Twist Human Sample ID Kit

An integrated single source solution for sample tracking

KEY BENEFITS

Streamlined Workflow

- Direct from whole blood or genomic DNA
- Sample to sequence ready libraries in 5 hours
- Easy to integrate into Twist NGS workflows

Robust Performance

- Measure 30 SNPs and the AMELX/Y Locus
- Optimized primer pairs for best results
- Efficient polymerase and proprietary buffer system
- High power of discrimination—as low as 1 in 150,000 of 96 well plates

Integrated Solution

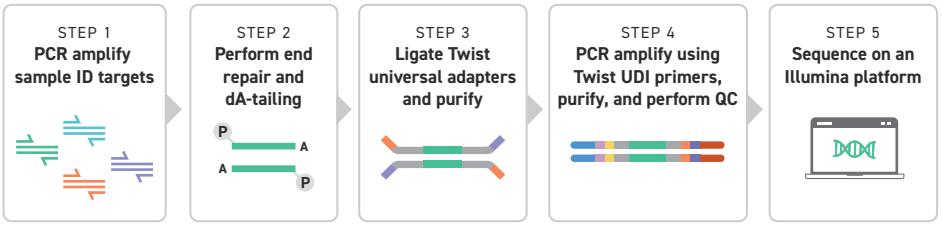
- Complete end-to end solution with single source reagents
- Validated with Twist EF library prep, UDIs and exome 2.0 panel
- Step-by-step bioinformatics guide for data analysis percentages available

Laboratories offering NGS services receive a high volume of incoming samples often with an expectation of fast turnaround time and accurate reporting. However, the high complexity of sequencing workflows can lead to inevitable sample mix-ups. This can result in costly re-runs and misinterpretation of data for critical samples. Safeguards against misidentification often require non-sequencing test methods and analysis, or do not enable tracking samples starting at arrival to the lab. The Twist Human Sample ID Kit offers a simple multiplex PCR approach coupled with an NGS workflow to ensure sample identification and tracking from an early step in the workflow.

Starting from whole blood, or purified genomic DNA, the Sample ID primer pool targets a combination of Single Nucleotide Polymorphisms (SNPs) (described by Pengelly, et al[1] and the EuroGentest committee[2]) and the AMELX/Y locus and pairs with genotypes of samples enriched with the Twist Exome 2.0 panel. Alternatively, a Sample ID target enrichment panel can be blended into a custom target enrichment panel so that samples can be matched with the Human Sample ID genotypes. SNPs included in the Sample ID were selected to ensure discrimination in any population. Using the minor allele frequency (MAF) in various populations, the calculated pairwise Power of Discrimination is one in 700 million individuals. In other words, the probability of encountering the same SNP profile on a 96-well plate is approximately 1 in 150,000 plates. Overall, with its direct-from-blood capability and power of discrimination, the Twist Human Sample ID kit provides high confidence that the right results are matched to the right samples.

Chemistry

SAMPLE ID LIBRARY PREPARATION WITH UNIVERSAL ADAPTERS AND UDI PRIMERS



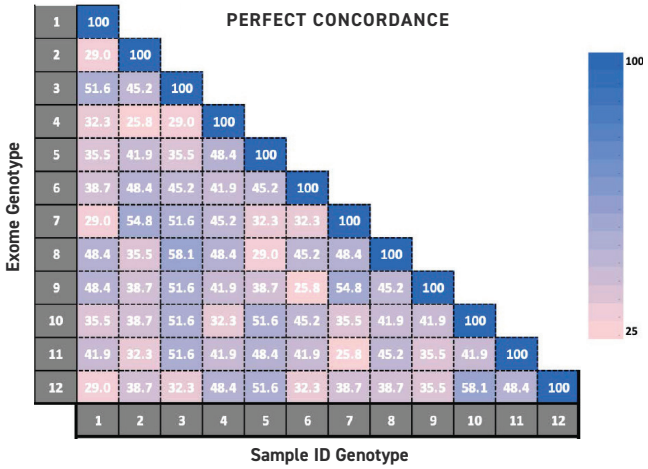
High Discrimination Power to Distinguish Two Samples

The genomic sites chosen for Twist Human Sample ID ensure that the power of discrimination remains high even in populations with relatively low diversity at these SNPs. The pairwise power of discrimination shows the per population estimates of the probability that any two nonidentical samples would be profiled as identical using the SNPs in this product. In practical terms, the chance that two patients on the same 96-well plate would have the same genotype and results in a “collision” have a very low probability and would only happen in 1 out of 150,000 plates processed in the least diverse population

POPULATION NAME FROM GNOMAD	PAIRWISE POWER OF DISCRIMINATION	NUMBER OF PLATES FOR ONE EXPECTED COLLISION
East Asian	3.71e-10	(1/675k)
African / African American	1.19e-10	(1/1694k)
Ashkenazi Jewish	2.32e-10	(1/934k)
European (non-Finnish)	1.45e-10	(1/1235k)
South Asian	1.40e-9	(1/150k)
Latino / Admixed American	1.96e-10	(1/1250k)
European (Finnish)	1.87e-10	(1/1086k)
Other	1.19e-10	(1/181k)
All	1.08e-10	(1/2083k)

Sample Identification

Here we show concordance between the targets in the Sample ID libraries and their counterparts in exome-enriched libraries. Sample ID libraries were pooled with their respective exome-enriched libraries, sequenced on the NextSeq 550, and downsampled to 1000X of the target size. SNPs from the Sample ID libraries were then compared to the SNP profile of their paired exome libraries.



REFERENCES

1. Pengelly, et al. A SNP profiling panel for sample tracking in whole-exome sequencing studies. Genome Medicine. 2013 (5):89
2. EuroGentest. Guidelines for diagnostic next generation sequencing. 2014. www.college-genetics.be/assets/recommendations/fr/guidelines/EuroGentest%20NGS_2014.pdf

Twist Human Sample ID kit with Enzymatic Fragmentation Library Prep kit (EF) and Universal adapter system (UDI) are all components of the Twist Bioscience portfolio of products for NGS Library Preparation.

LEARN MORE

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ORDERING INFORMATION

105889: Twist Human Sample ID Kit with EF 1.0 and UDI Plate A
105892: Twist Human Sample ID Kit with EF 1.0 and UDI Plate B
105893: Twist Human Sample ID Kit with EF 1.0 and UDI Plate C
105894: Twist Human Sample ID Kit with EF 1.0 and UDI Plate D
105895: Twist Human Sample ID Kit with EF 2.0 and UDI Plate A
105896: Twist Human Sample ID Kit with EF 2.0 and UDI Plate B
105897: Twist Human Sample ID Kit with EF 2.0 and UDI Plate C
105898: Twist Human Sample ID Kit with EF 2.0 and UDI Plate D

The individual SKUs mentioned above contains all necessary reagents to carry out complete workflow of Sample ID library preparation for 96 samples.