

SAMPLE INTEGRITY



Sample Integrity Solutions



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

Agena[®]
BIOSCIENCE

Rapid & Accurate Sample Management

1 IN EVERY 200 SAMPLES IS MIXED UP¹

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 CASES²

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.

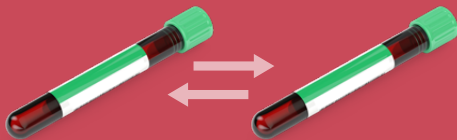
IPLEX PRO EXOME QC PANEL

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:

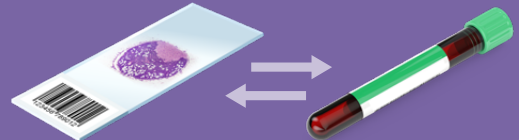
COMPARISON: Non-Tumor vs. Non-Tumor

Automated algorithm assigns a match score based on the SNP results of two samples. The software reporter will clearly identify matched and mismatched samples



COMPARISON: Tumor vs. Non-Tumor

Algorithm is adjusted to account for potential loss of heterozygosity (LOH) in tumor specimens. The software reporter will clearly identify matched and mismatched samples





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

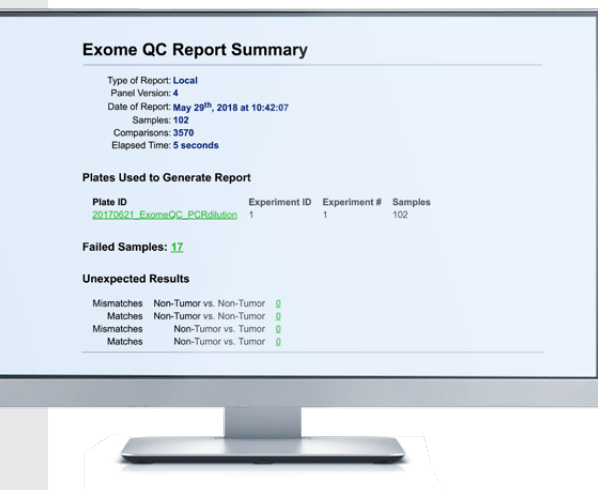
POLR2A	TPT1	rs10495563	rs4478844
POLR2A	VIM	rs1065457	rs586421
TBP	POLR2A	rs1127379	rs6420424
ALB	RPL27A	rs11998387	rs6977125
IPO8	HBS1L	rs1200349	rs7653897
POLR2A	TPT1	rs12594531	rs773901
POLR2A	IPO8	rs1344	rs9131
TBP	PDCD2	rs17548783	AHSP
IPO8	TBP	rs2246209	AMEL
HBG2	PPIA	rs2273171	ARSD
GUSB	HBS1L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

		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21
MATCH	A	CT	AG	AG	AG	CT	CT	AA	AG	CT	AG	CT	CT	TT	AC	AA	AG	GG	AC	AA	AA	AA
	B	CT	AG	AG	AG	CT	CT	AA	AG	CT	AG	CT	CT	TT	AC	AA	AG	GG	AC	AA	AA	AA
	Penalties	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0

		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21
MISMATCH	A	TT	AA	AA	AA	TT	TT	AC	AA	TT	AA	CC	TT	TT	AA	AA	GG	AA	AC	GG	CC	GG
	B	CT	AG	AG	AG	CC	CC	AA	GG	CT	AA	CC	CC	CC	CC	AG	AG	AG	CC	AA	AC	GG
	Penalties	3	4	4	3	16	16	3	16	4	0	0	16	16	16	4	3	3	4	16	3	0

- Match
- Heterozygous Mismatch
- Homozygous Mismatch
- Missing/Ignored SNP

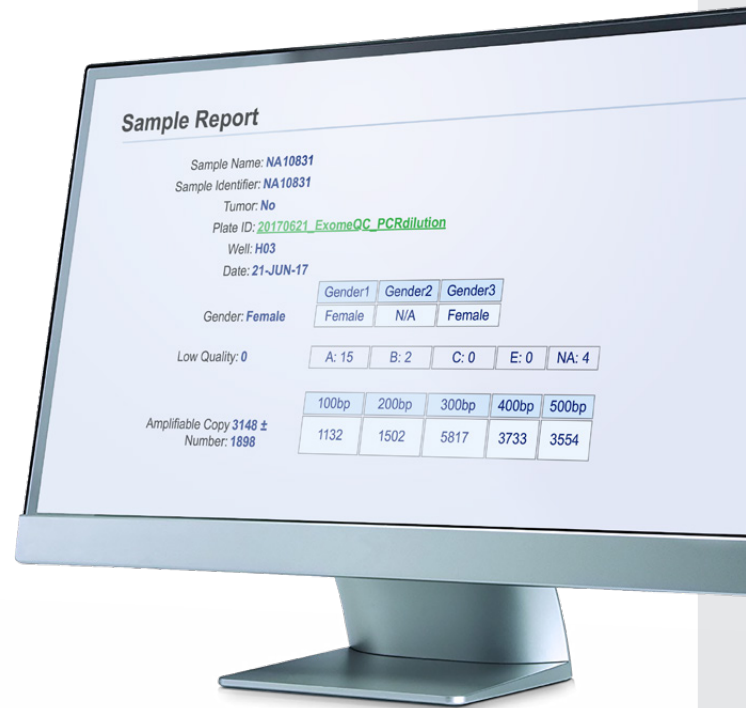


SAMPLE REPORT

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

QC REPORT

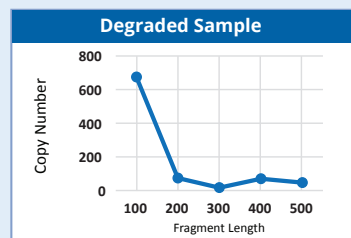
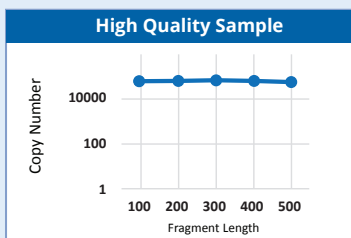
Both the iPLEX Sample ID and Exome QC panels use in-assay control to determine the number of amplifiable DNA copies present in a sample. The Exome QC panel provides this quality analysis at several DNA fragment lengths and generates a sample degradation curve.



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
13116F	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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