

PATIENT INFORMATION

First name	MI	Last name
Date of birth (MM/DD/YYYY)	Sex <input type="radio"/> M <input type="radio"/> F	MRN (medical record number)
Ancestry <input type="radio"/> Asian <input type="radio"/> Black/African American <input type="radio"/> White/Caucasian <input type="radio"/> Ashkenazi Jewish <input type="radio"/> Hispanic <input type="radio"/> Native American <input type="radio"/> Pacific Islander <input type="radio"/> Other:		
▶ Email address (for report access after release by medical professional)		
Phone	Is this patient deceased? <input type="radio"/> Yes <input type="radio"/> 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)

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

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 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.

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			
<input type="radio"/> 04102	Invitae Ciliopathies Panel	102	AH11, 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, DAAAF1, DAAAF2, DAAAF3, DAAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, DYNC2H1, DDX11, 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, RPL10A, RPL11, RPL13, RPL13A, RPL13B, RPL13C, RPL13D, RPL13E, RPL13F, RPL13G, RPL13H, RPL13I, RPL13J, RPL13K, RPL13L, RPL13M, RPL13N, RPL13O, RPL13P, RPL13Q, RPL13R, RPL13S, RPL13T, RPL13U, RPL13V, RPL13W, RPL13X, RPL13Y, RPL13Z, RPL14, RPL15, RPL16, RPL17, RPL18, RPL19, RPL20, RPL21, RPL22, RPL23, RPL24, RPL25, RPL26, RPL27, RPL28, RPL29, RPL30, RPL31, RPL32, RPL33, RPL34, RPL35, RPL36, RPL37, RPL38, 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CLINICAL AREA: PEDIATRIC AND RARE DISEASE (continued)

Test code	Test name	# gene(s)	Gene list
Congenital Heart Disease			
<input type="radio"/> 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
<input type="radio"/> 04201.1	Add-on preliminary-evidence genes	7	CFAP52, CRELD1, GATA6, HAND1, MYH6, SMAD6, ZFPM2
Cystic Fibrosis and Chronic Pancreatitis			
<input type="radio"/> 04714	Invitae Cystic Fibrosis Test	1	CFTR
<input type="radio"/> 04714.1	Add-on chronic pancreatitis genes	4	CASR, CTRC, PRSS1, SPINK1
<input type="radio"/> 01745	Invitae Chronic Pancreatitis Panel	5	CASR, CFTR, CTRC, PRSS1, SPINK1
Developmental Disorders			
<input type="radio"/> 04215	Invitae Alagille Syndrome Panel	2	JAG1, NOTCH2
<input type="radio"/> 04721	Invitae Alpha Thalassemia X-linked Intellectual Disability Test	1	ATRX
<input type="radio"/> 04741	Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel	2	ACTB, ACTG1
<input type="radio"/> 04724	Invitae Branchiootorenal Spectrum Disorders Panel	2	EYA1, SIX1
<input type="radio"/> 04724.1	Add-on Townes-Brocks syndrome gene	1	SALL1
<input type="radio"/> 04725	Invitae Carpenter Syndrome Panel	2	MEGF8, RAB23
<input type="radio"/> 01732	Invitae CASR-Related Conditions Test	1	CASR
<input type="radio"/> 04211	Invitae CHARGE Syndrome Test	1	CHD7
<input type="radio"/> 04743	Invitae CHOPS Syndrome Test	1	AFF4
<input type="radio"/> 04738	Invitae Coffin-Lowry Syndrome Test	1	RPS6KA3
<input type="radio"/> 04737	Invitae Cohen Syndrome Test	1	VPS13B
<input type="radio"/> 04727	Invitae Cornelia de Lange Syndrome Panel	5	HDAC8, NIPBL, RAD21, SMC1A, SMC3
<input type="radio"/> 04727.1	Add-on preliminary-evidence gene	1	EP300
<input type="radio"/> 04744	Invitae Glass Syndrome Test	1	SATB2
<input type="radio"/> 04730	Invitae GLI3-Related Disorders Test	1	GLI3
<input type="radio"/> 04745	Invitae HPRT1-Related Disorders test	1	HPRT1
<input type="radio"/> 04736	Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Panel	3	ANOS1, CHD7, FGFR1
<input type="radio"/> 04731	Invitae Kabuki Syndrome Panel	2	KDM6A, KMT2D
<input type="radio"/> 04746	Invitae KAT6B-Related Disorders Test	1	KAT6B
<input type="radio"/> 04747	Invitae KBG Syndrome Test	1	ANKRD11
<input type="radio"/> 04716	Invitae MED12-Related Disorders Test	1	MED12
<input type="radio"/> 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)			
<input type="radio"/> 01704	Invitae PTEN-Related Disorders Test	1	PTEN
<input type="radio"/> 04748	Invitae Renpenning Syndrome Test	1	PQBP1
<input type="radio"/> 04739	Invitae Rubinstein-Taybi Syndrome Panel	2	CREBBP, EP300
<input type="radio"/> 01739	Invitae Simpson-Golabi-Behmel Syndrome Test	1	GPC3
<input type="radio"/> 04740	Invitae Smith-Lemli-Opitz Syndrome Test	1	DHCR7
<input type="radio"/> 04214	Invitae Sotos Syndrome Test	1	NSD1
<input type="radio"/> 04735	Invitae van der Woude Syndrome Panel	2	GRHL3, IRF6
<input type="radio"/> 01716	Invitae von Hippel-Lindau Syndrome Test	1	VHL
<input type="radio"/> 04749	Invitae WAGR Syndrome Panel	2	PAX6, WT1
<input type="radio"/> 01740	Invitae Weaver Syndrome Test	1	EZH2
Disorders of Sex Development/Endocrinology (if available, please provide karyotype information)			
<input type="radio"/> 04411	Invitae Disorders of Male Sex Development Panel	8	AR, DHH, MAP3K1, NR0B1, NR5A1, SRD5A2, SRY, WT1
<input type="radio"/> 04411.1	Add-on Kallman syndrome genes	4	ANOS1, CHD7, FGFR1, HESX1
<input type="radio"/> 04411.2	Add-on alpha-thalassemia X-linked intellectual disability gene	1	ATRX
<input type="radio"/> 04411.3	Add-on campomelic dysplasia gene	1	SOX9
<input type="radio"/> 04411.4	Add-on Smith-Lemli-Opitz syndrome gene	1	DHCR7
<input type="radio"/> 04412	Invitae Androgen Insensitivity Panel	2	AR, SRD5A2
<input type="radio"/> 04736	Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Panel	3	ANOS1, CHD7, FGFR1
Epilepsy, Seizures, and Developmental Brain Abnormalities			
<input type="radio"/> 03401	Invitae Epilepsy Panel	125	ADSL, ALDH5A1, ALDH7A1, ALG13, ARHGEF9, ARX, ATP1A2, ATP1A3, ATRX, BRAT1, C12orf57, CACNA2D2, CASK, CDKL5, CHD2, CHRNA2, CHRNA4, CHRN2, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CNTNAP2, CSTB, CTSD, DEPDC5, DNAJC5, DNM1, 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, 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, SLC13A5, SLC19A3, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SNX27, SPATA5, SPTAN1, STX1B, STXBP1, SYN1, SYNJ1, SYNGAP1, SZT2, TBC1D24, TCF4, TSC1, TSC2, UBE3A, WWOX, ZDHHC9, ZEB2
<input type="radio"/> 03401.1	Add-on preliminary-evidence genes	58	ABAT, ARHGEF15, ATP6AP2, CACNA1A, CACNA1H, CACNB4, CARS2, CASR, CBL, CERS1, CLCN4, CNTN2, COQ4, CPA6, DIAPH1, DOCK7, FARS2, FASN, GABBR2, GABRB2, GABRD, GAL, GPHN, JMJ1D1C, KCNA1, KCND2, KCNH5, KCNMA1, KPNA7, LMNB2, MTOR, NECAP1, NEDD4L, NPRL3, PIGG, PIGQ, PIK3AP1, PRDM8, PRICKLE2, PRIMA1, RBFOX1, RBFOX3, RELN, RYR3, SCN5A, SETD2, SIK1, SLC12A5, SLC25A12, SLC35A3, SNAP25, SRPX2, ST3GAL3, ST3GAL5, STRADA, TBL1XR1, TPK1, WDR45
<input type="radio"/> 03401.2	Add-on genes for glycine encephalopathy	3	AMT, GCSH, GLDC
<input type="radio"/> 03401.3	Add-on FLNA gene	1	FLNA
<input type="radio"/> 03401.4	Add-on PTEN gene	1	PTEN
<input type="radio"/> 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)			
<input type="radio"/> 03407	Invitae Alternating Hemiplegia of Childhood Panel	2	ATP1A2, ATP1A3
<input type="radio"/> 03407.1	Add-on clinically overlapping genes	3	CACNA1A, SCN1A, SLC2A1
<input type="radio"/> 04741	Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel	2	ACTB, ACTG1
<input type="radio"/> 04422	Invitae Cerebral Caverosus Malformations Panel	3	CCM2, KRIT1, PDCD10
<input type="radio"/> 04211	Invitae CHARGE Syndrome Test	1	CHD7
<input type="radio"/> 03402	Invitae Early Infantile Epileptic Encephalopathy Panel	49	ALDH7A1, ARHGEF9, ARX, BRAT1, CACNA2D2, CASK, CDKL5, CHD2, DNM1, EEF1A2, FOLR1, FRRS1L, GABRA1, GABRB3, GNAO1, GRIN1, GRIN2B, HCN1, HNRNPU, IER3IP1, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNT1, PCDH19, PIGA, PIGN, PIGO, PLCB1, PNKP, PNPO, PURA, SCN1A, SCN2A, SCN8A, SCN9A, SLC2A1, SLC13A5, SLC25A22, SLC35A2, SLC6A1, SMC1A, SPTAN1, STXBP1, SYNGAP1, SZT2, TBC1D24, WWOX
<input type="radio"/> 03402.1	Add-on preliminary-evidence genes	14	ARHGEF15, CLCN4, COQ4, DOCK7, GABBR2, GPHN, KCNH5, MTOR, NECAP1, NEDD4L, SCN1B, SIK1, SLC12A5, ST3GAL3
<input type="radio"/> 04424	Invitae Holoprosencephaly Panel	5	GLI2, SHH, SIX3, TGIF1, ZIC2
<input type="radio"/> 04424.1	Add-on preliminary-evidence genes	4	CDON, FOXH1, NODAL, PTCH1
<input type="radio"/> 03406	Invitae Neurodegeneration with Brain Iron Accumulation Panel	11	ATP13A2, C19orf12, COASY, CP, DCAF17, FTL, FUCA1, PANK2, PLA2G6, SQSTM1, WDR45
<input type="radio"/> 03406.1	Add-on preliminary-evidence genes	3	FA2H, KIF1A, TRIM32
<input type="radio"/> 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
<input type="radio"/> 03404.1	Add-on preliminary-evidence genes	4	GABRD, TBL1XR1, JMJD1C, WDR45
<input type="radio"/> 01721	Invitae Tuberous Sclerosis Complex Panel	2	TSC1, TSC2
Eye Disorders			
<input type="radio"/> 04722	Invitae Aniridia Test	1	PAX6
<input type="radio"/> 03406	Invitae Axenfeld-Rieger Panel	2	FOXC1, PITX2
<input type="radio"/> 03406.1	Add-on aniridia gene	1	PAX6
<input type="radio"/> 04112	Invitae Bardet-Biedl Syndrome Panel	16	ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP
<input type="radio"/> 04211	Invitae CHARGE Syndrome Test	1	CHD7
<input type="radio"/> 05131	Invitae Choroideremia Test	1	CHM
<input type="radio"/> 05132	Invitae Congenital Cataracts Panel	32	AGK, BCOR, BFSP1, BFSP2, CRYAA, CRYAB, CRYBA1, 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
<input type="radio"/> 05132.1	Add-on preliminary-evidence genes	6	CHMP4B, CRYBA4, CRYGB, LIM2, TDRD7, VIM
<input type="radio"/> 04728	Invitae Duane-Radial Ray Syndrome Test	1	SALL4
<input type="radio"/> 05133	Invitae Early-Onset Glaucoma Panel	3	CYP1B1, FOXC1, PITX2
<input type="radio"/> 05142	Invitae Microphthalmia/Anophthalmia Disorders Panel	15	ALDH1A3, BCOR, BMP4, FOXE3, GDF6, MFRP, OTX2, PAX2, PRSS56, PXDN, RAX, SHH, SOX2, STRA6, VSX2
<input type="radio"/> 05142.1	Add-on preliminary-evidence genes	5	GDF3, HESX1, MAB21L2, RARB, VAX1
<input type="radio"/> 05143	Invitae Leber Congenital Amaurosis Panel	19	AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, OTX2, PRPH2, RD3, RDH12, RPE65, RPGRI1, SPATA7, TULP1
<input type="radio"/> 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)			
<input type="radio"/> 04213	Invitae Oculo-Facio-Cardio-Dental Syndrome Test	1	BCOR
<input type="radio"/> 01738	Invitae Retinoblastoma Test	1	RB1
<input type="radio"/> 04114	Invitae Senior-Loken Syndrome Panel	8	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, WDR19
Overgrowth Syndromes			
<input type="radio"/> 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
<input type="radio"/> 04501.1	Add-on preliminary-evidence genes	5	EED, DICER1, PDGFRB, RNF125, UPF3B
<input type="radio"/> 01736	Invitae Perlman Syndrome Test	1	DIS3L2
<input type="radio"/> 04502	Invitae Proteus Syndrome Test	1	AKT1
<input type="radio"/> 01704	Invitae PTEN-Related Disorders Test	1	PTEN
<input type="radio"/> 01739	Invitae Simpson-Golabi-Behmel Syndrome Test	1	GPC3
<input type="radio"/> 04214	Invitae Sotos Syndrome Test	1	NSD1
<input type="radio"/> 01740	Invitae Weaver Syndrome Test	1	EZH2
RASopathies (Noonan spectrum disorders)			
<input type="radio"/> 04151	Invitae RASopathies Comprehensive Panel	18	A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1
<input type="radio"/> 04163	Invitae Cardio-Facio-Cutaneous Syndrome Panel	6	BRAF, KRAS, MAP2K1, MAP2K2, SHOC2, SOS1
<input type="radio"/> 04164	Invitae Costello Syndrome Test	1	HRAS
<input type="radio"/> 04165	Invitae Legius Syndrome Test	1	SPRED1
<input type="radio"/> 04165.1	Add-on neurofibromatosis type 1 gene	1	NF1
<input type="radio"/> 01708	Invitae Neurofibromatosis Type 1 Test	1	NF1
<input type="radio"/> 01708.1	Add-on Legius syndrome gene	1	SPRED1
<input type="radio"/> 04161	Invitae Noonan Syndrome Panel	14	A2ML1, BRAF, CBL, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, RRAS, SHOC2, SOS1, SOS2
<input type="radio"/> 04161.1	Add-on Baraitser-Winter cerebrofrontofacial syndrome genes	2	ACTB, ACTG1
<input type="radio"/> 04162	Invitae Noonan Syndrome with Multiple Lentigines Panel	3	BRAF, PTPN11, RAF1
Skeletal Disorders			
<input type="radio"/> 04612	Invitae Antley-Bixler syndrome Test	1	POR
<input type="radio"/> 04612.1	Add-on craniosynostosis gene	1	FGFR2
<input type="radio"/> 04726	Invitae ARSE-Related Chondrodysplasia Punctata Test	1	ARSE
<input type="radio"/> 04726.1	Add-on NSDHL-related disorders gene	1	NSDHL
<input type="radio"/> 04712	Invitae Campomelic Dysplasia Test	1	SOX9
<input type="radio"/> 04423	Invitae Craniosynostosis Panel	9	ERF, FGFR1, FGFR2, FGFR3, GLI3, MEGF8, MSX2, RAB23, TWIST1
<input type="radio"/> 04423.1	Add-on 3MC and Treacher Collins syndromes genes	2	MASP1, TCOF1
<input type="radio"/> 04728	Invitae Duane-Radial Ray Syndrome Test	1	SALL4
<input type="radio"/> 04613	Invitae Ellis van Creveld and Weyers Acrofacial Dysostosis Panel	2	EVC, EVC2
<input type="radio"/> 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)			
<input type="radio"/> 04614	Invitae Hereditary Multiple Osteochondromas Panel	2	EXT1, EXT2
<input type="radio"/> 04614.1	Add-on Langer-Giedion syndrome gene	1	TRPS1
<input type="radio"/> 04212	Invitae Holt-Oram Syndrome Test	1	TBX5
<input type="radio"/> 04615	Invitae NSDHL-Related Disorders Test	1	NSDHL
<input type="radio"/> 04732	Invitae Osteogenesis Imperfecta Panel	4	COL1A1, COL1A2, CRTAP, P3H1
<input type="radio"/> 04103	Invitae Skeletal Ciliopathies Panel	17	CEP120, CSPP1, DYNC2H1, EVC, EVC2, IFT80, IFT122, IFT140, IFT172, KIAA0586, NEK1, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60
<input type="radio"/> 04103.1	Add-on FGFR3-related thanatophoric dysplasia gene	1	FGFR3
<input type="radio"/> 04616	Invitae Thrombocytopenia Absent Radius Syndrome Test	1	RBM8A
<input type="radio"/> 04733	Invitae Townes-Brocks Syndrome Test	1	SALL1
<input type="radio"/> 04734	Invitae Treacher Collins Syndrome Test	1	TCOF1
<input type="radio"/> 04617	Invitae Trichorhinophalangeal Syndrome Panel	2	EXT1, TRPS1
<input type="radio"/> 04618	Invitae Ulnar-Mammary Syndrome Test	1	TBX3
Skin Disorders			
<input type="radio"/> 04163	Invitae Cardio-Facio-Cutaneous Syndrome Panel	6	BRAF, KRAS, MAP2K1, MAP2K2, SHOC2, SOS1
<input type="radio"/> 05021	Invitae Ectodermal Dysplasia with or without Tooth Agenesis Panel	8	EDA, EDAR, EDARADD, LTBP3, MSX1, NFKBIA, PAX9, WNT10A
<input type="radio"/> 05021.1	Add-on Clouston syndrome and TP63-related disorder genes	2	GJB6, TP63
<input type="radio"/> 04165	Invitae Legius Syndrome Test	1	SPRED1
<input type="radio"/> 04165.1	Add-on neurofibromatosis type 1 gene	1	NF1
<input type="radio"/> 04162	Invitae Noonan Syndrome with Multiple Lentiginos Panel	3	BRAF, PTPN11, RAF1
<input type="radio"/> 01704	Invitae PTEN-Related Disorders Test	1	PTEN
<input type="radio"/> 05022	Invitae TP63-Related Disorders Test	1	TP63
<input type="radio"/> 04735	Invitae van der Woude Syndrome Panel	2	GRHL3, IRF6

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES

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

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

○ CHM	○ CTRC	○ EEF1A2	○ GABRG2	○ HRAS	○ KMT2D	○ MYH6	○ PANK2
○ CHMP4B	○ CTSD	○ EFHC1	○ GAL	○ HSF4	○ KPNA7	○ NECAP1	○ PAX2
○ CHRNA2	○ CUL4B	○ EHMT1	○ GALK1	○ IER3IP1	○ KPTN	○ NEDD4L	○ PAX6
○ CHRNA4	○ CYP1B1	○ ELN	○ GAMT	○ IFT122	○ KRAS	○ NEK1	○ PAX9
○ CHRNB2	○ DCAF17	○ EP300	○ GAS8	○ IFT140	○ KRIT1	○ NEK8	○ PCDH19
○ CLCN4	○ EPHA2	○ EPM2A	○ GATA4	○ IFT172	○ LCA5	○ NF1	○ PDCD10
○ CLN2 (TPP1)	○ DCDC2	○ ERF	○ GATA6	○ IFT80	○ LEFTY2	○ NF2	○ PDE6D
○ CLN3	○ DEPDC5	○ EVC	○ GATM	○ IMPDH1	○ LGI1	○ NFIX	○ PDGFRB
○ CLN5	○ DHCR7	○ EVC2	○ GCNT2	○ INPP5E	○ LIAS	○ NFKBIA	○ PHF6
○ CLN6	○ DHH	○ EXT1	○ GCSH	○ INVS	○ LIM2	○ NGLY1	○ PIGA
○ CLN8	○ DIAPH1	○ EXT2	○ GDF1	○ IQCB1	○ LMNB2	○ NHLRC1	○ PIGG
○ CNTN2	○ DICER1	○ EYA1	○ GDF3	○ IQSEC2	○ LRAT	○ NHS	○ PIGN
○ CNTNAP2	○ DNAAF1	○ FA2H	○ GDF6	○ IRF6	○ LRRC6	○ NIPBL	○ PIGO
○ COASY	○ DNAAF2	○ FAM126A	○ GJA1	○ ITPA	○ LTBP3	○ NKX2-5	○ PIGQ
○ COL1A1	○ DNAAF3	○ FARS2	○ GJA3	○ JAG1	○ MAB21L2	○ NKX2-6	○ PIK3AP1
○ COL1A2	○ DNAAF5	○ FASN	○ GJA8	○ JMJD1C	○ MAF	○ NME8	○ PIK3CA
○ COQ4	○ DNAH1	○ FGFR1	○ GJB6	○ KANSL1	○ MAP2K1	○ NMNAT1	○ PIK3R2
○ CP	○ DNAH11	○ FGFR2	○ GLDC	○ KAT6B	○ MAP2K2	○ NODAL	○ PITX2
○ CPA6	○ DNAH5	○ FGFR3	○ GLI2	○ KCNA1	○ MAP3K1	○ NOTCH1	○ PITX3
○ CRB1	○ DNAH8	○ FLNA	○ GLI3	○ KCNA2	○ MASP1	○ NOTCH2	○ PKD2
○ CREBBP	○ DNAI1	○ FOLR1	○ GLIS2	○ KCNB1	○ MBD5	○ NPHP1	○ PKHD1
○ CRELD1	○ DNAI2	○ FOXC1	○ GLRA1	○ KCNC1	○ MCIDAS	○ NPHP3	○ PLA2G6
○ CRTAP	○ DNAJCS	○ FOXE3	○ GNAO1	○ KCND2	○ MECP2	○ NPHP4	○ PLCB1
○ CRX	○ DNAL1	○ FOXP1	○ GOSR2	○ KCNH2	○ MED12	○ NPR2	○ PNKD
○ CRYAA	○ DNMT3A	○ FRRS1L	○ GPC3	○ KCNH5	○ MED13L	○ NPRL3	○ PNKP
○ CRYAB	○ DOCK7	○ FTL	○ GPHN	○ KCNJ10	○ MEF2C	○ NR0B1	○ PNPO
○ CRYBA1	○ DRC1	○ GRHL3	○ GRIN1	○ KCNJ13	○ MEGF8	○ NR2F2	○ POLG
○ CRYBA4	○ DYNC1H1	○ GRIN2A	○ GRIN2B	○ KCNMA1	○ MEIS2	○ NR5A1	○ POR
○ CRYBB1	○ DYX1C1	○ GUCY2D	○ HAND1	○ KCNQ2	○ MFRP	○ NRAS	○ PPT1
○ CRYBB2	○ EDA	○ HESX1	○ HCN1	○ KCNQ3	○ MFSB8	○ NRXN1	○ PQBP1
○ CRYBB3	○ EDAR	○ HNRNPU	○ HDAC8	○ KCNT1	○ MIP	○ NSD1	○ PRDM8
○ CRYGB	○ EDARADD	○ HPRT1	○ HESX1	○ KCTD7	○ MKKS	○ NSDHL	○ PRICKLE1
○ CRYGC	○ EED	○ KIF1A	○ HNF1B	○ KDM6A	○ MKS1	○ OCRL	○ PRICKLE2
○ CRYGD	○ GABRA1	○ KIF7	○ HNF1B	○ KIAA0586	○ MRE11A	○ OFD1	○ PRIMA1
○ CRYGS	○ GABRA6	○ KIF7	○ HNF1B	○ KIAA2022	○ MSX1	○ OTX2	○ PRPH2
○ CSPP1	○ GABRB2	○ KIF7	○ HNF1B	○ KIAA2022	○ MSX2	○ P3H1	○ PRRT2
○ CSTB	○ GABRB3	○ KIF7	○ HNF1B	○ KIAA2022	○ MSX2	○ PACS1	○ PRSS1
	○ GABRD	○ KIF7	○ HNF1B	○ KIAA2022	○ MSX2		

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

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

CLINICAL AREA: HEREDITARY CANCER

Test code	Test name	# gene(s)	Gene list
Pediatric Oncology			
<input type="radio"/> 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
<input type="radio"/> 01105	Invitae Pediatric Hematologic Malignancies Panel	16	ATM, BLM, CEBPA, EPCAM, GATA2, HRAS, MLH1, MSH2, MSH6, NBN, NF1, PMS2, RUNX1, TERC, TERT, TP53
<input type="radio"/> 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
<input type="radio"/> 01106.1	Add-on hereditary paraganglioma-pheochromocytoma genes	8	MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127

▶ PEDIATRIC ONCOLOGY INDIVIDUAL GENES

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