MEDICAL GENETICS

Lotto 3, piano 1

Director

Alessandra Renieri

Medical Doctors

Caterina Lo Rizzo, Anna Maria Pinto, Margherita Baldassarri

Biologists

Francesca Ariani, Mirella Bruttini, Chiara Fallerini, Roberta Mancini, Rossella Tita, Ilaria Meloni

Research Fellows/ RDTA/ Scholarship Holders

Susanna Croci, Sergio Daga, Andrea Degl'Innocenti

Biologists in Specialist Training

Giuseppe Castello, Mirjiam Lista, Loredaria Adamo, Enrica Antolini, Jasmine Covarelli, Deborah Maffeo, Elena Pasquinelli, Simona Basso, Giulia Rollo

Medical doctors in Specialist Training

Anna Carrer, Lorenzo Loberti, Pietro Ilardi

PhD Students

Simona Innamorato, Flavia Di Re, Martina Rozza, Gabriele Martelloni, Giulia Brunelli, Katia Mokabari, Sara Pastore, Omaima Belakhdar, Heynan Museynli

Nurse (Genetic Counsellors)

Valeria Luppoli

Administrative Staff

Lorella Benocci

Technicians

Elisabetta Casarotti, Olga Lorenza Colavecchio, Maria Nocerino

Il Servizio di consulenza è attivo dal lunedì al venerdì dalle ore 9 alle18 .
La prenotazione può essere effettuata tramite CUP: 0577 767676

Azienda ospedaliero-universitaria Senese

MEDICAL GENETICS

Servizio clinico di Genetica Medica

Laboratorio di Genetica Medica

Ricerca e sviluppo in Genetica Medica Biobanca Genetica

SST Servizio Sanitario della Toscana

European Reference Network
for rare or low prevalence complex diseases

Clinical service of Medical Genetics

The Clinical Service of Medical Genetics operates to provide the diagnostic framework for subjects affected by genetic diseases and to inform and define the reproductive risk of couples belonging to families with genetically determined pathologies, making use of genetic tests, including prenatal tests, where possible. The meaning, limitations, reliability and specificity of the test are clarified during genetic counseling.
During the counseling session, the user, or his guardian if a minor, is informed of the procedures that are necessary for carrying out the required tests, of the possibility of archiving the biological material at the end of the test and of the possible use even for research purposes.

Information is provided on the methods of processing personal data and on the right to privacy, for which consent must be provided which can be revoked at any time.

During the consultancy, information relating to the prognosis, treatment and prevention of the pathology in question is communicated to the interested party. For specific genetic diseases, reference centers are indicated and certificates are issued for requesting specific exemption codes. At the end of the genetic counseling meeting or, when carried out, at the time of conclusion of the genetic test, the user will be provided with a written report, which contains personal and family anamnestic data, any tests carried out with the relative outcome and the considerations connected to this outcome.

The service provides different types of genetic counseling: preconceptional, prenatal, postnatal, dysmorphological, oncological and presymptomatic. As part of the different types of consultancies, several meetings may be envisaged, including pre-test and post-test evaluations, controls and follow-up visits in subjects with specific pathologies and for some types of consultancy other professionals may also be present, such as the psychologist, the oncologist and the surgeon. Genetic oncological consultations can be booked by external users via CUR. The waiting time for the consultation appointment is between a few days, as in the case of urgent services, such as prenatal consultations, and one month.

Genetic tests, due to their peculiarity and the implications concerning the biological identity of the person and the family, must be carried out in authorized and/or accredited Medical Genetics facilities according to the following provisions (resolutions n. 145/2004 and n. 24 /2006 of the Regional Health Council and Resolution No. 887/2006 of the Tuscany Region Council).

Medical Genetics Laboratory

The Medical Genetics laboratory is divided into: pre- and post-natal Molecular Genetics laboratory and pre- and post-natal cytogenetics laboratory. The waiting times for the genetic tests indicated below vary from one to twelve months. The reports will be provided to the user as an attachment to the consultation responses.

1. Adult disease

- Alkaptonuria
- Hereditary aneurysms
- Hereditary arrhythmias
- Charcot-Marie-Tooth type I A (CMT I A)
- Keratoconus
- Huntington's chorea
- Arrhythmogenic right ventricular dysplasia
- Facioscapulohumeral muscular dystrophy
- Oculopharyngeal dystrophy
- Familial pulmonary fibrosis
- Glaucoma
- Ichthyosis due to steroid sulfatase deficiency, X-linked
- Lymphoedema
- Arteriovenous malformations (AVM)
- Peripheral neuropathies
- Parkinson's
- Polyneuropathy with pressure paralysis
- Alport syndrome, autosomal recessive and dominant
- Alport syndrome, X-linked
- X-linked Kallman syndrome

2. Pediatric diseases

- Autism
- Intellectual disabilities and microcephaly (array CGH/Exome analysis)
- Mitochondrial encephalopathies
- Leucoencephalopathies
- Cohen Syndrome
- Lesch Nyhan Syndrome
- Rett Syndrome
- Nicolaides-Baraitser and Coffin-Siris syndromes
- Prader-Willi/Angelmann Syndrome
- Retinoschisis
- Intellectual Disabilities (array CGH/Exomes)

3. Tumoral Diseases

- Liquid Biopsy (77 cancer driver genes)
- Hereditary Breast and Ovarian Cancer
- Gastric cancer
- Medullary carcinoma of the thyroid
- Familial adenomatous polyposis
- Retinoblastoma
- Li Fraumeni Syndrome
- Lynch Syndrome
- WAGR Syndrome
- Gastrointestinal stromal cancers (GIST)

The cytogenetic investigation can be performed in pregnancy and allows the study of the fetal karyotype on chorionic villi or amniotic fluid. The karyotype allows you to have an answer after 21 days. A rapid Molecular Genetics test has recently been developed, which allows for a response to be obtained in 48-72 hours, the QF-PCR. Prenatal analysis is offered to mothers with an increased risk of chromosomal pathology, parents with a previous
child affected by chromosomal anomaly or diagnosable genetic disease, parents with structural rearrangement or genetic disease, eco-evidenced fetal pathology, positive screening test. The postnatal cytogenetic investigation is offered to subjects with suspected chromosomal syndrome or genetic disease; parents and family members of subjects with chromosomal abnormalities; subjects with congenital defects, intellectual disability or growth retardation; couples with a child with suspected chromosomal syndrome; couples with multiple abortions; infertile subjects. The waiting time is one month.