

**Human Technopole
National Facility for Genomics
National Facility for Data Handling and Analysis
CALL FOR EXPRESSION OF INTEREST
LARGE-SCALE GENOMICS PROJECTS
26-EoI-G-LARGE-SCALE**

1. PURPOSE OF THE DOCUMENT

The purpose of this document is to solicit submissions of and provide guidelines for submitting **Expressions of Interest** to access the **Whole Genome Sequencing and Analysis** services offered by the Human Technopole National Facilities. We invite preliminary proposals for large-scale population genomics projects in the biomedical field that meet the following criteria:

- **Large-scale scope:** Projects must involve significant cohort sizes or sample numbers.
- **Translational potential:** Research should demonstrate a clear pathway toward clinical applications, such as new diagnostic tools or therapeutic interventions.
- **Commitment to data sharing:** To accelerate genomic research, there must be a clear and detailed commitment to make results accessible to the wider scientific community through data sharing frameworks, while ensuring compliance with relevant legal and ethical frameworks.

2. INTRODUCTION

Human Technopole (HT) currently has the capacity to support large-scale whole-genome sequencing of at least 10,000 samples per year through subsidized access to the relevant services offered by the National Facilities (NFs) for Genomics and for Data Handling and Analysis. The Standing Independent Evaluation Committee (SIEC) is exploring the potential of offering a capacity of up to 20,000 samples as a 'Special Project' to support large-scale population genomics in the biomedical field.

The goal is to provide whole genome sequencing for projects that require large cohorts, using comprehensive genetic data as the foundation for mechanistic understanding of physiological and pathological processes, to directly improve medical care. The objective here will be to generate genomic data and utilise multi-omics to – for example – pinpoint causal variants, map genotype-phenotype links for risk stratification, formulate testable hypotheses, create diagnostic biomarkers, therapeutic targets and personalized treatment plans.

This initiative is designed to pilot end-to-end data generation and standardized analysis, while exploring solutions to make the data available and accessible in a manner consistent with applicable legal and ethical requirements. The overarching goal is to advance genomic medicine in Italy by evaluating the infrastructure required to facilitate genomic and clinical data analysis, sharing and accessibility at scale.

Specifically, the initiative seeks to:

- Use **genomic data to benefit clinical care and drive discoveries** in Italy.
- Expand the **availability and accessibility of genomic data** from the Italian population.
- Broaden the **dissemination of integrated genomic, phenotypic data, and the associated metadata**.

A fundamental pillar of this initiative is the generation of human genomic datasets, which are made available to the wider scientific community in their entirety. These datasets will be

accompanied by demographic and phenotypic information, and where possible by related relevant biological samples.

3. SCOPE OF THE EXPRESSION OF INTEREST

The scope of this Expression of Interest (EoI) is to assess the needs and vision of the national scientific community and its potential contributions to genomic medicine in Italy through large-scale genomics studies.

Researchers are invited to submit an EoI for accessing the Whole Genome Sequencing (WGS) service offered by the NF for Genomics ([SID:G-001 – Whole Genome Sequencing](#)) for projects of study cohorts of up to 20,000 human samples over two years. WGS analysis (alignment to human reference genome and variant calling of each sample) will be performed by the NF Data Handling and Analysis according to standardized pipelines ([SID: NF62.02.01 – WGS Analysis](#)), and the output raw and processed data (FASTQ, BAM and VCF files) will be returned to the approved applicants.

The submission of an EoI involving multiple centers is encouraged.

EoIs will be considered by the SIEC to inform the drafting of a forthcoming call for Special Projects (26-SP-G-LARGE SCALE), which we are currently aiming to publish in Q3-Q4 2026.

EoIs will not be formally evaluated and do not represent requests for Access to the National Facilities. However, **submission of an EoI is a mandatory prerequisite** for participating in any resulting upcoming call(s) for Special Projects (26-SP-G-LARGE SCALE).

Please note that detailed requirements and evaluation criteria will be described in the resulting upcoming call for Special projects, if and when launched. For information regarding eligibility and NF access rules, please refer to the information available on the dedicated website ([National Facilities - Human Technopole](#)).

4. EXPRESSION OF INTEREST CONTENT AND FORMAT

The EoI, to be submitted through the application portal PICA ([link](#)) shall consist of the following four components:

1. Applicant's CV in NIH biosketch format.

Applicant is the Principal Investigator who submits the EoI on behalf of the entire team of collaborators, if any. Junior Researchers (refer to NF calls for Access [National Facilities - Human Technopole](#) for terms and definitions) are encouraged to participate.

The CV, to be uploaded in PDF in the dedicated section of the portal, shall be drafted in English, using the template available at this [link](#) and following the mandatory format: max 4 pages, page format: A4, Font type: Arial, Font size: 11 or above, Line spacing: single, Margins 2 cm side/ 1.5 bottom. For support in drafting the CV, please refer to NIH website: [Create Biosketches](#). Please note that eRA COMMONS USERNAME is NOT required. Accepted file formats: PDF. Max size: 30 MB - Name the file as APPLICATION ID_CV_Surname (e.g., ID123456_CV_Rossi).

2. List of collaborators (if any).

Collaborators (name, surname, job title, affiliation), shall be listed in the dedicated section of the PICA portal, together with their anticipated role in the project.

3. Expression of Interest.

Eols should clearly describe the proposed project's focus, aims, significance, approach and expected scientific and clinical impact. A strong emphasis should be placed on the translational potential of the research, whether in clinical practice or through mechanistic studies. An Eol should describe how the proposed project will advance population genomics and genomic medicine, deepen understanding of disease biology and/or human physiology, and contribute to broader biomedical and clinical research.

Eols shall be compiled in the dedicated section of the portal. A maximum of 6,000 characters is allowed.

4. Study cohort description, data storage, sharing and analysis.

- a. How many samples would the cohort be composed of?
- b. Would all the samples be already available?
 - If not, how many are available, and how many are to be collected, and when?
- c. Would clinical data be available on all samples (minimum data include year of birth, sex, disease according to standardized clinical vocabulary), year at sample collection, year at clinical assessment)?
 - If not, for how many samples would clinical data available?
- d. What kind of sample-related biological material would be available and accessible (e.g., tissue samples)?
- e. Would you require multi-omics studies on sample-related biological material?
 - If so, what type and how many samples?
- f. Currently, raw genomic data (e.g. FASTQ and BAM files) cannot be permanently stored at HT. Where would you store the raw data generated by the analysis (e.g., in a public repository, locally at the PI's or collaborator's host institution)? Please consider that the total capacity required for the storage of the raw data generated by the analysis of 10,000 samples is indicatively one (1) petabyte.
- g. In the future, should the HT infrastructure allow long-term storage of the raw data, where would you prefer storing them (e.g., in a public repository, centrally at HT, locally at the PI's or collaborator's host institution)?
- h. How would you share data (genomic and clinical data) with collaborators e.g., share individual level raw data (FASTQ or BAM files) from the entire cohort **in a hard drive (or equivalent)**; share raw data (FASTQ or BAM files) from the entire cohort **by allowing access to your storage/analysis system**; share summary level aggregated data (VCF file); other?
- i. In the future, should the HT infrastructure allow long-term storage of the raw data, would you make them available to the wider research community?
- j. Would you perform the analysis for joint variant calling of the entire cohort, and if so, where would you perform this analysis (e.g., centrally at HT if made available in the future, through a distributed/ federated processing approach, locally at the PI's or collaborator's host institution)? The analysis of 10,000

samples typically requires six months of computation time (using 240 CPUs) and 1800 GB of storage for the analysis results.

- k. Where would you perform your downstream analysis, e.g., genomic association studies (e.g., centrally at HT, if made possible, locally at the PI or collaborator's host institution)?
- l. Do you have, or anticipate having, appropriate patient consent for storing and sharing the data generated?
 - o If so, please indicate the scope of such consent.
- m. Overall, what is your vision in terms of data storage, sharing and accessibility for the wider scientific community (e.g., sharing raw or aggregated data, centralized storage at HT, local storage at the PI's or collaborator's host institution) taking into account applicable legal and ethical requirements?

Information regarding study cohort, data analysis and sharing shall be provided in the dedicated section of the portal.

5. EoI SUBMISSION METHODS AND DEADLINE

EoIs shall be submitted by the PI on behalf of the Collaborators(s) (if any) exclusively through the application portal PICA managed by CINECA and accessible at this [link](#), according to the indicated terms and methods.

This request for Expressions of Interest – Large Scale Genomics Projects (ID: 26-EoI-G-LARGE SCALE) will open on the 9th of February 2026 (13:00 CET) and will close on the 20th of April 2026 (13:00 CET).

4. CONTACTS

Requests for information and/or clarifications concerning the procedure for submitting the EoI may be sent to the dedicated e-mail address national.facilities@fht.org, indicating "26-EoI-G-LARGE SCALE" in the subject line.