



## **SERVICE CHARTER**

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### **R&I GENETICS srl.**

(previously RESEARCH & INNOVATION)

Genetic Diagnostic Laboratory

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## **PREMISES**

The purpose of this Service Charter is to help citizens learn about R&I Genetics and, in particular:

- to illustrate, in brief, the activities and services provided
- to illustrate the driving principles and values that guide and inspire the daily work
- to illustrate the underlying organization and activities as well as duties and conduct of the staff
- to make the services easy to access.



## **R&I GENETICS**

**R&I GENETICS**, a limited liability company (srl), is a certified clinical diagnostic laboratory that specializes in genetic testing, particularly in the area of rare diseases.

The laboratory, accredited within the Italian National Health Service (Servizio Sanitario Nazionale),

provides comprehensive diagnostic genetic services ranging from single gene sequencing to whole exome or genome analysis for specialist diagnosis of patients with a suspected genetic disorder causative of a rare constitutive disease in different medical fields (e.g. neurology, ophthalmology, pediatrics etc.)

To this end, the laboratory also pursues research activities, primarily focused on improving current test methodologies and introducing new genetic tests within the laboratory, with a constant focus on the clinical needs and technical-scientific advancements in the field

### The Laboratory and its Cutting-edge Technologies

R&I GENETICS is located in Padova in the 8<sup>th</sup> floor of the prestigious Pediatric Research Institute (IRP).

The workspace, consisting of approximately 500 square meters, comprises fully equipped, state of the art, molecular diagnostic laboratories (wet and dry), in addition to research and office spaces.

Available facilities, ensuring the most advanced diagnostic technologies, include high-throughput next-generation sequencing (NGS) technologies for exome and genome analysis, new cutting-edge bioinformatic computing techniques and unique, original, software programs and databases for data interpretation.

### Staff and Professionalism

With over 20 years' experience in molecular research and diagnostics, the staff at R&I Genetics, has been at the forefront of genetic innovation, pioneering new technologies (e.g. microarray, NGS sequencing) into application for the diagnosis of genetic diseases.

A characteristic of R&I is its high degree of staff specialization, continuous updating and professional experience. This is, in addition to hard work and passion, promoted and

supported through internal meetings and discussion, participation in international scientific congresses/ conferences/workshops and refresher courses.

## **Mission and Values**

R&I GENETICS seeks to achieve excellence in the area of advanced diagnostic genetic testing in rare diseases so as to best support healthcare professionals in advancing precision medicine in genetically determined conditions in a timely and appropriate manner, with continuous attention devoted to the needs of the patients and quality of the services delivered.

The values and principles which inspire and guide R&I include:

- Clinical needs and Patient focus
- Continuous quality improvement
- Service excellence
- Multidisciplinary versatile approach to genetic testing
- Applied research development and continuous up-date of current methodologies

### *Founding Principles and User/Patient Rights*

In perusal of the above, R&I GENETICS is committed to ensuring respect of the following *founding principles and patient rights*:

- *Equality*: all citizens/users are entitled to the most appropriate available services regardless of age, gender, ethnic origin, religious beliefs, nationality, culture, social status or political views;
- *IMPARTIALITY*: staff conduct towards all citizens/users is guided by criteria of objectivity, fairness, justice, impartiality and empathy;
- *CONTINUITY*: Each citizen/user is entitled to appropriate and continuous healthcare services, using the most advanced scientific knowledge along with the best technologies. In the event of malfunction or interruption of a service, measures are taken to provide users with as little discomfort as possible;
- *RIGHT TO CHOOSE*: each citizen/user has the right to choose their healthcare provider that best matches their needs. They are also free to request for second professional opinions or present suggestions/complaints without the fear of compromising the quality of the services provided;
- *INFORMATION AND PARTICIPATION*: consensus in the provision of a service must be guaranteed in line with current legislation through accurate, clear and complete

information. Users have the right to access information regarding the services provided and put forward suggestions or complaints on how to improve their quality.

- *QUALITY*: R&I GENETICS is committed to ensuring high quality services for its users/patients.

It does this by adopting appropriate measures to guarantee their continuity, efficiency

and effectiveness., in line with the latest national and international guidelines and up-to-date standards.

To this end, R&I GENETICS has implemented a QUALITY SYSTEM (UNI EN ISO 9001:2015) that establishes the standards and sets out the rules for verification of compliance by means of internal and external inspections regarding the quality of the services (genetic tests) delivered in accordance with the current ITALIAN legislation (D.L. 502/92 art. 12, comma 2, lett. a) and national (e.g. SIGU, Societa' Italiana di Genetica Medica,) and international (e.g. ACMG, American College of Medical Genetics) guidelines in the field.

- *EFFICIENCY AND EFFECTIVENESS*: the services are delivered in such a way as to ensure efficiency and effectiveness, employing the best optimal resources and modalities available to satisfy the healthcare need requested. The efficiency of the services also includes the safeguarding of resources, which are employed without wastage or unnecessary costs.

## **GENETIC TESTING SERVICES**

Research over the last decades has resulted in the identification of genes responsible for over 50% of the estimated 7,000 rare monogenic diseases. This achievement is the result of dramatic improvements in DNA-testing methodologies, particularly next generation sequencing technologies and the associated analyses. As this has expanded, so have the types and numbers of genetic tests increased in the last years

Using molecular genetic diagnostics, R&I Genetics support the referring physician in making a diagnosis, and in many cases, can also make recommendations for treatments and therapies. We can also provide information on disease progression and prognosis. Family members may also receive a risk assessment on the basis of heredity.

### *Information and Consent*

Given the peculiarity of genetic testing not only for the patient but also his/her family members, obtainment of informed consent is obligatory, following receipt of appropriate information about the risks, benefits, efficacy, and alternatives to the testing. This usually occurs upon consultation with a geneticist or other medical specialist in a pre-testing setting.

For R&I GENETICS, receipt of informed consent is mandatory for conduction of a genetic test. In addition, R&I GENETICS accepts referrals for genetic testing for clinical purposes within the Italian National Health Service solely from a clinical geneticist or other medical specialist operating within the healthcare system.

### *Privacy Issues*

R&I GENETICS takes great precaution to protect patient's privacy very seriously.

Special measures are taken in place to anonymous samples and mandate that all staff members maintain the confidentiality of patients' personal health information, which includes both medical information and individually identifiable information about any patient, such as address, phone number or Social Security number that they may come in contact with that could reveal the person's identity. Data security measures and protocols have been implemented so that the personal and sensitive details of the patients concerned is processed in an appropriate manner in accordance with current legislation in the field.

Genetic test results are solely provided to the requesting medical specialist authorized by the patient.

Otherwise, R&I GENETICS prohibits the unauthorized disclosure of any health or sensitive information except as required by law.

Patients have the possibility to exercise their rights in accordance with the current Italian legislation (GDPR 2016/679, art. 15-22)

### Genetic tests: types and characteristics

The arrays of DNA based tests currently available at R&I GENETICS for diagnosis of human genetic diseases include:

- Targeted mutation analysis:

These types of test search for a specific DNA variant in one gene. The selected variant is known to cause a disorder. It is often used to test family members of someone who is known to have a particular mutation, to determine whether the variant is inherited (family segregation studies) or whether other family members are affected.

- Single gene sequencing:

These tests look for any genetic changes in one gene. These tests are typically used to confirm (or rule out) a specific diagnosis, particularly when there are many variants in the gene that can cause the suspected condition.

- Gene Panels

These tests are designed to search for disease causing mutations (nucleotide and copy number variations) employing next generation sequencing technologies in any one of a number of genes that are known to be mutated in a specific type of disease. At difference with whole exome or genome sequencing (see below), gene panels are specifically used for the molecular diagnosis of diseases for which a small number of genes are causative of the pathological phenotype (i.e: hypercholesterolemia, Stargardt disease, neurofibromatosis, cystic fibrosis, etc...)

- Whole exome /whole genome sequencing and their analysis.

These tests sequence the bulk of an individual's DNA to find genetic variations employing next generation sequencing technologies. While exome sequencing involves sequencing of all the coding DNA regions, genome sequencing analyzes also the non-coding regions in one's genome.

Generally, exome sequencing is currently employed and targeted analysis is carried out, in relation to the clinical phenotype. In these cases, the whole exome is sequenced and analysis initially focuses on a subset of genes (virtual panel) potentially causative of a particular condition. This approach is particularly useful in establishing a molecular diagnosis in many genetic disorders characterized by clinical, genetic and phenotypical variability (see below)

- *comparative genomic hybridization (CGH) at high resolution (400 x)*

This test is a molecular cytogenetic method for analyzing gains or losses in DNA that are not detectable with routine chromosome analysis. CGH can detect small deletions and duplications, but not structural chromosomal changes such as balanced reciprocal translocations or inversions or changes in chromosomal copy number.

- Testing for repeat expansions

These genetic variations, not easily detectable, are highly important in human disease, particularly in the field of neurology. Because of this, R&I GENETICS has developed a number of different tests specific for different disorders. This includes testing for repeats in suspected C9orf72-related disorders, myotonic dystrophy 1, FRAXA, etc. Others are in progress.

It is estimated that the number of rare diseases due to gene defect exceeds 7,000. Many families with children with rare diseases experience the diagnostic odyssey that involves multiple specialist visits, laboratory and imaging studies and in some cases surgical procedures. Despite extensive testing in many cases diagnoses are not made or solely suspected. In some cases, establishing a correct diagnosis was complicated by unusual clinical presentation. In others, genetic heterogeneity of the disorder where defects in a number of different genes can give rise to the same clinical or syndromic manifestations, complicates reaching a diagnosis. At times, defects in a specific gene may lead to presence of different subsets of clinical manifestations in a specific patient. Also, for many genetic disorders the most characteristic features of that disorder are not present in young infants. Furthermore, defects in a specific gene may lead to presence of different subsets of clinical manifestations in a specific patient.

Genetic Tests: availability in different medical specialities

Medical Specialty	Gene Panel	Includes
Ophthalmology	Non-syndromic Retinal Diseases	retinitis pigmentosa, Stargardt diseases, vitreoretinal diseases, macular dystrophies, congenital night blindness, cone rod dystrophies, fundus albipunctatus
	Usher syndrome	
	Albinism	
	Waardenburg syndrome	
	Optic Atrophy	Mitochondrial mutations
	Stickler Syndrome	
	Piebaldism	
	Congenital Cataract	
	Coloboma	
	Congenital Glaucoma	
	Congenital Nystagmus	
	Microphthalmia and Anophtalmia	
	Anterior segment dysgenesis	
Neurology	Neurodevelopmental disorders	Syndromic and non-syndromic, autism

	Brain Malformations	
	Microcephaly	
	Macrocephaly	
	Craniosynostosis	
	Epilepsy	Syndromic and non-syndromic, developmental epilepsy, epileptic encephalopathy, focal epilepsy, neonatal benign epilepsy, temporal epilepsy, febrile epilepsy
	Motor-neuron diseases	ALS, spastic paraplegia, distal motor neuropathies, C9orf72 repeat expansion
	Neuropathies	Axonal, demyelinating, distal motor, autonomic
	Myopathies	Dystrophic, congenital, metabolic and mitochondrial limb-girdle
	Ataxia	
	Dystonia	DMPK repeat expansion
	Small Vessel Cerebral disease	Stroke, hemiplegic migraine
	Alzheimer	
	Parkinsonism	
	Dementia	C9orf72 repeat expansion
	Ciliopathies	Joubert syndrome, Meckel syndrome, Bardet-Biedl syndrome
	Leukoencephalopathy	
	Non syndromic hearing loss	Neurosensorial and conductive

Cardiology	Arrhythmias	Brugada syndrome, atrio-ventricular block, atrial fibrillation, LVNC, long and short QT
	Cardiomyopathies	Hypertrophic, dilatative, restrictive, LVNC
	Congenital heart defects	
	Rasopathies	Noonan disease, neurofibromatosis
Endocrinology - Metabolism	MODY	
	Diabetes	Neonatal diabetes, infancy diabetes, insulin resistance, hyperinsulinism
	Glycogen diseases	
	Glycosylation diseases	
	Hypercholesterolemia	
	Mitochondrial diseases	Nuclear genes affecting mitochondrial metabolism
	Hypogonadism	
	Disorders of sex differentiation	
Dermatology	Epidermolysis Bullosa	
	Congenital Ichthyosis	
	Peeling skin syndrome	
	Palmoplantar keratosis	
	Rasopathies	Noonan disease, neurofibromatosis
	Monilethrix	
Nephrology	Nephronophthisis	
	Autosomal Dominant Polycystic Renal disease	PKD1
	Polycystic hepato-renal	

	disease	
	Nephrotic syndrome	
	Tubulo-interstitial diseases	
	Tubulopathies	
	Proteinuria	Nephrotic syndrome, Alport Syndrome, syndromic syndromes
	Alport Syndrome	
	Uremic hemolytic anemia	
Gastroenterology	Cholestasis	
	Metabolic liver diseases	
	Glycogenosis	
	Alagille syndrome	
	Liver fibrosis	
	Polycystic hepato-renal disease	
	Pancreatitis	CFTR
	Very early onset inflammatory bowel disease	
	Congenital diarrhea	
Skeletal	Congenital Short Stature	
	Skeletal Malformations	
	Jeune disease	
	Craniosynostosis	
	Ospetopetrosis	
	Osteogenesis imperfecta	
	Arthrogryposis	
Immunology	Autoinflammation and Autoimmunity	Periodic fevers
	Immunodeficit	SCID and CVID
	Hypogammaglobulinemia	
	Deficit of the complement	

	system	
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### Test Reports

According to the Italian law and the recommendations of the Italian society of human genetics (SIGU), genetic reports are delivered to the requesting physician.

The report contain:

- Patient informations
- Genetic results summary
- Detailed information concerning the genetic variations found in the patient
- Comments and interpretation in which we summarize the current state of scientific literature for the variants found and discuss them in relation to the patient.
- Additional information: at the end of the report, we present technical information. This includes information such as technology used, limits and bioinformatic parameters.

Reports are available within 8-12 weeks after receiving the sample for multigene panels constituted of less than 200 genes, for genetic tests requiring > 200 genes referral times are 12-20 weeks.

### **COMPLAINTS**

If you have a concern or complaint about our work, we want to hear from you. We continuously try to improve the work we do and we value any feedback that will help us to do this. We take all feedback seriously and we will take action when appropriate to do so. All complaints will be dealt with in a timely and professional manner.

If you would like to formally raise a concern or complaint please write to:

R&I Genetics Srl

C.so Stati Uniti 4, int. F

35127 Padova (IT)

or e-mail us:

[info@rigenetics.com](mailto:info@rigenetics.com)

[genmed@rigenetics.com](mailto:genmed@rigenetics.com)

Including the following details will help us to effectively and quickly investigate your complaint:

- Your name and contact details: this is essential as we will not investigate anonymous complaints.
- The specific area, service or resource to which the complaint applies.

## **COMMITMENTS AND PROGRAMS**

Striving for excellence

Continuous improvement of the quality of services is one of the main objectives of R&I GENETICS.

To achieve and maintain its standards of excellence, at R&I GENETICS has developed an improvement process based on a rigorous quality plan modelled on some of the top national and international benchmarks (latest ISO standards, European guidelines etc.) now extended to all activities

Users/Patients may also actively contribute to our improvement process, by submitting suggestions (or reporting malfunctions), or participating in our patient satisfaction surveys.

## **ACCESS TO THE SERVICES**

According to the Italian law, genetic test can be requested only by medical specialists and genetic counsellor.

Blood or DNA samples must be accompanied by the compiled and signed consent form and request form available at [www.rigenetics.online](http://www.rigenetics.online).

Administrative and payment information are available upon request (see contact information)

## **CONTACTS**

R&I Genetics laboratories are in Padova, Corso Stati Uniti 4, int.F.

Opening hours: Monday to Friday from 8:30 a.m. to 5:30 p.m

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