

# **Twist Mitochondrial Panel**

The mitochondrial genome encodes factors critical to energy production, which directly affects the energy state of the cell, tissue, and human individual. Pathogenic mutations of Mitochondrial DNA (mtDNA) are often implicated in a group of complex human diseases which can be difficult to diagnose such as metabolic & neurologic disorders as well as cancer. Interrogation of the human mitochondrial genome by targeted Next-Generation Sequencing (NGS) can help investigators shed light on the genetic mechanism behind mitochondrial disorders.

The Twist Mitochondrial Panel is a fixed content panel designed to cover all 16,659 base pairs (bp) and 37 genes of the mitochondrial genome (Table 1). This panel can be used as a standalone panel or as a spike-in with Twist's Human Core Exome, Comprehensive Exome as well as Twist's Custom Panels. Providing industry-leading coverage, uniformity, and flexibility, the Twist Mitochondrial Panel can help investigators enrich for, sequence, and analyze mtDNA variants contributing to disease including incidences of heteroplasmy.

## **Revolutionizing Target Enrichment**

A critical component for successful targeted sequencing analysis is the quality of the capture reaction, which drives the uniformity of reads. A more uniform capture leads to more uniform sequencing, as indicated by lower fold80 base penalty scores. With higher uniformity, sequencing reads can effectively drive on target coverage, limiting wasted reads.

High uniformity is the combined result of the probe design algorithm, silicon-based synthesis of dsDNA probes, and calibration processes that ensure uniform amplification, GC content, and enrichment performance across all targets. NGS-based quality control of all Twist Panels ensures that all probes are present at the correct concentrations and that the reproducibility of results is independent of our production processes.

HUMAN MITOCHONDRIAL GENOME	
Bait Territory	16447
Design Efficiency	1.00
Mean Target Coverage	105
% Duplicates	0.01%
Uniformity (Fold-80 Base Penalty)	1.20
% TARGETED BASES AT	
10X	100%
20X	100%
30X	99.56%

Table 1. Twist Mitochondrial panel spiked into Twist Core Exome. Data obtained with 2.3 Mb (150X) sequencing. Complete coverage with excellent uniformity and sequencing efficiency is demonstrated.

### Flexibility as a Standalone or as Added Content

The Twist Mitochondrial Panel can serve as added content to the Twist Human Core & Comprehensive Exome Panels, a Twist Custom Panel, or used as a standalone panel (figure 1). Sequencing results with the Mitochondrial Panel as a spike-in with the Twist Human Core Exome at different concentrations demonstrate similar coverage indicative of uniformity irrespective of varying concentrations (figure 2).

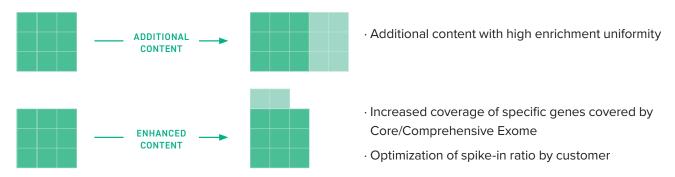


Figure 1. The Twist Mitochondrial Panel can be used in conjunction with the Twist Human Core Exome or a Twist Custom Panel to expand your range of targets while retaining high enrichment uniformity.

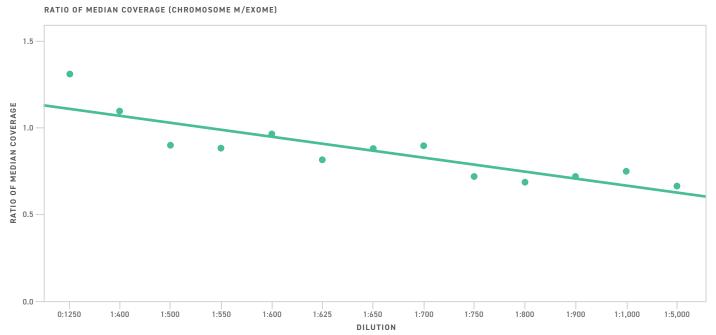


Figure 2: Ratio median target coverage of mtDNA/Exome at various dilutions demonstrate little variation in coverage.

The Twist Mitochondrial Panel is a component of the Twist portfolio of products for NGS Target Enrichment.

### **LEARN MORE**

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#### ORDERING INFORMATION

102038: Twist Mitochondrial Panel, 2 Reactions, Kit 102039: Twist Mitochondrial Panel, 12 Reactions, Kit 102040: Twist Mitochondrial Panel, 96 Reactions, Kit