

SOPHiA INTEGRATED

Enable equal access to effective genomic applications worldwide

APPLICATION LIST





Oncology

SOPHiA Myeloid Solution

30 genes^(1, 2)

SOPHiA Solid Tumor Solution

42 genes⁽¹⁾

SOPHiA mini Homologous Recombination Solution

4 genes

Tumor Hotspot Application

26 genes

NRAS, KRAS & BRAF Application

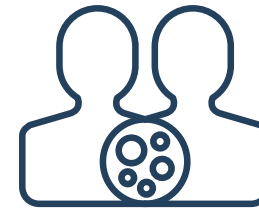
BRAF, *KRAS* and *NRAS* genes

BRCA1 & BRCA2 Application

BRCA1 and *BRCA2* genes⁽²⁾

Solid Tumor Applications

- panel of 15 genes
- panel of 56 genes
- panel of 170 genes^(1, 3)



Hereditary Cancer

SOPHiA Hereditary Cancer Solution

26 genes

SOPHiA Expanded Hereditary Cancer Solution

- panel of 33 genes
- panel of 56 genes

Hereditary Cancer Application

94 genes⁽¹⁾

BRCA1 & BRCA2 Application

BRCA1 and *BRCA2* genes

Lynch Syndrome (HNPCC) Application

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

Familial Adenomatous Polyposis (FAP) Application

APC and *MUTYH* 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

 kit + data analysis

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



Turnaround time⁽²⁾

From **10** to **20** days



Detected variants

SNVs, Indels and CNVs

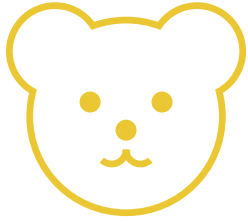


Raw material⁽²⁾

Blood, extracted DNA, saliva and buccal mucosa

 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

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

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

 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



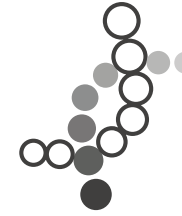
Metabolism

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

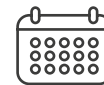
SNVs, Indels and CNVs



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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