel	2	KDM6A, KMT2D
○ 04746	Invitae KAT6B-Related Disorders Test	1	KAT6B
○ 04747	Invitae KBG Syndrome Test	1	ANKRD11
○ 04716	Invitae MED12-Related Disorders Test	1	MED12
○ 04213	Invitae Oculo-Facio-Cardio-Dental Syndrome Test	1	BCOR
○ 01704	Invitae PTEN-Related Disorders Test	1	PTEN
○ 04748	Invitae Renpenning Syndrome Test	1	PQBP1
○ 04739	Invitae Rubinstein-Taybi Syndrome Panel	2	CREBBP, EP300
○ 01739	Invitae Simpson-Golabi-Behmel Syndrome Test	1	GPC3
○ 04740	Invitae Smith-Lemli-Opitz Syndrome Test	1	DHCR7
○ 04214	Invitae Sotos Syndrome Test	1	NSD1
○ 04735	Invitae van der Woude Syndrome Panel	2	GRHL3, IRF6

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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)			
○ 01716	Invitae von Hippel-Lindau Syndrome Test	1	VHL
○ 04749	Invitae WAGR Syndrome Panel	2	PAX6, WT1
○ 01740	Invitae Weaver Syndrome Test	1	EZH2
Disorders of Sex Development/Endocrinology (if available, please provide karyotype information)			
○ 04411	Invitae Disorders of Male Sex Development Panel	8	AR, DHH, MAP3K1, NR0B1, NR5A1, SRD5A2, SRY, WT1
○ 04411.1	Add-on Kallman syndrome genes	4	ANOS1, CHD7, FGFR1, HESX1
○ 04411.2	Add-on alpha-thalassemia X-linked intellectual disability gene	1	ATRX
○ 04411.3	Add-on campomelic dysplasia gene	1	SOX9
○ 04411.4	Add-on Smith-Lemli-Opitz syndrome gene	1	DHCR7
○ 04413	Invitae Disorders of Female Sex Development Test	1	SRY
○ 04412	Invitae Androgen Insensitivity Panel	2	AR, SRD5A2
○ 04736	Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Panel	3	ANOS1, CHD7, FGFR1
Developmental Brain Abnormalities			
○ 03407	Invitae Alternating Hemiplegia of Childhood Panel	2	ATP1A2, ATP1A3
○ 03407.1	Add-on clinically overlapping genes	3	CACNA1A, SCN1A, SLC2A1
○ 04741	Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel	2	ACTB, ACTG1
○ 04422	Invitae Cerebral Cavernous Malformations Panel	3	CCM2, KRIT1, PDCD10
○ 04211	Invitae CHARGE Syndrome Test	1	CHD7
○ 03402	Invitae Early Infantile Epileptic Encephalopathy Panel	59	ALDH7A1, ARHGEF9, ARX, BRAT1, CACNA2D2, CASK, CDKL5, CHD2, CLCN4, DNM1, DOCK7, EEF1A2, FARS2, FOLR1, FRRS1L, GABBR2, GABRA1, GABRB3, GNAO1, GRIN1, GRIN2A, GRIN2B, HCN1, HNRNPU, IER3IP1, KCNA2, KCNB1, KCNMA1, KCNQ2, KCNQ3, KCNT1, PCDH19, PIGA, PIGN, PIGO, PLCB1, PNKP, PNPO, PURA, SCN1A, SCN2A, SCN8A, SCN9A, SIK1, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SMC1A, SPTAN1, STXBP1, SYNGAP1, SZT2, TBC1D24, WDR45, WWOX
○ 03402.1	Add-on preliminary-evidence genes	10	ARHGEF15, ATP1A2, COQ4, GPHN, KCNH5, MTOR, NECAP1, NEDD4L, SCN1B, ST3GAL3
○ 04424	Invitae Holoprosencephaly Panel	6	FGFR1, GLI2, SHH, SIX3, TGIF1, ZIC2
○ 04424.1	Add-on preliminary-evidence genes	4	CDON, FOXH1, NODAL, PTCH1
○ 03406	Invitae Neurodegeneration with Brain Iron Accumulation Panel	11	ATP13A2, C19orf12, COASY, CP, DCAF17, FTL, FUCA1, PANK2, PLA2G6, SQSTM1, WDR45
○ 03406.1	Add-on preliminary-evidence genes	3	FA2H, KIF1A, TRIM32
○ 03404	Invitae Rett and Angelman Syndromes and Related Disorders Panel	24	ADSL, ALDH5A1, ATRX, CDKL5, CNTNAP2, DYRK1A, EHMT1, FOXG1, GABBR2, IQSEC2, KANSL1, MBD5, MECP2, MEF2C, NGLY1, NRXN1, SATB2, SCN8A, SLC9A6, STXBP1, TCF4, UBE3A, WDR45, ZEB2
○ 03404.1	Add-on preliminary-evidence genes	4	GABRD, HDAC8, TBL1XR1, JMJD1C
○ 01721	Invitae Tuberous Sclerosis Complex Panel	2	TSC1, TSC2

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Eye Disorders			
○ 04722	Invitae Aniridia Test	1	PAX6
○ 04723	Invitae Axenfeld-Rieger Panel	2	FOXC1, PITX2
	○ 04723.1 Add-on aniridia gene	1	PAX6
○ 04112	Invitae Bardet-Biedl Syndrome Panel	16	ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP
○ 04211	Invitae CHARGE Syndrome Test	1	CHD7
○ 05131	Invitae Choroideremia Test	1	CHM
○ 05132	Invitae Congenital Cataracts Panel	34	AGK, BCOR, BFSP1, BFSP2, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, CTDP1, EPHA2, FAM126A, FOXC1, FYCO1, GALK1, GCNT2, GJA3, GJA8, HSF4, MAF, MIP, NHS, OCRL, PAX6, PITX2, PITX3, SIL1, TDRD7, VSX2
	○ 05132.1 Add-on preliminary-evidence genes	4	CHMP4B, CRYGB, LIM2, VIM
○ 04728	Invitae Duane-Radial Ray Syndrome Test	1	SALL4
○ 05133	Invitae Early-Onset Glaucoma Panel	3	CYP1B1, FOXC1, PITX2
○ 05143	Invitae Leber Congenital Amaurosis Panel	19	AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, OTX2, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1
	○ 05143.1 Add-on preliminary-evidence genes	2	BBS4, IMPDH1
○ 05142	Invitae Microphthalmia/Anophthalmia Disorders Panel	17	ALDH1A3, BCOR, BMP4, FOXE3, GDF6, MAB21L2, MFRP, OTX2, PAX2, PRSS56, PXDN, RARB, RAX, SHH, SOX2, STRA6, VSX2
	○ 05142.1 Add-on preliminary-evidence genes	4	GDF3, HESX1, SALL4, VAX1
○ 04213	Invitae Oculo-Facio-Cardio-Dental Syndrome Test	1	BCOR
○ 01738	Invitae Retinoblastoma Test	1	RB1
○ 04114	Invitae Senior-Loken Syndrome Panel	8	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, WDR19
Overgrowth Syndromes			
○ 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
○ 01736	Invitae Perlman Syndrome Test	1	DIS3L2
○ 04502	Invitae Proteus Syndrome Test	1	AKT1
○ 01704	Invitae PTEN-Related Disorders Test	1	PTEN
○ 01739	Invitae Simpson-Golabi-Behmel Syndrome Test	1	GPC3
○ 04214	Invitae Sotos Syndrome Test	1	NSD1
○ 01740	Invitae Weaver Syndrome Test	1	EZH2

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: HYPOPHOSPHATEMIA

The panels below cannot accept DNA as a specimen type (blood and saliva specimens only).

Test code	Test name	# gene(s)	Gene list
Hypophosphatemia			
○ 72038	Invitae X-Linked Hypophosphatemia Test	1	PHEX
○ 72039	Invitae Hypophosphatemia Panel	13	ALPL, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, PHEX, SLC34A3, VDR

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
RASopathies (Noonan spectrum disorders)			
○ 04151	Invitae RASopathies Comprehensive Panel	18	A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1
○ 04163	Invitae Cardio-Facio-Cutaneous Syndrome Panel	6	BRAF, KRAS, MAP2K1, MAP2K2, SHOC2, SOS1
○ 04164	Invitae Costello Syndrome Test	1	HRAS
○ 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
○ 04161	Invitae Noonan Syndrome Panel	14	A2ML1, BRAF, CBL, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, RRAS, SHOC2, SOS1, SOS2
○ 04161.1	Add-on Baraitser-Winter cerebrofrontofacial syndrome genes	2	ACTB, ACTG1
○ 04162	Invitae Noonan Syndrome with Multiple Lentigines Panel	3	BRAF, PTPN11, RAF1
Skeletal Disorders			
○ 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	9	ERF, FGFR1, FGFR2, FGFR3, GLI3, MEGF8, MSX2, RAB23, TWIST1
○ 04423.1	Add-on 3MC and Treacher Collins syndromes genes	2	MASP1, TCOF1
○ 04728	Invitae Duane-Radial Ray Syndrome Test	1	SALL4
○ 04613	Invitae Ellis van Creveld and Weyers Acrofacial Dysostosis Panel	2	EVC, EVC2
○ 04729	Invitae FGFR3-Related Disorders Test	1	FGFR3

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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)			
○ 04614	Invitae Hereditary Multiple Osteochondromas Panel	2	EXT1, EXT2
○ 04614.1	Add-on Langer-Giedion syndrome gene	1	TRPS1
○ 04212	Invitae Holt-Oram Syndrome Test	1	TBX5
○ 04615	Invitae NSDHL-Related Disorders Test	1	NSDHL
○ 04732	Invitae Osteogenesis Imperfecta Panel	4	COL1A1, COL1A2, CRTAP, P3H1
○ 04103	Invitae Skeletal Ciliopathies Panel	17	CEP120, CSPP1, DYNC2H1, EVC, EVC2, IFT80, IFT122, IFT140, IFT172, KIAA0586, NEK1, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60
○ 04103.1	Add-on skeletal dysplasia genes	3	FGFR1, FGFR2, FGFR3
○ 04616	Invitae Thrombocytopenia Absent Radius Syndrome Test	1	RBM8A
○ 04733	Invitae Townes-Brocks Syndrome Test	1	SALL1
○ 04734	Invitae Treacher Collins Syndrome Test	1	TCOF1
○ 04617	Invitae Trichorhinophalangeal Syndrome Panel	2	EXT1, TRPS1
○ 04618	Invitae Ulnar-Mammary Syndrome Test	1	TBX3
Skin Disorders			
○ 04163	Invitae Cardio-Facio-Cutaneous Syndrome Panel	6	BRAF, KRAS, MAP2K1, MAP2K2, SHOC2, SOS1
○ 05021	Invitae Ectodermal Dysplasia with or without Tooth Agenesis Panel	8	EDA, EDAR, EDARADD, LTBP3, MSX1, NFKBIA, PAX9, WNT10A
○ 05021.1	Add-on Clouston syndrome and TP63-related disorder genes	2	GJB6, TP63
○ 04165	Invitae Legius Syndrome Test	1	SPRED1
○ 04165.1	Add-on neurofibromatosis type 1 gene	1	NF1
○ 04162	Invitae Noonan Syndrome with Multiple Lentigines Panel	3	BRAF, PTPN11, RAF1
○ 01704	Invitae PTEN-Related Disorders Test	1	PTEN
○ 05022	Invitae TP63-Related Disorders Test	1	TP63
○ 04735	Invitae van der Woude Syndrome Panel	2	GRHL3, IRF6

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INVITAE

Patient's first name

Patient's last name

PEDIATRIC GENETICS, METABOLIC, AND NEPHROLOGY
FM170-4

PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES

○ A2ML1	○ ATP6AP2	○ CCDC65	○ COL1A2	○ DICER1	○ EVC	○ GATA4	○ IER3IP1
○ ABAT	○ ATRX	○ CCM2	○ COQ4	○ DIS3L2	○ EVC2	○ GATA6	○ IFT122
○ ACTB	○ B9D1	○ CCNO	○ CP	○ DNAAF1	○ EXT1	○ GATM	○ IFT140
○ ACTC1	○ B9D2	○ CDKL5	○ CPA6	○ DNAAF2	○ EXT2	○ GCNT2	○ IFT172
○ ACTG1	○ BBS1	○ CDKN1C	○ CPLANE1	○ DNAAF3	○ EYA1	○ GCSH	○ IFT80
○ ACVR2B	○ BBS10	○ CDON	○ CRB1	○ DNAAF4	○ EZH2	○ GDF1	○ IMPDH1
○ ADSL	○ BBS12	○ CEP104	○ CREBBP	○ DNAAF5	○ FA2H	○ GDF3	○ INPP5E
○ AFF4	○ BBS2	○ CEP120	○ CRELD1	○ DNAH1	○ FAM126A	○ GDF6	○ INVS
○ AGK	○ BBS4	○ CEP164	○ CRTAP	○ DNAH11	○ FARS2	○ GJA1	○ IQCB1
○ AHI1	○ BBS5	○ CEP290	○ CRX	○ DNAH5	○ FASN	○ GJA3	○ IQSEC2
○ AIPL1	○ BBS7	○ CEP41	○ CRYAA	○ DNAH8	○ FGFR1	○ GJA8	○ IRF6
○ AKT1	○ BBS9	○ CEP83	○ CRYAB	○ DNAI1	○ FGFR2	○ GJB6	○ ITPA
○ AKT2	○ BCOR	○ CERS1	○ CRYBA1	○ DNAI2	○ FGFR3	○ GLDC	○ JAG1
○ AKT3	○ BFSP1	○ CFAP52	○ CRYBA4	○ DNAJC5	○ FLNA	○ GLI2	○ JMJD1C
○ ALDH1A3	○ BFSP2	○ CFAP53	○ CRYBB1	○ DNAL1	○ FOLR1	○ GLI3	○ KANSL1
○ ALDH5A1	○ BMP4	○ CFAP298	○ CRYBB2	○ DNM1	○ FOXC1	○ GLIS2	○ KAT6B
○ ALDH7A1	○ BRAF	○ CFTR	○ CRYBB3	○ DNMT3A	○ FOXE3	○ GLRA1	○ KCNA1
○ ALG13	○ BRAT1	○ CHD2	○ CRYGB	○ DOCK7	○ FOXG1	○ GNAO1	○ KCNA2
○ ALMS1	○ C12orf57	○ CHD7	○ CRYGC	○ DRC1	○ FOXH1	○ GOSR2	○ KCNB1
○ AMT	○ C19orf12	○ CHM	○ CRYGD	○ DYNC1H1	○ FRRS1L	○ GPC3	○ KCNC1
○ ANKRD11	○ CACNA1A	○ CHMP4B	○ CRYGS	○ DYNC2H1	○ FTL	○ GPHN	○ KCND2
○ ANKS6	○ CACNA1H	○ CHRNA2	○ CSPP1	○ DYRK1A	○ FUCA1	○ GRHL3	○ KCNH2
○ ANOS1	○ CACNA2D2	○ CHRNA4	○ CSTB	○ EDA	○ FYCO1	○ GRIN1	○ KCNH5
○ AR	○ CACNB4	○ CHRNB2	○ CTDP1	○ EDAR	○ GABBR2	○ GRIN2A	○ KCNJ10
○ ARHGEF15	○ CARS2	○ CLCN4	○ CTRC	○ EDARADD	○ GABRA1	○ GRIN2B	○ KCNJ13
○ ARHGEF9	○ CASK	○ CLN2 (TPP1)	○ CTSD	○ EED	○ GABRA6	○ GUCY2D	○ KCNMA1
○ ARL13B	○ CASR	○ CLN3	○ CUL4B	○ EEF1A2	○ GABRB2	○ HAND1	○ KCNQ2
○ ARL6	○ CBL	○ CLN5	○ CYP1B1	○ EFHC1	○ GABRB3	○ HCN1	○ KCNQ3
○ ARMC4	○ CC2D2A	○ CLN6	○ DCAF17	○ EHMT1	○ GABRD	○ HDAC8	○ KCNT1
○ ARSE	○ CCDC103	○ CLN8	○ DCDC2	○ ELN	○ GABRG2	○ HESX1	○ KCTD7
○ ARX	○ CCDC114	○ CNTN2	○ DEPDC5	○ EP300	○ GAL	○ HNRNPU	○ KDM6A
○ ATP13A2	○ CCDC151	○ CNTNAP2	○ DHCR7	○ EPHA2	○ GALK1	○ HPRT1	○ KIAA0586
○ ATP1A2	○ CCDC39	○ COASY	○ DHH	○ EPM2A	○ GAMT	○ HRAS	○ KIF1A
○ ATP1A3	○ CCDC40	○ COL1A1	○ DIAPH1	○ ERF	○ GAS8	○ HSF4	○ KIF7

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES (continued)

○ KMT2D	○ MRE11A	○ NRAS	○ PNKD	○ RDH12	○ SHOC2	○ SRPX2	○ TSC1
○ KPNA7	○ MSX1	○ NRXN1	○ PNKP	○ RELN	○ SIK1	○ SRY	○ TSC2
○ KPTN	○ MSX2	○ NSD1	○ PNPO	○ RIT1	○ SIL1	○ ST3GAL3	○ TTC21B
○ KRAS	○ MTOR	○ NSDHL	○ POLG	○ RNF125	○ SIX1	○ ST3GAL5	○ TTC8
○ KRIT1	○ MYH6	○ NUS1	○ POR	○ ROGDI	○ SIX3	○ STRA6	○ TULP1
○ LCA5	○ NECAP1	○ OCRL	○ PPT1	○ RPE65	○ SLC12A5	○ STRADA	○ TWIST1
○ LEFTY2	○ NEDD4L	○ OFD1	○ PQBP1	○ RPGR	○ SLC13A5	○ STX1B	○ UBE3A
○ LGI1	○ NEK1	○ OTX2	○ PRDM8	○ RPGRIP1	○ SLC19A3	○ STXBP1	○ UPF3B
○ LIAS	○ NEK8	○ P3H1	○ PRICKLE1	○ RPGRIP1L	○ SLC25A12	○ SYN1	○ VAX1
○ LIM2	○ NEXMIF	○ PACS1	○ PRICKLE2	○ RPS6KA3	○ SLC25A22	○ SYNGAP1	○ VHL
○ LMNB2	○ NF1	○ PANK2	○ PRIMA1	○ RRAS	○ SLC2A1	○ SYNJ1	○ VIM
○ LRAT	○ NF2	○ PAX2	○ PRPH2	○ RSPH1	○ SLC35A2	○ SZT2	○ VPS13B
○ LRRC6	○ NFIX	○ PAX6	○ PRRT2	○ RSPH3	○ SLC35A3	○ TBC1D24	○ VSX2
○ LTBP3	○ NFKBIA	○ PAX9	○ PRSS1	○ RSPH4A	○ SLC6A1	○ TBL1XR1	○ WDPCP
○ MAB21L2	○ NGLY1	○ PCDH19	○ PRSS56	○ RSPH9	○ SLC6A8	○ TBX1	○ WDR19
○ MAF	○ NHLRC1	○ PDCD10	○ PTCH1	○ RYR3	○ SLC9A6	○ TBX3	○ WDR34
○ MAP2K1	○ NHS	○ PDE6D	○ PTEN	○ SALL1	○ SMAD6	○ TBX5	○ WDR35
○ MAP2K2	○ NIPBL	○ PDGFRB	○ PTPN11	○ SALL4	○ SMC1A	○ TCF4	○ WDR45
○ MAP3K1	○ NKX2-5	○ PHF6	○ PURA	○ SATB2	○ SMC3	○ TCOF1	○ WDR60
○ MASP1	○ NKX2-6	○ PIGA	○ PXDN	○ SCARB2	○ SNAP25	○ TCTN1	○ WNT10A
○ MBD5	○ NME8	○ PIGG	○ QARS	○ SCN1A	○ SNX27	○ TCTN2	○ WT1
○ MCIDAS	○ NMNAT1	○ PIGN	○ RAB23	○ SCN1B	○ SOS1	○ TCTN3	○ WWOX
○ MECP2	○ NODAL	○ PIGO	○ RAD21	○ SCN2A	○ SOS2	○ TDRD7	○ XPNPEP3
○ MED12	○ NOTCH1	○ PIGQ	○ RAF1	○ SCN3A	○ SOX2	○ TGIF1	○ ZDHHC9
○ MED13L	○ NOTCH2	○ PIK3AP1	○ RANBP2	○ SCN5A	○ SOX9	○ TMEM138	○ ZEB2
○ MEF2C	○ NPHP1	○ PIK3CA	○ RARB	○ SCN8A	○ SPAG1	○ TMEM216	○ ZFPM2
○ MEGF8	○ NPHP3	○ PIK3R2	○ RASA1	○ SCN9A	○ SPATA5	○ TMEM231	○ ZIC2
○ MEIS2	○ NPHP4	○ PITX2	○ RAX	○ SDCCAG8	○ SPATA7	○ TMEM237	○ ZIC3
○ MFRP	○ NPR2	○ PITX3	○ RB1	○ SEMA3E	○ SPINK1	○ TMEM67	○ ZMYND10
○ MFSD8	○ NPRL3	○ PKD2	○ RBFOX1	○ SERPINI1	○ SPRED1	○ TP63	○ ZNF423
○ MIP	○ NR0B1	○ PKHD1	○ RBFOX3	○ SETD2	○ SPTAN1	○ TPK1	
○ MKKS	○ NR2F2	○ PLA2G6	○ RBM8A	○ SGCE	○ SQSTM1	○ TRIM32	
○ MKS1	○ NR5A1	○ PLCB1	○ RD3	○ SHH	○ SRD5A2	○ TRPS1	

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INVITAE

Patient's first name

Patient's last name

PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: EPILEPSY

Test code	Test name		# gene(s)	Gene list
Epilepsy and Seizures				
○ 03401	Invitae Epilepsy Panel		151	ADSL, ALDH5A1, ALDH7A1, ALG13, ARG1, ARHGEF9, ARX, ATP1A2, ATP1A3, ATRX, BRAT1, C12orf57, CACNA1A, CACNA2D2, CARS2, CASK, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN4, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CNTNAP2, CSTB, CTSD, DDC, DEPDC5, DNAJC5, DNM1, DOCK7, DYRK1A, EEF1A2, EFHC1, EHMT1, EPM2A, FARS2, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GLRA1, GNAO1, GOSR2, GRIN1, GRIN2A, GRIN2B, HCN1, HNRNPU, IER3IP1, IQSEC2, ITPA, JMJD1C, KANSL1, KCNA2, KCNB1, KCNC1, KCNH2, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, LGI1, LIAS, MBD5, MECP2, MEF2C, MFSD8, MOCS1, MOCS2, MTOR, NEDD4L, NEXMIF, NGLY1, NHLRC1, NPRL3, NRXN1, PACS1, PCDH19, PIGA, PIGN, PIGO, PLCB1, PNKD, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRIMA1, PRRT2, PURA, QARS, RELN, ROGDI, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SERPINI1, SGCE, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SNX27, SPATA5, SPTAN1, ST3GAL5, STRADA, STX1B, STXBP1, SUOX, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TPK1, TSC1, TSC2, UBE3A, WDR45, WWOX, ZDHHC9, ZEB2
	○ 03401.1	Add-on preliminary-evidence genes	35	ABAT, ARHGEF15, ATP6AP2, CACNA1H, CACNB4, CASR, CERS1, CNTN2, CPA6, DIAPH1, FASN, GABRD, GAL, GPHN, KCNA1, KCND2, KCNH5, KPNA7, LMNB2, NECAP1, PIGG, PIGQ, PIK3AP1, PRDM8, PRICKLE2, RBFOX1, RBFOX3, RYR3, SCN5A, SETD2, SLC35A3, SNAP25, SRPX2, ST3GAL3, TBL1XR1
	○ 03401.2	Add-on genes for glycine encephalopathy	3	AMT, GCSH, GLDC
	○ 03401.3	Add-on FLNA gene	1	FLNA
	○ 03401.4	Add-on PTEN gene	1	PTEN
	○ 03401.5	Add-on RANBP2 gene	1	RANBP2

EPILEPSY AND SEIZURES INDIVIDUAL GENES

○ ABAT	○ CACNA1A	○ CLN5	○ EFHC1	○ GAMT	○ JMJD1C	○ LGI1	○ NRXN1
○ ADSL	○ CACNA1H	○ CLN6	○ EHMT1	○ GATM	○ KANSL1	○ LIAS	○ PACS1
○ ALDH5A1	○ CACNA2D2	○ CLN8	○ EPM2A	○ GCSH	○ KCNA1	○ LMNB2	○ PCDH19
○ ALDH7A1	○ CACNB4	○ CNTN2	○ FARS2	○ GLDC	○ KCNA2	○ MBD5	○ PIGA
○ ALG13	○ CARS2	○ CNTNAP2	○ FASN	○ GLRA1	○ KCNB1	○ MECP2	○ PIGG
○ AMT	○ CASK	○ CPA6	○ FLNA	○ GNAO1	○ KCNC1	○ MEF2C	○ PIGN
○ ARG1	○ CASR	○ CSTB	○ FOLR1	○ GOSR2	○ KCND2	○ MFSD8	○ PIGO
○ ARHGEF15	○ CDKL5	○ CTSD	○ FOXG1	○ GPHN	○ KCNH2	○ MOCS1	○ PIGQ
○ ARHGEF9	○ CERS1	○ DDC	○ FRRS1L	○ GRIN1	○ KCNH5	○ MOCS2	○ PIK3AP1
○ ARX	○ CHD2	○ DEPDC5	○ GABBR2	○ GRIN2A	○ KCNJ10	○ MTOR	○ PLCB1
○ ATP1A2	○ CHRNA2	○ DIAPH1	○ GABRA1	○ GRIN2B	○ KCNMA1	○ NECAP1	○ PNKD
○ ATP1A3	○ CHRNA4	○ DNAJC5	○ GABRB2	○ HCN1	○ KCNQ2	○ NEDD4L	○ PNKP
○ ATP6AP2	○ CHRNB2	○ DNM1	○ GABRB3	○ HNRNPU	○ KCNQ3	○ NEXMIF	○ PNPO
○ ATRX	○ CLCN4	○ DOCK7	○ GABRD	○ IER3IP1	○ KCNT1	○ NGLY1	○ POLG
○ BRAT1	○ CLN2 (TPP1)	○ DYRK1A	○ GABRG2	○ IQSEC2	○ KCTD7	○ NHLRC1	○ PPT1
○ C12orf57	○ CLN3	○ EEF1A2	○ GAL	○ ITPA	○ KPNA7	○ NPRL3	○ PRDM8

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PEDIATRIC GENETICS TEST CATALOG

CLINICAL AREA: EPILEPSY

EPILEPSY AND SEIZURES INDIVIDUAL GENES (continued)

○ PRICKLE1	○ RBFOX1	○ SCN1B	○ SGCE	○ SLC35A2	○ SPATA5	○ SUOX	○ TPK1
○ PRICKLE2	○ RBFOX3	○ SCN2A	○ SIK1	○ SLC35A3	○ SPTAN1	○ SYN1	○ TSC1
○ PRIMA1	○ RELN	○ SCN3A	○ SLC12A5	○ SLC6A1	○ SRPX2	○ SYNGAP1	○ TSC2
○ PRRT2	○ ROGDI	○ SCN5A	○ SLC13A5	○ SLC6A8	○ ST3GAL3	○ SYNJ1	○ UBE3A
○ PTEN	○ RYR3	○ SCN8A	○ SLC19A3	○ SLC9A6	○ ST3GAL5	○ SZT2	○ WDR45
○ PURA	○ SATB2	○ SCN9A	○ SLC25A12	○ SMC1A	○ STRADA	○ TBC1D24	○ WWOX
○ QARS	○ SCARB2	○ SERPINI1	○ SLC25A22	○ SNAP25	○ STX1B	○ TBL1XR1	○ ZDHHC9
○ RANBP2	○ SCN1A	○ SETD2	○ SLC2A1	○ SNX27	○ STXBP1	○ TCF4	○ ZEB2

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			
○ 01106	Invitae Pediatric Nervous System/Brain Tumors Panel	26	AIP, ALK, APC, DICER1, EPCAM, HRAS, LZTR1, MEN1, MLH1, MSH2, MSH6, NF1, NF2, PHOX2B, PMS2, PRKAR1A, PTCH1, PTEN, RB1, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL
	○ 01106.1 Add-on hereditary paraganglioma-pheochromocytoma genes	8	MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127
○ 01104	Invitae Pediatric Solid Tumors Panel	53	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, 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
○ 01105	Invitae Pediatric Hematologic Malignancies Panel	16	ATM, BLM, CEBPA, EPCAM, GATA2, HRAS, MLH1, MSH2, MSH6, NBN, NF1, PMS2, RUNX1, TERC, TERT, TP53

PEDIATRIC ONCOLOGY INDIVIDUAL GENES

○ AIP	○ CDC73	○ EXT1	○ MLH1	○ PRSS1	○ SDHAF2	○ STK11	○ WRN
○ ALK	○ CDKN1C	○ EXT2	○ MSH2	○ PTCH1	○ SDHB	○ SUFU	○ WT1
○ APC	○ CEBPA	○ FH	○ MSH6	○ PTEN	○ SDHC	○ TERC	
○ ATM	○ CFTR	○ GATA2	○ NBN	○ RB1	○ SDHD	○ TERT	
○ AXIN2	○ CPA1	○ GPC3	○ NF1	○ RECQL4	○ SMAD4	○ TMEM127	
○ BAP1	○ CTRC	○ HRAS	○ NF2	○ REST	○ SMARCA4	○ TP53	
○ BLM	○ DICER1	○ LZTR1	○ PHOX2B	○ RET	○ SMARCB1	○ TSC1	
○ BMPR1A	○ DIS3L2	○ MAX	○ PMS2	○ RUNX1	○ SMARCE1	○ TSC2	
○ CASR	○ EPCAM	○ MEN1	○ PRKAR1A	○ SDHA	○ SPINK1	○ VHL	

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INVITAE

Patient's first name

Patient's last name

PEDIATRIC GENETICS, METABOLIC, AND NEPHROLOGY
FM170-4

METABOLIC 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 specimen tube for each clinical area. Each clinical area represents an individual billable event and report. Please contact Client Services with any questions. For Invitae's full test menu, please visit www.invitae.com.

CLINICAL AREA: HYPOPHOSPHATEMIA

The panels below cannot accept DNA as a specimen type (blood and saliva specimens only).

Test code	Test name	# gene(s)	Gene list
Hypophosphatemia			
○ 72038	Invitae X-Linked Hypophosphatemia Test	1	PHEX
○ 72039	Invitae Hypophosphatemia Panel	13	ALPL, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, PHEX, SLC34A3, VDR

CLINICAL AREA: PORPHYRIAS

Test code	Test name	# gene(s)	Gene list
Porphyrias			
○ 06226	Invitae Acute Hepatic Porphyrias Panel	4	ALAD, CPOX, HMBS, PPOX

PORPHYRIA INDIVIDUAL GENES

○ ALAD	○ CPOX	○ HMBS	○ PPOX

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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METABOLIC TEST CATALOG

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test code	Test name		# gene(s)	Gene list
Metabolic Disorders Newborn Screening Confirmation				
○ 06102	Invitae Metabolic Disorders Newborn Screening Confirmation Panel		93	ABCD1, ABCD4, ACAD8, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACSF3, AHCY, ALDH4A1, ARG1, ASL, ASS1, AUH, BCKDHA, BCKDHB, BTD, CBS, CD320, CFTR, CPS1, CPT1A, CPT2, DBT, DNAJC19, ETFA, ETFB, ETFDH, ETHE1, FAH, FTCD, G6PD, GAA, GALE, GALK1, GALT, GCDH, GCH1, GLA, GNMT, GSS, HADH, HADHA, HADHB, HCFC1, HLCS, HMGCL, HPD, HSD17B10, IDUA, IVD, LMBRD1, MAT1A, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, NAGS, OAT, OPA3, OTC, PAH, PCBD1, PC, PCCA, PCCB, PPM1K, PRODH, PTS, QDPR, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC52A1, SLC52A2, SLC52A3, SMPD1, SPR, SUCLA2, SUCLG1, TAT, TAZ, TMEM70
	○ 06102.01	Add-on 2,4-dienoyl-CoA reductase deficiency genes	1	NADK2
	○ 06102.02	Add-on cerebral creatine deficiency genes	3	GAMT, GATM, SLC6A8
	○ 06102.03	Add-on congenital disorders of glycosylation genes	102	ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, C1GALT1C1, CHST14, CHST3, CHST6, CHSY1, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, DSE, EOGT, EXT1, EXT2, FKRP, FKTN, G6PC3, GALNT3, GFPT1, GMPPA, GMPPB, GNE, ISPD, LARGE1, LFNG, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, NUS1, PAPSS2, PGM1, PGM3, PIGA, PIGL, PIGM, PIGN, PIGO, PIGQ, PIGT, PIGV, PIGW, PMM2, POFUT1, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RFT1, RPN2, RXYLT1, SEC23A, SEC23B, SLC26A2, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, TMEM165, TRIP11, TUSC3, XYLT1
	○ 06102.04	Add-on generalized leukodystrophies genes	6	ARSA, ASPA, GALC, GM2A, HEXA, HEXB
	○ 06102.05	Add-on glucose transporter type 1 (GLUT1) deficiency gene	1	SLC2A1
	○ 06102.06	Add-on glycine encephalopathy genes	6	AMT, GCSH, GLDC, LIAS, NFU1, SLC6A9
	○ 06102.07	Add-on mucopolysaccharidosis type II (MPSII) gene	1	IDS
	○ 06102.08	Add-on Niemann-Pick type C genes	2	NPC1, NPC2
	○ 06102.09	Add-on pyridoxal 5'-phosphate-dependent epilepsy gene	1	PNPO
	○ 06102.10	Add-on pyridoxine-responsive epilepsy gene	1	ALDH7A1
	○ 06102.11	Add-on Smith-Lemli-Opitz syndrome gene	1	DHCR7
	○ 06102.12	Add-on cerebrotendinous xanthomatosis gene	1	CYP27A1
	○ 06102.13	Add-on 3-hydroxy-3-methylglutaryl-CoA (HMG-CoA) lyase deficiency gene	1	HMGCS2
	○ 06102.14	Add-on neuronal ceroid lipofuscinosis genes	10	ATP13A2, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1
	○ 06102.15	Add-on succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency gene	1	OXCT1
	○ 06102.16	Add-on elevated very long chain fatty acids genes	14	ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26
○ 06171	Invitae Lysosomal Storage Disorders Newborn Screening Panel		6	GAA, GALC, GLA, IDS, IDUA, SMPD1
○ 06210	Invitae X-Linked Adrenoleukodystrophy Newborn Screening Confirmation Test		1	ABCD1
	○ 06210.1	Add on peroxisomal acyl-CoA oxidase (ACOX1) deficiency gene	1	ACOX1
	○ 06210.2	Add-on elevated very long chain fatty acids genes	13	HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26

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METABOLIC TEST CATALOG

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Panels by Analyte			
○ 06103	Invitae Low C0 Test	1	SLC22A5
○ 06104	Invitae Elevated C0/(C16+C18) Test	1	CPT1A
○ 06105	Invitae Elevated C3 Panel	15	ABCD4, BTD, CD320, HCFC1, HLCS, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MUT, PCCA, PCCB, TCN2
	○ 06105.1 Add-on ACSF3 gene	1	ACSF3
○ 06106	Invitae Elevated C3-DC Test	1	MLYCD
○ 06107	Invitae Elevated C4 Panel	3	ACAD8, ACADS, ETHE1
	○ 06107.1 Add-on limited evidence gene	1	FTCD
○ 06127	Invitae Elevated C4-DC Panel	2	SUCLA2, SUCLG1
○ 06108	Invitae Elevated C4-OH Panel	2	HADH, HIBCH
○ 06109	Invitae Elevated C4 & C5 Panel	7	ETFA, ETFB, ETFDH, ETHE1, SLC52A1, SLC52A2, SLC52A3
○ 06110	Invitae Elevated C5 Panel	2	ACADSB, IVD
○ 06111	Invitae Elevated C5-DC Test	1	GCDH
○ 06112	Invitae Elevated C5-OH Panel	13	ACAT1, AUH, BTD, CLPB, DNAJC19, HLCS, HMGCL, HSD17B10, MCCC1, MCCC2, OPA3, SERAC1, TAZ
○ 06113	Invitae Elevated C6, C8 & C10 Test	1	ACADM
○ 06114	Invitae Elevated C14 & C14:1 Test	1	ACADVL
○ 06115	Invitae Elevated C16-OH, C16:1-OH, C18-OH & C18:1-OH Panel	2	HADHA, HADHB
○ 06116	Invitae Elevated C16, C16:1, C18, & C18:1 Panel	2	CPT2, SLC25A20
○ 06117	Invitae Elevated Arginine Test	1	ARG1
○ 06118	Invitae Elevated Citrulline Panel	4	ASL, ASS1, PC, SLC25A13
	○ 06118.1 Add-on dihydrolipoamide dehydrogenase deficiency gene	1	DLD
○ 06123	Invitae Low Citrulline Panel	3	CPS1, NAGS, OTC
	○ 06123.1 Add-on limited evidence gene	1	ALDH18A1
○ 06124	Invitae Elevated Glycine Panel	6	AMT, GLDC, GCSH, LIAS, NFU1, SLC6A9
	○ 06124.1 Add-on organic acidemia genes	56	ACAD8, ACADSB, ACAT1, ACSF3, ASPA, AUH, BCKDHA, BCKDHB, BTD, D2HGDH, DBT, DHTKD1, DLD, DNAJC19, ETFA, ETFB, ETFDH, ETHE1, FBP1, FH, FTCD, GCDH, GSS, HIBCH, HLCS, HMGCL, HSD17B10, IDH2, IVD, L2HGDH, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MUT, NFU1, OGDH, OPA3, OPLAH, OXCT1, PCCA, PCCB, POLG, PPM1K, SERAC1, SLC13A5, SLC25A1, SLC25A19, SUCLA2, SUCLG1, TAZ, TMEM70
○ 06119	Invitae Elevated Leucine Panel	5	BCKDHA, BCKDHB, DBT, DLD, PPM1K
○ 06125	Invitae Elevated Methionine Panel	4	AHCY, CBS, GNMT, MAT1A
	○ 06125.1 Add-on additional causes of elevated methionine genes	2	FAH, SLC25A13
○ 06120	Invitae Elevated Phenylalanine Panel	6	GCH1, PAH, PCBD1, PTS, QDPR, SPR

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METABOLIC TEST CATALOG

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Panels by Analyte (continued)			
○ 06121	Invitae Elevated Proline Panel	2	ALDH4A1, PRODH
○ 06122	Invitae Elevated Succinylacetone Test	1	FAH
○ 06126	Invitae Elevated Tyrosine Panel	3	FAH, HPD, TAT
Aminoacidopathies			
○ 06140	Invitae Alkaptonuria Test	1	HGD
○ 06141	Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
○ 06142	Invitae Cystinuria Panel	3	PREPL, SLC3A1, SLC7A9
○ 06148	Invitae Disorders of Serine Biosynthesis Panel	3	PHGDH, PSAT1, PSPH
○ 06143	Invitae Glycine Encephalopathy Panel	6	AMT, GCSH, GLDC, LIAS, NFU1, SLC6A9
○ 06144	Invitae Homocystinuria Panel	4	CBS, MTHFR, MTR, MTRR
○ 06144.1	Add-on combined methylmalonic acidemia and homocystinuria genes	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
○ 06144.2	Add-on elevated methionine genes	4	AHCY, CBS, GNMT, MAT1A
○ 06145	Invitae Hyperphenylalaninemia Panel	6	GCH1, PAH, PCBD1, PTS, QDPR, SPR
○ 06146	Invitae Hyperprolinemia Panel	2	ALDH4A1, PRODH
○ 06147	Invitae Maple Syrup Urine Disease Panel	4	BCKDHA, BCKDHB, DBT, PPM1K
○ 06147.1	Add-on DLD gene	1	DLD
○ 06149	Invitae Tyrosinemia Panel	3	FAH, HPD, TAT
Carbohydrate Disorders			
○ 06152	Invitae Galactosemia Panel	3	GALE, GALK1, GALT
○ 06153	Invitae Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency Test	1	G6PD
○ 06154	Invitae Glucose Transporter Type 1 (GLUT1) Deficiency Syndrome Test	1	SLC2A1
○ 06156	Invitae Comprehensive Glycogen Storage Disease Panel	23	AGL, ALDOA, ENO3, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKA2, PHKB, PHKG2, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4
○ 06156.1	Add-on fatty acid oxidation genes	21	ACADM, ACADS, ACADSB, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, NADK2, SLC22A5, SLC25A20, SLC52A1, SLC52A2, SLC52A3
○ 06156.2	Add-on limited evidence genes	2	PGM1, POLG
○ 06157	Invitae Liver Glycogen Storage Disease Panel	11	AGL, FBP1, G6PC, GBE1, GYS2, PHKA2, PHKB, PHKG2, PYGL, SLC2A2, SLC37A4
○ 06158	Invitae Muscle Glycogen Storage Disease Panel	14	ALDOA, ENO3, GAA, GBE1, GYG1, GYS1, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKB, PYGM, RBCK1
○ 06158.1	Add-on fatty acid oxidation genes	21	ACADM, ACADS, ACADSB, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, NADK2, SLC22A5, SLC25A20, SLC52A1, SLC52A2, SLC52A3
○ 06158.2	Add-on limited evidence genes	2	PGM1, POLG
○ 06159	Invitae Hereditary Fructose Intolerance Test	1	ALDOB
○ 06160	Invitae Rare Carbohydrate Disorders Panel	2	FBP1, SLC5A1

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METABOLIC TEST CATALOG

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test code	Test name		# gene(s)	Gene list
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		50	ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, B3GLCT, CHST14, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, G6PC3, GFPT1, GMPPA, GMPPB, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PGM3, PMM2, RFT1, SEC23B, SLC35A1, SLC35A2, SLC35C1, SRD5A3, SSR4, ST3GAL5, TMEM165, TRIP11, TUSC3
	○ 06155.1	Add-on preliminary-evidence genes	11	ALG14, B4GALT1, DDOST, NUS1, PIGM, RPN2, SEC23A, SLC35A3, ST3GAL3, STT3A, STT3B
	○ 06155.2	Add-on disorders of O-mannosylation genes	13	B3GALNT2, B4GAT1, FKRP, FKTN, GNE, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1
	○ 06155.3	Add-on glycosylation genes not involved in N-glycosylation	29	B3GALT6, B3GAT3, B4GALNT1, B4GALT7, C1GALT1C1, CHST3, CHST6, CHSY1, DSE, EOGT, EXT1, EXT2, GALNT3, LFNG, PAPSS2, PIGA, PIGL, PIGM, PIGN, PIGO, PIGQ, PIGT, PIGV, PIGW, POFUT1, POGLUT1, SLC26A2, SLC35D1, XYLT1
Creatine Biosynthesis Disorders				
○ 06162	Invitae Cerebral Creatine Deficiency Panel		3	GAMT, GATM, SLC6A8
Cystic Fibrosis				
○ 06220	Invitae Cystic Fibrosis Newborn Screening Confirmation Test		1	CFTR
Fatty Acid Oxidation Defects				
○ 06165	Invitae Fatty Acid Oxidation Defects Panel		17	ACADM, ACADS, ACADSB, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, SLC22A5, SLC25A20
	○ 06165.1	Add-on 2,4-dienoyl-CoA reductase deficiency gene	1	NADK2
	○ 06165.2	Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
	○ 06165.3	Add-on muscle glycogen storage disorders	14	ALDOA, ENO3, GAA, GBE1, GYG1, GYS1, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKB, PYGM, RBCK1
○ 06168	Invitae Ketogenesis Disorders Panel		2	HMGCL, HMGCS2
○ 06169	Invitae Ketolysis Disorders Panel		2	ACAT1, OXCT1
○ 06166	Invitae Medium Chain Acyl-CoA Dehydrogenase Deficiency Test		1	ACADM
○ 06197	Invitae Multiple Acyl-CoA Dehydrogenase Deficiency Panel		3	ETFA, ETFB, ETFDH
	○ 06197.1	Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
○ 06167	Invitae Very Long Chain Acyl-CoA Dehydrogenase Deficiency Test		1	ACADVL
Lysosomal Storage Disorders				
○ 06170	Invitae Comprehensive Lysosomal Storage Disorders Panel		48	AGA, ARSA, ARSB, ASAH1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSD, CTSK, FUCA1, GAA, GALC, GALNS, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, KCTD7, LAMP2, LIPA, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1
	○ 06170.1	Add-on chitotriosidase deficiency gene	1	CHIT1
	○ 06170.2	Add-on preliminary-evidence gene	1	ATP13A2
	○ 06170.3	Add-on adult-onset neuronal ceroid lipofuscinoses genes	3	CTSF, DNAJC5, GRN

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METABOLIC TEST CATALOG

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Lysosomal Storage Disorders (continued)			
○ 06172	Invitae Cystinosis Test	1	CTNS
○ 06179	Invitae Farber Lipogranulomatosis Test	1	ASAH1
○ 02266	Invitae Fabry Disease Test	1	GLA
○ 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
○ 06174.1	Add-on generalized leukodystrophies genes	4	ASPA, GALC, HEXA, HEXB
○ 06184	Invitae Mucolipidosis Panel	4	GNPTAB, GNPTG, MCOLN1, NEU1
○ 06185	Invitae Comprehensive Mucopolysaccharidoses (MPS) Panel	11	ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH
○ 06185.1	Add-on mucolipidosis and oligosaccharidoses genes	12	AGA, CTSA, CTSK, FUCA1, GNPTAB, GNPTG, MAN2B1, MANBA, MCOLN1, NAGA, NEU1, SLC17A5
○ 06186	Invitae Mucopolysaccharidosis Type I (MPS I) Test	1	IDUA
○ 06186.1	Add-on clinically overlapping lysosomal storage disorder genes	5	ARSB, GNPTAB, GUSB, IDS, SUMF1
○ 06175	Invitae Mucopolysaccharidosis Type II Test	1	IDS
○ 06175.1	Add-on clinically overlapping genes	4	GNPTAB, GUSB, IDUA, SUMF1
○ 06187	Invitae Mucopolysaccharidosis Type III (MPS III) Panel	4	GNS, HGSNAT, NAGLU, SGSH
○ 06188	Invitae Mucopolysaccharidosis Type IV (MPS IV) Panel	2	GALNS, GLB1
○ 06188.1	Add-on multiple sulfatase deficiency gene	1	SUMF1
○ 06189	Invitae Multiple Sulfatase Deficiency Test	1	SUMF1
○ 06189.1	Add-on mucolipidosis and mucopolysaccharidosis genes	15	ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MCOLN1, NAGLU, NEU1, SGSH
○ 03405	Invitae Comprehensive Neuronal Ceroid Lipofuscinoses Panel	9	CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1
○ 03405.1	Add-on preliminary-evidence gene	1	ATP13A2
○ 03405.2	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
○ 06200	Invitae Oligosaccharidoses Panel	8	AGA, CTSA, CTSK, FUCA1, MAN2B1, MANBA, NAGA, SLC17A5
○ 06200.1	Add-on mucolipidosis and mucopolysaccharidosis genes	15	ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MCOLN1, NAGLU, NEU1, SGSH

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METABOLIC TEST CATALOG

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Lysosomal Storage Disorders (continued)			
○ 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
○ 06201	Invitae Prosaposin Deficiency Test	1	PSAP
○ 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
○ 04719.1	Add-on Sandhoff disease gene	1	HEXB
Metal Transport Disorders			
○ 06182	Invitae ATP7A-Related Disorders Test	1	ATP7A
○ 06202	Invitae Copper Metabolism Disorders Panel	5	AP1S1, ATP7A, ATP7B, CP, SLC33A1
○ 06183	Invitae Wilson Disease Test	1	ATP7B
Neurotransmitter Disorders			
○ 06203	Invitae Neurotransmitter Disorders Panel	27	ABAT, ALDH5A1, ALDH7A1, AMT, ARHGEF9, DBH, DDC, GAD1, GCH1, GCSH, GLDC, GLRA1, GLRB, GPHN, MAOA, PCBD1, PHGDH, PNPO, PSAT1, PSPH, PTS, QDPR, SLC25A22, SLC6A3, SLC6A5, SPR, TH
○ 06203.1	Add-on neurodegeneration with brain iron accumulation genes	10	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45
○ 06204	Invitae Hereditary Hyperekplexia Panel	6	ARHGEF9, CLPB, GLRA1, GLRB, GPHN, SLC6A5
Organic Acidemias			
○ 06191	Invitae Organic Acidemias Panel	49	ACAD8, ACADSB, ACAT1, ACSF3, ASPA, AUH, BCKDHA, BCKDHB, BTD, D2HGDH, DBT, DNAJC19, ETFA, ETFB, ETFDH, ETHE1, FBP1, FTCD, GCDH, GSS, HIBCH, HLCS, HMGCL, HSD17B10, IDH2, IVD, L2HGDH, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MUT, OPA3, OPLAH, OXCT1, PCCA, PCCB, POLG, PPM1K, SERAC1, SLC25A1, SUCLA2, SUCLG1, TAZ, TMEM70
○ 06191.1	Add-on Kreb cycle defect genes	7	DHTKD1, DLD, FH, NFU1, OGDH, SLC13A5, SLC25A19
○ 06205	Invitae 2-Hydroxyglutaric Aciduria Panel	4	D2HGDH, IDH2, L2HGDH, SLC25A1
○ 06192	Invitae 3-Methylcrotonyl CoA Carboxylase Panel	2	MCCC1, MCCC2
○ 06206	Invitae 3-Methylglutaconic Aciduria Panel	8	AUH, CLPB, CPS1, DNAJC19, OPA3, SERAC1, TAZ, TMEM70
○ 06193	Invitae Barth Syndrome Test	1	TAZ
○ 06194	Invitae Biotinidase Deficiency Test	1	BTD
○ 04713	Invitae Canavan Disease Test	1	ASPA
○ 06195	Invitae Glutaric Acidemia Type I Test	1	GCDH
○ 06141	Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2
○ 06196	Invitae Methylmalonic Acidemia Panel	7	MMAA, MMAB, MMADHC, MCEE, MUT, SUCLA2, SUCLG1
○ 06196.1	Add-on combined malonic and methylmalonic acidemia gene	1	ACSF3
○ 06196.2	Add-on combined methylmalonic acidemia and homocystinuria genes	10	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, TCN1, TCN2

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METABOLIC TEST CATALOG

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Organic Acidemias (continued)			
○ 06197	Invitae Multiple Acyl-CoA Dehydrogenase (MAD) Deficiency Panel	3	ETFA, ETFB, ETFDH
○ 06197.1	Add-on riboflavin transporter deficiency genes	3	SLC52A1, SLC52A2, SLC52A3
○ 06198	Invitae Multiple Carboxylase Deficiency Panel	2	BTD, HLCS
○ 06199	Invitae Propionic Acidemia Panel	2	PCCA, PCCB
○ 06199.1	Add-on methylmalonic acidemia genes	5	MMAA, MMAB, MMADHC, MMACHC, MUT
○ 06199.2	Add-on multiple carboxylase deficiency genes	2	BTD, HLCS
Peroxisomal Disorders			
○ 06207	Invitae Adult Refsum Disease Panel	2	PEX7, PHYH
○ 06208	Invitae Rhizomelic Chondrodysplasia Punctata Spectrum Panel	3	AGPS, GNPAT, PEX7
○ 06209	Invitae X-linked Adrenoleukodystrophy (X-ALD) Test	1	ABCD1
○ 06209.1	Add-on peroxisomal acyl-CoA oxidase (ACOX1) deficiency gene	1	ACOX1
○ 06209.2	Add-on elevated very long chain fatty acids genes	13	HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26
○ 06211	Invitae Zellweger Spectrum Disorder Panel	15	ACOX1, AMACR, HSD17B4, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6
Purine Metabolism Disorders			
○ 06213	Invitae Purine Metabolism Disorders Panel	9	ADA, ADSL, AMPD1, HPRT1, GPHN, MOCOS, MOCS1, PNP, XDH
○ 06213.1	Add-on sulfite oxidase deficiency gene	1	SUOX
○ 06214	Invitae Lesch-Nyhan Syndrome Test	1	HPRT1
Pyruvate Metabolism and Tricarboxylic Acid Cycle Defects			
○ 06215	Invitae 2-Ketoglutarate Dehydrogenase Deficiency Panel	3	DLD, OGDH, SLC25A19
○ 06215.1	Add-on alpha-ketoadipic acid dehydrogenase deficiency gene	1	DHTKD1
○ 06216	Invitae Citrate Transporter Deficiency Test	1	SLC13A5
○ 06217	Invitae Dihydrolipoamide Dehydrogenase Deficiency Test	1	DLD
○ 06218	Invitae Fumarase Deficiency Test	1	FH
○ 06219	Invitae Pyruvate Carboxylase Deficiency Test	1	PC
○ 06221	Invitae Pyruvate Dehydrogenase Deficiency Panel	8	DLAT, DLD, LIAS, MPC1, PDHA1, PDHB, PDHX, PDP1

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METABOLIC TEST CATALOG

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
Symptom-based Metabolic Panels			
○ 06230	Invitae Hyperammonemia Panel	58	ABCD4, ACADM, ACADVL, ALDH18A1, ARG1, ASL, ASS1, BCKDHA, BCKDHB, BTD, CA5A, CPS1, CPT1A, CPT2, DBT, DLAT, DLD, ETFA, ETFB, ETFDH, GLUD1, GLUL, HADHA, HADHB, HCFC1, HLCS, HMGCL, IVD, LMBRD1, MCCC1, MCCC2, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, NAGS, OAT, OTC, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC7A7, TAZ, TMEM70, UMPS
○ 06228	Invitae Mendelian Disorders with Psychiatric Symptoms Panel	88	ABCD1, ADSL, ALDH5A1, AMACR, AMT, AP1S1, ARG1, ARSA, ASL, ASS1, ATP7B, ATP13A2, BCKDHA, BCKDHB, BCKDK, C19orf12, CA5A, CBS, CLN3, CLN5, CLN6, CLN8, COASY, CP, CPS1, CTSD, CYP27A1, DBH, DBT, DCAF17, DDC, DLD, FAH, FTL, FUCA1, GALC, GAMT, GATM, GCH1, GCSH, GLA, GLB1, GLDC, GM2A, GNS, GSS, HEXA, HEXB, HGSNAT, HMGCL, HPRT1, MAN2B1, MANBA, MAOA, MFSD8, MMACHC, MTHFR, MTR, NAGLU, NAGS, NPC1, NPC2, OTC, PAH, PANK2, PCCA, PCCB, PLA2G6, POLG, PPT1, PRODH, PSAP, PTS, QDPR, SGSH, SLC6A8, SLC7A7, SLC25A13, SLC25A15, SLC52A1, SLC52A2, SLC52A3, SPR, SUMF1, TBX1, TH, TPP1, WDR45
	○ 06228.1 Add-on adult-onset neuronal ceroid lipofuscinoses genes	3	CTSF, DNAJC5, GRN
○ 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
Treatable Disorders			
○ 06222	Invitae Treatable Neurometabolic Disorders Panel	102	ABCD1, ABCD4, ACAT1, AGA, ALDH5A1, ALDH7A1, AMN, AMT, ARG1, ARHGEF9, ARSA, ASL, ASS1, ATP7A, ATP7B, AUH, BCKDHA, BCKDHB, BTD, CBS, CD320, CLN2 (TPP1), CP, CPS1, CUBN, CYP27A1, DBT, DHCR7, DLAT, DLD, ETFA, ETFB, ETFDH, ETHE1, GALC, GAMT, GATM, GCDH, GCH1, GCSH, GIF, GLA, GLDC, GLRA1, GLRB, GLUD1, GUSB, HCFC1, HLCS, HMGCL, HMGCS2, HSD17B10, IDS, IDUA, IVD, LIPA, LMBRD1, MAN2B1, MCCC1, MCCC2, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MTHFR, MTR, MTRR, MUT, NAGS, NPC1, NPC2, OTC, OXCT1, PAH, PANK2, PCBD1, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PHGDH, PNPO, PPM1K, PSAT1, PSPH, PTS, QDPR, SGSH, SLC19A3, SLC25A13, SLC25A15, SLC2A1, SLC6A5, SLC6A8, SPR, TAT, TCN1, TCN2, TH
	○ 06222.1 Add-on neurometabolic conditions genes	42	ABAT, ADSL, AP1S1, ATP13A2, BCKDK, C19orf12, CLN3, CLN5, CLN6, CLN8, CLPB, COASY, CTSD, D2HGDH, DBH, DCAF17, DDC, FA2H, FTL, GAD1, GNS, GPHN, HEXA, HEXB, HGSNAT, HPRT1, IDH2, KCTD7, L2HGDH, MAOA, MFSD8, MOCOS, NAGLU, PLA2G6, POLG, PPT1, SLC13A5, SLC33A1, SLC6A3, SUOX, WDR45, XDH
○ 06223	Invitae Biotin-Thiamine-Responsive Basal Ganglia Disease (BTBGD) Test	1	SLC19A3
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
○ 06224	Invitae Arginase Deficiency Test	1	ARG1
○ 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

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INVITAE

Patient's first name

Patient's last name

PEDIATRIC GENETICS, METABOLIC, AND NEPHROLOGY
FM170-4

METABOLIC TEST CATALOG

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

METABOLIC NEWBORN SCREENING AND IMMUNOLOGY INDIVIDUAL GENES

ABAT	ALG9	CBS	DBH	FTL	GNPTAB	LAMP2	NADK2
ABCD1	AMACR	CD320	DBT	FUCA1	GNPTG	LARGE1	NAGA
ABCD4	AMN	CFTR	DCAF17	G6PC	GNS	LDHA	NAGLU
ABCG5	AMPD1	CHIT1	DDC	G6PC3	GPHN	LFNG	NAGS
ABCG8	AMT	CHST14	DDOST	G6PD	GRN	LIAS	NEU1
ACAD8	AP1S1	CHST3	DECR1	GAA	GSS	LIPA	NFU1
ACADM	ARG1	CHST6	DHCR7	GAD1	GUSB	LMBRD1	NGLY1
ACADS	ARHGEF9	CHSY1	DHDDS	GALC	GYG1	MAGT1	NPC1
ACADSB	ARSA	CLN2 (TPP1)	DHTKD1	GALE	GYS1	MAN1B1	NPC2
ACADVL	ARSB	CLN3	DLAT	GALK1	GYS2	MAN2B1	NUS1
ACAT1	ASAH1	CLN5	DLD	GALNS	HADH	MANBA	OAT
ACOX1	ASL	CLN6	DNAJC19	GALNT3	HADHA	MAOA	OGDH
ACSF3	ASPA	CLN8	DNAJC5	GALT	HADHB	MAT1A	OPA3
ADA	ASS1	CLPB	DOLK	GAMT	HCFC1	MCCC1	OPLAH
ADSL	ATP13A2	COASY	DPAGT1	GATM	HEXA	MCCC2	OTC
AGA	ATP6V0A2	COG1	DPM1	GBE1	HEXB	MCEE	OXCT1
AGL	ATP7A	COG2	DPM2	GCDH	HGD	MCOLN1	PAH
AGPS	ATP7B	COG4	DPM3	GCH1	HGSNAT	MFSD8	PANK2
AHCY	AUH	COG5	DSE	GCSH	HIBCH	MGAT2	PAPSS2
ALDH18A1	B3GALNT2	COG6	ENO3	GFPT1	HLCS	MLYCD	PC
ALDH4A1	B3GALT6	COG7	EOGT	GIF	HMGCL	MMAA	PCBD1
ALDH5A1	B3GAT3	COG8	ETFA	GLA	HMGCS2	MMAB	PCCA
ALDH7A1	B3GLCT	CP	ETFB	GLB1	HPD	MMACHC	PCCB
ALDOA	B4GALNT1	CPS1	ETFDH	GLDC	HPRT1	MMADHC	PDHA1
ALDOB	B4GALT1	CPT1A	ETHE1	GLRA1	HSD17B10	MOCOS	PDHB
ALG1	B4GALT7	CPT2	EXT1	GLRB	HSD17B4	MOCS1	PDHX
ALG11	B4GAT1	CTNS	EXT2	GLUD1	HYAL1	MOGS	PDP1
ALG12	BCKDHA	CTSA	FA2H	GLUL	IDH2	MPC1	PEX1
ALG13	BCKDHB	CTSD	FAH	GM2A	IDS	MPDU1	PEX10
ALG14	BCKDK	CTSF	FBP1	GMPPA	IDUA	MPI	PEX12
ALG2	BTD	CTSK	FH	GMPPB	ISPD	MTHFR	PEX13
ALG3	C19orf12	CUBN	FKRP	GNE	IVD	MTR	PEX14
ALG6	C1GALT1C1	CYP27A1	FKTN	GNMT	KCTD7	MTRR	PEX16
ALG8	CA5A	D2HGDH	FTCD	GNPAT	L2HGDH	MUT	PEX19

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METABOLIC TEST CATALOG

CLINICAL AREA: METABOLIC NEWBORN SCREENING AND IMMUNOLOGY

METABOLIC NEWBORN SCREENING AND IMMUNOLOGY INDIVIDUAL GENES (continued)

○ PEX2	○ PHYH	○ POGLUT1	○ PYGL	○ SLC22A5	○ SLC35D1	○ SRD5A3	○ TCN2
○ PEX26	○ PIGA	○ POLG	○ PYGM	○ SLC25A1	○ SLC37A4	○ SGSH	○ TH
○ PEX3	○ PIGL	○ POMGNT1	○ QDPR	○ SLC25A13	○ SLC3A1	○ SSR4	○ TMEM165
○ PEX5	○ PIGM	○ POMGNT2	○ RBCK1	○ SLC25A15	○ SLC52A1	○ ST3GAL3	○ TMEM70
○ PEX6	○ PIGN	○ POMK	○ RFT1	○ SLC25A19	○ SLC52A2	○ ST3GAL5	○ TRIP11
○ PEX7	○ PIGO	○ POMT1	○ RPN2	○ SLC25A20	○ SLC52A3	○ STT3A	○ TUSC3
○ PFKM	○ PIGQ	○ POMT2	○ RXYLT1	○ SLC25A22	○ SLC5A1	○ STT3B	○ UMPS
○ PGAM2	○ PIGT	○ PPM1K	○ SLC52A1	○ SLC26A2	○ SLC6A3	○ SUCLA2	○ WDR45
○ PGM1	○ PIGV	○ PPT1	○ SEC23A	○ SLC2A1	○ SLC6A5	○ SUCLG1	○ XDH
○ PGM3	○ PIGW	○ PREPL	○ SEC23B	○ SLC2A2	○ SLC6A8	○ SUMF1	○ XYLT1
○ PHGDH	○ PLA2G6	○ PRODH	○ SERAC1	○ SLC33A1	○ SLC6A9	○ SUOX	
○ PHKA1	○ PMM2	○ PSAP	○ SGSH	○ SLC35A1	○ SLC7A7	○ TAT	
○ PHKA2	○ PNP	○ PSAT1	○ SLC13A5	○ SLC35A2	○ SLC7A9	○ TAZ	
○ PHKB	○ PNPO	○ PSPH	○ SLC17A5	○ SLC35A3	○ SMPD1	○ TBX1	
○ PHKG2	○ POFUT1	○ PTS	○ SLC19A3	○ SLC35C1	○ SPR	○ TCN1	

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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 specimen tube for each clinical area. Each clinical area represents an individual billable event and report. Please 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	102	AHI1, ANKS6, ARL13B, ARL6, ARMC4, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP298, CPLANE1, CSPP1, DCDC2, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, DYNC2H1, EVC, EVC2, GAS8, GLIS2, IFT122, IFT140, IFT172, IFT80, INPP5E, INVS, IQCB1, KIAA0586, KIF7, LRRC6, MCIDAS, MKKS, MKS1, MRE11A, NEK1, NEK8, NME8, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PKD2, PKHD1, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SDCCAG8, SPAG1, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRIM32, TTC21B, TTC8, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZMYND10, ZNF423
○ 04103	Invitae Skeletal Ciliopathies Panel	17	CEP120, CSPP1, DYNC2H1, EVC, EVC2, IFT80, IFT122, IFT140, IFT172, KIAA0586, NEK1, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60
○ 04103.1	Add-on FGFR3-related thanatophoric dysplasia gene	3	FGFR1, FGFR2, FGFR3
○ 04101	Invitae Primary Ciliary Dyskinesia Panel	34	ARMC4, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFAP298, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, GAS8, LRRC6, MCIDAS, NME8, OFD1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10
○ 04101.1	Add-on preliminary-evidence gene	1	INVS
○ 04101.2	Add-on clinically overlapping gene	1	CFTR
○ 04112	Invitae Bardet-Biedl Syndrome Panel	16	ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP
○ 04111	Invitae Joubert and Meckel-Gruber Syndromes Panel	31	AHI1, ARL13B, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, MRE11A, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423
○ 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
○ 04117	Invitae Oral-Facial-Digital Syndrome Type 1 Test	1	OFD1
○ 04115	Invitae Polycystic Kidney Disease Type 2 Panel	2	PKD2, PKHD1
○ 04114	Invitae Senior-Loken Syndrome Panel	8	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, WDR19

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES

○ AHI1	○ BBS2	○ CCDC39	○ CEP83	○ CSPP1	○ DNAH5	○ EVC	○ IFT122
○ ANKS6	○ BBS4	○ CCDC40	○ CEP104	○ DCDC2	○ DNAH8	○ EVC2	○ IFT140
○ ARL6	○ BBS5	○ CCDC65	○ CEP120	○ DNAAF1	○ DNAH11	○ FGFR1	○ IFT172
○ ARL13B	○ BBS7	○ CCDC103	○ CEP164	○ DNAAF2	○ DNAI1	○ FGFR2	○ INPP5E
○ ARMC4	○ BBS9	○ CCDC114	○ CEP290	○ DNAAF3	○ DNAI2	○ FGFR3	○ INVS
○ B9D1	○ BBS10	○ CCDC151	○ CFAP298	○ DNAAF4	○ DNAL1	○ GAS8	○ IQCB1
○ B9D2	○ BBS12	○ CCNO	○ CFTR	○ DNAAF5	○ DRC1	○ GLIS2	○ KIAA0586
○ BBS1	○ CC2D2A	○ CEP41	○ CPLANE1	○ DNAH1	○ DYNC2H1	○ IFT80	○ KIF7

*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.*

NEPHROLOGY TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES (continued)							
○ LRRC6	○ NEK8	○ PDE6D	○ RSPH1	○ SPAG1	○ TMEM138	○ TTC8	○ WDR35
○ MCIDAS	○ NME8	○ PKD2	○ RSPH3	○ TCTN1	○ TMEM216	○ TTC21B	○ WDR60
○ MKKS	○ NPHP1	○ PKHD1	○ RSPH4A	○ TCTN2	○ TMEM231	○ WDPCP	○ XPNPEP3
○ MKS1	○ NPHP3	○ RPGR	○ RSPH9	○ TCTN3	○ TMEM237	○ WDR19	○ ZMYND10
○ MRE11A	○ NPHP4	○ RPGRIP1L	○ SDCCAG8	○ TMEM67	○ TRIM32	○ WDR34	○ ZNF423
○ NEK1	○ OFD1						

CLINICAL AREA: NEPHROLITHIASIS

The panel below cannot accept DNA as a specimen type (blood and saliva specimens only).

Test code	Test name	# gene(s)	Gene list
Nephrolithiasis			
○ 72037	Invitae Nephrolithiasis Panel	35	ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, CA2, CASR, CLCN5, CLDN16, CLDN19, CYP24A1, FAM20A, GPHN, GRHPR, HOGA1, HPRT1, KCNJ1, MOCOS, MOCS1, OCRL, PREPL, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1, UMOD, VDR, XDH

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