OPENING HOURS AND ACCESS

The service activity takes place from Monday to Friday:

- From 8 a.m. to 3.30 p.m.: therapy;
- From 8 a.m. to 9.30 a.m.: blood sampling (for patients undergoing blood transfusion);
- From 8.30 a.m. to 5.30 p.m.: outpatient visits.

Secretariat hours are:

- Mondays, Wednesdays and Thursdays: from 8.30 a.m. to 5.30 p.m;
- Tuesday and Friday: from 8.30 a.m. to 3 p.m.

Users can access the Centre only by appointment following a prescription for "First internal medicine visit at Rare Diseases Centre" (DM 89.7).

Access to the outpatient clinic is possible by two different methods:

- CUP or regional toll-free number or Lombardy Region portal exclusively for first visits;
- direct management of bookings by the Centre (in attendance during secretarial hours, by telephone at 0392339555 or electronically: malattie.rare@irccs-sangerardo.it).

PRATICAL TIPS

- Please bring with you all the clinical documentation, a list of your current therapies and your prescriptions.
- If you cannot show up for the appointment, please inform us in advance so that we can book another appointment.
- At the end of the examination, the following appointment will be arranged directly with our Secretariat.
- If you need a certificate of attendance for work purposes, you can request it at the end of your appointment.

WHAT'S YOUR OPINION?

We invite you to fill in the satisfaction questionnaire using the following QR code.

We would like to inform you that our activity is supported by the Association for the Study of Hemochromatosis and Iron Overload Diseases ETS: www.emocromatosi.it

A volunteer of the Association is present on Thursdays from 9 a.m. to 1 p.m.

Sistema Socio Sanitario

Fondazione IRCCS San Gerardo dei Tintori

Regione Lombardia

SSD RARE DISEASES

4° Floor

Palazzina Accoglienza

Medical Area

Director: Dr.ssa Raffaella Mariani

Medical Doctors: Dr.ssa Mara Botti
Dr.ssa Anna Commone

Nursing Coordinator: Laura Bonfanti

Nurses: Laura Cantoreggi, Antonella Lainà, Alice Ripamonti

CONTACTS

Secretariat: 039 233 9555
Mon-Wed-Thu: 1.30-4.30 p.m.
Tue: 10 a.m.-12 p.m.

Email @: malattie.rare@irccs-sangerardo.it

Mod. n°4052 MRare-FE-OO1 Rev.0 in inglese

TREATED DISEASES

Iron related disorders:

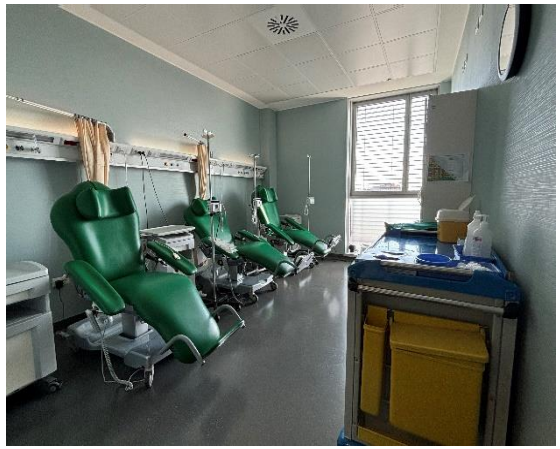
- Hemochromatosis type 1, 2a and 2b, 3 and 4;
- Ferroportin deficiency;
- Aceruloplasminemia;
- Hereditary hypo-transferrinemia;
- Hereditary hyperferritinemia-cataract;
- Hereditary benign hyperferritinemia;
- Secondary iron overload (chronic liver disease, post-transfusion, porphyria cutanea tarda, exposure to welding fumes);
- Dysmetabolic hyperferritinemia;
- Iron Refractory Iron Deficiency Anaemia (IRIDA) and other complex iron deficiency anaemias.

Hereditary Anaemias:

- Transfusion- and non-transfusion-dependent thalassaemia;
- Sickle cell disease (SS, SThal, SC);
- Congenital dyserythropoietic anaemia;
- Blackfan-Diamond anaemia;
- Congenital and acquired sideroblastic anaemia.

Hereditary metabolic disorders:

- Disorders of Carbohydrate metabolism (Glycogenosis, Galactosaemia, Fructose intolerance, Fructose-1,6-bisphosphatase deficiency), Hyperinsulinism Hyperammonemia);
- Disorders of Mitochondrial Fatty Acids oxidation and Energy metabolism (Carnitine deficiency (CUD), Multiple Acyl carnitine CoA dehydrogenase deficiency (MADD), Carnitine acyl carnitine translocase (CACT), Acyl carnitine palmitoyl transferase (CPT2) deficiency, Pyruvate dehydrogenase deficiency, Very long chain acyl-CoA dehydrogenase deficiency (VLCAD));
- Disorders of amino acid metabolism and transport (Tyrosinaemia, Argininosuccinic aciduria, 3-Methyl crotonyl glycinuria (3MCC), Alkaptonuria, Citrullinaemia, Hyperargininaemia, MSUD, Lysinuric protein intolerance (IPL) Methylmalonic aciduria with homocystinuria type cblC, Methylmalonic acidemia, propionic acidemia, Ornithine transcarbamoylase deficiency (OTC), Glutaric aciduria type 1 and 2, Gyrate Atrophy of the choroid, Homocystinuria, Hyperhomocysteinemia, HHH syndrome);
- Disorders of nucleic acids (xanthine oxidase deficiency, Lesch Nyhan syndrome);
- Lysosomal, peroxisomal and Golgi system disorders (Mucopolysaccharidosis, Niemann Pick type B, Mucolipidosis 3, Gangliosidosis, Mannosidosis, Sialidosis, Protein Glycosylation Disorder, Gaucher type III with epilepsy), vitamin-responsive disorders (Biotinidase deficiency, severe MTHFR deficiency);
- Disorders of Lipid metabolism (Abetalipoproteinemia,Hypoabetalipoproteinemia), Neurotransmitter and small peptide disorders pyridoxine-sensitive epilepsy.



SERVICES

Clinical activities mainly involve outpatients with the provision of the following services:

- first visits;
- check-ups;
- providing medical certifications for rare diseases and treatment plans for diseases of interest;
- genetic testing related to the disease;
- patient education on therapies;
- blood transfusions in patients suffering from hereditary anemia;
- drug infusions (including innovative drugs);
- venesections for diseases secondary to iron metabolism disorders;
- diagnostic and therapeutic Complex Outpatient Macro Activities (MAC).

EuroBloodNet

European Reference Network | MetabERN
European Reference Network for Hereditary Metabolic Disorders

The SSD is part of the **regional network for rare diseases** of the Lombardy region and is part of the **European Reference Networks** (ERNs) for **non-oncological haematological diseases** (**EuroBloodNet**) and for **hereditary metabolic diseases** (**MetabERN**).