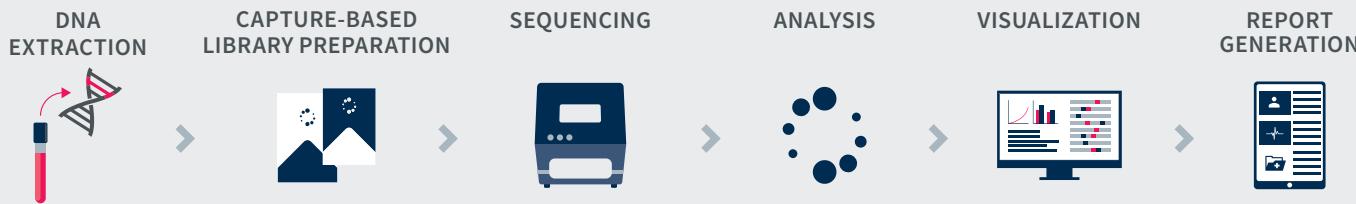

COMMUNITY PANELS
HIGHLIGHTS

- Uniform coverage of target regions
- High quality probe design to optimize on-target rate
- Developed with genomic experts in Rare and Inherited Diseases
- Simple and reliable data analysis and interpretation

Accelerate your analysis with pre-designed and tested panels

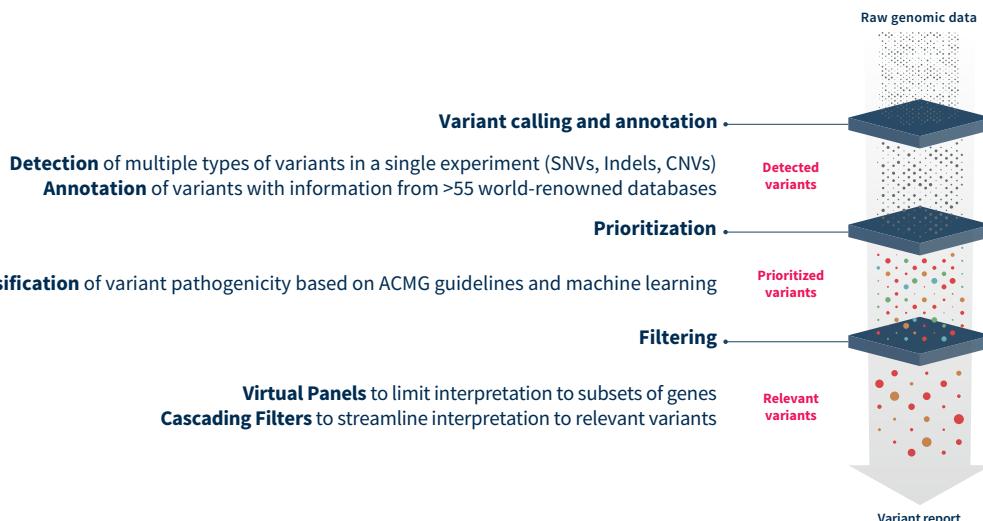
Designing, creating, and testing a new gene panel takes considerable time and effort. **SOPHiA DDM™ Community Panels** are **targeted, capture-based NGS panels** developed and tested by genomic experts to minimize set-up challenges and accelerate your research. These panels cover a wide range of Rare and Inherited Diseases, with the flexibility to add or remove genes to meet your unique requirements.

In combination with the analytical and interpretation capabilities of the SOPHiA DDM™ Platform and Alamut™ Visual Plus, our Community Panels help you to gain accurate and cost-effective insights from your target regions of interest.



For a fast and worry-free transition to routine analysis, the **SOPHiA GENETICS™ Set Up Program** provides full set-up assistance.

The SOPHiA DDM™ **Community Panels** leverage on the SOPHiA DDM™ Platform and Alamut™ Visual Plus to ensure accurate variant detection and streamlined variant assessment.



Discover our Community Panels for **Cardiovascular Diseases**, **Hereditary Cancers**, **Metabolic Disorders**, **Neurological Disorders**, and **Pediatric Diseases** including Autoinflammatory Diseases, Dermatological Diseases, and Developmental Disorders, in this booklet.

COMMUNITY PANELS FOR CARDIOVASCULAR DISEASES

Panel, sequencer, and panel size	Associated diseases*	Genes covered
CCAS_109 NextSeq® 398 kb	Arrhythmias, cardiomyopathies, channelopathies, and muscular dystrophies: Brugada syndrome, Danon disease, Duchenne muscular dystrophy, Emery-Dreifuss syndrome, Holt-Oram syndrome, Fabry disease, glycogen storage disease, lipodystrophy, long QT syndrome	109 genes: ABCC9, ACTA1, ACTC1, ACTN2, ANK2, ANKRDI, APOA1, BAG3, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CDH2, CRYAB, CSR3, DES, DMD, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EMD, FHL1, FHL2, FLCN, GAA, GATA4, GJA1, GJA5, GJC1, GLA, GPD1L, HCNA4, JPH2, JUP, KCNA5, KCNAB2, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LAMA4, LAMP2, LDB3, LMNA, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX25, NPPA, NUP155, PDLIM3, PKP2, PLN, PPA2, PRDM16, PRKAG2, RBM20, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SGCD, SLC22A5, SLC2A5, SLC8A1, SNTA1, TAZ, TBX5, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, VCL
CCAS_163 MiSeq® and NextSeq® 567 kb	Arrhythmias, cardiomyopathies, channelopathies, and muscular dystrophies: atrial fibrillation, Brugada syndrome, Danon disease, Duchenne muscular dystrophy, Emery-Dreifuss syndrome, heart hand syndrome, Holt-Oram syndrome, Fabry disease, glycogen storage disease, lipodystrophy, long QT syndrome	163 genes: ABCC9, ACTA1, ACTC1, ACTN2, ANK2, ANKRDI, APOA1, BAG3, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CDH2, CRYAB, CSR3, DES, DMD, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EMD, FHL1, FHL2, FLCN, GAA, GATA4, GJA1, GJA5, GJC1, GLA, GPD1L, HCNA4, JPH2, JUP, KCNA5, KCNAB2, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LAMA4, LAMP2, LDB3, LMNA, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NPPA, NUP155, PDLIM3, PKP2, PLN, PPA2, PRDM16, PRKAG2, RBM20, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SGCD, SLC22A5, SLC4A3, SLC8A1, SNTA1, TAZ, TBX5 TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, VCL
CD_9 MiSeq® 33 kb	Familial hypercholesterolemia, hypobetalipoproteinemia, sitosterolemia, atherosclerosis susceptibility	9 genes: ABCG5, ABCG8, APOB, APOE, LDLR, LDLRAP1, LIPA, PCSK9, STAP1

COMMUNITY PANELS FOR HEREDITARY CANCERS

Panel, sequencer, and multiplexing	Associated cancers*	Genes covered
HC_55 MiSeq® v2, 24 samples MiSeq® v3, 32 samples	Breast cancer, colorectal cancer and polyposis syndromes, gastric cancer, hereditary breast and ovarian cancer, hereditary melanoma, neuroendocrine tumors, ovarian cancer, pancreatic cancer, renal tumors	55 genes: AIP, AIRE, AP2S1, APC, ATM, BAP1, BMPR1A, BRCA1, BRCA2, BRIP1, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, EPCAM, FH, FLCN, GATA3, GCM2, GNA11, GREM1, MAX, MEN1, MET, MLH1, MSH2, MSH6, MUTYH, NTHL1, PALB2, PMS2, PMS2CL, POLD1, POLE, PRKAR1A, PTEN, PTH, RAD51C, RAD51D, RET, RNF43, SCG5, SDHAF2, SDHB, SDHC, SDHD, SMAD4, STK11, TBCE, TMEM127, TP53, VHL
HC_60 MiSeq® v2, 16 samples	Breast cancer, colorectal cancer and polyposis syndromes, gastric cancer, hereditary breast and ovarian cancer, hereditary melanoma, neuroendocrine tumors, ovarian cancer, pancreatic cancer, renal tumors	60 genes: APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, DDB2, CDC73, CDK1, CDKN1B, CDKN2A, EGFR, EGR1, EPCAM, ERCC2, ERCC4, ERCC5, FANCA, FANCC, FH, FLCN, GATA3, GCM2, GNA11, GREM1, MAX, MEN1, MET, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PMS2, POLD1, POLE, POLH, PTCH1, PTEN, RAD51C, RAD51D, RB1, RET, SMAD4, TSC1, STK11, TP53, TSC2, VHL, WT1, XPA, XPC
HC_66 MiSeq® v3, 24 samples	Breast cancer, colorectal cancer and polyposis syndromes, gastric cancer, hereditary breast and ovarian cancer, hereditary melanoma, neuroendocrine tumors, neurofibromatosis 1, ovarian cancer, pancreatic cancer, renal tumors, thyroid cancer	66 genes: ABRAVAS1, AP2S1, APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, CTNNAI, Dicer1, EPCAM, FH, FLCN, GCM2, GNA11, GREM1, MAX, MEN1, MET, MLH1, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PIK3CA, PMS2, PMS2CL, POLD1, POLE, POT1, PRKAR1A, PTEN, RAD50, RAD51C, RAD51D, RB1, RET, SCG5, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, STK11, TMEM127, TP53, TSC1, TSC2, VHL, XRCC2
HC_117 NextSeq® 500/550 mid-output, 48 samples NextSeq® 500/550 high-output, 144 samples NextSeq® 1000/2000 P2, 144 samples MiniSeq™ high-output, 10 samples NovaSeq® 6000 SP 1 lane, 144 samples NovaSeq® 6000 S1 1 lane, 288 samples	Breast cancer, colorectal cancer and polyposis syndromes, cutaneous tumors, Fanconi anemia, gastric cancer, hereditary breast and ovarian cancer, hereditary melanoma, leukemia, nervous system tumors, neuroendocrine tumors, neurofibromatosis 1, ovarian cancer, pancreatic cancer, prostate cancer, renal tumors, sarcomas, skin cancer, thyroid cancer	117 genes: ACD, AIP, AKT1, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CEBPA, CHEK2, CREBBP, CTNNAI, Dicer1, DIS3L2, EGFR, EGLN1, EPCAM, ERBB2, ERCC2, EXT1, EXT2, FANCC, FANCG, FANCM, FH, FLCN, GATA2, GATA3, GREM1, HNF1A, HOXB13, KIF1B, KIT, LZTR1, MAX, MC1R, MDH2, MEN1, MET, MIF1, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NSD1, NTHL1, PALB2, PDGFRA, PHOX2B, PMS2, PMS2CL, POLD1, POLE, POLH, POT1, PRKAR1A, PRSS1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL, RECQL4, RET, RHBDF2, RNFA43, RPS20, RUNX1, SCG5, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TERC, TERF2IP, TERT, TMEM127, TP53, TSC1, TSC2, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2, YAP1
HC_144 NextSeq® mid-output, 64 samples	Breast cancer, colorectal cancer and polyposis syndromes, cutaneous tumors, Fanconi anemia, gastric cancer, hereditary breast and ovarian cancer, hereditary melanoma, leukemia, nervous system tumors, neuroendocrine tumors, neurofibromatosis 1, ovarian cancer, pancreatic cancer, prostate cancer, renal tumors, sarcomas, skin cancer, thyroid cancer	144 genes: AIP, ALK, APC, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CEBPA, CEP57, CHEK2, CTC1, CYLD, DDB2, Dicer1, DIS3L2, DKK1, EGFR, EGLN1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, EZH2, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCM, FH, FLCN, GATA2, GATA3, GREM1, HNF1A, HOXB13, KIF1B, KIT, KM2D2, MAX, MC1R, MDH2, MEN1, MET, MERTK, MRE11A, MIF1, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NHP2, NOP10, NSD1, PALB2, PDGFRA, PHOX2B, PMS1, PMS2, POLD1, POLE, POLH, POT1, PRF1, PRKAR1A, PRSS1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL, RET, RHBDF2, RUNX1, SBD5, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TINF2, TMEM127, TERC, TERT, TP53, TSC1, TSC2, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2, YAP1

*The disorders covered by the Community Panels include, but are not limited to, those listed here.

For some Community Panels, additional permissions may be required. Contact us at info@sophiagenetics.com to discuss the panel that suits your needs.



COMMUNITY PANELS FOR METABOLIC DISORDERS

Panel, sequencer, and panel size	Associated disorders*	Genes covered
MD_55 MiSeq® 103 kb	Bile and liver disorders: Bile conjugation defects, bile malabsorption, bile synthesis defect, cholestasis, Dubin-Johnson syndrome, gallbladder disease, Gilbert's Syndrome, hyperbilirubinemia Metabolism, bone, and joint disorders: Arterial calcification, arthrogryposis, citrullinemia, mitochondrial trifunctional protein deficiency (MTPD), dominant hypophosphatemia with nephrolithiasis or osteoporosis, pseudoxanthoma elasticum, sitosterolemia, vitamin D-dependent rickets	55 genes: ABCB11, ABCB4, ABCC2, ABCC3, ABCC4, ABCF3, ABCG2, ABCG5, ABCG8, ALAS1, AQP8, ATP8B1, BAAT, CLDN1, ELOB, F11R, FABP6, GPBAR1, HADHA, HSD3B7, LTBP4, MCM3AP, MRPS9, MYO5B, NPC1L1, NR1H4, PCSK9, PEMT, PFDN6, PNPLA3, PPIH, PPP4R2, PPP5C, RAB11A, RABEP1, SLC10A1, SLC10A2, SLC15A4, SLC25A13, SLC42A2, SLC9A3R1, SLC01A2, SLC01B1, SLC02B1, SRY, SULT2A1, TJP2, UBE2Z, UGT1A1, USP4, VDR, VIPAS39, VPS33B
MD_50 MiSeq® 105 kb	Dislipidemias: arterial calcification, atherosclerosis, Danon disease, Fabry disease, familial hypercholesterolemia, fucosidosis, Gaucher disease, GM1 gangliosidosis, hepatic lipase deficiency, Hurler syndrome, hypoalphalipoproteinemia, mucopolysaccharidosis, myopathy, neuraminidase deficiency, pseudoxanthoma elasticum, Scheie syndrome, sitosterolemia, Tangier disease	50 genes: ABCA1, ABCC6, ABCG5, ABCG6, ANGPTL3, ANGPTL4, ANGPTL8, APOA1, APOA5, APOB, APOC2, APOC3, APOE, ARSB, ARSH, ASAHI, CETP, CYP27A1, ENPP1, FUC1, GAA, GALNS, GBA, GGCX, GLA, GLB1, GM2A, GPIHBP1, IDS, IDUA, LAMP2, LCAT, LDLR, LDLRAP1, LIPA, LIPC, LMF1, LPL, MTTP, MYLIP, NAGLU, NEU1, NPC1, NPC2, PCSK9, PSAP, SAR1B, SMPD1, SUMF1, VEGFA

COMMUNITY PANELS FOR NEUROLOGICAL DISORDERS

Panel, sequencer, and panel size	Associated disorders*	Genes covered
ND_83 MiSeq® and NextSeq® 221 kb	Epilepsy	83 genes: ALDH7A1, ALG13, ARHGEF9, ASAHI, ATN1, ATP6V1A, BRAT1, CAMK2A, CDKL5, CERS1, CHD2, CLN3, CLN5, CLN6, CLN8, CSTB, CTSA, CTD5, CTSF, DNAJC5, DNMT1, EEF1A2, EPM2A, FGF12, GABRA1, GABRG2, GBA, GNAO1, GOSR2, GRIN1, GRIN2A, GRIN2B, GRN, HCN1, HNRNPU, HTT, IQSEC2, KCNA2, KCNB1, KCNC1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD7, MEF2C, MFSD8, NEU1, NEXMIF, NHLC1, NPC1, NPC2, NTRK2, PACS2, PCDH19, PIGA, PNKP, PNPO, PPT1, PRICKLE2, PRRT2, QARS, SACS, SCARB2, SCN1A, SCN2A, SCN8A, SLC25A22, SLC2A1, SLC35A2, SLC35A3, SLC6A1, SPTAN1, ST3GL3, STXBP1, SYNGAP1, SQT2, TBC1D24, TPP1, UBA5, WDR45, WWOX
ND_286 MiSeq® 867 kb	Intellectual disabilities: Angelman syndrome, autism, cerebellar ataxia, congenital disorders of glycosylation, Cornelia de Lange syndrome, developmental and epileptic encephalopathy, intellectual development disorders, Kabuki syndrome, KBG syndrome Neurodevelopmental disorders: Pettigrew syndrome, Sotos syndrome, spastic paraplegia, Temtamy syndrome, Timothy syndrome, Tonne-Kalscheuer syndrome	286 genes: ACSL4, ACY1, ADSL, AFF2, AGO1, AGTR2, ALDH18A1, ALDH5A1, ALG6, AMT, ANK3, ANKRD11, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, ARHGEF6, ARHGEF9, ARID1B, ARIH1, ARX, ASPM, ATP6AP2, ATP7A, ATRX, AUTS2, BCKDK, BCOR, BRWD3, C12orf57, CAB, CACNA1C, CACNA1F, CACNG2, CAMTA1, CASK, CC2D1A, CCDC22, CDH15, CDK5RAP2, CDKL5, CENPJ, CHD8, CHRNA7, CLCN4, CLIC2, CNKS2, CNTNAP2, CRBN, CREBBP, CTNNB1, CUL4A, CYFIP1, CYP2U1, DCX, DHD2, DPB2, DISC1, DKC1, DLG2, DLG3, DMD, DOCK8, DYNC1H1, DYRK1A, EHMT1, EIF2S3, ELP2, EP300, EPB41L1, ERLIN2, FGD1, FLNA, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FRMPD4, FTCD, FTSJ1, GABRB3, GAMT, GATA2B, GDI1, GK, GPC3, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, HAL, HCCS, HCF1, HDAC4, HDAC8, HEpacam, HIST1H4B, HIVEP2, HPRT1, HRAS, HSD17B10, HUWE1, IDS, IGBP1, IGF1, IGF1R, IKBKG, IL1RAPL1, IQSEC2, KANSL1, KATNAL2, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCTD13, KDM5A, KDM5C, KDM6A, KIF1A, KLF8, KLHL15, KMT2A, KMT2D, L1CAM, LAMC3, LAMP2, LAMTOR3, LASL1, LIMK1, LRP2, MAGT1, MAN1B1, MANBA, MAOA, MBDS, MBTPS2, MCPH1, MECP2, MED12, MED13L, MED17, MED23, MEF2C, MIDI, MIR222, MTHFR, MTNR1A, MTNR1B, MYT1L, NAA10, NDP, NDUF1, NEXMIF, NFX1, NHS, NIPBL, NLGN3, NLGN4X, NPC2, NRXN1, NRXN2, NSD1, NSDHL, NSUN2, NTNG1, NXFS, OCRL, OFD1, OPHN1, OTC, PAX3, PRK1, PAFAH1B1, PAK3, PCDH19, PCDH8, PDHA1, PDHX, PEX7, PHF6, PIGV, PLP1, PMM2, PNPK, POGZ, PORCN, PQB1, PRKR4, PRODH, PRPS1, PRRT2, PRSS12, PTCHD1, PTEN, RAB39B, RAB40AL, RAD21, RAI1, RBM10, RELN, RLIM, RNF135, RPL10, RPS6KA3, SATB2, SCNA1, SCNA2, SCNA8, SETBP1, SETD5, SHANK1, SHANK2, SHANK3, SHROOM4, SKI, SLC16A2, SLC2A1, SLC46A1, SLC6A1, SLC6A17, SLC6A8, SLC9A6, SMARCA2, SMC1A, SMC3, SMS, SOBP, SOX3, SOX5, SPTAN1, SRD5A3, SRGAP3, SRPX2, ST3GL3, STIL, STXBP1, SYN1, SYNGAP1, SYT, TBL1XR1, TBR1, TCF4, TECPR2, TECR, TIMM8A, TRAPP9, TRIO, TSC2, TSPAN7, TUBA1A, TUBB2B, TUSC3, UBE2A, UBE3A, UBE3B, UBR1, UPB1, UPF3B, UROC1, USP9X, VLDLR, VPS13B, WDR45B, WDR62, Y1, ZBTB20, ZC3H14, ZC4H2, ZCHCH12, ZDHHC15, ZDHHC9, ZEB2, ZNF292, ZNF41, ZNF526, ZNF674, ZNF711, ZNF81
ND_45 Ion55 240 kb	Autism	45 genes: ADNP, ANK2, ANKRD11, ARID1B, ASH1L, CHD2, CHD8, CNTNAP2, DDX3X, DSCAM, DYNC1H1, DYRK1A, FOXP1, GRIA3, GRIN1, GRIN2B, HOMER1, KMT2A, KMT2C, KMT5B, LAMC3, MECP2, MED13L, MEF2C, NAA15, NLGN3, NLGN4X, NRXN1, NRXN2, NRXN3, OPHN1, PAX5, POGZ, PTEN, RAB39B, SCN2A, SHANK1, SHANK2, SHANK3, SYNGAP1, TBL1XR1, TBR1, TRIO, TRIP12, WDFY3
ND_117 MiSeq® 435 kb	Hypoacusis	117 genes: ACTG1, ADCY1, ADGRV1, BDP1, BSN, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A2, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, FY4A, FOXJ1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HAR52, HGF, HOMER2, HSD17B4, ILDR1, KARS, KCNE1, KCNQ1, KCNQ4, KITLG, LARS2, LHFP1, LOXHD1, LRTO1, MARVELD2, MCM2, MET, MIR182, MIR183, MIR96, MITF, MSR83, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OSBPL2, OTOA, OTOF, OTOG, OTOG1, P2RX2, PCDH15, PDZD7, PVK, PNPT1, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RIPOR2, S1PR2, SERPINB6, SIK3, SIX1, SLC17A8, SLC22A4, SLC26A4, SLTRK6, SMPX, SOX10, STRC, SYNE4, TBC1D24, TECTA, TIMM8A, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOPB, TSPEAR, TWNK, USH1C, USH1G, USH2A, WBP2, WFS1, WHRN

*The disorders covered by the Community Panels include, but are not limited to, those listed here.

For some Community Panels, additional permissions may be required. Contact us at info@sophiagenetics.com to discuss the panel that suits your needs.



COMMUNITY PANELS FOR PEDIATRIC DISEASES

Including Autoinflammatory Diseases, Dermatological Diseases, and Developmental Disorders

Panel, sequencer, and panel size	Associated diseases*	Genes covered
Autoinflammatory Diseases		
PD_17 MiSeq® 40 kb	Familial fever: Autoinflammatory syndrome, Blau syndrome, familial Mediterranean fever, inflammatory bowel disease, periodic fever	17 genes: ADA2, CARD14, ELANE, IL10RA, IL10RB, IL1RN, LPIN2, MEFV, MVK, NLRP12, NLRP3, NLRP7, NOD2, PSMB8, PSTPIP1, TNFRSF11A, TNFRSF1A
PD_61 MiSeq® and NextSeq® 125 kb	Autoinflammatory disorders: Acne inversa, Aicardi-Goutieres syndrome, Alzheimer's disease, auto-immune diseases, auto-inflammation syndromes, chilblain lupus, Crohn's disease, ectodermal dysplasia with immunodeficiency-1, immunodeficiencies, juvenile arthritis, familial fever syndromes, Kosaki overgrowth syndrome, lymphoproliferative syndrome	61 genes: ACP5, ADA2, ADAR, ADGRE2, ARPC1B, CARD14, CDC42, COPA, CTLA4, DDX58, DNASE1, DNASE1L3, DNASE2, FOXP3, IFIH1, IL1RN, IL36RN, ISG15, JAK1, LACC1, LPIN2, LRBA, MEFV, MVK, NCSTN, NFKB1, NLRC4, NLRP1, NLRP3, NOD2, OTULIN, PDGFRB, PLCG2, POLA1, POMP, PRKCD, PSEN1, PSENEN, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, RBC1, RNASEH2A, RNASEH2B, RNASEH2C, RNF31, SAMHD1, SERPING1, SHARPIN, STAT1, STAT3, TMEM173, TNFAIP3, TNFRSF1A, TREX1, WDR1, WNT7
Dermatological Diseases		
PD_45 MiSeq® 184 kb	Epidermolysis bullosa: Dystrophic epidermolysis bullosa, epidermolysis bullosa simplex, transient bullous dermolysis of the newborn (TBDN) Genodermatoses: Acrokeratosis verruciformis, Darier-White disease Ectodermal disorders: Amelogenesis imperfecta, ankyloblepharon-ectodermal defects-cleft lip/palate (AEC) syndrome, dermatopathia pigmentosa reticularis, epidermolytic hyperkeratosis, epithelial recurrent erosion dystrophy (ERED), Rapp-Hodgkin Syndrome (RHS)	45 genes: ATP2A2, BMS1, CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSC3, DSG1, DSG2, DSG3, DSG4, DSP, DST, EDA, EXPH5, FERM1, FREM1, GRIP1, ITGA3, ITGA6, ITGB4, JUP, KIT, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT5, LAMA3, LAMB3, LAMC1, LAMC2, NID1, PKP1, PLCG2, PLEC, SERPINB8, SLC39A4, TGM5, TP63, UROD
Developmental Disorders		
PD_104 MiSeq® and NextSeq® 353 kb	Short stature disorders: Coffin-Siris syndrome, Cornelia de Lange syndrome, epiphyseal dysplasia, hypogonadism, microcephaly, microphthalmia, Noonan syndrome, pituitary hormone deficiency, Russell-Silver syndrome, Seckel syndrome	104 genes: ACAN, ALMS1, ANKRD11, ARID1A, ARID1B, ARNT2, ATR, ATRIP, BLM, BRAF, CBL, CCDC8, CENPJ, CEP152, CEP63, CHD7, COL10A1, COL2A1, COL9A1, COL9A2, COMP, CREBBP, CRIP, CUL7, DNA2, DLL1, EP300, ERCC8, FBN1, FGDI1, FGF8, FGFR1, FGFR3, GH1, GHR, GHRHR, GLI2, GLI3, GNAS, GPR161, HDAC8, HESX1, HMG2A, HRAS, HSPG2, IGF1, IGF1R, IGF2, IGFALS, IGSF1, IHH, KDM6A, KMT2D, KRAS, LARP7, LHX3, LHX4, LMNA, MATN3, NIPBL, NPR2, NRAS, NSMCE2, OBSL1, OTX2, PAPPA2, PCNT, PDE4D, PITX2, POC1A, POU1F1, PRKAR1A, PROK2, PROKR2, PTPN11, RAD21, RAF1, RBBP8, RIT1, RNPCC3, ROR2, SHH, SHOC2, SHOX, SMARCA4, SMARCA1, SMARCB1, SMARCE1, SMC1A, SMC3, SOCS1, SOS1, SOX11, SOX2, SOX3, SOX9, SRCAP, STAT5B, TRAI1, TRIM37, WDR11, WNT5A, XRCC4
PD_61b MiSeq® 203 kb	Imprinting disorders: Bloom syndrome, intra-uterine growth retardation, Meier-Gorlin syndrome, osteogenesis defects, retinal dystrophy, Russell-Silver syndrome, short stature syndrome	61 genes: ACAN, BLM, CCDC8, CDC6, CDKN1C, CDT1, COL1A1, CUL7, Dicer1, DIS3L2, DLK1, EED, EZH2, GHR, GPC3, GRB10, HMGA1, HMGA2, HOXA4, HRAS, IGF1, IGF1R, IGF2, IGF2BP3, IGF2R, IGFALS, IGFBP2, IGFBP3, IRS1, IRS2, MEST, NBN, NFIX, NLRP2, NLRP5, NLRP7, NSD1, OBSL1, OOF1, ORC1, ORC4, ORC6, PAD16, PAPPA, PAPPA2, PCNT, PIK3R1, PLAG1, PLAGL1, PLAGL2, POC1A, POLE, PTPN11, RNF125, RNF135, SETD2, SRCAP, STAT5B, STK11, TRIM37, UHRF1

*The disorders covered by the Community Panels include, but are not limited to, those listed here.

For some Community Panels, additional permissions may be required. Contact us at info@sophiagenetics.com to discuss the panel that suits your needs.

About us

SOPHiA GENETICS™ (Nasdaq: SOPH) is a software company dedicated to establishing the practice of data-driven medicine as the standard of care and for life sciences research. We are the creator of the SOPHiA DDM™ Platform, a cloud-native platform capable of analyzing data and generating insights from complex multimodal data sets and different diagnostic modalities. The SOPHiA DDM™ Platform and related applications, modules, and services are currently used by a broad network of hospital, laboratory, and biopharma institutions globally.

Where others see data, we see answers.

Want to know more? Contact us at: info@sophiagenetics.com

