SERVICE CHARTER

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of

R&I Genetics srl

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ORGANISATION, PRINCIPLES AND PROGRAMMES

Through this Service Charter, R&I Genetics addresses all interested parties to present its principles, programmes and the services it undertakes to guarantee.

Through this Service Charter, R&I Genetics aims to provide those who request its services with an instrument for participation and for the protection of their rights.

The Service Charter describes the organisation, programmes and commitments of R&I Genetics as a whole.

Further information is available on the company website www.rigenetics.com, from which it is also possible to download the documents relating to the analyses and, for physicians, to access the portal for completing requests and consulting reports.

FUNDAMENTAL PRINCIPLES

R&I Genetics recognises the right to health, as set out in the Italian Constitution, and the right of citizens to the care and services provided by its facility, and undertakes to guarantee them, in accordance with the regulations in force, on the basis of the following fundamental principles:

Equality
The services of R&I Genetics are provided to every citizen/user according to the same rules for all, without discrimination on grounds of age, sex, language, religion, geographical origin, social status, political opinions or health condition.

Impartiality
Staff act objectively and fairly towards every citizen/user, who is assisted with care, courtesy and attention, with respect for the person and their dignity.

Continuity
Services are provided continuously and without interruption (except during major public holidays), in accordance with national and regional provisions or company regulations.

Freedom of choice
Where the law allows, users may choose, among the providers of the testing service, the one they consider best able to meet their needs, within the limits set for organisational reasons.

Citizen participation and information
Citizens have the right to obtain the information that concerns them and that enables them to give their consent properly; they have the right to make suggestions and observations, contributing to the improvement of the Service, and to submit complaints whenever they consider it necessary.

Quality of services
R&I Genetics is accredited to UNI EN ISO 15189, operates a Quality Management System in accordance with UNI EN ISO 9001:2015 and systematically participates in external quality assessment (EQA) schemes specific to genetic testing.

In order to verify the efficiency of the laboratory and maintain a high level of quality in service delivery, R&I Genetics takes part in technical and regulatory update meetings promoted by the relevant scientific

societies, such as the Italian Society of Human Genetics (SIGU) and the Italian Society of Clinical Biochemistry and Clinical Molecular Biology (SIBioC), as well as in specialist conferences, where it also presents, as a speaker, the results obtained in collaboration with clinical centres.

R&I Genetics also provides a complaints procedure, so that users can report any shortcomings and R&I Genetics can continuously improve its standards.

Effectiveness and efficiency
Services are provided through the optimal use of resources, in accordance with the most up-to-date quality standards, adopting all appropriate measures to meet the needs of citizens/users as quickly as possible and avoiding waste that would be to the detriment of the community.

RIGHTS

R&I Genetics encourages citizens/users to participate and collaborate in the process of improving the services offered and to put forward new proposals and initiatives, and undertakes to recognise the Rights set out below.

Right to assistance
Citizens/users have the right to appropriate assistance, care and prevention, provided with care and attention, with full respect for human dignity and without any discrimination on grounds of racial or ethnic origin, provenance, culture, religious beliefs, sex, or social and economic status.

Right to recognition
Citizens/users have the right to be addressed by their first name and surname and to be protected in the exercise of their health rights and of the rights provided for by the laws in force. They also have the right to be able to identify immediately the staff with whom they are dealing.

Right to information and education
Citizens/users have the right to obtain complete and understandable information about their state of health and to receive the information that enables them to give genuinely informed consent before undergoing the diagnostic test. This information also covers the possible risks or discomfort arising from the results.

Right to privacy
R&I Genetics ensures that personal data are processed with full respect for the rights and dignity of the data subject, with particular regard to confidentiality, personal identity and the right to the protection of special categories of data, including genetic data, in accordance with Regulation (EU) 2016/679 (GDPR).

Right to quality
Citizens/users have the right to fair access to waiting lists and to know the criteria that govern them, as well as, for the services they request, to know the related fees and the booking and delivery arrangements.

Right to transparency
Every citizen/user has the right to receive clear and transparent information at every stage of their relationship with the provider of the service.

INFORMATION AND CONSENT

Every diagnostic procedure proposed to a patient is preceded by adequate information, provided by specifically trained staff, followed by the patient's agreement and consent.

For genetic tests, given the nature of the results and their possible implications for family members, the patient is given in advance all information on the type of investigation, its purpose and limitations, the possible results and the implications for the family.

Informed consent is obtained in writing, after the patient has been given all relevant information. For minors, consent is given by the holders of parental responsibility.

PRIVACY

R&I Genetics undertakes to ensure respect for the rights and dignity of the citizens/users who come into contact with its various professionals.

Staff are required to respect patients' privacy, and the necessary measures are implemented to prevent the disclosure of users' sensitive or personal data (for example, the use of an internal code).

Information concerning health status is provided only to the requesting physician and to the persons designated by the patient on a specific form.

Before any information is collected, users are informed of how the requested data will be used and of how they will be collected and stored.

Personal data, including genetic data, collected in paper and digital form are processed with the necessary security and confidentiality measures. With regard to such data, citizens may exercise the rights provided for by Articles 15–22 of Regulation (EU) 2016/679.

R&I GENETICS AND ITS ORGANISATIONAL STRUCTURE

R&I Genetics S.r.l. (R&I Genetics) is a clinical testing laboratory based in Padua, Corso Stati Uniti 4 int. F, which since the start of its activities (2008) has been dedicated to genetic testing for diagnostic purposes in hereditary diseases, both rare and common.

Since 2015, R&I Genetics has been accredited by the Veneto Region as a provider for the Italian National Health Service (SSN), under which it operates by agreement.

The premises cover a total area of approximately 650 m² on the fifth and eighth floors of the Torre della Ricerca, home of the Istituto di Ricerca Pediatrica Città della Speranza (Pediatric Research Institute).

The staff, currently made up of 30 professionals, most of whom have more than ten years' experience in genetic testing, work in three distinct areas:

laboratory area with advanced genetics and genomics platforms (e.g. high-resolution array-CGH, Sanger and NGS sequencers), including the bioinformatics infrastructure for the analysis of exome and genome data;

research area equipped for in vitro experimentation;

office area, which also includes a waiting room and the in-house sample collection room, used only for SSN-covered activity.

Since May 2022, R&I Genetics has also had an external sample collection point at the Poliambulatorio Legnaro for activity not covered by the SSN.

Since March 2025, the Technical Director of R&I Genetics has been Dr Alice Bruson, specialist in Medical Genetics.

ACTIVITIES

Molecular diagnostics

For more than fifteen years, R&I Genetics has operated in the field of genomic diagnostics, providing a nationwide genetic testing service that keeps pace with available technological advances.

This includes classical genetic tests, such as targeted variant analysis and single-gene sequencing. In 2015 R&I Genetics was among the first laboratories in Italy to make diagnostic solutions based on next-generation sequencing (NGS) available to specialist physicians for the diagnosis of genetic diseases, rare diseases in particular. Today, three analyses out of four are based on **whole exome sequencing (WES), with phenotype-driven analysis of all genes currently associated with the suspected conditions**: the analysis can be extended to other genes from the same data, without a new sample or new sequencing. In the most complex cases, reverse phenotyping is used, reassessing the clinical picture together with the physician in the light of the variants identified.

Research

Research has always been of primary importance in our activities. Several research projects are currently under way, aimed mainly at implementing innovative methodologies. These are targeted translational research projects in the field of genomic applications in healthcare, based on advanced technological and bioinformatics platforms.

Today, within its Research activities, R&I Genetics also offers whole genome analysis, the latest frontier of human genetics.

TYPES AND FEATURES OF THE SERVICES

R&I Genetics is a laboratory dedicated exclusively to the diagnosis of hereditary diseases, both rare and common. Our analyses, available throughout Italy, range from single-gene and targeted panel analysis to exome and genome sequencing, and include methods for the study of copy number variations (array-CGH, MLPA, Real-Time PCR) and of repeat expansions (TP-PCR).

R&I Genetics provides genetic testing services to specialist physicians working within the National Health Service and at major regional and national hospital centres.

The core values of R&I Genetics are high quality and continuous innovation, made possible by full and constant collaboration with treating clinicians.

In this perspective, we are available for pre-test discussion of prescriptions, so that they are not only appropriate but also have the greatest likelihood of success. We also stress the importance of a concise description of the proband's clinical picture, which is essential to guide the interpretation of the exome and genome.

Interaction and dialogue with the clinical staff requesting the analysis continue throughout the interpretation phase and, after the report is issued, through post-test support for the interpretation of the results.

The available analyses cover 14 clinical areas, the most representative of which are:

- Paediatrics (rare diseases in children)
- Neurology
- Ophthalmology
- Oncology
- Cardiology
- Nephrology

The full list of tests (see www.rigenetics.com), the methods used and the equipment employed are periodically and constantly updated in line with the requirements of continuous improvement.

ACCESS TO TESTING

Analyses provided under the SSN agreement for outpatients must be requested by specialist physicians working in public healthcare facilities.

Test requests for inpatients must be approved by the Medical Directorate of the requesting facility.

Prescriptions must be accompanied by the request form, the test-specific informed consent and the consent to the processing of personal data, signed by the patient or by the holders of parental responsibility; for outpatients, also by the SSN prescription forms (impegnative). The forms of the requesting facility or those downloadable from www.rigenetics.com may be used.

The request can also be completed online with SmartRequest, the function of the physicians' portal dedicated to completing requests: guided test selection, pre-filled data where possible, automatic completion of the consent forms and generation of PDF documents ready for signature.

Please contact our front office on +39 049 870 5062 or on the toll-free number 800 031 066 (from Italy only) for information on how to complete prescriptions or for any further information.

For SSN-covered activity, samples can be collected at our sample collection room at Corso Stati Uniti 4, Padua, by appointment booked through our front office.

For activity not covered by the SSN, samples are collected only at the Poliambulatorio Legnaro collection point, again by appointment booked through our front office.

Samples may also be collected at a healthcare facility anywhere in Italy. Sample pick-up must be arranged by telephone with R&I Genetics, which bears the full cost.

Sample: peripheral blood in EDTA (purple-top tubes), two tubes of at least 3 ml each; a single tube where it is not possible to collect two (e.g. paediatric patients). Pending pick-up, store at +4 °C; do not freeze.

Shipping: at room temperature only if the sample reaches the laboratory within 24 hours of collection; otherwise at controlled temperature (+4 °C). Avoid scheduling pick-up on Fridays and on the day before public holidays.

The payment methods currently available, including for patient co-payments (ticket) where due, are bank transfer or card payment at the R&I Genetics administrative office.

COMPLAINTS AND REPORTS

R&I Genetics guarantees all its clients the possibility of submitting complaints, observations or reports, with the aim of constantly improving the quality of the services offered.

Complaints and reports may be submitted through any of the following channels:

- **by e-mail**, writing to info@rigenetics.com;
- **by post,** writing to R&I Genetics srl, C.so Stati Uniti, 4, 35127 Padua, Italy
- **by telephone**, calling the laboratory on +39 049 870 5062.

For a complaint to be handled effectively and promptly, please state:

- Name and contact details, as anonymous complaints cannot be considered;
- The area, service or person to which the complaint refers.

Every communication received is recorded and handled in accordance with the laboratory's internal procedure, and a response is provided within 30 working days of receipt.

OUR COMMITMENT TO QUALITY AND INNOVATION

R&I Genetics directs all its activities towards the continuous improvement of quality, in line with regional guidelines, the requirements of the National Health Service and the needs of patients, physicians and healthcare facilities.

Quality is a founding and shared value that guides every strategic and operational choice of the laboratory.

In this perspective, R&I Genetics has identified the following areas for improvement, on which it is currently working.

1. Reporting times for neurodevelopmental analyses

Multigene analyses for neurodevelopmental disorders cover more than 2,000 genes and require longer interpretation times than other analyses. The laboratory has introduced organisational measures to bring at least 70% of these analyses within the target time of 240 days.

2. Organisational development

R&I Genetics promotes the development of its organisation through the recruitment of new professionals and the strengthening of in-house expertise, with the aim of consolidating an efficient, competent and service-quality-oriented organisation.

3. Appropriateness of services

The laboratory is committed to the constant improvement of the appropriateness of diagnostic services, including through the involvement of specialist physicians, continuous staff training and the adoption of interpretation workflows based on scientific evidence and good laboratory practice.

4. Development of information systems

R&I Genetics invests in the development of integrated information systems to improve sample traceability, process efficiency and the quality of analytical data. The aim is to foster technological innovation and strengthen bioinformatics capabilities, ensuring increasingly accurate and reproducible results.

5. Institutional accreditation, quality and patient safety

The laboratory maintains high quality standards, in full compliance with the requirements for institutional accreditation. All activities are aimed at ensuring patient safety, the clinical validity of the analyses and compliance with the applicable regulatory and technical criteria.

6. User satisfaction

R&I Genetics promotes initiatives aimed at user satisfaction, fostering access to services, transparency of information and constant dialogue with patients and professionals. Listening to users' needs is considered an integral part of the quality management system.

SAMPLE COLLECTION, REPORTING TIMES AND REPORT DELIVERY

Activity covered by the SSN

Sample collection is by appointment only and on prescription by the referring physician, by calling the telephone number given in the laboratory's contact details.

Opening hours: Monday to Friday, **9.30 a.m. to 4.30 p.m.**.

At registration, the patient must present:

- a valid identity document;
- the medical prescription;
- the Italian health insurance card (tessera sanitaria).

Sample collection centre:

R&I Genetics Srl

Torre della Ricerca

Corso Stati Uniti, 4 int. F – 35127 Padua, Italy

Activity not covered by the SSN

For activity not covered by the SSN, the patient – after booking an appointment, which is mandatory, with the R&I Genetics front office – may go to the external collection point:

R&I Genetics external collection point

at Poliambulatorio Legnaro

Piazzetta del Donatore, 3 – Legnaro (PD), Italy

Opening hours: Monday to Friday, **9.30 a.m. to 12.30 p.m.**.

In this case too, the patient must present:

- a valid identity document;
- the medical prescription, if any;
- the Italian health insurance card (tessera sanitaria).

Reporting times

The reporting time is calculated from the date of acceptance of the request, complete with sample and documentation, to the date the report is issued: incomplete documentation delays acceptance and, consequently, reporting.

For each type of analysis, the laboratory has set a target time:

Type of analysis	Target time
NGS analysis for neurodevelopmental disorders	240 days
NGS analysis for other rare diseases	120 days
Single-gene NGS, array-CGH, MLPA and methylation array	90 days
Sanger sequencing and TP-PCR	60 days

For critically ill paediatric inpatients, an urgent service is available on the physician's justified request: trio exome analysis with reporting within 10 days.

Quality target and results

The laboratory undertakes to report at least 70% of analyses within the target time. In the period 1 January – 30 September 2026, the following share of analyses was reported within the target time:

Type of analysis	Analyses reported within the target time
NGS analysis for neurodevelopmental disorders	see note
NGS analysis for other rare diseases	96.8%
Single-gene NGS, array-CGH, MLPA and methylation array	76.3%
Sanger sequencing and TP-PCR	81.7%

Overall, 79.1% of analyses were reported within the target time, with a median reporting time of 71 days.

Note – Neurodevelopment: see section "Our commitment to quality and innovation", item 1 "Reporting times for neurodevelopmental analyses".

Report delivery and consultation

The genetic testing service is guaranteed throughout the year (12 months out of 12).

Samples are accepted Monday to Friday, from 9.00 a.m. to 5.00 p.m.

The laboratory keeps the referring physician or the healthcare facility constantly informed of the progress of the analyses.

The report is made available to the requesting physician on the physicians' portal as soon as it is issued, with e-mail notification; on request, a paper copy is sent by courier to the physician or to the healthcare facility.

Access to the portal is reserved for physicians and is permitted only with the personal credentials provided by the R&I Genetics front office.

The requesting physician can make use of pre- and post-test support: a telephone discussion with the Technical Director or specialist staff to choose the most appropriate analysis and, once the report has been issued, to interpret the results.

For cases already analysed in our laboratories, the physician may request a reassessment of the data in the light of the most up-to-date scientific knowledge, on terms agreed with the laboratory.

CONTACTS

For any information or clarification, you may contact R&I Genetics staff, Monday to Friday from 9.00 a.m. to 5.00 p.m., at:

telephone: +39 049 870 5062

fax: +39 049 870 6696

e-mail: info@rigenetics.com.

For general information on our services, please also visit our website: www.rigenetics.com.

The Data Protection Officer (DPO), pursuant to the EU General Data Protection Regulation (Regulation (EU) 2016/679), is Dr Andrea Orsi, who can be reached at the contact details above.

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