

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 DDM™ Nephropathies Solution covers the complete coding sequence (\pm 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, GRHR, 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

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

The platform can also provide access to Alamut™ Visual Plus, a full-genome browser that integrates numerous curated genomic and literature databases, guidelines, missense and slicing predictors, thus enabling a deeper variant exploration. 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 data storage

Access to the SOPHiA DDM™ Platform is restricted to registered users only. The Platform keeps data safe by applying the highest industrial standards of encryption in compliance with your local data security policies.

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 time³ 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%	

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.