SOPHiA GENETICS™

Nephropathies Solution

The genomic application that combines a capture-based target enrichment kit with the analytical capabilities and advanced features of the SOPHiA DDM™ platform.

Main Features

SOPHiA Nephropathies Solution covers the complete coding sequence (± 5bp of exon-flanking regions) of 44 genes (target region of 105.8 kb) related to a broad range of nephropathies such as nephrotic syndromes, polycystic kidney disorders, Bartter syndromes, Alport syndrome, CAKUT and tubulopathies. Probe design is optimized to guarantee high on-target rate and coverage uniformity even in GC-rich regions, including the first exon.

Gene Panel	Variants Called	Recommendations	Wet Lab
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	SNVs Indels CNVs Differentiation of gene and pseudogene variants in *PKD1*[1]	**Starting material** 200 ng DNA **Sample type** Blood **Samples per run for > 250x coverage depth / Sequencer (Flow Cell / Ion Chip Kit)** 32 for Illumina MiSeq® v3 (2x300bp) 48 for Thermo Fisher Scientific Ion S5™ Ion 540 24 for Illumina MiSeq® v2 (2x250bp) 96[2] for Illumina NextSeq® 500/550 Mid Output Kit v2 (2x125bp) and High Output Kit v2 (2x150bp) 32 for Thermo Fisher Scientific Ion Proton™ Ion P1	**Day 1:** Library Preparation **Day 2:** Capture and Sequencing **Total library preparation time: 1.5 days**

Analytical Performance

The SOPHiA DDM™ platform analyzes complex NGS data by detecting, annotating and pre-classifying multiple types of genomic variants in all the genes of the panel.

Analysis time[3] from FASTQ: 4 hours

	Observed	Lower 95% CI
Sensitivity	100%	82.21%
Accuracy	100%	84.21%
Precision	99.99%	99.97%
Repeatability	100%	100%
Reproducibility	100%	100%
Average on-target rate[4]	75%	
Coverage uniformity	97.55%	
Average % of target region with depth >200x	95.09%	

One Simple Intuitive Platform: Beyond Analytics

Accelerated assessment and reporting of genomic variants

Dedicated features in SOPHiA DDM™ reduce the complexity of determining the significance of genomic variants and facilitate the interpretation process, thus reducing turnaround time:

- **Dual Variant Pre-classification** - Improve assessment of variant pathogenicity with both ACMG scores and SOPHiA GENETICS' machine learning-based predictions
- **Virtual Panels** - Restrict the interpretation to sub-panels of genes of interest using the HPO or OMIM® browser
- **Cascading Filters** - Apply custom filtering options for quicker screening of relevant variants and save strategies for future analyses

After the interpretation, you can generate a customizable variant report, including valuable information to support decision making.

Global support at every step

We offer local support anywhere in the world. Our dedicated bioinformaticians help save time and resources, ensuring fast resolution of workflow disruptions. In addition, our Set Up Program provides assistance with assay set up for fast and worry-free transition to routine testing.

Secure and unlimited data storage

The SOPHiA DDM™ platform provides unlimited and unrestricted storage, while keeping data safe by applying the highest industrial standards of encryption in compliance with local data security policies.

Access to the SOPHiA GENETICS community

In the SOPHiA DDM™ platform, experts from hundreds of healthcare institutions interpret their 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. Due to high gene conversion rates, a definite location in *PKD1* and its pseudogenes cannot be assigned in homologous regions of exon 5.
2. Maximum number of indices available.
3. Analysis time may vary depending on the number of samples multiplexed and server load.
4. The number of off-target high coverage regions is particularly high because of the presence of pseudogenes in the panel.

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