

Panel Testing

- [Cerebral Cavemous Malformations Panel](#)

CCM2, KRIT1, MAP3K3, PDCD10, PIK3CA

- [Childhood Interstitial Lung Disease \(chILD\) Panel](#)

ABCA3, ACVRL1, AP3B1, ATP13A3, BMPR1B, BMPR2, CAV1, CCBE1, COPA, CSF2RA, CSF2RB, DICER1, DKC1, EIF2AK4, ELMOD2, ENG, FAT4, FGF10, FLCN, FLNA, FLT4, FOXC2, FOXF1, FOXP1, GATA2, GDF2, HPS1, HPS4, ITGA3, KCNK3, MARS1, MUC5B, NKX2-1, OAS1, PARN, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC7A7, SMAD4, SMAD9, SMPD1, STAT3, STING1, STRA6, TBX4, TERC, TERT, TINF2

- [Congenital Hyperinsulinism Panel](#)

ABCC8, FOXA2, GCK, GLUD1, HADH, HNF1A, HNF4A, INSR, KCNJ11, SLC16A1

- [Craniosynostosis Panel](#)

ACTB, ACTG1, ADAMTSL4, AHDC1, ALPL, ALX4, ASXL1, ASXL3, ATR, B3GAT3, CDC45, CHD5, CHD7, COLEC10, COLEC11, CTSK, CYP26B1, EFNB1, ERF, FAM20C, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FREM1, GLI3, GPC3, HNRNPK, HUWE1, IFT122, IFT43, IGF1R, IL11RA, IRX5, JAG1, KAT6A, KAT6B, KMT2D, KRAS, MAN2B1, MASP1, MEGF8, MSX2, P4HB, PHEX, POR, PTCH1, RAB23, RECQL4, RUNX2, SIX1, SKI, SLC25A24, SMAD6, SOX6, SPECC1L, STAT3, TCF12, TFAP2B, TGFB1, TGFB2, TMCO1, TRAF7, TWIST1, WDR19, WDR35, ZEB2, ZIC1, ZNF462

- [Differences in Sex Development Panel](#)

AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, CBX2, CDKN1C, CHD7, CUL7, CYP11A1, CYP17A1, CYP19A1, DHH, DMRT1, DMRT2, FEZF1, FGF8, FGFR1, FSHB, GATA4, GNRH1, GNRHR, HESX1, HSD17B3, HSD3B2, IL17RD, KISS1R, LHCGR, LHX3, LHX4, MAMLD1, MAP3K1, NROB1, NR5A1, NSMF, PCSK1, POR, PROK2, PROKR2, PROP1, PSMC3IP, RSPO1, SEMA3A, SOX3, SOX9, SRD5A2, SRY, STAR, TAC3, TACR3, WDR11, WNT4, WT1, WWOX, ZFPM2

- [Encephalocraniocutaneous Lipomatosis \(ECCL\) and Oculoectodermal Syndrome \(OES\) Panel](#)

FGFR1, KRAS

- [Hereditary Hemorrhagic Telangiectasia Panel](#)

ACVRL1, ENG, GDF2, SMAD4, RASA1, EPHB4

- [ImmuneSeq Panel – see LabMan entry for gene list \(≥700 genes\)](#)

- [Interstitial Pseudo-Obstruction Panel](#)
ACTA2, ACTG2, CHD8, CLMP, ERBB3, FLNA, LIG3, LMOD1, MYH11, MYL9, MYLK, POLG, POLG2, RAD21, SGO1, TYMP
- [MODY Panel](#)
ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, INS-IGF2, KCNJ11, KLF11, NEUROD1, PAX4, PDX1
- [Neonatal Diabetes Panel](#)
ABCC8, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1B, IER3IP1, INS, INS-IGF2, KCNJ11, MNX1, NEUROD1, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, WFS1, ZFP57
- [Rett/Angelman Syndrome Panel](#)
CDKL5, CLCN4, CNTNAP2, DYRK1A, EHMT1, FOXG1, MBD5, MECP2, MEF2C, NRXN1, SATB2, SLC9A6, TCF4, UBE3A, WDR45, ZEB2
- [VanSeq Vascular Anomalies Panel](#) – subpanel info available: [SCH VANseq Panel Descriptions](#)
ACVRL1, ANGPT2, ARAF, BRAF, CCBE1, CCM2, CELSR1, CTNNA1, DCHS1, ELMO2, ENG, EPHB4, FAT4, FGFR1, FLT4, FOXC2, GATA2, GDF2, GJC2, GLMN, GNA11, GNA14, GNAQ, HGF, HRAS, IDH1, IDH2, KIF11, KRAS, KRIT1, MAP2K1, MAP3K3, MDFIC, MET, NRAS, PDCD10, PDGFRB, PIEZO1, PIK3CA, PIK3R1, PTEN, PTPN14, RASA1, SMAD4, SOX18, TEK, VEGFC

Sanger Gene Testing

- *ACADM*
- *ACADVL*
- *AGXT*
- *ALDH7A1*
- *ATP7B*
- *FAH*
- *GAA*
- *GALT*
- *GBA*
- *GJB2*
- *GJB6*
- *HADHA*
- *HADHB*
- *LIPA*
- *POLG*
- *POLG2*
- *SLC26A4*