

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	RPGRI1L	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	RPGRI1L	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	RPGR	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 PEDIATRIC GENE PANEL TESTS (continued)

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) PQBPI
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 Disorders of Female Sex Development Test (1 gene)

Primary panel (1 gene)

SRY

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 (133 genes)

ADSL ALDH5A1 ALDH7A1 ALG13 ARHGEF9 ARX ATP1A2 ATP1A3 ATRX BRAT1 C12orf57 CACNA1A
CACNA2D2 CASK CDKL5 CHD2 CHRNA2 CHRNB2 CLCN4 CLN2 (TPP1) CLN3 CLN5 CLN6
CLN8 CNTNAP2 CSTB CTSD DEPDC5 DNAJC5 DNMT1 DOCK7 DYRK1A EEF1A2 EFHC1 EHMT1
EPM2A FOLR1 FOXP1 FRRS1L GABRA1 GABRB3 GABRG2 GAMT GATM GLDC GLRA1 GNAO1
GOSR2 GRIN1 GRIN2A GRIN2B HCN1 HNRNPU IER3IP1 IQSEC2 ITPA KANSL1 KCNA2 KCNB1
KCNK1 KCNH2 KCNJ10 KCNQ2 KCNQ3 KCNT1 KCTD7 KIAA2022 LGI1 LIAS MBD5 MECP2
MEF2C MFSB8 MTOR NEDD4L NGLY1 NHLRC1 NRXN1 PACS1 PCDH19 PIGA PIGN PIGO
PLCB1 PNKD PNKP PNPO POLG PPT1 PRICKLE1 PRRT2 PURA QARS ROGD1 SATB2
SCARB2 SCN1A SCN1B SCN2A SCN3A SCN8A SCN9A SERPINI1 SGCE SLC12A5 SLC13A5 SLC19A3
SLC25A22 SLC2A1 SLC35A2 SLC6A1 SLC6A8 SLC9A6 SNX27 SPATA5 SPTAN1 ST3GAL5 STRADA STX1B
STXBP1 SYN1 SYNGAP1 SYNJ1 SZT2 TBC1D24 TCF4 TSC1 TSC2 UBE3A WWOX ZDHHC9
ZEB2

Add-on preliminary-evidence genes (50 genes)

ABAT ARHGEF15 ATP6AP2 CACNA1H CACNB4 CARS2 CASR CBL CERS1 CNTN2 COQ4 CPA6
DIAPH1 FARS2 FASN GABBR2 GABRB2 GABRD GAL GPHN JMJDC1 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

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 65 genes)

Primary panel (53 genes)

ALDH7A1 ARHGEF9 ARX BRAT1 CACNA2D2 CASK CDKL5 CHD2 CLCN4 DNMT1 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 SLC13A5 SLC25A22 SLC2A1 SLC35A2 SLC6A1 SMC1A SPTAN1
STXBP1 SYNGAP1 SZT2 TBC1D24 WWOX

Add-on preliminary-evidence genes (12 genes)

ARHGEF15 ATP1A2 COQ4 GABBR2 GPHN KCNH5 MTOR NECAP1 NEDD4L SCN1B SIK1 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)

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) AZML1 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) AZML1 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 Lentiginos 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

INVITAE PEDIATRIC GENE PANEL TESTS (continued)

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