

# THE SCIENCE OF ] WHAT SAMPLE IS THIS?



How many biobank samples are mis-annotated?

Over 2%



Make sure your results are valid

## Reliable sample identification and quantification.

A sample can be mislabeled at point of origin or mixed up during transfer from one laboratory to another. It is estimated that ~2% of samples stored within biobanks and other repositories are incorrectly annotated. The challenge of the research community is to ensure that the correct samples are used and reported in research studies.<sup>1,2</sup>

The iPLEX® Pro Sample ID Panel provides a highly accurate and rapid research method for identification and quantification. It is also ideal for qualifying samples prior to PCR-based assay methods on the MassARRAY® System or next generation sequencing platforms.

- 44 SNPs ensure the chances of a random match between any two samples is  $\leq 1.8 \times 10^{-19}$ .
- Spike-in controls with known copy number for estimating amplifiable copies (500-18,000 copies).
- Suitable for a variety of DNA sample types including cell lines and formalin-fixed, paraffin-embedded (FFPE) tissues.
- A cost-effective single-well assay that requires  $\leq 10$  nanograms of DNA per sample.

The panel contains 44 SNPs and 3 gender markers for sample identification, and 5 DNA copy number controls for sample quantification.

### SNPS AND GENES IN THE iPLEX PRO SAMPLE ID PANEL

albumin_1	rs11781516	rs1994997	rs3819854
albumin_10	rs13050660	rs2010253	rs717302
albumin_5	rs1335873	rs2040411	rs727811
albumin_8	rs1357617	rs2046361	rs729172
albumin_9	rs1360288	rs2056277	rs740910
AMEL_XY	rs136337	rs2076848	rs8037429
ARSD_X_Y	rs1382387	rs214054	rs826472
TGIF2L_XY	rs1413212	rs2247221	rs876724
rs1005533	rs1454361	rs2518968	rs891700
rs1024116	rs1463729	rs251934	rs901398
rs1028528	rs1468118	rs2714854	rs914165
rs10495407	rs1493232	rs2831700	rs9583190
rs10771010	rs1982986	rs354439	rs964681

**Disclaimer:** The panel includes 44 SNPs (designated by reference sequence [rs]), 5 DNA copy number assays (albumin), and 3 gender markers (AMEL, ARSD, and TGIF2L). SNP assays were selected with 45-55% heterozygosity across six major populations in the HapMart tool.

The iPLEX Pro Sample ID Panel is not intended for forensic, parentage, or ethnicity identification. The panel is also not intended for differentiating certain individuals (monozygotic twins).



## KEY FEATURES OF THE IPLEX PRO SAMPLE ID REPORT SOFTWARE

The Sample ID Panel uses Typer 4.0 for standard plate setup, data acquisition, and signal processing, and for generating a database from which you can quickly manage sample data and interpret relationships. An HTML-based Sample ID report tool offers Summary, Plate, Match, and Sample Reports.

- Samples can be queried against a database to quickly determine matches and mismatches.
- Automated reports provide the SNP profile, gender ID, and copy number for every sample run.
- A historical comparison queries samples against the entire database.
- A local comparison queries a sample against other samples within the same run.

## HISTORICAL COMPARISON

A historical comparison determines sample relationships via global search comparisons within the database and accommodates a growing number of samples for medium-to-large scale biobanks. You can determine whether or not two samples originate from the same individual, establish a chain of custody, and manage a large number of samples, including any replicated samples.

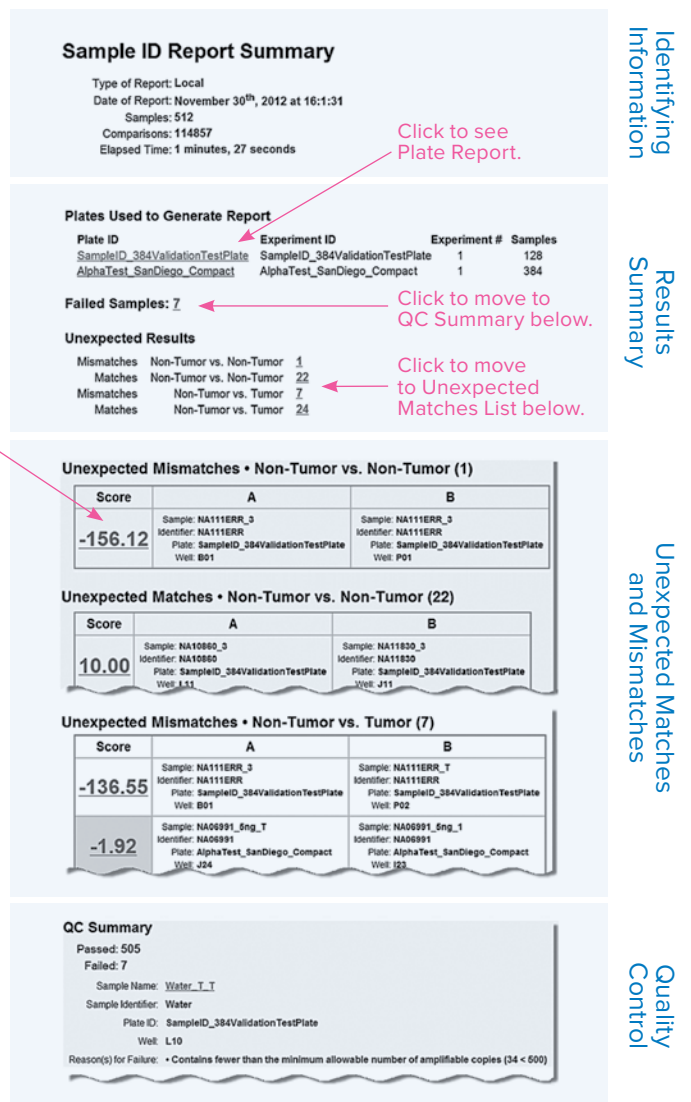
## LOCAL COMPARISON

A local comparison quickly analyzes samples within a specific run, and is ideal for applications in which you want to determine whether samples are derived from the same patient (tumor vs. normal). It will account for allele dropout in the analysis, as these types of samples may be subject to loss-of-heterozygosity (LOH).

## SUMMARY REPORT

A summary report shows the number of comparisons and an overview of any unexpected matches or unexpected mismatches.

Figure 1. Sample ID Report Summary



Identifying Information

Results Summary

Unexpected Matches and Mismatches

Quality Control



Amplify

Rapidly identify sample mismatches



Detect

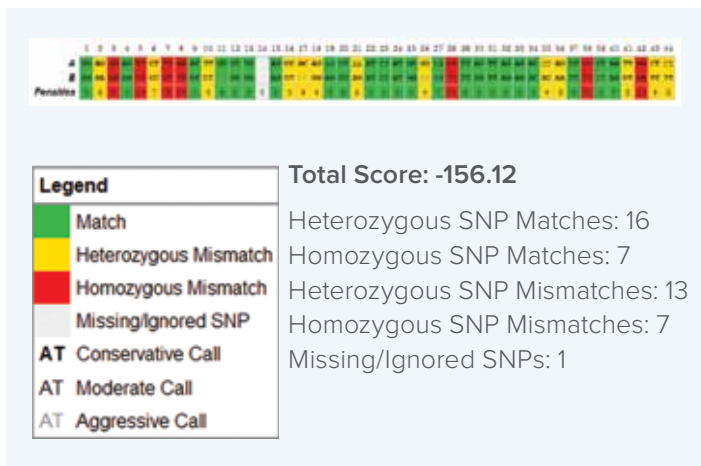
### PLATE REPORT

The Plate Report lists results for every well in the plate, including the number of SNP calls, number of intact amplifiable copies, gender, whether it passed quality control, and the number of matches and mismatches found.

### MATCH REPORT

The Match Report lists identifying information for the two samples involved in a comparison, details of the SNP calls for each sample, and the matching score. Figure 2 shows an excerpt from an unexpected mismatch report.

Figure 2. Sample ID Match Report







### SAMPLE REPORT

Each Sample Report (Figure 3) contains results of the five copy number assays, the three gender assays, and the 44 SNP assays. It also reports the details of each match or mismatch with other samples.

Figure 3. Sample ID Sample Report



## iPLEX PRO SAMPLE ID PANEL COMPONENTS

<b>AMPLIFY</b>		PCR Enzyme PCR Accessory Set Sample ID PCR Primers Sample ID Q-Mix
<b>EXTEND</b>		iPLEX Pro Reagent Set Sample ID Extend Primers
<b>DETECT</b>		SpectroCHIP® Array and Resin Kit (10 x 96, 2 x 384)
<b>ANALYZE</b>		Sample ID HTML report tool (Typer)

## ORDERING INFORMATION

CAT NO	FORMAT	SAMPLES/KIT
<b>25094</b>	2 x 384	768
<b>25093</b>	10 x 96	960



Analyze

## REFERENCES

- Sanchez J, et al. A multiplex assay with 52 single nucleotide polymorphisms for human identification. *Electrophoresis* 2006;27:1713–1724.
- Demichelis F, et al. SNP panel identification assay (SPIA): A genetic-based assay for the identification of cell lines. *Nucleic Acids Research* 2008;1-11.

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