Seattle Children's
HOSPITAL • RESEARCH • FOUNDATION

Panel Testing

- Cerebral Cavernous 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, TGFBR1, TGFBR2, 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, NR0B1, 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)
  Genes with known pseudogenes or repetitive regions that may impact ability to detect variants & result in low coverage are marked with (*). Before requesting a gene with these known limitations, please contact our Lab Genetic Counseling Team for approval: LabGC@seattlechildrens.org or (206) 987-5400.

- 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, CTNNB1, 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

Biochemical Genetics Genes

- *MCAD (ACADM) Sequencing*
- *VLCAD (ACADVL) Sequencing*
- *Wilson Disease (ATP7B) Sequencing*
- *Pompe (GAA) Sequencing*
- *Galactosemia (GALT) Sequencing*