SOPHiA GENETICS™ | GeneMind

Distributed by PROQUINORTE close to you | cerca de ti

One connected germline workflow

Choose your scope. Sequence with GeneMind. Analyse with **SOPHiA DDM™**.

HCS

Hereditary cancer

Guideline-recommended genes + healthcare service portfolio

WES/CES

Whole & clinical exome

Mitochondrial DNA
>200 non-coding regions

Enhanced WES/CES

Dedicated modules

HCS • Carrier screening
Pharmacogenomics



GeneMind SURFSeq™ 5000

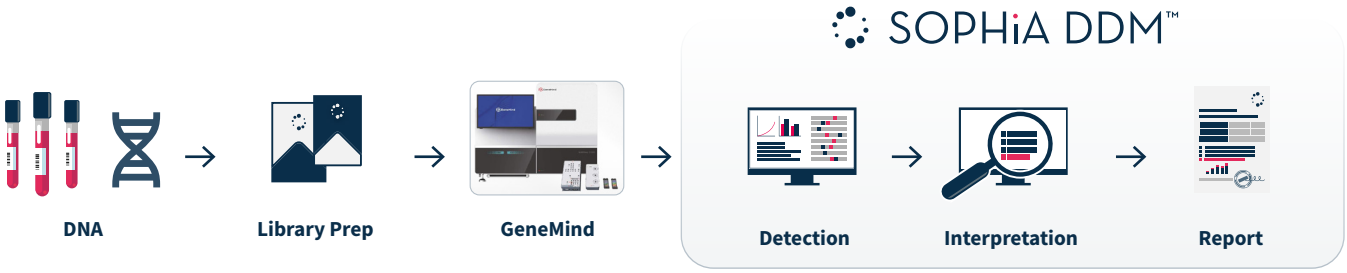
CMS™ chemistry
3 flow cell capacities

PE150 <16 h with FCM[1]

>95% Q40 • **>70%** Q50

GeneMind

From DNA to an expert-reviewed report



Advanced bioinformatics modules for challenging analyses

Genomic + mitochondrial DNA • SNVs • Indels • CNVs

Hereditary cancer

PMS2/PMS2CL gene conversion
MSH2 inversion
Alu insertions

Whole & clinical exome

SMN1/SMN2
HBA1/HBA2
CYP21A2/A1P
Conversions • Deletions
CFTR repeats

Dedicated modules

Star allele calling
CYP2D6/CYP2D7 gene conversion

Also available: **GBA/GBAP1** gene conversion in selected applications.

Capabilities depend on application/module. HBA1/HBA2: large deletions. MSH2: Boland inversion.
[1]Loading to basefile generation; longer with FCH/FCP. Quality specifications: manufacturer data; results vary.

SOPHiA DDM™ Solutions are for Research Use Only (RUO); RUO results must not be used for clinical diagnosis or patient management without further validation by the laboratory.

ES-GN-260001

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From variants to clearer answers

Connected analysis. Expert interpretation. Shared knowledge.

Your interpretation workspace

Family & phenotype
Inheritance segregation
• Family analysis

Focused review
HPO/OMIM® panels
Filters • Transcripts

Evidence in view
Alamut™ Visual Plus
Community knowledge

Three innovations. Three clear tasks.

VPR

Prioritise variants
Ranks variant + phenotype evidence.
Point-based ACMG classification.

Flash View

View Review & report
Predefined review strategies.
Direct-to-report workflows.

AI Scribe

Draft interpretation text
AI drafts text from trusted evidence.
Expert reviews the draft.

LIMS / EHR → **SOPHiA DDM™** → **Reviewed report**

Connectivity options • MaxCare set-up support • Automation scripts

Voices from the laboratory

> ***...reduce hands-on time, turnaround times, retesting, and resampling***



Arantxa Hervás, PhD
CGT Dx Director
Vitrolife Group

> ***...a community in which you can compare your data with others***



Gorka Alkorta-Aranburu, PhD
Laboratory Director
Clínica Universidad de Navarra

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