Quick start your HiFi sequencing projects with pre-designed panels curated by leading researchers

Access many benefits of PacBio® HiFi long-read sequencing at a fraction of the cost with Twist Bioscience long-read sequencing panels. Twist Bioscience’s target enrichment technology provides high capture efficiency and exceptional uniformity across target regions. Combined with HiFi sequencing, this enables highly accurate variant calling, including structural variants (SVs) and complex variants, as well as direct haplotype phasing, for your regions of interest. Develop your own fully customizable and scalable panel now, or get started quickly with pre-designed Twist Alliance panels that have been developed by researchers at leading institutions for specific applications. Learn more at twistbioscience.com/products/ngs/Long-Read-Sequencing-Panels.

Long-read sequencing at scale:
• Probes optimized for high uniformity and sequencing efficiency
• High-accuracy variant calling of SNPs, SVs, and indels
• Unambiguous haplotype phasing without the need for trios
• Cost-effective and high-throughput, compatible with PacBio Sequel® IIe and Revio™ systems

Twist Alliance Dark Genes panel

Several medically-relevant genes lie in so-called next-generation sequencing (NGS) “dark regions” of the genome; that is, areas of the genome that are difficult or impossible to sequence or map with short-read sequencing. Due to high sequence similarity with pseudogenes or paralogs, or the presence of repeat elements and segmental duplications, short-read sequencing may result in poor mapping, read depth, and sequencing yield. Discover what you’ve been missing with the Twist Alliance Dark Genes panel, a comprehensive 22 Mb panel of 389 medically-relevant dark genes, including CYP2D6, GBA, SMN1/2, and PMS2 (figure 1). This panel includes genes missed by short-read NGS that might typically require several platforms to adequately capture, allowing for assay consolidation and cost-effectiveness. A full list of genes can be found on the Twist Long Read Sequencing Panels website.


“The Dark Genes panel enables the accurate detection of genetic diversity in a large and unique set of medically relevant genes. I am confident this workflow will generate significant and novel insights in disease genomics and population-wide genetic studies.”

– Fritz Sedlazeck, PhD
Associate Professor Human Genome Sequencing Center,
Baylor College of Medicine
Pharmacogenomic (PGx) profiles have an impact on medication safety and efficacy. With the Twist Alliance Long-Read Pharmacogenomics panel, you can leverage the benefits of HiFi sequencing in a robust, scalable, and cost-effective PGx assay. Stuart Scott, PhD, FACMG, Director of the Stanford Medicine Clinical Genomics Laboratory, highlighted the importance of phasing enabled by the panel, noting “really clean characterization of haplotypes and complete star allele architecture.” This is a comprehensive 2 Mb panel that includes 49 genes, the majority of which are captured full-length (introns and exons) (table 1). This panel was designed to capture all 20 current genes with Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines, as well as FDA PGx genes and genes of PGx research interest.

**Why pharmacogenomics with PacBio?**

- Comprehensive coverage includes SNVs, indels, and SVs
- Direct phasing for unambiguous star allele assignment
- Ancestry-agnostic coverage of actionable PGx genes

**Panels at a glance**

<table>
<thead>
<tr>
<th>Twist Alliance panel</th>
<th>Number of genes</th>
<th>Samples per Sequel Ile SMRT® Cell 8M</th>
<th>Samples per Revio SMRT Cell</th>
<th>Mean target coverage</th>
<th>Target bases ±10-fold read depth</th>
</tr>
</thead>
<tbody>
<tr>
<td>Long-read PGx</td>
<td>49</td>
<td>24</td>
<td>72</td>
<td>190-fold</td>
<td>99%</td>
</tr>
<tr>
<td>Dark Genes</td>
<td>389</td>
<td>4</td>
<td>12</td>
<td>75-fold</td>
<td>93%</td>
</tr>
</tbody>
</table>

**RESOURCES**

1. Sample data for both panels can be found at [https://www.pacb.com/connect/datasets/#targeted-datasets](https://www.pacb.com/connect/datasets/#targeted-datasets)
2. Twist long-read resources [https://www.twistbioscience.com/products/ngs/Long-Read-Sequencing-Panels](https://www.twistbioscience.com/products/ngs/Long-Read-Sequencing-Panels)

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