SOPHiA INTEGRATED

Enable equal access to effective genomic applications worldwide

APPLICATION LIST

Oncology

Exome Solutions

Hereditary cancer

Metabolism

Cardiology

Pediatrics

SOPHiA™

Oncology

SOPHiA Myeloid Solution CE IVD

30 genes[1,2]

NRAS, KRAS & BRAF Application

BRAF, *KRAS* and *NRAS* genes

SOPHiA Solid Tumor Solution

42 genes[1]

BRCA1 & BRCA2 Application

BRCA1 and *BRCA2* genes[2]

SOPHiA mini Homologous Recombination Solution

4 genes

Solid Tumor Applications

- panel of 15 genes
- panel of 56 genes
- panel of 170 genes[1,3]

Tumor Hotspot Application

26 genes

Turnaround time[4]
From **10** to **30** days

Detected variants
SNVs and Indels

Raw material[4]
FFPE, blood, plasma, bone marrow and extracted DNA

CE IVD kit + data analysis

(1) Fusion detection available
(2) CNV detection available for all the genes
(3) Available soon
(4) Depending on the application
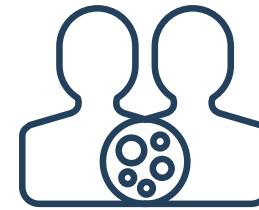
Hereditary Cancer

SOPHiA Hereditary Cancer Solution CE IVD

26 genes

BRCA1 & BRCA2 Application

BRCA1 and *BRCA2* genes

SOPHiA Expanded Hereditary Cancer Solution

- panel of 33 genes
- panel of 56 genes

Lynch Syndrome (HNPCC) Application

MLH1, MSH2, MSH6 (excluding exon1), *PMS2* and the 3' UTR of *EPCAM* gene

Hereditary Cancer Application

94 genes[1]

Familial Adenomatous Polyposis (FAP) Application

APC and *MUTYH* genes

Turnaround time[2]
From **10** to **20** days
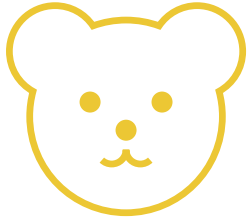
Detected variants
SNVs, Indels and CNVs

Raw material[2]
Blood, extracted DNA, saliva and buccal mucosa

CE IVD kit + data analysis

(1) CNV detection only available for 11 genes
(2) Depending on the application

Pediatrics

SOPHiA Nephropathies Solution
44 genes

Cystic Fibrosis Application CE IVD
CFTR gene

Stargardt Application
ABCA4, CNGB3 and *ELOVL4* genes[1]

Marfan Application
FBN1 gene[1]

Alport Application
COL4A3, COL4A4 and *COL4A5* genes[1]

Thalassemia Application
3 genes

Familial Mediterranean Fever Application CE IVD
MEFV gene[1]

Duchenne Muscular Dystrophy Application
DMD gene

Turnaround time[2]
From **10** to **20** days

Detected variants
SNVs, Indels and CNVs

Raw material[2]
Blood, extracted DNA, saliva and buccal mucosa

CE IVD   kit + data analysis

(1) CNV detection currently not available
(2) Depending on the application

Cardiology

SOPHiA Cardio Solution
31 genes

Cardio Application
174 genes[2]

SOPHiA Extended Cardio Solution
128 genes[1]

Primary Electrical Disorders Application
51 genes

Familial Hypertrophic Cardiomyopathy Application
MYBPC3, MYH7, MYL2, TNNI3 and *TNNT2* genes[2]

Hypercholesterolemia Application Cardio Solution
APOB (exon26 partially covered), *APOE, LDLR* and *PCSK9* genes + *12 SNPs*

Turnaround time[3]
From **10** to **20** days

Detected variants
SNVs, Indels and CNVs

Raw material[3]
Blood, extracted DNA, saliva and buccal mucosa

(1) CNV detection available, except from exon 47 of *FLNC* and exons 172-197 of *TTN*
(2) CNV detection currently not available
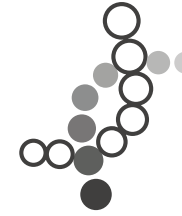(3) Depending on the application

Metabolism

SOPHiA Dyslipidemia Solution
31 genes[1]

Maturity Onset Diabetes of the Young Application
ABCC8, GCK, HNF1A, HNF4A, HNF1B, INS and *KCNJ11* genes[1]

Exome Solutions

SOPHiA Hereditary Disorder Solution
569 genes

SOPHiA Whole Exome Solution
> 19,000 genes[1]

SOPHiA Clinical Exome Solution
4,490 genes[1]

Whole Exome Application
> 21,000 genes

Turnaround time[2]
From **10** to **20** days

Detected variants
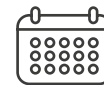SNVs, Indels and CNVs

Raw material[2]
Blood, extracted DNA, saliva and buccal mucosa

(1) CNV detection currently not available
(2) Depending on the application

Turnaround time[2]
From **20** to **30** days

Detected variants
SNVs and Indels

Raw material[2]
Blood, extracted DNA, saliva and buccal mucosa

(1) CNV detection available
(2) Depending on the application

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