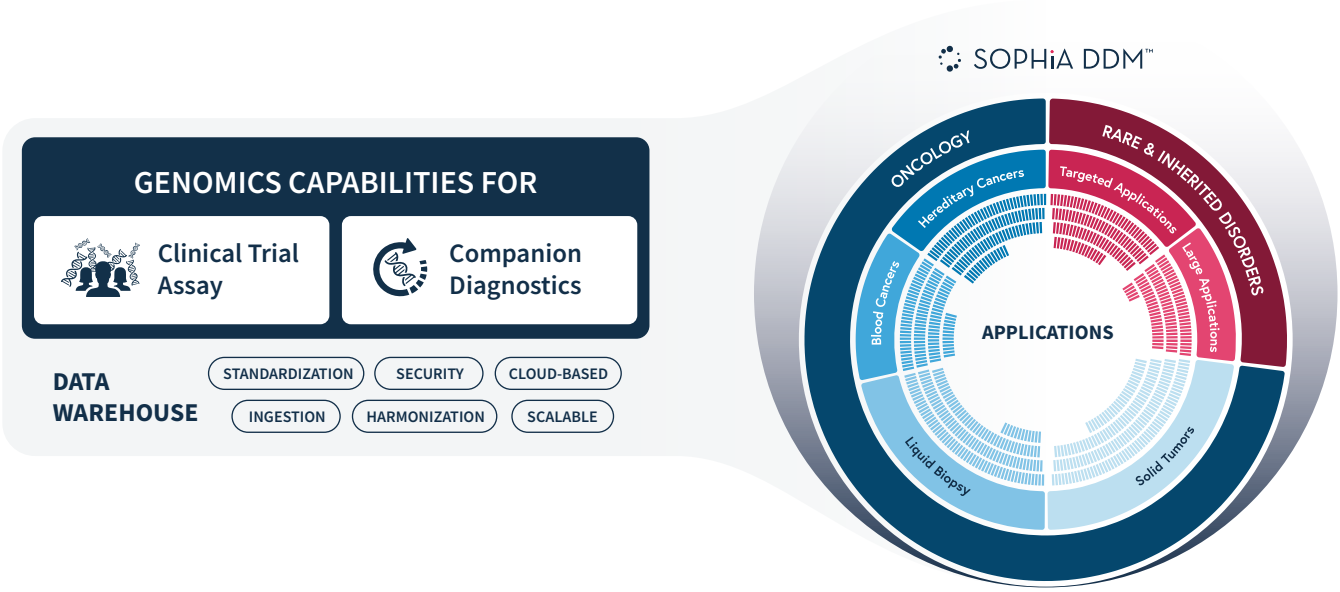
SOPHiA GENETICS™

Genomics applications tailored to your needs

Charting new paths in genomics analysis with SOPHiA DDM™

A comprehensive portfolio of applications across disease areas

SOPHiA GENETICS™ offers a broad portfolio of applications to help accelerate confident decision-making in Oncology and Rare & Inherited Disorders with end-to-end genomics workflows for streamlined sequencing data analysis and interpretation.



Transforming precision medicine with next generation genomics solutions

Tailored solutions

Benefit from proven genomics expertise in designing, developing, and optimizing solutions customized to your unique requirements.

Regulatory support

Advance the development of genomic solutions with in-house regulatory expertise to successfully obtain IUO, or IVD approval.

Decentralized network

Enable sustainable patient access by leveraging the vast global SOPHiA DDM™ network to maximize the decentralization of your newly developed genomic application.

Cost-effective development

Rely on the universally compatible, technology-agnostic, and scalable SOPHiA DDM™ Platform to expedite the development and deployment of highly accurate genomic solutions.

Want to know more about our BioPharma offerings? GET IN TOUCH >

SOPHiA GENETICS™ Research Use Only (RUO) products can be further validated into Investigational Use Only (IUO) and/or In Vitro Diagnostic (IVD) products

Flagship Oncology Solutions

	Genomic Solution	Number of genes	Variants called	Associated Diseases	Sequencing Platform
Liquid Biopsy (LBx)	SOPHiA DDM™ LBx Community Solution (CLBS)*	21	SNVs, Indels, CNVs[1], gene fusions, MSI	• Pan-cancer	NextSeq® NovaSeq™
	MSK-ACCESS® powered with SOPHiA DDM™	147	SNVs, Indels, CNVs[1], gene fusions, *TERT* promoter, *MET* exon 14 skipping	• Pan-cancer	NextSeq® NovaSeq™
Targeted Solid Tumors	SOPHiA DDM™ Dx RNAtarget Oncology Solution (ROS) CE IVD	11	Gene fusions, exon skipping	• Lung cancer	NextSeq®
	SOPHiA DDM™ Solid Tumor Solution (STS) CE IVD	42	SNVs, Indels, gene amplifications[2,3], *TERT* promoter[2,3], *MET* large deletions[2], MSI[2,3], exon 14 skipping[3], gene fusions[3]	• Solid tumors, including lung, colorectal, skin, and brain cancers	MiSeq® MiniSeq®[2,3] NextSeq®[3] Ion S5™[2] MGI[2]
	SOPHiA DDM™ Community Solid Tumor Solution	57	SNVs, Indels, CNVs[1], MSI, gene fusions, gene expression, exon skipping	• Solid tumors, including glioma, melanoma, gastric, ovarian, colorectal, thyroid, breast, lung, and endometrial cancers	NextSeq®
	SOPHiA DDM™ RNAtarget PanCancer Solution	143	SNVs, Indels, gene fusions, *CD274* gene expression	• Solid tumors, including cholangiocarcinoma, lung, sarcoma, and thyroid cancers	NextSeq®
	Homologous Recombination Solutions (HRS)				
	SOPHiA DDM™ Homologous Recombination Deficiency Solution (HRD) CE IVD	28	SNVs, Indels, CNVs[1], GI	• Ovarian cancer	NextSeq®
	SOPHiA DDM™ Homologous Recombination Solution (HRS)	4 (miniHRS), 16, 28 (extHRS)	SNVs, Indels, CNVs[1], GI[4]	• Ovarian, breast, prostate, colon, lung, and pancreatic cancers	MiSeq® NextSeq® MiniSeq® Ion S5™
Comprehensive Genomic Profiling	MSK-IMPACT® Flex powered with SOPHiA DDM™ *	533	SNVs, Indels, CNVs[1], gene fusions, gene expression, MSI, TMB, *TERT* promoter, *MET* exon 14 skipping, exon skipping, HRD, matched T/N	• Rare and common cancers	NextSeq® NovaSeq™ Element MGI
	MSK-IMPACT® powered with SOPHiA DDM™	505	SNVs, Indels, CNVs, *TERT* promoter, *MET* exon 14 skipping, gene fusions, TMB, MSI, matched T/N	• Rare and common cancers	NextSeq® NovaSeq™
	SOPHiA DDM™ Cancer Profiling Solution (CPS)	556	SNVs, Indels, CNVs[1], gene fusions, exon skipping, TMB, MSI, *TERT* promoter, GI[4]	• Pan-cancer, solid tumors	NextSeq® NovaSeq™ MGI
Blood Cancers	SOPHiA DDM™ Residual Acute Myeloid Solution (RAM)	23	Measurable Residual Disease (MRD) markers	• Acute Myeloid Leukemia (AML)	NextSeq® NovaSeq™
	SOPHiA DDM™ Community CLL Clonality Chronic Solution (CLL)	23	SNVs, Indels, CNVs, IGH rearrangements	• Chronic lymphocytic leukemia (CLL)	MiSeq® MiniSeq®
	SOPHiA DDM™ Myeloid Solution (MYS) CE IVD	30	SNVs, Indels, CNVs[2], *FLT3* ITDs, gene fusions[3]	• Acute Myeloid Leukemia (AML) and related neoplasms • Myelodysplastic syndromes (MDS) • Myeloproliferative neoplasms (MPN)	MiSeq® NextSeq® MiniSeq® Ion S5™ MGI
	SOPHiA DDM™ Blood Cancer Community Solution	51	SNVs, Indels, CNVs, gene amplifications	• Leukemia, including Acute Myeloid Leukemia (AML) • Myelodysplastic syndromes (MDS) • Myeloproliferative neoplasms (MPN)	MiSeq®
	SOPHiA DDM™ Lymphoma Solution (LYS)	54	SNVs, Indels, CNVs, gene amplifications	• B- and T-cell lympohmas • Diffuse large B-cell lymphoma (DLBCL) • Follicular, Mantle cell, and Burkitt lymphomas	MiSeq® NextSeq®
	SOPHiA DDM™ Extended Myeloid Solution (ExtMYS)	98	SNVs, Indels, CNVs, *FLT3* ITDs, *KMT2A* PTDs	• Acute Myeloid Leukemia (AML) and related neoplasms • Myelodysplastic syndromes (MDS) • Myeloproliferative neoplasms (MPN)	NextSeq®
Hereditary Cancer	SOPHiA DDM™ Hereditary Cancer Solution (HCS) CE IVD	27 (v1.0), 83 (v2.0)	SNVs, Indels, CNVs[2,5], Alu insertions[2,5], *PMS2* vs *PMS2CL* variants[2,5], Boland inversion[2,5]	• Hereditary breast and ovarian cancers • Intestinal cancers, including Lynch and various intestinal polyposis syndromes	MiSeq® NextSeq®[2,5] Ion S5™[2] MGI[2,5]

Flagship Rare & Inherited Disorders Solutions

	Genomic Solution	Number of genes	Variants called	Associated Diseases	Sequencing Platform
Rare Disorders	SOPHiA DDM™ Enhanced Exome Solutions	CES or WES	SNVs, Indels, CNVs, mtDNA	• Rare and inherited disorders	NextSeq® NovaSeq™
	SOPHiA DDM™ Clinical Exome Solution (CES)	5,500	SNVs, Indels, CNVs, mtDNA	• Rare and inherited disorders	MiSeq® NovaSeq™ NextSeq® MGI
	SOPHiA DDM™ Whole Exome Solution (WES)	19,425	SNVs, Indels, CNVs, mtDNA	• Rare and inherited disorders	NextSeq® MGI NovaSeq™
	SOPHiA DDM™ Whole Genome Solution (WGS)±	30x & lpWGS	SNVs, Indels, CNVs, structural variants	• Rare and inherited disorders	NovaSeq™
Pharmacogenomics	SOPHiA DDM™ Community Pharmacogenomics Solution	28	SNVs, Indels, CNVs, Star alleles (*CYP2D6* only)	• Pharmacogenomics	NextSeq®

SNVs, single nucleotide variants; Indels, insertions/deletions; CNVs, copy number variants; IGH, immunoglobulin heavy locus, ITDs, internal tandem duplications; PTDs, partial tandem duplications; TMB, tumor molecular burden; MSI, microsatellite instability; GI, genomic instability; T/N, tumor-normal; mtDNA, mitochondrial DNA; lpWGS, low-pass Whole Genome Sequencing. * New product, launched in 2025. [1] Including whole gene amplifications and deletions. [2] Only available in the RUO version
[3] Only available in the plus version (SOPHiA DDM™ Myeloid Plus Solution, SOPHiA DDM™ Extended Myeloid Plus Solution, and SOPHiA DDM™ Solid Tumor Plus Solution). [4] Available as add-on. [5] Only available on SOPHiA DDM™ Hereditary Cancer Solution (HCS) v2.0.
± SOPHiA DDM™ Whole Genome Solution is under development. SOPHiA GENETICS™ products are for Research Use Only and not for use in diagnostic procedures unless specified otherwise. SOPHiA DDM™ Dx Hereditary Cancer Solution, SOPHiA DDM™ Dx RNAtarget Solution, and SOPHiA DDM™ Dx Homologous Recombination Deficiency Solution are available as CE-IVD products for In Vitro Diagnostic Use in the European Economic Area (EEA), the United Kingdom (UK), and Switzerland. SOPHiA DDM™ Dx Myeloid Solution and SOPHiA DDM™ Dx Solid Tumor Solution are available as CE-IVD products for In Vitro Diagnostic Use in the EEA, the United Kingdom, Switzerland, and Israel.