INVITAE

ORDER ONLINE at www.invitae.com/signin.

ORDER ID
For Invitae internal use only

Requisition Form
PEDIATRIC GENETICS TRF906-10

PATIENT INFORMATION

First name | MI | Last name

Date of birth (MM/DD/YYYY) | Sex ◯ M ◯ F | 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 report access after release by medical professional)

Phone | Is this patient deceased? ◯ Yes ◯ No Deceased date:

Address | City

State | ZIP code | Country

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

Specimen type : ◯ Blood ◯ Saliva ◯ Assisted saliva ◯ DNA - source:

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

We are unable to accept blood/saliva from patients with:

- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

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.*

Special cases : ◯ History of/current hematologic malignancy ◯ Resubmission

REASON FOR TESTING

Primary indication:

ICD-10 codes | **Previous results**

Testing for a personal history of disease? ◯ **Yes** ◯ **No** If yes, describe below.
Age at diagnosis: _____

ORGANIZATION INFORMATION

Organization name and address

Organization name

Phone | Fax

Address | City

State | ZIP code | Country

Primary clinical contact

Name | Role/title

Phone | NPI

Email address (for report access)

Ordering physician

◯ **Same as primary clinical contact**

Name | NPI

Email address (for report access)

Additional clinical or laboratory contact (optional)

Name | Email address (for report access)

Letter of Medical Necessity (LMN)

◯ I have attached an LMN and/or other documentation for insurance billing purposes.

◯ I agree to allow Invitae to transfer the information from this requisition to an LMN and/or other documentation using the ordering physician's name as the signature for insurance billing.

Family history? ◯ **Yes** ◯ **No** If yes, describe in detail below or attach pedigree. If there is a known familial variant, indicate here.

◯ INSURANCE BILLING (U.S. ONLY)

◯ **I have attached a copy of the patient's card**

Insurance company name | Member ID#

Patient relation to policy holder: ◯ Self ◯ Child ◯ Spouse ◯ Other

Policy holder name | Prior-authorization #

◯ PATIENT PAY BILLING

Invitae will send an electronic invoice to the patient email listed above

◯ INSTITUTIONAL BILLING

◯ **Send invoice to organization address above**

Billing contact name | Phone | Fax

Billing email address

Billing address | City

State | ZIP code | Country

◯ OTHER BILLING

Invitae partner code:

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/patient-consent), and has been informed that Invitae may notify them of clinical updates related to genetic test results (in consultation with the ordering medical professional as indicated). The Patient has further been informed and hereby authorizes Invitae Corporation ("Invitae") and its designees to release information concerning testing to their insurer in order to process and/or appeal claims on behalf of the Patient. For amounts received directly, the Patient agrees to remit payment to Invitae for testing services rendered. I acknowledge that I offered pre-test Genetic Counseling to the Patient, if required by their insurer. In addition to the above, I attest that I am the ordering physician, or I am authorized by the ordering physician to order this test, or I am authorized under applicable state law to order this test.

Medical professional signature | **Date**

HEADQUARTERS & LABORATORY | 1400 16th Street, San Francisco, CA 94103 | www.invitae.com | IN THE US | clientservices@invitae.com | p: 800-436-3037 | f: 415-276-4164
OUTSIDE THE US | globalsupport@invitae.com or visit www.invitae.com/contact |

ORDER INSTRUCTIONS

Select a pre-curated test, combine multiple tests, or customize your own test for each patient. Invitae's pricing is per clinical area for initial order and re-requisition. All tests on this form are organized by clinical area. If your order contains tests from multiple clinical areas, you will need to send in two sample tubes and your order will represent two billable events. Your test results will be delivered as two reports. Please contact Client Services with any questions. For Invitae's full test menu, please visit www.invitae.com.

RE-REQUISITION

Invitae offers one re-requisition at no additional charge within 90 days for genes within the original clinical area. For more information and to request online, please visit www.invitae.com/re-requisition.

FAMILY VARIANT TESTING

Invitae offers Family Variant Testing at no additional charge within 90 days for the genes in which the original family member's variant was identified. In such cases, please use the Family Variant Testing/VUS Resolution requisition form (TRF920), available at www.invitae.com/forms.

To request a complimentary specimen collection kit visit www.invitae.com/request-a-kit

SHIPPING INSTRUCTIONS

Please ship specimen overnight in insulated containers:

Attn: Invitae Client Services
1400 16th Street
San Francisco, CA 94103
USA

PRELIMINARY-EVIDENCE GENES

Invitae's primary panels contain genes for which there is definitive evidence that variants in these genes cause specific diseases. Preliminary-evidence genes are genes for which there is only early evidence of a relationship between variants in these genes and specific diseases. All preliminary-evidence genes are indicated as such on the requisition form below.

ASSAY

Invitae is a CAP-accredited and CLIA-certified clinical diagnostic laboratory performing full-gene sequencing and deletion/duplication analysis using next-generation sequencing technology (NGS). Search for details on the analysis of any gene in our test catalog at www.invitae.com/physician/search.

Invitae continually updates its panels based on the most recent evidence. Please note that if an order is placed using an older version of this form, Invitae reserves the right to upgrade any ordered panel(s) to the current version(s). To avoid confusion, please consider placing your order using our online test catalog.

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, C21orf59, C5orf42, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CSPP1, DCDC2, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, DYNC2H1, DYX1C1, 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	1	FGFR3
○ 04101	Invitae Primary Ciliary Dyskinesia Panel	34	ARMC4, C21orf59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, 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	30	AHI1, ARL13B, B9D1, B9D2, C5orf42, CC2D2A, CEP104, CEP290, CEP41, 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

CLINICAL AREA: PEDIATRIC AND RARE DISEASE (continued)

Test code	Test name	# gene(s)	Gene list
Congenital Heart Disease			
○ 04201	Invitae Congenital Heart Defects and Heterotaxy Panel	82	ACTC1, ACVR2B, ALMS1, ANKS6, ARMC4, BBS10, BCOR, BRAF, C21orf59, CBL, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP290, CFAP53, CHD7, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, 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
Cystic Fibrosis and Chronic Pancreatitis			
○ 04714	Invitae Cystic Fibrosis Test	1	CFTR
	○ 04714.1 Add-on chronic pancreatitis genes	4	CASR, CTRC, PRSS1, SPINK1
○ 01745	Invitae Chronic Pancreatitis Panel	5	CASR, CFTR, CTRC, PRSS1, SPINK1
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	5	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

CLINICAL AREA: PEDIATRIC AND RARE DISEASE (continued)

Test code	Test name	# gene(s)	Gene list
Developmental Disorders (continued)			
○ 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
○ 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
Epilepsy, Seizures, and Developmental Brain Abnormalities			
○ 03401	Invitae Epilepsy Panel	133	ADSL, ALDH5A1, ALDH7A1, ALG13, ARHGEF9, ARX, ATP1A2, ATP1A3, ATRX, BRAT1, C12orf57, CACNA1A, CACNA2D2, CASK, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN4, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CNTNAP2, CSTB, CTSD, DEPDC5, DNAJC5, DNM1, DOCK7, DYRK1A, EEF1A2, EFHC1, EHMT1, EPM2A, FOLR1, FOXG1, FRRS1L, GABRA1, GABRB3, GABRG2, GAMT, GATM, GLRA1, GNAO1, GOSR2, GRIN1, GRIN2A, GRIN2B, HCN1, HNRNPU, IER3IP1, IQSEC2, ITPA, KANSL1, KCNA2, KCNB1, KCNC1, KCNH2, KCNJ10, KCNQ2, KCNQ3, KCNT1, KCTD7, KIAA2022, LGI1, LIAS, MBD5, MECP2, MEF2C, MFSD8, MTOR, NEDD4L, NGLY1, NHLRC1, NRXN1, PACS1, PCDH19, PIGA, PIGN, PIGO, PLCB1, PNKD, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRRT2, PURA, QARS, ROGDI, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SERPINI1, SGCE, SLC12A5, SLC13A5, SLC19A3, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SNX27, SPATA5, SPTAN1, ST3GAL5, STRADA, STX1B, STXBP1, SYN1, SYNJ1, SYNGAP1, SZT2, TBC1D24, TCF4, TSC1, TSC2, UBE3A, WWOX, ZDHHC9, ZEB2
	○ 03401.1 Add-on preliminary-evidence genes	50	ABAT, ARHGEF15, ATP6AP2, CACNA1H, CACNB4, CARS2, CASR, CBL, CERS1, CNTN2, COQ4, CPA6, DIAPH1, FARS2, FASN, GABBR2, GABRB2, GABRD, GAL, GPHN, JMJD1C, KCNA1, KCND2, KCNH5, KCNMA1, KPNA7, LMNB2, NECAP1, NPRL3, PIGG, PIGQ, PIK3AP1, PRDM8, PRICKLE2, PRIMA1, RBFOX1, RBFOX3, RELN, RYR3, SCN5A, SETD2, SIK1, SLC25A12, SLC35A3, SNAP25, SRPX2, ST3GAL3, TBL1XR1, TPK1, WDR45
	○ 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

CLINICAL AREA: PEDIATRIC AND RARE DISEASE (continued)

Test code	Test name	# gene(s)	Gene list
Epilepsy, Seizures, and Developmental Brain Abnormalities (continued)			
○ 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	53	ALDH7A1, ARHGEF9, ARX, BRAT1, CACNA2D2, CASK, CDKL5, CHD2, CLCN4, DNM1, DOCK7, EEF1A2, FOLR1, FRRS1L, GABRA1, GABRB3, GNAO1, GRIN1, GRIN2A, GRIN2B, HCN1, HNRNPU, IER3IP1, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNT1, PCDH19, PIGA, PIGN, PIGO, PLCB1, PNKP, PNPO, PURA, SCN1A, SCN2A, SCN8A, SCN9A, SLC12A5, SLC2A1, SLC13A5, SLC25A22, SLC35A2, SLC6A1, SMC1A, SPTAN1, STXBP1, SYNGAP1, SZT2, TBC1D24, WWOX
○ 03402.1	Add-on preliminary-evidence genes	12	ARHGEF15, ATP1A2, COQ4, GABBR2, GPHN, KCNH5, MTOR, NECAP1, NEDD4L, SCN1B, SIK1, ST3GAL3
○ 04424	Invitae Holoprosencephaly Panel	5	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	22	ADSL, ALDH5A1, ATRX, CDKL5, CNTNAP2, DYRK1A, EHMT1, FOXG1, IQSEC2, KANSL1, MBD5, MECP2, MEF2C, NGLY1, NRXN1, SATB2, SCN8A, SLC9A6, STXBP1, TCF4, UBE3A, ZEB2
○ 03404.1	Add-on preliminary-evidence genes	4	GABRD, TBL1XR1, JMJD1C, WDR45
○ 01721	Invitae Tuberous Sclerosis Complex Panel	2	TSC1, TSC2
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	33	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, VSX2
○ 05132.1	Add-on preliminary-evidence genes	5	CHMP4B, CRYGB, LIM2, TDRD7, VIM
○ 04728	Invitae Duane-Radial Ray Syndrome Test	1	SALL4
○ 05133	Invitae Early-Onset Glaucoma Panel	3	CYP1B1, FOXC1, PITX2
○ 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	3	GDF3, HESX1, VAX1
○ 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

CLINICAL AREA: PEDIATRIC AND RARE DISEASE (continued)

Test code	Test name	# gene(s)	Gene list
Eye Disorders (continued)			
○ 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	20	AKT2, AKT3, CDKN1C, CUL4B, DIS3L2, DNMT3A, EZH2, GLI3, GPC3, KPTN, MED12, 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
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

CLINICAL AREA: PEDIATRIC AND RARE DISEASE (continued)

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 FGFR3-related thanatophoric dysplasia gene	1	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

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES

○ A2ML1	○ AKT1	○ ANOS1	○ ATP1A3	○ BBS7	○ C5orf42	○ CCDC114	○ CEP120
○ ABAT	○ AKT2	○ AR	○ ATP6AP2	○ BBS9	○ CACNA1A	○ CCDC151	○ CEP164
○ ACTB	○ AKT3	○ ARHGEF15	○ ATRX	○ BCOR	○ CACNA1H	○ CCDC39	○ CEP290
○ ACTC1	○ ALDH1A3	○ ARHGEF9	○ B9D1	○ BFSP1	○ CACNA2D2	○ CCDC40	○ CEP41
○ ACTG1	○ ALDH5A1	○ ARL13B	○ B9D2	○ BFSP2	○ CACNB4	○ CCDC65	○ CEP83
○ ACVR2B	○ ALDH7A1	○ ARL6	○ BBS1	○ BMP4	○ CARS2	○ CCM2	○ CERS1
○ ADSL	○ ALG13	○ ARMC4	○ BBS10	○ BRAF	○ CASK	○ CCNO	○ CFAP52
○ AFF4	○ ALMS1	○ ARSE	○ BBS12	○ BRAT1	○ CASR	○ CDKL5	○ CFAP53
○ AGK	○ AMT	○ ARX	○ BBS2	○ C12orf57	○ CBL	○ CDKN1C	○ CFTR
○ AHI1	○ ANKRD11	○ ATP13A2	○ BBS4	○ C19orf12	○ CC2D2A	○ CDON	○ CHD2
○ AIPL1	○ ANKS6	○ ATP1A2	○ BBS5	○ C21orf59	○ CCDC103	○ CEP104	○ CHD7

CLINICAL AREA: PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES (continued)

○ CHM	○ CTDP1	○ EEF1A2	○ GABRG2	○ HRAS	○ KMT2D	○ MYH6	○ PANK2
○ CHMP4B	○ CTRC	○ EFHC1	○ GAL	○ HSF4	○ KPNA7	○ NECAP1	○ PAX2
○ CHRNA2	○ CTSD	○ EHMT1	○ GALK1	○ IER3IP1	○ KPTN	○ NEDD4L	○ PAX6
○ CHRNA4	○ CUL4B	○ ELN	○ GAMT	○ IFT122	○ KRAS	○ NEK1	○ PAX9
○ CHRNB2	○ CYP1B1	○ EP300	○ GAS8	○ IFT140	○ KRIT1	○ NEK8	○ PCDH19
○ CLCN4	○ DCAF17	○ EPHA2	○ GATA4	○ IFT172	○ LCA5	○ NF1	○ PDCD10
○ CLN2 (TPP1)	○ DCDC2	○ EPM2A	○ GATA6	○ IFT80	○ LEFTY2	○ NF2	○ PDE6D
○ CLN3	○ DEPDC5	○ ERF	○ GATM	○ IMPDH1	○ LGI1	○ NFIX	○ PDGFRB
○ CLN5	○ DHCR7	○ EVC	○ GCNT2	○ INPP5E	○ LIAS	○ NFKBIA	○ PHF6
○ CLN6	○ DHH	○ EVC2	○ GCSH	○ INVS	○ LIM2	○ NGLY1	○ PIGA
○ CLN8	○ DIAPH1	○ EXT1	○ GDF1	○ IQCB1	○ LMNB2	○ NHLRC1	○ PIGG
○ CNTN2	○ DICER1	○ EXT2	○ GDF3	○ IQSEC2	○ LRAT	○ NHS	○ PIGN
○ CNTNAP2	○ DIS3L2	○ EYA1	○ GDF6	○ IRF6	○ LRRC6	○ NIPBL	○ PIGO
○ COASY	○ DNAAF1	○ EZH2	○ GJA1	○ ITPA	○ LTBP3	○ NKX2-5	○ PIGQ
○ COL1A1	○ DNAAF2	○ FA2H	○ GJA3	○ JAG1	○ MAB21L2	○ NKX2-6	○ PIK3AP1
○ COL1A2	○ DNAAF3	○ FAM126A	○ GJA8	○ JMJD1C	○ MAF	○ NME8	○ PIK3CA
○ COQ4	○ DNAAF5	○ FARS2	○ GJB6	○ KANSL1	○ MAP2K1	○ NMNAT1	○ PIK3R2
○ CP	○ DNAH1	○ FASN	○ GLDC	○ KAT6B	○ MAP2K2	○ NODAL	○ PITX2
○ CPA6	○ DNAH11	○ FGFR1	○ GLI2	○ KCNA1	○ MAP3K1	○ NOTCH1	○ PITX3
○ CRB1	○ DNAH5	○ FGFR2	○ GLI3	○ KCNA2	○ MASP1	○ NOTCH2	○ PKD2
○ CREBBP	○ DNAH8	○ FGFR3	○ GLIS2	○ KCNB1	○ MBD5	○ NPHP1	○ PKHD1
○ CRELD1	○ DNAI1	○ FLNA	○ GLRA1	○ KCNC1	○ MCIDAS	○ NPHP3	○ PLA2G6
○ CRTAP	○ DNAI2	○ FOLR1	○ GNAO1	○ KCND2	○ MECP2	○ NPHP4	○ PLCB1
○ CRX	○ DNAJC5	○ FOXC1	○ GOSR2	○ KCNH2	○ MED12	○ NPR2	○ PNKD
○ CRYAA	○ DNAL1	○ FOXE3	○ GPC3	○ KCNH5	○ MED13L	○ NPRL3	○ PNKP
○ CRYAB	○ DNM1	○ FOXG1	○ GPHN	○ KCNJ10	○ MEF2C	○ NR0B1	○ PNPO
○ CRYBA1	○ DNMT3A	○ FOXH1	○ GRHL3	○ KCNJ13	○ MEGF8	○ NR2F2	○ POLG
○ CRYBA4	○ DOCK7	○ FRRS1L	○ GRIN1	○ KCNMA1	○ MEIS2	○ NR5A1	○ POR
○ CRYBB1	○ DRC1	○ FTL	○ GRIN2A	○ KCNQ2	○ MFRP	○ NRAS	○ PPT1
○ CRYBB2	○ DYNC1H1	○ FUCA1	○ GRIN2B	○ KCNQ3	○ MFSD8	○ NRXN1	○ PQBP1
○ CRYBB3	○ DYNC2H1	○ FYCO1	○ GUCY2D	○ KCNT1	○ MIP	○ NSD1	○ PRDM8
○ CRYGB	○ DYRK1A	○ GABBR2	○ HAND1	○ KCTD7	○ MKKS	○ NSDHL	○ PRICKLE1
○ CRYGC	○ DYX1C1	○ GABRA1	○ HCN1	○ KDM6A	○ MKS1	○ OCRL	○ PRICKLE2
○ CRYGD	○ EDA	○ GABRA6	○ HDAC8	○ KIAA0586	○ MRE11A	○ OFD1	○ PRIMA1
○ CRYGS	○ EDAR	○ GABRB2	○ HESX1	○ KIAA2022	○ MSX1	○ OTX2	○ PRPH2
○ CSPP1	○ EDARADD	○ GABRB3	○ HNRNPU	○ KIF1A	○ MSX2	○ P3H1	○ PRRT2
○ CSTB	○ EED	○ GABRD	○ HPRT1	○ KIF7	○ MTOR	○ PACS1	○ PRSS1

CLINICAL AREA: PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES (continued)

○ PRSS56	○ RDH12	○ SCARB2	○ SLC12A5	○ SOX9	○ SYNJ1	○ TP63	○ WDR34
○ PTCH1	○ RELN	○ SCN1A	○ SLC13A5	○ SPAG1	○ SZT2	○ TPK1	○ WDR35
○ PTEN	○ RIT1	○ SCN1B	○ SLC19A3	○ SPATA5	○ TBC1D24	○ TRIM32	○ WDR45
○ PTPN11	○ RNF125	○ SCN2A	○ SLC25A12	○ SPATA7	○ TBL1XR1	○ TRPS1	○ WDR60
○ PURA	○ ROGDI	○ SCN3A	○ SLC25A22	○ SPINK1	○ TBX1	○ TSC1	○ WNT10A
○ PXDN	○ RPE65	○ SCN5A	○ SLC2A1	○ SPRED1	○ TBX3	○ TSC2	○ WT1
○ QARS	○ RPGR	○ SCN8A	○ SLC35A2	○ SPTAN1	○ TBX5	○ TTC21B	○ WWOX
○ RAB23	○ RPGRIP1	○ SCN9A	○ SLC35A3	○ SQSTM1	○ TCF4	○ TTC8	○ XPNPEP3
○ RAD21	○ RPGRIP1L	○ SDCCAG8	○ SLC6A1	○ SRD5A2	○ TCOF1	○ TULP1	○ ZDHHC9
○ RAF1	○ RPS6KA3	○ SEMA3E	○ SLC6A8	○ SRPX2	○ TCTN1	○ TWIST1	○ ZEB2
○ RANBP2	○ RRAS	○ SERPINI1	○ SLC9A6	○ SRY	○ TCTN2	○ UBE3A	○ ZFPM2
○ RARB	○ RSPH1	○ SETD2	○ SMAD6	○ ST3GAL3	○ TCTN3	○ UPF3B	○ ZIC2
○ RASA1	○ RSPH3	○ SGCE	○ SMC1A	○ ST3GAL5	○ TDRD7	○ VAX1	○ ZIC3
○ RAX	○ RSPH4A	○ SHH	○ SMC3	○ STRA6	○ TGIF1	○ VHL	○ ZMYND10
○ RB1	○ RSPH9	○ SHOC2	○ SNAP25	○ STRADA	○ TMEM138	○ VIM	○ ZNF423
○ RBFOX1	○ RYR3	○ SIK1	○ SNX27	○ STX1B	○ TMEM216	○ VPS13B	
○ RBFOX3	○ SALL1	○ SIL1	○ SOS1	○ STXBP1	○ TMEM231	○ VSX2	
○ RBM8A	○ SALL4	○ SIX1	○ SOS2	○ SYN1	○ TMEM237	○ WDPCP	
○ RD3	○ SATB2	○ SIX3	○ SOX2	○ SYNGAP1	○ TMEM67	○ WDR19	

CLINICAL AREA: HEREDITARY CANCER

Test code	Test name	# gene(s)	Gene list
Pediatric Oncology			
○ 01104	Invitae Pediatric Solid Tumors Panel	48	ALK, APC, AXIN2, BAP1, BLM, BMPR1A, CDC73, CDKN1C, DICER1, DIS3L2, EPCAM, FH, GPC3, HRAS, MAX, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PHOX2B, PMS2, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, 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
○ 01106	Invitae Pediatric Nervous System/Brain Tumors Panel	24	ALK, APC, DICER1, EPCAM, HRAS, 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

PEDIATRIC ONCOLOGY INDIVIDUAL GENES

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

