

# Mitochondrial DNA Sequencing Panel

Mitochondrial Diseases  
: Metabolic and neurological disorders and cancers

## DESCRIPTION

Celemics has specifically designed capture probes and adjusted the concentration of the panel for each respective use with our own proprietary rebalancing technologies to provide complete, consistent coverage of the whole mtDNA genome while taking into consideration small target regions. This enables the same high level of target capture efficiency regardless of small target sizes even with a stand-alone panel.

## KEY FEATURES

|   |  |
|---|--|
| 1. High-fidelity sequencing               | Guarantees maximum capture efficiency in custom panels without affecting target specificity  |
| 2. Highly uniform coverage and mean depth | High coverage and uniformity across the entire human mitochondrial genome  |
| 3. Flexible customization                 | Convenient addition to other Celemics target enrichment panels such as G-Mendeliome panels for further mtDNA-derived rare disease analysis |

## SPECIFICATION

|                         |  |
|-------------------------|--|
| Covered region*         | Whole mitochondrial genome   |
| Target size             | 16.6 kb  |
| Mutation type           | SNV, Indel   |
| Sample type (amount)    | Blood (> 50 ng of fragmented DNA)  |
| Platform                | All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore          |
| Bioinformatics pipeline | Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report) |

## PERFORMANCE

| NGS Sequencing Amount | On-target Base Ratio | Mean Depth | Coverage |        |        |
|-----------------------|----------------------|------------|----------|--------|--------|
|                       |                      |            | 10x      | 50x    | 100x   |
| 10Mb                  | 97.93%               | 493x       | 99.98%   | 99.91% | 99.87% |

## IGV EXAMPLE OF CELEMICS mtDNA SEQUENCING PANEL



Celemics mtDNA Sequencing Panel shows 99% coverage with uniformity

## PACKAGE COMPOSITION

| Package name      | Compositions               |                    | Package option           | Options         |                     |
|-------------------|----------------------------|--------------------|--------------------------|-----------------|---------------------|
| Target Enrichment | Target capture Probe       | -                  | Pooling method           | Single Reaction | Pre-capture Pooling |
| Standard          | Target Enrichment reagents | Library prep Kit   | Library Preparation kits | Standard Kit    | EP-kit              |
| All-in-One        |                            | Beads / Polymerase | Hybridization Enhancer   | Included        | Not included        |

