INVITAE PEDIATRIC GENE PANEL TESTS

Our testing menu contains a broad variety of panels curated by medical genetics experts for conditions that have pediatric onset, including epilepsies, brain malformations, ciliopathies, RASopathies, overgrowth syndromes, skeletal disorders, developmental disorders, and disorders of sex development.

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

CILIOPATHIES

Invitae Ciliopathies Panel (102 genes)

Primary panel (102 genes)

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						

Invitae Skeletal Ciliopathies Panel (up to 18 genes)

Primary panel (17 genes)

CEP120	CSPP1	DYNC2H1	EVC	EVC2	IFT80	IFT122	IFT140	IFT172	KIAA0586	NEK1	TCTN3
TTC21B	WDR19	WDR34	WDR35	WDR60							

Add-on FGFR3-related thanatophoric dysplasia gene (1 gene)

FGFR3

Invitae Primary Ciliary Dyskinesia Panel (up to 36 genes)

Primary panel (34 genes)

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		

Add-on preliminary-evidence gene (1 gene)

INVS

Add-on clinically overlapping gene (1 gene)

CFTR

Invitae Bardet-Biedl Syndrome Panel (16 genes)

Primary panel (16 genes)

ARL6	BBS1	BBS10	BBS12	BBS2	BBS4	BBS5	BBS7	BBS9	CEP290	MKKS	MKS1
SDCCAG8	TRIM32	TTC8	WDPCP								

Invitae Joubert and Meckel-Gruber Syndromes Panel (30 genes)

Primary panel (30 genes)

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						

Invitae Nephronophthisis Panel (27 genes)

Primary panel (27 genes)

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									

Invitae Oral-Facial-Digital Syndrome Type 1 Test (1 gene)

Primary panel (1 gene)

OFD1

Invitae Polycystic Kidney Disease Type 2 Panel (2 genes)

Primary panel (2 genes)

PKD2 PKHD1

Invitae Senior-Loken Syndrome Panel (8 genes)

Primary panel (8 genes)

CEP290	INVS	IQCB1	NPHP1	NPHP3	NPHP4	SDCCAG8	WDR19

CONGENITAL HEART DEFECTS

Invitae Congenital Heart Defects and Heterotaxy Panel (up to 89 genes)

Primary panel (82 genes)

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		

Add-on preliminary-evidence genes (7 genes)

CFAP52	CRELD1	GATA6	HAND1	MYH6	SMAD6	ZFPM2

CYSTIC FIBROSIS AND CHRONIC PANCREATITIS

Invitae Cystic Fibrosis Test (up to 5 genes)

Primary panel (1 gene)

CFTR

Add-on chronic pancreatitis genes (4 genes)

CASR CTRC PRSS1 SPINK1

Invitae Chronic Pancreatitis Panel (5 genes)

Primary panel (5 genes)

CASR CFTR CTRC PRSS1 SPINK1

INVITAE

DEVELOPMENTAL DISORDERS

Invitae Alagille Syndrome Panel (2 genes)	Primary panel (2 genes) JAG1 NOTCH2
Invitae Alpha Thalassemia X-linked Intellectual Disability Test (1 gene)	Primary panel (1 gene) ATRX
Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel (2 genes)	Primary panel (2 genes) ACTB ACTG1
Invitae Branchiootorenal Spectrum Disorders Panel (up to 3 genes)	Primary panel (2 genes) EYA1 SIX1
	Add-on Townes-Brocks syndrome gene (1 gene) SALL1
Invitae Carpenter Syndrome Panel (2 genes)	Primary panel (2 genes) MEGF8 RAB23
Invitae CASR-Related Conditions Test (1 gene)	Primary panel (1 gene) CASR
Invitae CHARGE Syndrome Test (1 gene)	Primary panel (1 gene) CHD7
Invitae CHOPS Syndrome Test (1 gene)	Primary panel (1 gene) AFF4
Invitae Coffin-Lowry Syndrome Test (1 gene)	Primary panel (1 gene) RPS6KA3
Invitae Cohen Syndrome Test (1 gene)	Primary panel (1 gene) VPS13B
Invitae Cornelia de Lange Syndrome Panel (up to 6 genes)	Primary panel (5 genes) HDAC8 NIPBL RAD21 SMC1A SMC3
	Add-on preliminary-evidence genes (1 gene) EP300
Invitae Glass Syndrome Test (1 gene)	Primary panel (1 gene) SATB2
Invitae GLI3-Related Disorders Test (1 gene)	Primary panel (1 gene) GLI3
Invitae HPRT1-Related Disorders Test (1 gene)	Primary panel (1 gene) HPRT1
Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Panel (3 genes)	Primary panel (3 genes) ANOS1 CHD7 FGFR1
Invitae Kabuki Syndrome Panel (2 genes)	Primary panel (2 genes) KDM6A KMT2D
Invitae KAT6B-Related Disorders Test (1 gene)	Primary panel (1 gene) KAT6B
Invitae KBG Syndrome Test (1 gene)	Primary panel (1 gene) ANKRD11
Invitae MED12-Related Disorders Test (1 gene)	Primary panel (1 gene) MED12
Invitae Oculo-Facio-Cardio-Dental Syndrome Test (1 gene)	Primary panel (1 gene) BCOR
Invitae PTEN-Related Disorders Test (1 gene)	Primary panel (1 gene) PTEN
Invitae Renpenning Syndrome Test (1 gene)	Primary panel (1 gene) PQBP1
Invitae Rubinstein-Taybi Syndrome Panel (2 genes)	Primary panel (2 genes) CREBBP EP300
Invitae Simpson-Golabi-Behmel Syndrome Test (1 gene)	Primary panel (1 gene) GPC3
Invitae Smith-Lemli-Opitz Syndrome Test (1 gene)	Primary panel (1 gene) DHCR7
Invitae Sotos Syndrome Test (1 gene)	Primary panel (1 gene) NSD1
Invitae van der Woude Syndrome Panel (2 genes)	Primary panel (2 genes) GRHL3 IRF6
Invitae von Hippel-Lindau Syndrome Test (1 gene)	Primary panel (1 gene) VHL
Invitae WAGR Syndrome Panel (2 genes)	Primary panel (2 genes) PAX6 WT1
Invitae Weaver Syndrome Test (1 gene)	Primary panel (1 gene) EZH2

INVITAE PEDIATRIC GENE PANEL TESTS (continued)

DISORDERS OF SEX DEVELOPMENT/ENDOCRINOLOGY

Invitae Disorders of Male Sex Development Panel (up to 15 genes)

Primary panel (8 genes)
AR DHH MAP3K1 NR0B1 NR5A1 SRD5A2 SRY WT1

Add-on Kallman syndrome genes (4 genes)
ANOS1 CHD7 FGFR1 HESX1

Add-on alpha-thalassemia X-linked intellectual disability (1 gene)
ATRX

Add-on campomelic dysplasia gene (1 gene)
SOX9

Add-on Smith-Lemli-Opitz syndrome gene (1 gene)
DHCR7

Invitae Androgen Insensitivity Panel (2 genes)

Primary panel (2 genes)
AR SRD5A2

Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Panel (3 genes)

Primary panel (3 genes)
ANOS1 CHD7 FGFR1

EPILEPSY, SEIZURES, AND DEVELOPMENTAL BRAIN ABNORMALITIES

Invitae Epilepsy Panel (up to 189 genes)

Primary panel (125 genes)

ADSL	ALDH5A1	ALDH7A1	ALG13	ARHGEF9	ARX	ATP1A2	ATP1A3	ATRX	BRAT1	C12orf57	CACNA2D2
CASK	CDKL5	CHD2	CHRNA2	CHRNA4	CHRNB2	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	GLDC	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
SNX27	SPATA5	SPTAN1	STX1B	STXBP1	SYN1	SYNGAP1	SYNJ1	SZT2	TBC1D24	TCF4	TSC1
TSC2	UBE3A	WWOX	ZDHHC9	ZEB2							

Add-on preliminary-evidence genes (58 genes)

ABAT	ARHGEF15	ATP6AP2	CACNA1A	CACNA1H	CACNB4	CARS2	CASR	CBL	CERS1	CLCN4	CNTN2
COQ4	CPA6	DIAPH1	DOCK7	FARS2	FASN	GABBR2	GABRB2	GABRD	GAL	GPHN	JMJD1C
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		

Add-on genes for glycine encephalopathy (3 genes)
AMT GCSH GLDC

Add-on FLNA gene (1 gene)
FLNA

Add-on PTEN gene (1 gene)
PTEN

Add-on RANBP2 gene (1 gene)
RANBP2

Invitae Alternating Hemiplegia of Childhood Panel (up to 5 genes)

Primary panel (2 genes)
ATP1A2 ATP1A3

Add-on clinically overlapping genes (3 genes)
CACNA1A SCN1A SLC2A1

Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel (2 genes)

Primary panel (2 genes)
ACTB ACTG1

Invitae Cerebral Cavernous Malformations Panel (3 genes)

Primary panel (3 genes)
CCM2 KRIT1 PDCD10

Invitae CHARGE Syndrome Test (1 gene)

Primary panel (1 gene)
CHD7

Invitae Early Infantile Epileptic Encephalopathy Panel (up to 63 genes)

Primary panel (49 genes)

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											

Add-on preliminary-evidence genes (14 genes)

ARHGEF15	CLCN4	COQ4	DOCK7	GABBR2	GPHN	KCNH5	MTOR	NECAP1	NEDD4L	SCN1B	SIK1
SLC12A5	ST3GAL3										

Invitae Holoprosencephaly Panel (up to 9 genes)

Primary panel (5 genes)
GLI2 SHH SIX3 TGIF1 ZIC2

Add-on preliminary-evidence genes (4 genes)
CDON FOXH1 NODAL PTCH1

INVITAE PEDIATRIC GENE PANEL TESTS (continued)

EPILEPSY, SEIZURES, AND DEVELOPMENTAL BRAIN ABNORMALITIES (continued)

Invitae Neurodegeneration with Brain Iron Accumulation Panel (up to 14 genes)

Primary panel (11 genes)
ATP13A2 C19orf12 COASY CP DCAF17 FTL FUCA1 PANK2 PLA2G6 SQSTM1 WDR45

Add-on preliminary-evidence genes (3 genes)
FA2H KIF1A TRIM32

Invitae Rett/Angelman and Related Disorders Panel (up to 26 genes)

Primary panel (22 genes)
ADSL ALDH5A1 ATRX CDKL5 CNTNAP2 DYRK1A EHMT1 FOXG1 IQSEC2 KANSL1 MBD5 MECP2
MEF2C NGLY1 NRXN1 SATB2 SCN8A SLC9A6 STXBP1 TCF4 UBE3A ZEB2

Add-on preliminary-evidence genes (4 genes)
GABRD JMJD1C TBL1XR1 WDR45

Invitae Tuberous Sclerosis Complex Panel (2 genes)

Primary panel (2 genes)
TSC1 TSC2

EYE DISORDERS

Invitae Aniridia Test (1 gene)

Primary panel (1 gene)
PAX6

Invitae Axenfeld-Rieger Panel (2 genes)

Primary panel (2 genes)
FOXC1 PITX2

Add-on aniridia gene (1 gene)
PAX6

Invitae Bardet-Biedl Syndrome Panel (16 genes)

Primary panel (16 genes)
ARL6 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 CEP290 MKKS MKS1
SDCCAG8 TRIM32 TTC8 WDPCP

Invitae CHARGE Syndrome Test (1 gene)

Primary panel (1 gene)
CHD7

Invitae Choroideremia Test (1 gene)

Primary panel (1 gene)
CHM

Invitae Congenital Cataracts Panel (up to 38 gene)

Primary panel (32 genes)
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

Add-on preliminary-evidence genes (6 genes)
CHMP4B CRYBA4 CRYGB LIM2 TDRD7 VIM

Invitae Duane-Radial Ray Syndrome Test (1 gene)

Primary panel (1 gene)
SALL4

Invitae Early-Onset Glaucoma Panel (3 genes)

Primary panel (3 genes)
CYP1B1 FOXC1 PITX2

Invitae Microphthalmia/Anophthalmia Disorders Panel (up to 20 genes)

Primary panel (15 genes)
ALDH1A3 BCOR BMP4 FOXE3 GDF6 MFRP OTX2 PAX2 PRSS56 PXDN RAX SHH
SOX2 STRA6 VSX2

Add-on preliminary-evidence genes (5 genes)
GDF3 HESX1 MAB21L2 RARB VAX1

Invitae Leber Congenital Amaurosis Panel (up to 21 genes)

Primary panel (19 genes)
AIPL1 CEP290 CRB1 CRX GDF6 GUCY2D IQCB1 KCNJ13 LCA5 LRAT NMNAT1 OTX2
PRPH2 RD3 RDH12 RPE65 RPGRIP1 SPATA7 TULP1

Add-on preliminary-evidence genes (2 genes)
BBS4 IMPDH1

Invitae Oculo-Facio-Cardio-Dental Syndrome Test (1 gene)

Primary panel (1 gene)
BCOR

Invitae Retinoblastoma Test (1 gene)

Primary panel (1 gene)
RB1

Invitae Senior-Loken Syndrome Panel (8 genes)

Primary panel (8 genes)
CEP290 INVS IQCB1 NPHP1 NPHP3 NPHP4 SDCCAG8 WDR19

OVERGROWTH SYNDROMES

Invitae Overgrowth and Macrocephaly Syndromes Panel (up to 25 genes)

Primary panel (20 genes)
AKT2 AKT3 CDKN1C CUL4B DIS3L2 DNMT3A EZH2 GLI3 GPC3 KPTN MED12 NF1
NFIX NPR2 NSD1 PHF6 PIK3R2 PTEN SETD2 SPRED1

Add-on preliminary-evidence genes (5 genes)
DICER1 EED PDGFRB RNF125 UPF3B

Invitae Perlman Syndrome Test (1 gene)

Primary panel (1 gene)
DIS3L2

Invitae Proteus Syndrome Test (1 gene)

Primary panel (1 gene)
AKT1

Invitae PTEN-Related Disorders Test (1 gene)

Primary panel (1 gene)
PTEN

INVITAE PEDIATRIC GENE PANEL TESTS (continued)

OVERGROWTH SYNDROMES (continued)

Invitae Simpson-Golabi-Behmel Syndrome Test (1 gene)	Primary panel (1 gene) GPC3
Invitae Sotos Syndrome Test (1 gene)	Primary panel (1 gene) NSD1
Invitae Weaver Syndrome Test (1 gene)	Primary panel (1 gene) EZH2

RASOPATHIES (NOONAN SPECTRUM DISORDERS)

Invitae RASopathies Comprehensive Panel (18 genes)	Primary panel (18 genes) A2ML1 BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1 NRAS PTPN11 RAF1 RASA1 RIT1 RRAS SHOC2 SOS1 SOS2 SPRED1
Invitae Cardio-Facio-Cutaneous Syndrome Panel (6 genes)	Primary panel (6 genes) BRAF KRAS MAP2K1 MAP2K2 SHOC2 SOS1
Invitae Costello Syndrome Test (1 gene)	Primary panel (1 gene) HRAS
Invitae Legius Syndrome Test (up to 2 genes)	Primary panel (1 gene) SPRED1 Add-on Legius syndrome gene (1 gene) NF1
Invitae Neurofibromatosis Type 1 Test (up to 2 genes)	Primary panel (1 gene) NF1 Add-on neurofibromatosis type 1 gene (1 gene) SPRED1
Invitae Noonan Syndrome Panel (up to 16 genes)	Primary panel (14 genes) A2ML1 BRAF CBL KRAS MAP2K1 MAP2K2 NRAS PTPN11 RAF1 RIT1 RRAS SHOC2 SOS1 SOS2 Add-on Baraitser-Winter cerebrofrontofacial syndrome genes (2 genes) ACTB ACTG1
Invitae Noonan Syndrome with Multiple Lentigines Pane (3 genes)	Primary panel (3 genes) BRAF PTPN11 RAF1

SKELETAL DISORDERS

Invitae Antley-Bixler Syndrome Test (up to 2 genes)	Primary panel (1 gene) POR Add-on craniosynostosis gene (1 gene) FGFR2
Invitae ARSE-Related Chondrodysplasia Punctata Test (up to 2 genes)	Primary panel (1 gene) ARSE Add-on NSDHL-related disorders gene (1 gene) NSDHL
Invitae Campomelic Dysplasia Test (1 gene)	Primary panel (1 gene) SOX9
Invitae Craniosynostosis Panel (up to 11 genes)	Primary panel (9 genes) ERF FGFR1 FGFR2 FGFR3 GLI3 MEGF8 MSX2 RAB23 TWIST1 Add-on 3MC and Treacher-Collins syndromes genes (2 genes) MASP1 TCOF1
Invitae Duane-Radial Ray Syndrome Test (1 gene)	Primary panel (1 gene) SALL4
Invitae Ellis van Creveld and Weyers Acrofacial Dysostosis Panel (2 genes)	Primary panel (2 genes) EVC EVC2
Invitae FGFR3-Related Disorders Test (1 gene)	Primary panel (1 gene) FGFR3
Invitae Hereditary Multiple Osteochondromas Panel (up to 3 genes)	Primary panel (2 genes) EXT1 EXT2 Add-on Langer-Giedion syndrome gene (1 gene) TRPS1
Invitae Holt-Oram Syndrome Test (1 gene)	Primary panel (1 gene) TBX5
Invitae NSDHL-Related Disorders Test (1 gene)	Primary panel (1 gene) NSDHL
Invitae Osteogenesis Imperfecta Panel (4 genes)	Primary panel (4 genes) COL1A1 COL1A2 CRTAP P3H1

SKELETAL DISORDERS (continued)

Invitae Skeletal Ciliopathies Panel (up to 18 genes)	Primary panel (17 genes) CEP120 CSPP1 DYNC2H1 EVC EVC2 IFT80 IFT122 IFT140 IFT172 KIAA0586 NEK1 TCTN3 TTC21B WDR19 WDR34 WDR35 WDR60 Add-on FGFR3-related thanatophoric dysplasia gene (1 gene) FGFR3
Invitae Thrombocytopenia Absent Radius Syndrome Test (1 gene)	Primary panel (1 gene) RBM8A
Invitae Townes-Brocks Syndrome Test (1 gene)	Primary panel (1 gene) SALL1
Invitae Treacher-Collins Syndrome Test (1 gene)	Primary panel (1 gene) TCOF1
Invitae Trichorhinophalangeal Syndrome Panel (2 genes)	Primary panel (2 genes) EXT1 TRPS1
Invitae Ulnar-Mammary Syndrome Test (1 gene)	Primary panel (1 gene) TBX3

SKIN DISORDERS

Invitae Cardio-Facio-Cutaneous Syndrome Panel (6 genes)	Primary panel (6 genes) BRAF KRAS MAP2K1 MAP2K2 SHOC2 SOS1
Invitae Ectodermal Dysplasia with or without Tooth Agenesis Panel (up to 10 genes)	Primary panel (8 genes) EDA EDAR EDARADD LTBP3 MSX1 NFKBIA PAX9 WNT10A Add-on Clouston syndrome and TP63-related disorder genes (2 genes) GJB6 TP63
Invitae Legius Syndrome Test (up to 2 genes)	Primary panel (1 gene) SPRED1 Add-on neurofibromatosis type 1 gene (1 gene) NF1
Invitae Noonan Syndrome with Multiple Lentigines Panel (3 genes)	Primary panel (3 genes) BRAF PTPN11 RAF1
Invitae PTEN-Related Disorders Test (1 gene)	Primary panel (1 gene) PTEN
Invitae TP63-Related Disorders Test (1 gene)	Primary panel (1 gene) TP63
Invitae van der Woude Syndrome Panel (2 genes)	Primary panel (2 genes) GRHL3 IRF6

CLINICAL AREA: HEREDITARY CANCER

PEDIATRIC ONCOLOGY

Invitae Pediatric Solid Tumors Panel (48 genes)	Primary panel (48 genes) 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
Invitae Pediatric Hematologic Malignancies Panel (16 genes)	Primary panel (16 genes) ATM BLM CEBPA EPCAM GATA2 HRAS MLH1 MSH2 MSH6 NBN NF1 PMS2 RUNX1 TERC TERT TP53
Invitae Pediatric Nervous System/Brain Tumors Panel (up to 32 genes)	Primary panel (24 genes) ALK APC DICER1 EPCAM HRAS MEN1 MLH1 MSH2 MSH6 NF1 NF2 PHOX2B PMS2 PRKAR1A PTCH1 PTEN RB1 SMARCB1 SMARCE1 SUFU TP53 TSC1 TSC2 VHL Add-on hereditary paraganglioma pheochromocytoma genes (8 genes) MAX RET SDHA SDHAF2 SDHB SDHC SDHD TMEM127

INVITAE

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