User and deployment guide

How to install, sign in, record a patient and export. Covers the CRM module and the stand-alone TribroPheno file.

1. Install

- CRM: upload the TribroOMICS-CRM folder (version 9). “Patients & phenotypes” appears in the menu. Existing CRM data is kept.
- TribroLIMS or stand-alone: upload the TribroLIMS folder and open TribroPheno.html. It uses the same sign-in as the CRM but keeps its own separate encrypted data.
- Serve over HTTPS only. A current Chrome, Edge, Firefox or Safari is needed.
- New HPO release: request a new build. Old annotations keep their release; run “Check against HPO” per record.

2. First steps

- Open Module settings: enter your name (used as annotator), choose the acting role, set the patient ID prefix.
- Click “Load synthetic cases” to explore with six test patients.
- Turn on the authenticator app in Settings before entering real data.

3. Record a patient

- New patient → Registration. Sex and date of birth are needed before a record can be submitted.
- History, Family & pedigree: add entries with “Add”; everything saves automatically (see “Saved” at the top).
- Organ systems: pick a system and set a status for each relevant finding. Leave untouched what was not asked.
- HPO annotation: search any term or add a free-text observation and use “Suggest HPO”.
- Notes & extraction: paste a letter, click “Extract”, then accept, change or reject each suggestion.
- Prioritisation: “Rank genes and panels”. Read the warning about non-specific phenotypes.
- Submit, then Verify. A verified record is locked; use Amend and give a reason to change it.

4. Export

Export	Use
Print / save as PDF	Clinical phenotype summary for the file or referral
HPO TSV / CSV	Spreadsheets and phenotype tools

Phenopacket v2 JSON	Exomiser, LIRICAL and other GA4GH tools
FHIR R4 bundle	Hospital systems (mapped, not profile-validated)
PED	Family structure for variant pipelines
TribroLIMS subject JSON	Subject, HPO terms, test orders and suggested panels for import into TribroLIMS
Cohort TSV / CSV / Phenopackets	De-identified research data sets
Backup (Settings)	Full copy of all data; store securely

5. TribroLIMS hand-over format

The file "..._lims_subject.json" has format "tribro.lims.subject/1" with: subject (id, research key, family ID, sex, age group, case type, indication), hpo (present, excluded, suspected identifiers), orders (test type, panel code, assembly, status, date), suggestedPanels, candidateGenes and consent. A TribroLIMS import route can read it directly; the route itself is part of the LIMS code base.

6. Maintenance

- Back up after each working session.
- Review the audit trail monthly.
- Re-run "Extraction check" and the synthetic cases after every update.
- Contact: info@tribro-omics.com

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