

# HEMATOLOGY UNIT

## Clinical activities

The Unit is an advisor, a reference centre and a coordinator for the studies and the treatments of Marrow failures diseases for AIEOP (Italian Association of Pediatric Hemato–Oncology).

This activity is expressed through the provision of the following services to other Italian Centres:

- Formal clinical advice.
- Implementation of the ELA 2 mutation study.
- Implementation of the HAX 1 mutation study.
- Implementation of the TERC and TERT and other telomere biology disease mutation analysis.
- Large NGS panel for bone marrow failure disease, neutropenias (acquired and constitutional), immune cytopenias, immunodysregulations.
- Possibility of WES/WGS analyses within research programs.
- Functional studies aimed to confirm the effect of new variants/ VUS.
- Haematopoietic progenitor cell cultures.
- Performance of neutrophil antibody detection by indirect and methodical tests and cytofluorimetric systems.
- Execution in Italy of complementation analysis with retroviral vectors for Fanconi Anaemia.

Every year about 150 new patients are supervised and treated, whereas 420 overall patients with Hematologic diagnoses (acquired aplastic anemia, leukaemia, neutropenia, Fanconi Anaemia, Blackfan-Diamond anaemia, thrombocytopenia and other constitutional marrow failure diseases, immune cytopenias, thalassemia, sickle cell disease) are in charge.

Experimental Clinical Trials on new drugs in Leukemias and Sickle cell are conducted in the Unit.

Nation Registry of Fanconi Anemia, Neutropenia and ALPS have seat in the Unit.



## Director

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