NEPHROPATHIES SOLUTION™ BY SOPHiA GENETICS

The Nephropathies Solution (NES) by SOPHiA GENETICS is a molecular diagnostic application that bundles a capture-based target enrichment kit with the analytical power of SOPHiA™ AI and full access to the SOPHiA DDM® platform.

Smart Kit Design

SOPHiA™

The AI Democratizing Data-Driven Medicine

SaaS Analytical Platform

The NES panel covers the coding regions and splicing junctions (± 5bp) of 44 most clinically relevant genes (target region of 105.8 kb) related to a broad range of nephropathies such as nephrotic syndromes, polycystic kidney diseases, Bartter syndromes, Alport syndrome, CAKUT or tubulopathies. It guarantees high on-target rate and coverage uniformity even in GC-rich regions, including the first exon.

Gene panel

AGXT, AQP2, ATP6V0A4, ATP6V1B1, AVPR2, BSND, CASR, CEP290, CLCN5, CLCNKB, COL4A3, COL4A4, COL4A5, CRB2, CTNS, CUBN, CYP24A1, DSTYK, EMP2, EYA1, FN1, FOXC1, GRHPR, HNF1b, KANK2, KCNJ1, LAMB2, NPHS2, NR3C2 , OCRL, PAX2, PHEX, PKD1, PKD2, PKHD1, SIX1, SLC12A1, SLC12A3, SLC34A1, SLC4A1, SLC4A4, TTC21B, UMOD, WT1

Recommendations

Starting material: 200 ng
Sample source: Blood
Samples per run: Depending on sequencing platform(1)

Sequencer	Flow Cell / Ion Chip Kit	Recommended samples per run (for 250x median coverage depth)
Illumina MiSeq®	v3 (2x300bp)	32
Ion S5™	Ion 540	48

Wet lab

Day 1: Library Preparation
Day 2: Capture and Sequencing
Total hands-on time: 8 hours

SOPHiA analyzes complex genomic NGS data by detecting, annotating and pre-classifying genomic variants to help clinicians better diagnose their patients.

- SNVs, Indels and CNVs are accurately detected in all genes of the panel
- Pseudogene variants are efficiently differentiated from the ones in the *PKD1* gene(2)

SOPHiA reaches excellent clinical-grade analytical performance:

	Observed	Lower 95% CI
Sensitivity	100%	82.21%
Specificity	100%	100%
Accuracy	100%	100%
Precision	100%	84.21%
Repeatability	99.99%	99.97%
Reproducibility	99.99%	99.97%
Average on-target rate(3)	75%	
Coverage uniformity	97.55%	
Average percentage of target region with depth >200x	95.09%	

Analysis time from FASTQ files: 4 hours(4)

The results are presented in SOPHiA DDM, the platform of choice for clinicians performing routine diagnostic testing. Its intuitive user interface and advanced features facilitate the visualization and interpretation of genomic variants. Patient's data is kept safe by applying the highest industrial standards of encryption.

Main features

Dedicated features in SOPHiA DDM reduce the complexity of determining the clinical significance of genomic variants.

- **Dual variant pre-classification:** Improve assessment of variants pathogenicity with the pre-classification of both ACMG guidelines and SOPHiA's prediction.
- **Virtual Panels:** Restrict the interpretation to sub-panels of genes (e.g. focus on Alport syndrome or polycystic diseases)
- **Variant Filter Builder:** Define and edit custom filters for efficient analysis

Access to SOPHiA's Community

In SOPHiA DDM, experts from hundreds of healthcare institutions interpret the results and flag the pathogenicity level of variants according to their knowledge and experience. This highly valuable information feeds the variant knowledge base and is anonymously and safely shared among the members of the community.

(1) Sequencing recommendations and specifications for other sequencing kits and instruments available upon request. Delivery time may vary according to the selected sequencing platform
(2) Due to high gene conversion rates, a definite location in *PKD1* and its pseudogenes cannot be assigned in homologous regions of exon 5
(3) The number of off-target high coverage regions is particularly high because of the presence of pseudogenes in the panel.
(4) Analysis time may vary depending on the number of samples multiplexed and server load