SAMPLE INTEGRITY

# Sample Integrity Solutions

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Not for use in diagnostic procedures.

Agena® BIOSCIENCE

# Rapid & Accurate Sample Management

## 1 IN EVERY 200 SAMPLES IS MIXED UP[1]

It is estimated that 1 in every 200 samples is mixed up. Many of these mistakes originate outside the lab and traditional quality checks are not enough to detect them. Sample mix-ups lead to incorrect results and can damage a lab's reputation.

## DOWNSTREAM MOLECULAR ANALYSIS IS UNSUCCESSFUL IN 22.5% OF CASES[2]

Many samples fail downstream analysis due to low quantity or poor-quality DNA; meaning wasted time and no results. DNA quality measurements based on absorbance detect nucleic acid rather than amplifiable DNA fragments and often provide inaccurate results.

**Agena can help. The iPLEX® Pro Sample Integrity Panels provide sample identification, DNA quantity and DNA quality information.**

- **FOR MOLECULAR DIAGNOSTIC LABS** – Prevent sample mix-ups and ensure DNA is of sufficient quality before expensive downstream analysis.
- **FOR BIOBANKS** – Verify sample origin to prevent erroneous study findings.
- **FOR TOXICOLOGY LABS** – Detect fraudulent samples by matching submissions to a reference genetic fingerprint.

# Three sample quality measurements from a single assay:

**SAMPLE IDENTIFICATION**

## Is this the correct sample?

Verify sample identity by comparing genetic fingerprints. Prevent mix-ups and mismatches even if they originated outside of the lab.

**DNA QUANTITY**

## Is there enough DNA?

Understand how much DNA is present within a sample. Ensure there is enough for all applications.


**DNA QUALITY**

## Is the DNA of sufficient quality?

Decide if the DNA in a sample is suitable for downstream analysis by measuring the number of amplifiable DNA fragments rather than free nucleotides and other contaminants.

# Panel Selection

Each of the iPLEX® Pro Sample Integrity panels enable rapid and highly accurate sample identification and DNA quality measurements. SNPs and biological gender markers are used to generate a sample's unique genetic fingerprint. The automated reporter compares this to a reference profile and generates an easy to interpret Match or Mismatch result.  In the same reaction, amplification is compared to built-in controls to determine how many amplifiable DNA copies are present in the sample.

These pre-designed panels are available for on-demand ordering to get you testing samples quickly. Each leverages the proven iPLEX Pro chemistry and can be processed in the same run with other applications.

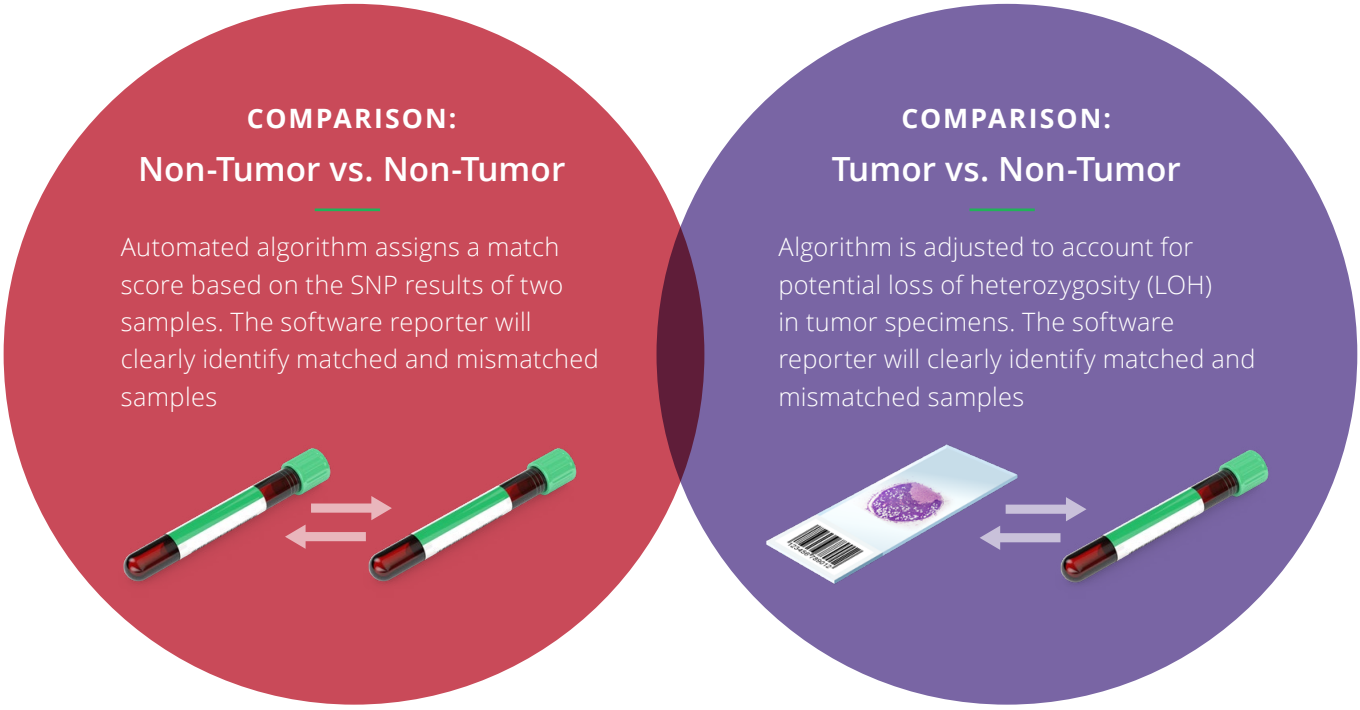
## IPLEX PRO SAMPLE ID PANEL

This panel determines a sample's identity, DNA quality and DNA quantity. It targets 44 SNPs with high minor allele frequency across major HapMap populations and 3 biological gender markers.

## IPLEX PRO EXOME QC PANEL

This panel determines a sample's identity, DNA quality and DNA quantity. To screen for sample degradation, it also provides the number of intact, amplifiable DNA copies across a size range of 100, 200, 300, 400 and 500 nucleotides. It targets 21 SNPs in exonic regions of the genome and 3 biological gender markers.

### Comparing DNA eliminates misidentification:



## Sample Integrity Panel Comparison

| | iPLEX Pro Sample ID Panel | iPLEX Pro Exome QC Panel |
|---|---|---|
| **Sample Identification** | 44 SNPs, 3 biological gender markers | 21 SNPs, 3 biological gender markers |
| **DNA Quantity** | 5 controls | 25 controls |
| **DNA Quality** | Assay measures amplifiable copies of DNA | Assay measures amplifiable copies of DNA at several fragment lengths |
| **DNA Input** | 5 ng+ | 5 ng+ |
| **Genomic Region Targeted** | Intronic & Exonic Regions | Exonic Regions |
| **Automated Reporting** | Included | Included |

## iPLEX Pro Sample ID Panel - SNPs & Genes

| | | | |
|---|---|---|---|
| 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_XY | 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 |

## iPLEX Pro Exome QC Panel - SNPs & Genes

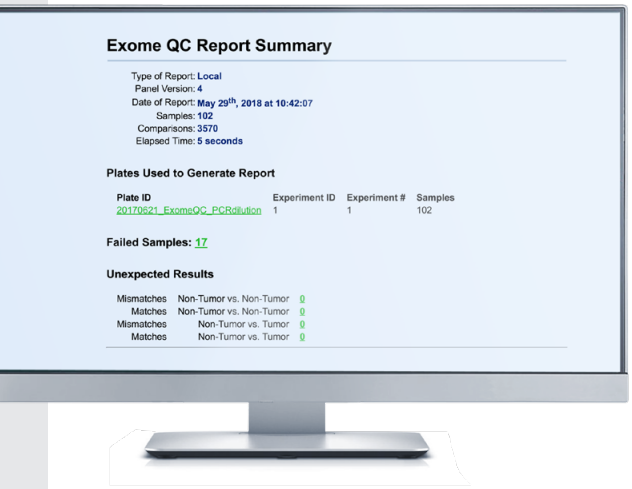
| | | | |
|---|---|---|---|
| POLR2A | TPT1 | rs10495563 | rs4478844 |
| POLR2A | VIM | rs1065457 | rs586421 |
| TBP | POLR2A | rs1127379 | rs6420424 |
| ALB | RPL27A | rs11998387 | rs6977125 |
| IPO8 | HBS1L | rs1200349 | rs7653897 |
| POLR2A | TPT1 | rs12594531 | rs773901 |
| POLR2A | IPO8 | rs1344 | rs9131 |
| TBP | PDCD2 | rs17548783 | AHSP |
| IPO8 | TBP | rs2246209 | AMEL |
| HBG2 | PPIA | rs2273171 | ARSD |
| GUSB | HBS1L | rs2301771 | |
| AHSP | PDCD2 | rs3743165 | |
| RPL27A | rs1045728 | rs3884596 | |

# iPLEX Pro Sample Integrity Reporting Software

The iPLEX Pro Sample Integrity panels are accompanied by reporting software that makes sample identity and DNA quality determination quick and easy. Automated reports clearly show sample information, biological gender, the number of amplifiable DNA copies and a match/mismatch analysis.

## SUMMARY REPORT

The summary report provides a quick overview, highlighting any unexpected sample matches or mismatches. It also lists samples that may have failed quality control due to poor quality or low quantity DNA.

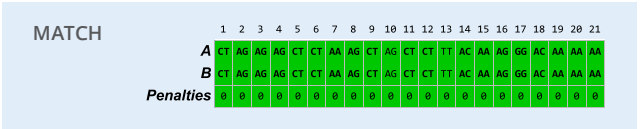


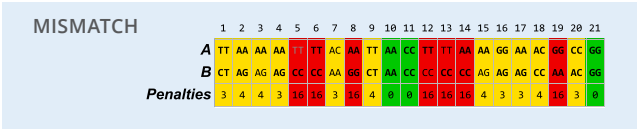

- **RUN INFORMATION** – Displays details of the run including name, date and number of samples analyzed.
- **QC STATUS** – Lists samples that may have failed QC due to poor quality or low quantity DNA
- **MATCH / MISMATCH INFORMATION** – Clearly identifies samples did not agree with the prescribed matching criteria

## MATCH REPORT

The Match Report lists identifying information, SNP call and matching score for two samples involved in a comparison.

- **MATCH / MISMATCH INFORMATION** – Clearly identifies samples did not agree with the prescribed matching criteria
- **HISTORICAL DATABASE** – Performs a global search across the entire database to identify sample relationships.
- **LOCAL COMPARISON** – Allows for a quick analysis of sample relationships within an individual run.

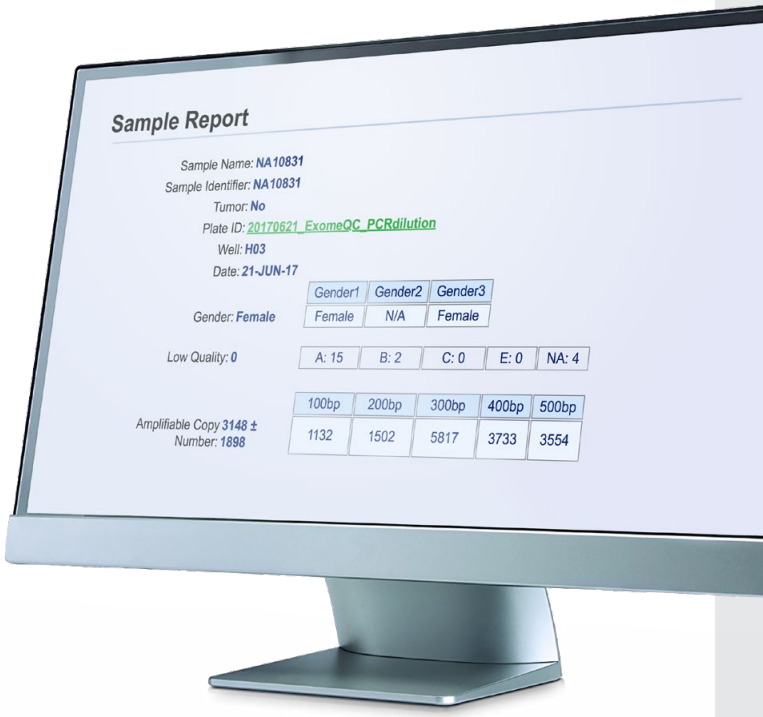



Match
Heterozygous Mismatch
Homozygous Mismatch
Missing/Ignored SNP

## SAMPLE REPORT

Each Sample Report contains identifying information, DNA copy number, biological gender, SNP calls and Unexpected Match/Mismatches.



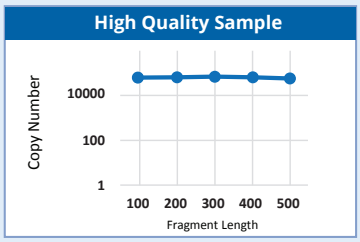

## QC REPORT

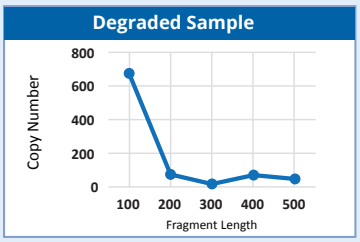
Both the iPLEX Sample ID and Exome QC panels use in-assay control to determine the number of amplifiable DNA copies present in a sample. The Exome QC panel provides this quality analysis at several DNA fragment lengths and generates a sample degradation curve.

### DNA Quality Data

Amplifiable Copy Number: **4334 ± 549** | 4188 | 3737 | 3964 | 4706 | 5076

### Exome QC Degradation Curves




## ASK ABOUT OUR OTHER APPLICATIONS

The MassARRAY® is a versatile genetic analysis tool, not limited to a single application. Ask for information regarding our other applications including Oncology, Pharmacogenetics and Hereditary Genetic Testing.

## ORDERING INFORMATION

| Catalog No. | Item | Chip format | # of Samples |
|---|---|---|---|
| 13176D | iPLEX Pro Sample ID Panel Set - CPM (2x96) | 96 CPM | 192 |
| 13116 | iPLEX Pro Sample ID Panel Set (2x96) | 96 | 192 |
| 25093F | iPLEX Pro Sample ID Panel Set - CPM (10x96) | 96 CPM | 960 |
| 25093 | iPLEX Pro Sample ID Panel Set (10x96) | 96 | 960 |
| 25094D | iPLEX Pro Sample ID Panel Set - CPM (2x384) | 384 CPM | 768 |
| 25094 | iPLEX Pro Sample ID Panel Set (2x384) | 384 | 768 |
| 25095 | iPLEX Pro Sample ID Panel Set (10x384) | 384 | 3,840 |
| 25095D | iPLEX Pro Sample ID Panel Set - CPM (10x384) | 384 CPM | 3840 |
| 13175F | iPLEX Pro Exome QC Panel Set - CPM (10x96) | CPM 96 | 960 |
| 13175 | iPLEX Pro Exome QC Panel Set (10x96) | 96 | 960 |
| 13176D | iPLEX Pro Exome QC Panel Set - CPM (10x384) | 384 CPM | 3840 |
| 13176 | iPLEX Pro Exome QC Panel Set (10x384) | 384 | 3,840 |

## References


1. Marberger M, McConnell JD, Fowler I, et al. Biopsy Misidentification Identified by DNA Profiling in a Large Multicenter Trial. Journal of Clinical Oncology. 2011;29(13):1744-1749. doi:10.1200/JCO.2010.32.1646.
2. Hussam Al-Kateb et al. Identification of major factors associated with failed clinical molecular oncology testing performed by next generation sequencing (NGS). Molecular Oncology, 9 (2015) 1737-1743.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