Gene List for Deletion/Duplication by Array

- Cerebral Cavernous Malformations Panel

  CCM2, KRIT1, PDCD10

- Childhood Interstitial Lung Disease (chILD) Panel

  ABCA3, ACVRL1, 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, MARS, NKX2-1, OAS1, PARN, RASA1, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC7A7, SMAD4, SMAD9, SMPD1, STAT3, STRA6, TBX4, TERC, TERT, TINF2

- Congenital Hyperinsulinism Panel

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

- Craniosynostosis Panel

  AHDC1, ALPL, ALX4, ASXL1, ATR, CDC45, COLEC11, CTSK, CYP26B1, EFNB1, ERF, FAM20C, FGFR1, FGFR2, FGFR3, FLNA, FREM1, GLI3, GPC3, HUWE1, IFT122, IFT43, IGF1R, IL11RA, IRX5, JAG1, KAT6A, KMT2D, KRAS, MASP1, MEGF8, MSX2, PHEX, POR, RAB23, RECQL4, RUNX2 , SKI, SPECC1L, STAT3, TCF12, TGFBR1, TGFBR2, TMCO1, TWIST1, WDR19, WDR35, ZEB2, ZIC1

- 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

- Interstitial Pseudo-Obstruction Panel

  ACTA2, ACTG2, CHD8, CLMP, FLNA, LMOD1, MYH11, MYLK, 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, SLCA2, 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, 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, MET, NRAS, PDCD10, PDGFRB, PIEZO1, PIK3CA, PTEN, PTPN14, RASA1, SMAD4, SOX18, TEK, VEGFC

Additional genes

- *ACADM*
- *AGXT*
- *ALDH7A1*
- *ATP7B*
- *FAH*
- *GJB2*
- *GJB6*
- *HADHA*
- *HADHB*
- *SLC26A4*