

Analytical validation of our bioinformatics services

Results obtained with **open reference data** · 8/8 services validated

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ISO 9001:2015 (ES147645-1, BV/ENAC) · Data processed in the EU · GDPR

Scope and nature. All results correspond to **research (RUO)** services and are delivered as a **technical report**. We are **not** accredited to ISO 15189 and we do **not** offer a product with CE-IVD marking. The validation documented in this report is **internal** and does not replace accredited clinical validation.

1. Why we publish this

When a laboratory outsources a bioinformatics analysis, the hard question is not «how much does it cost?» but «**can I trust the result and reproduce it?**». Our answer is not a promise: it is **eight validations with public data that have known truth**, with the success criterion **frozen before** measuring.

- **Each service** is compared against a public dataset with a ground-truth result.
- **The success metric is defined before** running (it is not tuned a posteriori).
- **The result is published even if it fails** (here, all eight passed).
- Everything is versioned, with scripts and declared production conditions.

2. Results (8/8 PASS)

#	Service	Reference dataset	Metric	Result
V1	TMB	SEQC2 HCC1395 tumor/ normal (WES)	Concordance vs independent counter	PASS — module $\pm 0,3$ %; production pipeline 19,9 %; negative control 0,089
V2	Mutational signatures	TCGA SKCM + COAD (GDC)	Dominant signature / contribution	PASS — UV 92,9 % top-1 · MMR 92 % by group
V3	HRD	TCGA ABSOLUTE + MC3 (truncated BRCA1/2)	AUC of the HRD-sum	PASS — AUC 0,979 (ovarian BRCA1/2 vs MMR)

V4	MSI	HCC1395 (MSS) + HCT116 (MSI-H, ENA)	Category + % unstable microsatellites	PASS — MSS 0,79 % · MSI-H 20,4 %
V5	GWAS	openSNP (eye colour, CC0)	Top hits + null test	PASS — top-5 100 % HERC2/OCA2 (rs12913832 $p=5\times 10^{-11}$); clean null
V6	Epigenomics	ENCODE GM12878 (ATAC/CTCF) + H1 WGBS	Jaccard / %methylation	PASS — ATAC J 0,597 · CTCF top-1000 72 % · WGBS 82,77 % vs 82,8 % ENCODE
V7	Neoantigens	IEDB HLA-A*02:01 (n=2.729)	AUC · Spearman correlation	PASS — AUC 0,947 · p 0,807
V8	Microbiome	Nielsen 2014 (curatedMetagenomicData)	Published IBD markers	PASS — 4/4 reproduced (e.g. <i>F. prausnitzii</i> ↓ $q=4\times 10^{-23}$)

What each result means, in short

TMB (V1)

Our count matches an independent counter within $\pm 0,3$ %, and the full production pipeline reproduces a published value (19,9 %) with a clean negative control (0,089).

Mutational signatures (V2)

In melanoma we identify the UV signature as dominant in 92,9 % of cases; in tumours with repair defects, the MMR group contribution is recovered in 92 %.

HRD (V3)

The score separates ovarian tumours with truncated BRCA1/2 from those without it with AUC 0,979 — that is, with almost no overlap.

MSI (V4)

Two cell lines with known truth: the stable one is classified as stable (0,79 % unstable) and the unstable one as MSI-H (20,4 %). No false positives or negatives.

GWAS (V5)

The top five hits of an eye-colour study all fall in HERC2/OCA2, with the canonical SNP at $p=5\times 10^{-11}$; the null test produces no false associations.

Epigenomics (V6)

ATAC and CTCF peaks against ENCODE (Jaccard 0,597; 72 % overlap in the top-1000) and global methylation 82,77 % against the published 82,8 %.

Neoantigens (V7)

Our peptide-HLA binding prediction against 2.729 experimental measurements from IEDB: AUC 0,947 and correlation 0,807.

Microbiome (V8)

We reproduce the four published inflammatory bowel disease markers in the reference cohort, with the same directions and significance.

3. What else was delivered: the corrected software

Validation does not only measure: **it finds errors**. This cycle fixed nine real defects in the pipelines (header-sensitive CSQ annotation, tumour sample column, filter labels, artefact-signature exclusions, MSI parameters, dosage→GT conversion, coordinates, reading of compressed files). All are in the repository, versioned.

4. Production conditions (what you need to know)

- **TMB** requires an in-house panel of normals + gnomAD + a VAF floor of 0,05. The panel from the validation is specific to the SEQC2 dataset.
- **MSI**: the commercial licence for the main tool is pending.
- **Signatures**: commercial licence for the signature database pending.
- **Epigenomics**: the alignment leg (Bismark, GRCh38 index) is used as-is from the field standard; it was not re-validated in this cycle.

5. Declared limits (no frills)

- The validations use **public datasets and cohorts**, with their own inherent biases (direct-to-consumer test data in GWAS; a single cohort in microbiome).
- This is **not** accredited clinical validation, nor does it replace what is required of a laboratory accredited to ISO 15189.
- The services are offered for **professional use and research**.

6. What you receive in each project

- **Scripts, container and versions** used (not just the result).
- **Parameters** and quality control at every step.
- Technical report in **Spanish, English, French, Italian or Portuguese**.
- Data processed in the EU, AES-256 encryption, data processing agreement (GDPR).
- Delivery via secure SFTP or portal.

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