SISTEMA SANITARIO REGIONALE
AZIENDA OSPEDALIERO-UNIVERSITARIA POLICLINICO UMBERTO I
SAPIENZA UNIVERSITÀ DI ROMA

Sara Amitrano
Biologist specialized in Medical Genetics
Roma, 11.12.2024

Polo GGB has provided top-level services to Universities, Industries and Diagnostic laboratories in genomic, genetic and bioinformatics fields since 2011.

Polo GGB has recently expanded its spectrum of activities by creating three different core facilities

Polo GGB DIVISIONS

Genomics & Bioinformatics Laboratory Authorized for Clinical Diagnosis 1.

Genetics & Ecology Research Center 2.

Immunology Laboratory 3.

BREAKING DOWN THE KNOWLEDGE BARRIERS

SUPPORT THE HEALTH DIAGNOSTIC NEEDS

IMPLEMENTATION OF NEW GENETIC TOOLS

CREATE VALUE FOR SOCIETY

SIENA **1.**

POLO GGB

Authorizations &Certifications

Diagnostic Service Laboratory

The Laboratory for Diagnostics, Genomics and Computational Analysis based in SIENA within the Toscana Life Sciences Incubator combines basic genetics techniques with the most advanced Next Generation Sequencing techniques for human DNA and RNA analysis.

- **Genetics of inherited diseases**
- **Genetics of polygenic/multifactorial diseases**
- **Personalized medicine**
- **Genetic counseling**
- **Oncology**
- **Pharmacogenetics**
- **Reproduction**

SPOKE 8 Clinical Exploitation

""Extensive sequencing for the molecular characterization of tumor samples and liquid biopsies and comparison with in-house techniques"

TITOLO: sUCCESs "Utilità clinica e sostenibilità economica di servizi esternalizzati di Comprehensive Genomic Profiling (CGP) per tumori solidi nella pratica clinica oncologica."

POLO GGB

AIM: corroborate the effectiveness and efficiency of an innovative approach, named extended genomic profiling, as a key enabling technology in the field of precision medicine. The results will provide elements to establish a new personalized clinical practice in oncology.

TIMETABLE 2024-2026:

1. Collection data from patients who cannot take advantage of standard diagnostics (ex: 'MTB Patients': rare tumours, tumours with unexpected fast progressions etc, depletion of any line of treatment which preserve the wellness of the patients; urgency of the investigation; requirement of liquid biopsy in the absence of any other kind of tissue).

2.1 Clinical benefits: establish which biomarkers are not detected with standard methods? Which is the degree of operability (ex: ESCAT)?

2.2 Assembly of the Repository and definition of Artificial Intelligence models.

2.3 Timing comparison of the report assessment vs the standard *in-house* method.

2.4 Assessment of the number of patients for whom liquid biopsy provides treatment guidance.

3 Price comparison between outsourced service vs standard method *in house*.

4 Guidance document that takes into account the cost-benefit emerging in points 2 and 3.

	1 M	2 M	3 M	4 M	5 M	6 M	7 M	8 M	9 M	10 M	11 M	12 M	13 M	14 M	15 M
WP	mag-24	giu-24	lug-24	ago-24	set-24	ott-24	nov-24	dic-24	gen-25	feb-25	mar-25	apr-25	mag-25	giu-25	lug-25
WP 1															
WP 2.1															
WP 2.2															
WP 2.3															
WP 2.4															
WP 3															
WP 4															

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STATE OF THE ART:

- Creation of a case report form (CRF) for the collection of clinical, anamnestic and pharmacogenetic data, useful for the design of machine learning and AI algorithms.
- Centers involved in the collection of samples:
  - Santa Chiara - Oncologia Medica – Pisa
  - Università Studi - Verona
  - IRE-IFO IRCCS Regina Elena – Roma
  - Ospedali Riuniti – Foggia.