# Twist CNV Backbone Spike-in Panels

## Enhance whole exome sequencing with copy number variation (CNV) analysis

Exome sequencing offers sequencing coverage of protein-coding targets and other specific regions of interest. Typically, exome sequencing only covers about 1-2% of the bases in the genome.[1,2] While this makes analysis of an individual's genetics more efficient than sequencing the whole genome, it leaves large regions between these genes and genetic elements out of downstream analysis. This can make detecting copy number variations (CNVs), an important driver in many genetic diseases, difficult from exome sequencing alone where probes are not regularly tiled.

Twist's CNV Backbone Spike-in Panels provide a simple way of adding CNV analysis to your exome sequencing by increasing the resolution of CNV calls.

The CNV backbone panels are offered in three different probe densities of 100 kb, 50 kb, and 25 kb of increasing resolution, respectively. This allows for tuning to the desired exome resolution. These panels are designed to pair with our best-in-class Exome 2.0 + Comprehensive Spike-in Panel and are also compatible with Twist's full suite of DNA library preparation and target enrichment workflows.

### KEY BENEFITS

**Tune the resolution of your copy number analysis for exome sequencing**
- Available in 100 kb (lower resolution), 50 kb (intermediate resolution), and 25 kb (higher resolution).
- Probes are tiled to regions between genes (intergenic regions) at regular intervals.
- Easily add this panel as a spike-in to your exome panel to enhance CNV calling capabilities of whole exome sequencing.

**Enables new capabilities to Twist's exome sequencing solutions**
- Designed to pair with our best-in-class Exome 2.0 plus Comprehensive Spike-in panel.
- Available in 2-reaction (16 samples) and 12-reaction sizes (96 samples).

## Enhance your exome sequencing

The Twist CNV Backbone Spike-in Panels provide a simple means to offer read coverage at regular intervals across the genome, improving the signal for detecting CNV events (**Table 1**). The spike-in panel's probes target single nucleotide polymorphisms (SNPs) that are known to be polymorphic across populations. The sites selected for the panel designs are positioned across both intergenic and intronic regions, with three density intervals of 100 kb, 50 kb, and 25 kb from lowest to highest resolution, respectively.

Twist's spike-in panels are simple to integrate into standard exome and target enrichment protocols. Simply combine an equal volume of the exome panel and the CNV Spike-in panel and use this blended panel according to Twist's standard target enrichment workflow protocols. Twist CNV Backbone Spike-in Panels are one of the several options for customization of whole exome sequencing.

| CNV BACKBONE SPIKE-IN PANEL | 25 KB | 50 KB | 100 KB |
|---|---|---|---|
| Panel Design Size (Mb) | 8.32 | 3.33 | 1.39 |
| SNVs & INDELs Called | 265,138 | 207,765 | 181,956 |
| All CNVs Called | 472 | 446 | 411 |
| CNVs <50 kb Called | 417 | 385 | 361 |
| Mean Target Coverage | 150x | 171x | 161x |

**Table 1. Example data of Twist CNV Backbone Spike-in Panels.** A highly characterized sample set known to contain CNVs[3] and a baseline set of 12 healthy individuals were sequenced with 2x150 PE reads on an Illumina NovaSeq 6000. The average number of SNVs, INDELs, and CNVs called and sequencing depth at each probe density was determined for each panel when spiked into Twist Exome 2.0 plus Comprehensive Spike-in. CNV calling was performed with a commercially available software solution.[4]

*For Research Use Only. Not for use in diagnostic procedures.*

*REFERENCES*

*1. Warr, Amanda et al. "Exome Sequencing: Current and Future Perspectives." G3 (Bethesda, Md.) vol. 5,8 1543-50. 2 Jul. 2015, doi:10.1534/g3.115.018564*

*2. Rehder, Catherine et al. "Next-generation sequencing for constitutional variants in the clinical laboratory, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG)." Genetics in medicine : official journal of the American College of Medical Genetics vol. 23,8 (2021): 1399-1415. doi:10.1038/s41436-021-01139-4*

*3. Coriell Institute's CNVPANEL01 - Human CNV Reference Panel*

*4. eVai Platform (secondary workflow), enGenome Software.*

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**ORDERING INFORMATION**

**Higher Resolution:**
**110756:** Twist 25 kb CNV Backbone Spike-in Panel, 2 Reaction kit
**110757:** Twist 25 kb CNV Backbone Spike-in Panel, 12 Reaction kit

**Intermediate Resolution:**
**110758:** Twist 50 kb CNV Backbone Spike-in Panel, 2 Reaction kit
**110759:** Twist 50 kb CNV Backbone Spike-in Panel, 12 Reaction kit

**Lower Resolution:**
**110760:** Twist 100 kb CNV Backbone Spike-in Panel, 2 Reaction kit
**110761:** Twist 100 kb CNV Backbone Spike-in Panel, 12 Reaction kit