SOPHiA GENETICS™

Accelerate your analysis with pre-designed and tested panels

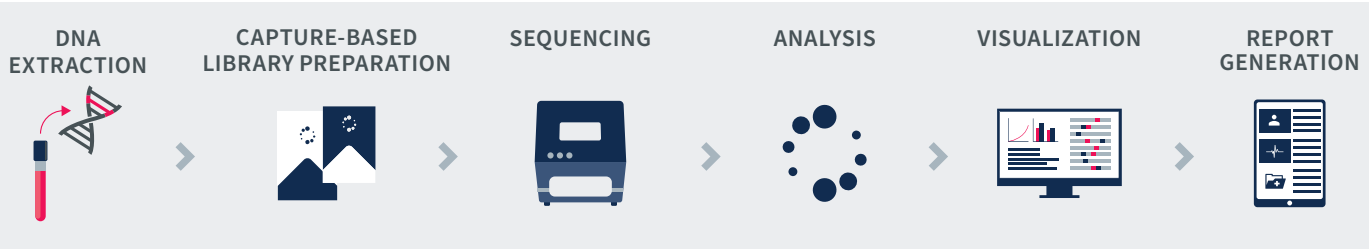
COMMUNITY SOLUTIONS

HIGHLIGHTS

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

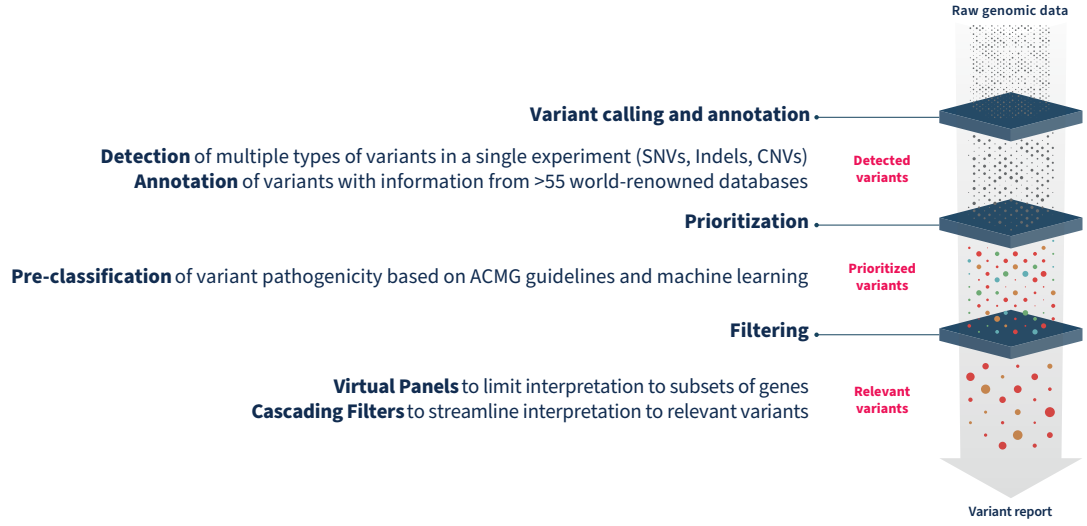
Designing, creating, and testing a new gene panel takes considerable time and effort. **SOPHiA DDM™ Community Solutions are targeted, capture-based NGS panels** developed and tested by genomic experts to minimize set-up challenges and accelerate your research. These panels cover a wide range of Rare and Inherited Disorders, 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 Solutions 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 DDM™ MaxCare Program** provides full set-up assistance.

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



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

COMMUNITY SOLUTIONS FOR CARDIOVASCULAR DISEASES

Panel, sequencer, and panel size	Associated diseases*	Genes covered
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, ANKRD1, APOA1, BAG3, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CDH2, CRYAB, CSRP3, DES, DMD, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EMD, FHL1, FHL2, FLNC, GAA, GATA4, GJA1, GJA5, GJC1, GLA, GPD1L, HCN4, 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 SOLUTIONS 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, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCC, FH, FLCN, GALNT12, HOXB13, HDAC2, MEN1, MET, MITF, 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:** *ABRAXAS1, AP2S1, APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, CTNNA1, DICER1, EPCAM, FH, FLCN, GCM2, GNA11, GREM1, MAX, MEN1, MET, MLH1, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, 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, CDKN1C, CDKN2A, CEBPA, CHEK2, CREBBP, CTNNA1, DICER1, DIS3L2, EGFR, EGLN1, EPCAM, ERBB2, ERCC2, EXT1, EXT2, FANCC, FANCG, FANCM, FH, FLCN, GALNT12, GATA2, GREM1, HNF1A, HOXB13, HRAS, KIF1B, KIT, LZTR1, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, 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, RNF43, RPS20, RUNX1, SCG5, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TERC, TERF2IP, TERT, TMEM127, TP53, TSC1, TSC2, VHL, 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, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CTC1, CYLD, DDB2, DICER1, DIS3L2, DKC1, EGFR, EGLN1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, EZH2, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GATA2, GPC3, GREM1, HNF1A, HOXB13, HRAS, KIF1B, KIT, KMT2D, MAX, MC1R, MDH2, MEN1, MET, MERTK, MRE11A, MITF, MLH1, MLH3, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NHP2, NOP10, NSD1, PALB2, PDGFRA, PHOX2B, PMS1, PMS2, POLD1, POLE, POLH, POT1, PRF1, PRKAR1A, PRSS1, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RUNX1, SBDS, 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, HDAC2, TERT_5UTR, GAA, GBA, GLA, IDUA, TYR, LZTR1, GALNT12, NTHL1, MSH3, CTNNA1*

COMMUNITY SOLUTIONS FOR METABOLIC DISORDERS

Panel, sequencer, and panel size	Associated disorders*	Genes covered
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, ABCG8, ANGPTL3, ANGPTL4, ANGPTL8, APOA1, APOA5, APOB, APOC2, APOC3, APOE, ARSB, ARSH, ASAH1, CETP, CYP27A1, ENPP1, FUCA1, 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*

*The disorders covered by the Community Solutions include, but are not limited to, those listed here.
For some Community Solutions, additional permissions may be required. Contact us at info@sophiagenetics.com to discuss the panel that suits your needs.

COMMUNITY SOLUTIONS FOR PEDIATRIC DISEASES

Including Autoinflammatory 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, RBCK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF31, SAMHD1, SERPING1, SHARPIN, STAT1, STAT3, TMEM173, TNFAIP3, TNFRSF1A, TREX1, WDR1, WNT6*
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, COL9A3, COMP, CREBBP, CRIPT, CUL7, DNA2, DVL1, EP300, ERCC8, FBN1, FGD1, FGF8, FGFR1, FGFR3, GH1, GHR, GHRHR, GLI2, GLI3, GNAS, GPR161, HDAC8, HESX1, HMGA2, 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, RNPC3, ROR2, SHH, SHOC2, SHOX, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SMC1A, SMC3, SOCS1, SOS1, SOX11, SOX2, SOX3, SOX9, SRCAP, STAT5B, TRAIP, 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, OOEP, ORC1, ORC4, ORC6, PADI6, PAPPA, PAPPA2, PCNT, PIK3R1, PLAG1, PLAGL1, PLAGL2, POC1A, POLE, PTPN11, RNF125, RNF135, SETD2, SRCAP, STAT5B, STK11, TRIM37, UHRF1*

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About us

SOPHiA GENETICS (Nasdaq: SOPH) is a cloud-native healthcare technology company on a mission to expand access to data-driven medicine by using AI to deliver world-class care to patients with cancer and rare disorders across the globe. It is the creator of the SOPHiA DDM™ Platform, which analyzes complex genomic and multimodal data and generates real-time, actionable insights for a broad global network of hospital, laboratory, and biopharma institutions. For more information, visit **SOPHiAGENETICS.COM** and connect with us on **LinkedIn**.

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