SERVICE CHARTER

Version 15 of 05/08/2023

Performance Catalogue and Product Standards

Personal Genomics Srl
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1. Presentation

Personal Genomics is a Medical Genetics Laboratory (LGM) that has been providing tests and services in the world of research and clinical diagnostics since 2015, applying various technologies in the healthcare field to serve the diagnosis and treatment of patients, promoting appropriate behaviour and delivering effective healthcare.

1.1. History

Personal Genomics was founded in 2010 as a spin-off of the Functional Genomics Centre of the Departments of Science, Biotechnology and Medicine of the University of Verona; since 2013 it has also been a spin-off of the University of Bologna. Personal Genomics was awarded among the best business ideas in the Veneto region during the 2010 edition of Start Cup Veneto (competition between innovative business ideas organised by the Universities of Verona, Padua and Venice Ca' Foscari and with the collaboration of Veneto Innovazione) and has been involved in international translational research projects in the diagnostic field. Since May 2017, Personal Genomics S.r.l. has become part of the SOL Group S.p.A. Founded in 1927 in Italy, the SOL Group operates in the production, applied research and marketing of technical and medicinal gases, home care, biotechnology and energy production from renewable sources.

1.2. Mission

The laboratory aims to bridge the gap between the exponential scientific progress developed in genomic research laboratories and their application in the field of health care and preventive and precision medicine. The endless advances in genetics research have revolutionised the concept of medicine, providing new approaches for the prevention, diagnosis and treatment of diseases. The opportunities arising from these discoveries provide information that was not available just a few years ago and are crucial for the diagnosis and prevention of diseases.

1.3. Principles

Personal Genomics complies with the principles laid down in the Prime Ministerial Decree of 27/01/94:

- Equality and impartiality: services are provided equally to all, without discrimination of age, gender, language, religion, social status, political opinions, health conditions through impartial behaviour.
- Continuity of service: the service is provided with continuity according to the following timetables, although it does not operate
  in an emergency; any temporary interruption is communicated to users in good time.
- Right of free choice: the user has the right to choose his or her facility of choice, according to the regulations in force.
- Efficiency and effectiveness: services are provided according to the latest quality standards by qualified and constantly updated staff.

1.4. Process certification and quality control

Personal Genomics has a Quality Management System designed to offer guarantees to patients, doctors and laboratory staff. To this end, the laboratory has been UNI EN ISO 9001and SIGUCERT (Quality Management System for Medical Genetics Laboratories) certified since 2017 and Veneto Region Accredited (accreditation code 768225) since 2020.

Personal Genomics is on the list of authorised laboratories for the Veneto Region and the Ministry of Health
the performance of molecular diagnostics on swabs according to specific Real Time PCR protocols for SARS-CoV-2.

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The quality of results is ensured by performing internal quality controls and participating in international External Quality Assessment (EQA) circuits. Processes are kept under constant control through the application of product and process indicators identified by the laboratory itself and/or according to the indications of the Scientific Societies of the sector. Data available from the laboratory.

1.5. European Projects

Personal Genomics has been a partner in several European projects in recent years:

- NGS-PTL (Next Generation Sequencing platform for targeted Personalised Therapy of Leukemia): a European project with 10 partners for the period 2013-2015 focused on the identification of new prognostic biomarkers for acute and chronic leukaemia by means of exomic, transcriptomic and miRNAseq analyses.
- PANINI (Physical Activity and Nutrition INfluences In ageing): a European multidisciplinary training project with 8 world-leading beneficiaries on healthy ageing and 10 non-academic partners carefully selected for quality and range of fields. PANINI's vision was to implement cross-sectoral multidisciplinary research and training of a new cohort of researchers taking a radically innovative approach to overcoming the challenge of healthy ageing.
- PROPAG-AGEING: a project aimed at exploiting recent evidence that both physiological ageing and neurodegenerative diseases are propagating phenomena, using omics measurements of the gut microbiota and circulating biological fluids; identifying the combination of molecular, cellular and signalling pathway alterations that mark the transition between physiological ageing and Parkinson's disease. Project to identify markers for early diagnosis of Parkinson's disease before motor symptoms manifest themselves, using whole blood or other accessible biological fluids.
- INTCATCH: Horizon 2020 environmental programme whose main aim was to assemble, validate and develop a range of innovative devices and monitoring services for river and lake water quality in a cost-effective and efficient way and to cover large monitoring areas with an innovative and technologically advanced system. Development of a replicable and harmonised business model for water monitoring in the period 2020-2050.

To date, it is a partner in two European projects of the Horizon 2020 programme.

- The European project KATY: whose main aim is to use Artificial Intelligence and Bioinformatics in the fight against cancer. It aims to build personalised medicine systems to help doctors make decisions about the best therapy for the patient. KATY's artificial intelligence-based system is at the forefront of new cancer treatments. It is built around two main components: a distributed knowledge graph (DGK) and a set of predictive systems based explainable artificial intelligence (XAIP). The data organised through the distributed knowledge graph are used by the prediction systems to derive the best therapy for the specific patient.
- The ECLIPSE (ECL-based Infectious Pathogen (bio)Sensor) project aims to develop and produce a platform for pathogen detection. The project envisages detecting the presence of specific pathogens by exploiting the potential of electrochemiluminescence (ECL), a highly sensitive method, combined with the use of nanotechnology, for signal amplification, and the use of genetically modified bacteriophages. This new technology will make it possible to develop a highly reliable method with reduced costs and analysis times.

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1.6. Staff

Personal Genomics' staff consists of graduates with a high degree of specialisation in the fields of Medical Genetics, Biotechnology and Bioinformatics, and qualified administrative personnel. All personnel keep up to date by attending courses and conferences as part of Continuing Medical Education, where applicable. Personal Genomics also relies on the support of a Scientific Committee made up of university professors and professionals with years of experience in the field of molecular biology, and on the SOL Group for logistics and a widespread presence in the area. The strong synergy between Personal Genomics staff and SOL Group colleagues also enables our company to rely on a high-level technical and organisational staff, both in the legal, safety and environmental fields, always with a view to scientific research in the medical-health field.

1.7. Instrumentation

Personal Genomics is equipped with state-of-the-art equipment (NGS sequencers, Sanger sequencer, automatic extractors, etc.) for performing the proposed analyses. All the instrumentation is appropriate for the type and load of work performed and is constantly maintained, checked and updated in accordance with the provisions of the supplier companies, leaders in the field, and in compliance with the law. The list of instrumentation is available at the Laboratory.

1.8. Information Systems

Personal Genomics is equipped with validated and/or certified computer systems that guarantee full traceability of each sample stage: pre-analytical, analytical and post-analytical. The servers with the databases and the laboratory LIMS are installed in a computer centre with ISO 9001 and ISO 27001 certification and in compliance with Regulation 2016/679 - GDPR. All data and systems are backed up daily with geographical replication and a *disaster recovery* procedure accessible via VPN connection.

2. Service information

2.1. Addresses and contact details

The Medical Genetics Laboratory Personal Genomics S.r.l. is located in via Roveggia 43/B, 37136 Verona.
Tel: 045/8202351
E-mail: info@personalgenomics.it
PEC: pg@pec.personalgenomics.it
Website: www.personalgenomics.it

2.2. Opening Hours

2.2.1. Pre-test counselling and Point of Collection

At Personal Genomics, a pre-test counselling service is available to clarify the modalities, advantages and limitations of a genetic test the patient wishes to undergo. This is followed by the acquisition of informed consent and the documentation required to start the genetic test.

At the Personal Genomics blood collection point, it is also possible to take the necessary venous blood sample to the analysis. Fasting is not required for samples concerning genetic examinations.
The counselling clinic and the Testing Point are located at Via Roveggia 43/B, 37136 Verona and are open Monday to Friday from 9:00 to 12:00. Access to the facility to use the pre-test counselling service and/or to take biological material is by appointment only. Waiting times, understood as the time elapsed between the request

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and the appointment for taking the sample, do not exceed 5 days and are in any case agreed with the user on the basis of specific needs.

2.2.2. Workshop

Workshop activities are held from Monday to Thursday from 8:30 a.m. to 5:30 p.m. and on Fridays from 8:30 a.m. to 4:30 p.m., via Roveggia 43/B 37136 Verona. Within these hours, staff are available for information.

2.3. Information and booking

For information, the Customer Care staff is available Monday to Friday from 9 a.m. to 5 p.m. at 039/2396468. E-mail: customercare@personalgenomics.it

The secretariat for bookings is available to users Monday to Friday from 9:00 a.m. to 12:30 p.m. at 045/8202351.

2.3.1. Payment Methods

For examinations not in agreement with the Regional Health System, you can access privately and the available payment methods are cash, debit/credit card or bank transfer. Further information can be obtained from the Customer Care staff.

2.4. Acceptance of biological samples from external centres

Personal Genomics accepts biological samples (peripheral blood, nasopharyngeal swabs, saliva or DNA) from hospitals, clinics and private laboratories/clinics. Samples must be received accompanied by all required documentation duly completed and signed: Specific Request Form, Informed Consent Form, Personal Data Processing Consent Form.
Samples must be transported using a suitable transport system to ensure proper storage and delivery in time to guarantee reliable results. To this end, Personal Genomics provides the necessary instructions and the triple-wrapped collection kit for transport and, when required, takes charge of collecting and transporting the samples by a contracted courier service, in accordance with the regulations in force.
Fasting is not required for genetic tests.
Biological samples are accepted daily from Monday to Thursday from 8:30 a.m. to 5 p.m. and on Fridays from 8:30 a.m. to 5:30 p.m.

2.5. Collection of reports

The reports of the requested examinations are made available to the requesting practitioner or patient, on the basis of the regulations in force and what is indicated on the Request Form, on the Personal Genomics certified web portal (subject to the consent of the person concerned) or by collection at the office during the opening hours of the secretariat.
In accordance with Regulation 2016/679 - GDPR, access to the portal is only possible using temporary credentials provided to the user by Personal Genomics (password change expected at first access).
Whatever the result, it is recommended that the results be handed over to the patient by a doctor, possibly during a genetic counselling session, for an explanation of the meaning and any further investigations to be carried out. The authorised Personal Genomics staff is available for any clarification of the methodology used and the analytical significance of the test.
In the case of direct access to the laboratory by the patient, the pick-up of the report takes place at the laboratory after booking in the office or by telephone.

In case of loss of the report it is possible to request a certified copy by e-mail at info@personalgenomics.it. The copy of the report can be collected from Personal Genomics during office opening hours or can be sent by certified mail within 7 working days of receipt of the completed duplicate report request form.

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3. User information

3.1. Protection of privacy

Personal Genomics is committed to ensuring respect for the rights and dignity of citizens/users who come into contact with its various professionals.
In order to ensure the confidentiality of the persons performing the tests, Personal Genomics pseudonymises the sample upon arrival at the laboratory by assigning a unique code and undertakes to process the data in accordance with Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016, on the protection of natural persons with regard to the processing of personal data and on the free movement of such data and repealing Directive 95/46/EC (General Data Protection Regulation) as well as the guidelines of the Data Protection Supervisor and in absolute privacy and confidentiality.
Before information is collected, the user is informed about the use of the data being requested and about the ways of collecting and storing them.

3.2. Complaints, praise and reports

For an optimal management of the activity, with a view to constant improvement, Personal Genomics considers the contribution of everyone to be fundamental in order to promptly report any inefficiencies, inaccuracies, problems, lack of clarity or non-compliance with quality standards. Reports should be made using the form available on the website https://www.personalgenomics.it/segnalazioni/.
A complaint will result, within 24 hours, in the opening of a Non-Compliance with the definition of the appropriate corrective action to be taken.

3.3. Customer satisfaction

Personal Genomics monitors the level of satisfaction with the service provided through the administration of satisfaction questionnaires to patients who consent to the survey. The questionnaires are web forms whose link is sent by e-mail and whose completion is free and anonymous. The results are analysed by the Management and used for the continuous improvement of the service.

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4. Performance and product standards (examination characteristics; technical notes; technique used; reporting times; mode of storage of samples, data and reports)

4.1. Antenatal screening

Examination	Biological material	Technique used	Conditions examined / Targets analysed	Reporting times[1]
NIPT Personal Vision Basic	Peripheral blood (plasma)	NGS	Tests for Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome), Trisomy 13 (Patau syndrome), Turner syndrome, Trisomy X, Klinefelter syndrome, Jacobs syndrome, foetal sex (optional)	3-6 days
NIPT Personal Vision Medium	Peripheral blood (plasma)	NGS	Tests for Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome), Trisomy 13 (Patau syndrome), Turner syndrome, Trisomy X, Klinefelter syndrome, Jacobs syndrome, non-sex chromosome aneuploidies, foetal sex (optional).	3-6 days
NIPT Personal Vision Full	Peripheral blood (plasma)	NGS	Tests for Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome), Trisomy 13 (Patau syndrome), Turner syndrome, Trisomy X, Klinefelter syndrome, Jacobs syndrome, Non-sex chromosome aneuploidies, Foetal sex (optional). The test also identifies the following deletion syndromes if they exceed 7Mb: Cri-du-Chat syndrome, Prader Willi/Angelman syndrome, Wolf-Hirschhorn syndrome, 1p36 deletion syndrome.	3-6 days
NIPT Personal Vision Platinum	Peripheral blood (plasma)	NGS	Tests for Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome), Trisomy 13 (Patau syndrome), Turner syndrome, Trisomy X, Klinefelter syndrome, Jacobs syndrome, Non-sex chromosome aneuploidies, Foetal sex (optional). The test also identifies partial deletion/duplication syndromes if they exceed 7 Mb, with particular attention to regions where the genes implicated in the following syndromes are present: DiGeorge syndrome, Cri-du-Chat syndrome, Prader Willi/Angelman syndrome, Wolf-Hirschhorn syndrome, 1p36 deletion syndrome, Jacobsen syndrome, Langer-Giedion syndrome, Smith-Magenis syndrome, Brachydactyly syndrome - cognitive impairment, Alagille syndrome - JAG1, CHARGE syndrome - CHD7, Cornelia de Lange syndrome type 1 - NIPBL, Nevo - Sotos syndrome type 1 - NSD1, Bohring-Opitz syndrome - ASXL1, Schinzel-Giedion syndrome - SETBP1, Osteogenesis imperfecta type I, II, III, IV - COL1A1, Achondrogenesis imperfecta type 2 - COL2A1, Osteogenesis imperfecta type II, III, IV - COL1A2, Achondroplasia - FGFR3, Crouzon syndrome with acanthosis nigricans - FGFR3, Hypochondroplasia - FGFR3,	3-10 days

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Examination	Biological material	Technique used	Conditions examined / Targets analysed	Reporting times[1]
			Muenke syndrome - FGFR3, Thanatophore dysplasia type I and II - FGFR3, CATSHL syndrome - FGFR, Deletion syndrome 1p32-p31, Deletion syndrome 1p31, Deletion syndrome 1q41-q42, Deletion syndrome 2p16.1-p15, Deletion syndrome 2q31.1, Hand and foot cleft malformation (SHFM5) 2q31, Deletion syndrome 2q33.1, Duplication syndrome 2q35, Deletion syndrome 3pter-p25, Deletion syndrome 3q13.31, Dandy-Walker syndrome 3q22-q24, Deletion syndrome 3q29, Duplication syndrome 3q29, Deletion syndrome 4q21, Triplication syndrome 4q32.1-q32.2, Deletion syndrome 5q12, Deletion syndrome 5q14.3, Deletion syndrome 6pter-p24, Deletion syndrome 6q24- q25, Deletion syndrome 8q22.1, Deletion syndrome 8q22.1, Deletion syndrome 8p23.1, Deletion syndrome 8p23.1, Deletion syndrome 10q22.3- q23.2 10q23, Deletion syndrome 10q26, WAGR syndrome 11p13-p12, Potocki-Shaffer syndrome 11p11.2, Deletion syndrome 12q14, Deletion syndrome 13q14, Deletion syndrome 14q22.1-q22.3, Deletion syndrome 15q14, Deletion syndrome 15q25, Deletion syndrome 16p13.3, Deletion syndrome 16q22, Yuan-Harel-Lupski syndrome 17p12-p11.2, Potocki-Lupski syndrome 17p11.2, Deletion syndrome 17q12, Duplication syndrome 17q12, Deletion syndrome 17q23.1-q23.2, De Grouchy syndrome - Monosomy 18p, Syndrome deletion 19q13.11	
Fetal RH	Blood peripheral (plasma)	Real Time PCR	Search for specific traits of exons 5 and 7 of the RHD gene in free DNA circulating in maternal plasma, by Real Time PCR.	13 days
BITEST	Peripheral blood (serum)	TRACE	HCGb- free and PAPP-A + Risk calculation for Trisomy 21, Trisomy 18, Trisomy 13	3-5 days
Pre-eclampsia screening	Peripheral blood (serum)	TRACE	PLGF plus and sFlT-1	7 days

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4.2. Molecular genetics

Examination	Biological material	Technique used	Genes or regions analysed	Reporting times[1]
Mutational analysis of tumour DNA	Circulating Free DNA (Liquid Biopsy)/Fresh Tissue/Frozen Tissue/F FFPE/Agoaspirate	NGS	ALK (T1151-S1172; S1172-P1215; R1248-R1279), BRAF (K439-G478; N581-M620), EGFR (P576-G627; L688-K728; G729-D761 E762-K823; G824-K875; V96-L119; V250-R297), ERBB2 (E770-G787, E832-G853; E247-C275), FGFR3 (A359-G380), HRAS (M1-E37; D38-R97), IDH1 (R49-Q138_U2; S41-N48, IDH2 (E125-Q178), KIT (K550-G592; P627-G664; G664-C714; S715-G745; C788-N828; A829-G866; R449-E514; Y4-E37; D38-R97; R97-Q150), MET (D981-D1028; E1232-V1284), NRAS (M1-E37; D38-R97; R97-Q150), PDGFRA (K552-G596; P631-G668; C814-S854), PIK3CA (N521-S553; M1-G118_U2; R979-*1069; G118-F139_U2; N338-K353_U2; N418-D434_U2), RET (R587-D627; D627-L712; E762-G798; L870-Q910; G911-V934), ROS1 (V2015-T2045)	10-15 days
Gene fusions analysis on tumour tissue	Fresh/frozen fabric/fabric FFPE/aspirated needle	NGS	ALK, FGFR2, FGFR3, NTRK1, NTRK2, NTRK3, MET (skipping exon 14), PPARG, RET, ROS1	10-15 days
Alpha Thalassaemia	Peripheral blood	MLPA	HBA1 and HBA2	20 days
Spinal Muscular Atrophy (SMA)	Peripheral blood	MLPA	SMN1	15 days

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Arrhythmias; Long QT syndrome; Short QT syndrome; Brugada Syndrome; Ventricular Tachycardia; Sinus Node Disease; Familial atrial fibrillation; Inter-atrial defects	Peripheral blood	NGS	ABCC9, ACTC1, ACTN2, AKAP9, ANK2, CACNA1C, CACNB2, CASQ2, CAV3, DTNA, GPD1L, HCN4, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNQ1, MYH6, MYH7, MYPN, PKP2, SCN1B, SCN3B, SCN4B, SCN5A, SNTA1	45 days

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Examination	Biological material	Technique used	Genes or regions analysed	Reporting times[1]
Beta Thalassaemia	Peripheral blood	Sanger	HBB	20 days
Cardiomyopathies	Peripheral blood	NGS	ABCC9, ACTC1, ACTN2, ADCK3, AGK, ANKRD1, BAG3, CALR3, CAV3, COA3, COA5, COQ7, COQ8A, COX14, COX20, CRYAB, CSRP3, CTNNA3, DES, DMD, DNAJC19, DSC2, DSG2, DSP, DTNA, ELAC2, EYA4, FLNC, FXN, GATAD1, GLA, GTPBP3, ILK, JPH2, JUP, LAMA4, LDB3, LMNA, MCEE, MMAA, MMAB, MMACHC, MMADHC, MMUT, MRPL3, MRPL44, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NDUFAF5, NEXN, PCCA, PCCB, PKP2, PLN, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RYR2, SCN5A, SDHA, SDHD, SGCD, SLC19A2, TCAP, TGFB3, TGFBR1, TGFBR2, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL	45 days
Dilated Cardiomyopathy	Peripheral blood	NGS	ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CRYAB, CSRP3, DES, DMD, DSG2, DTNA, EYA4, GATAD1, LAMA4, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYPN, NEXN, PKP2, PLN, PRDM16, PSEN1, PSEN2, RAF1, RBM20, SCN5A, SDHA, SGCD, TCAP, TMP, TNNC1, TNNI3, TNNT2, TTN, VCL	45 days
Hypertrophic cardiomyopathy	Peripheral blood	NGS	ACTC1, ACTN2, CALR3, CAV3, CSRP3, DTNA, FLNC, GLA, ILK, JPH2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, PLN, RBM20, TCAP, TNNI3, TNNT2, TPM1, TTN	45 days
Arrhythmogenic and Non-arrhythmogenic Cardiomyopathy Tablet	Peripheral blood	NGS	CTNNA3, DSC2, DSG2, DSP, GATAD1, JUP, MYH6, MYH7, MYPN, PKP2, PSEN1, PSEN2, RYR2, TCAP, TGFB3, TGFBR1, TGFBR2, TMEM43	45 days
Disease Cardiomyopathy Mitochondrial	Peripheral blood	NGS	ADCK3, AGK, COA3, COA5, COQ7, COQ8A, COX14, COX20, DNAJC19, ELAC2, FXN, GTPBP3, MCEE, MMAA, MMAB, MMACHC, MMADHC, MRPL3, MRPL44, MMUT, NDUFAF5, PCCA, PCCB, SDHD, SLC19A2	45 days
Congenital Heart Disease	Peripheral blood	NGS	ACTC1, ACTN2, DTNA, GLA, TTR	45 days
Carrier woman	Peripheral blood	NGS	CFTR, DMD, GJB2, GJB6, HBA1/HBA2, HBB, PAH and SMN1	30 days
Carrier man	Peripheral blood	NGS	CFTR, GJB2, GJB6, HBA1/HBA2, HBB, PAH and SMN1	30 days

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Mineralisation defects	Peripheral blood	NGS	ABCC6, ACVR1, AHSG, ALPL, ANKH, ANO6, BGLAP, BMP2, BMP2K, BMP4, BMP6, BMPR1A, BMPR1B, BMPR2, CCN3, CDH11, CLCN5, CLEC3B, COL1A1, COL1A2, CYP27B1, CYP2R1, DCN, DMP1, DSPP, ECM1, EFNB2, ENPP1, EPHB4, FAH, FAM20C, FBN1, FGF23, FGFR1, FKBP11, FN1, FOXC2, FZD9, GALNT3, GGCX, GGPS1, HNF4A, HNRNPC, IBSP, IFITM5, ITGB1, ITGB1BP1, KCNMA1, KL, LUM, MEPE, MGP, NT5E, OCRL, OGN, OMD, OSTN, PGAP2, PGAP3, PHEX, PHOSPHO1, PIGO, PIGV, PIGW, PIGY, PORCN, PPA1, SAMD9, SLC34A1, SLC34A3, SLC8A1, SLC9A3R1, SPP1, THBS2, TNFRSF11B, VDR	30 days

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Examination	Biological material	Technique used	Genes or regions analysed	Reporting times[1]
Congenital parathyroid disorders	Peripheral blood	NGS	ACADM, AIRE, AP2S1, CASR, CDC73, CDKN1A, CDKN1B, CDKN2B, CDKN2C, CHD7, FAM111A, FHL1, GATA3, GCM2, GNA11, GNAS, HADHA, HADHB, HLA-DRA, MEN1, PDE3A, PDE4D, PRKAR1A, PTH, PTH1R, RET, SEMA3E, SOX3, STX16, TBCE, TBX1, TRPV6	30 days
Congenital bone dysplasias	Peripheral blood	NGS	ABL1, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AFF4, AGA, AGPS, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, APC, APC2, AQP1, ARHGAP31, ARSB, ARSL, ATP6V0A2, ATR, B3GALT6, B3GAT3, B4GALT7, B9D1, B9D2, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, C2CD3, CA2, CANT1, CC2D2A, CCDC8, CCN6, CDC45, CDC6, CDKN1C, CDT1, CENPJ, CEP120, CEP152, CEP290, CEP63, CFAP410, CHRNA1, CHRND, CHRNG, CHST14, CHST3, CHSY1, CLCN5, CLCN7, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COMP, CPLANE1, CREB3L1, CREBBP, CRTAP, CTSA, CTSK, CUL7, CYP26B1, CYP27B1, CYP2R1, DCN, DDR2, DDRGK1, DDX59, DHODH, DLL3, DLL4, DLX3, DLX5, DLX6, DMP1, DNA2, DOCK6, DSPP, DVL1, DVL3, DYM, DYNC2H1, DYNC2LI1, EBP, EFNB1, EFTUD2, EIF2AK3, EIF4A3, ENPP1, EOGT, EP300, ERF, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FBLN1, FBN1, FBN2, FBXW4, FERMT3, FGD1, FGF10, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FUCA1, GALNS, GALNT3, GDF3, GDF5, GDF6, GJA1, GLB1, GLI3, GMNN, GNAS, GNPAT, GNPTAB, GNPTG, GORAB, GPC6, GUSB, HDAC8, HES7, HNRNPA1, HNRNPA2B1, HNRNPC, HOXA11, HOXA13, HOXD13, HPGD, HSPG2, ICK, IDH1, IDH2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT80, IFT81, IHH, IKBKG, IL11RA, IMPAD1, INPPL1, KAT6B, KDM6A, KIAA0586, KIAA0753, KIF22, KIF7, KL, KMT2D, LARP7, LBR, LEMD3, LFNG, LIFR, LMBR1, LMNA, LMX1B, lncRNA-RMRP, LONP1, LRP4, LRP5, LTBP3, MAB21L2, MAFB, MAN2B1, MAP3K7, MASP1, MATN3, MBTPS2, MCM5, MECOM, MEGF8, MEOX1, MESP2, MGP, MIR17HG, MKS1, MMP13, MMP2, MMP9, MSX2, MTAP, MYCN, MYO18B, NANS, NBR1, NEK1, NEU1, NFIX, NIN, NIPBL, NKX3-2, NOG, NOTCH1, NOTCH2, NPR2, NSD1, NSDHL, NSMCE2, OBSL1, OCRL, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PCNT, PCYT1A, PDE3A, PDE4D, PEX7, PGAP2, PGAP3, PHEX, PIEZO2, PIGO, PIGV, PIGW, PIGY, PITX1, PLEKHM1, PLOD2, PLS3, POC1A, POLR1A, POLR1C, POLR1D, POP1, POR, PORCN, PPIB, PRKAR1A, PTDSS1, PTH1R, PTHLH, PTPN11, PYCR1, RAB23, RAB33B, RAD21, RBBP8, RBM8A, RBPJ, RECQL4, RIPPLY2, RNU4ATAC, ROR2, RPGRIP1L, RSPO2, RUNX2, SALL4, SAMD9, SBDS, SCARF2,	30 days

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			SEC24D, SEM1, SERPINF1, SERPINH1, SF3B4, SFRP4, SGMS2, SH3BP2, SH3PXD2B,	

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Examination	Biological material	Technique used	Genes or regions analysed	Reporting times[1]
			SHOX, SKI, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35D1, SLC39A13, SLC9A3R1, SLCO2A1, SMAD4, SMAD6, SMARCAL1, SMC1A, SMC3, SMOC1, SMPD3, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, SQSTM1, SRCAP, SUMF1, TAPT1, TBCE, TBX15, TBX22, TBX4, TBX5, TBX6, TBXAS1, TCF12, TCIRG1, TCOF1, TCTEX1D2, TCTN2, TCTN3, TGDS, TGFB1, TMEM107, TMEM216, TMEM231, TMEM38B, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TP63, TPM2, TRAIP, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TTC21B, TWIST1, TYROBP, VCP, VDR, WDR19, WDR34, WDR35, WDR60, WNT1, WNT10B, WNT3, WNT3A, WNT5A, WNT7A, XYLT1, XYLT2, YY1AP1, ZIC1, ZMPSTE24, ZNF687	
Muscular dystrophy (Duchenne and Becker)	Peripheral blood	MLPA	DMD	15 days
Autism Spectrum Disorders	Peripheral blood	NGS	SFARI Gene database	80 days
Haemochromatosis - level 1	Peripheral blood	PCR and sequencing	HFE (C282Y, H63D, S65C)	10 days
Premature ovarian exhaustion	Peripheral blood	NGS	BMP15, DACH2, ERCC6, FANCM, FIGLA, FLJ22792, FMR1, FOXL2, FOXO3, FSHR, GDF9, HFM1, INHA, MCM9, MRPS22, MSH5, NANOS3, NOBOX, NR5A1, NUP107, PGRMC1, POU5F1, PSMC3IP, SOHLH1, SOHLH2, SPIDR, STAG3, SYCE1, SYNE1, TGFBR3	30 days
Familial exome	Peripheral blood	NGS	About 20000 known genes	80 days
Cystic fibrosis level 1	Peripheral blood	NGS	Around 130 known variants on the CFTR gene	13 days
Cystic fibrosis whole gene	Peripheral blood	NGS	Whole CFTR gene sequencing	30 days
Family genome	Peripheral blood	NGS	Entire genome	120 days
Hypophosphatasia of the adult	Peripheral blood	NGS	ALPL	30 days
Y chromosome microdeletions	Peripheral blood	Amplification and separation of amplified products with capillary electrophoresis	AZF Region	15 days

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FSH/LH receptor variants	Peripheral blood	PCR-RFLP	FSHR (T307A, N680S), LHR (R554*)	15 days

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Examination	Biological material	Technique used	Genes or regions analysed	Reporting times[1]
PAI	Peripheral blood	Real Time PCR	SERPINE1 (4G/5G)	15 days
Thrombophilic panel extended	Peripheral blood	Real Time PCR	FII (G20210A), FV (R506Q, H1299R), MTHFR (C677T, A1298C), PAI (4G/5G), FGB (-455G>A), HPA1 (1°/b), ACE (Ins/Del), ApoB (R3500Q), ApoE, FXIII (V34L).	15 days
Basic thrombophilic panel	Peripheral blood	Real Time PCR	FII (G20210A), FV (R506Q), MTHFR (C677T, A1298C)	10 days
Pathologies Cardiovascular	Peripheral blood	NGS	ANKRD1, APOB, LDLR, PCSK9, LDLRAP1	45 days
HLA-G polymorphism	Peripheral blood	Real Time PCR	HLA-G (Insertion/Deletion)	20 days
Searching for familiar known variants	Peripheral blood	Sanger/MLPA	Known variant	30 days
X- syndrome Fragile (FRAXA)	Peripheral blood	PCR and analysis of fragments	FMR1 (CGG triplet in 5'-UTR)	35 days
Whole gene congenital deafness Deletion congenital deafness	Peripheral blood	Sanger Detection of D13S1830 deletion	GJB2 GJB6	15 days 15 days
Typing molecular HLA for coeliac disease	Peripheral blood	Real-Time PCR	HLA alleles DQ2 and DQ8	15 days
Endometrial cancer	Peripheral blood	NGS	EPCAM (3' UTR deletion), MLH1, MSH2, MSH6, PTEN, STK11, TP53	30 days
Breast/ovarian cancer - Extended	Peripheral blood	NGS	ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, NBN, PALB2, PTEN, STK11, TP53	30 days
Breast Cancer - Second level	Peripheral blood	NGS	ATM, BARD1, CDH1, CHEK2, NBN, PALB2, PTEN, STK11, TP53	30 days
Tumour Breast/ovary	Peripheral blood	NGS	BRCA1 and BRCA2	30 days
Ovarian cancer	Peripheral blood	NGS	BRCA1, BRCA2, BRIP1, EPCAM (3' UTR deletion), MLH1, MSH2, MSH6, PMS2, RAD51C, RAD51D, STK11, TP53	30 days

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4.3. Classical and molecular cytogenetics

Examination	Biological material	Conditions examined	Reporting times[1]
Karyotype	Peripheral blood, Amniotic fluid, Abortion material	Chromosomal abnormalities, both numerical and structural	25 days (20 days for liquid amniotic)
QF-PCR	Amniotic fluid, foetal blood	Numerical abnormalities of chromosomes 21, 13, 18, X and Y	5 days
Low CGH arrays resolution(15k)	Amniotic fluid, Abortion material	Copy number alterations of chromosome portions	20 days
High-resolution CGH array (60k)	Amniotic fluid, Abortion material	Copy number alterations of chromosome portions	20 days
FISH/Chromosome painting	Peripheral blood, Amniotic fluid, Abortion material	Selected cases for the characterisation of karyotype abnormalities	20-30 days

4.4. Molecular biology

Examination	Biological material	Technique used	Reporting times[1]
Rapid antigen swab	Nasopharyngeal swab	Immunochromatography/Immunofluorescence	-
HPV DNA High Risk	Vaginal swab, Thin prep	Real-Time PCR	8 days
HPV DNA High and Low Risk	Vaginal swab, Thin prep	Real-Time PCR	15 days
Sexually Transmitted Diseases	Vaginal swab, Thin prep	Real Time PCR	7 days

[1] Reporting times are in working days from receipt of the sample in the laboratory.

Based on the laboratory's needs, some analyses may be performed at accredited, qualified and certified external laboratories appointed by Personal Genomics as suppliers, located at
- Via Pavoni, 18 - Castenedolo (BS);
- Via Castel Giubileo, 62 - Rome (RM);
- Keilaranta 16 A-B, 02150 Espoo - Finland.

The details and price for each service can be found in the Catalogue and the Schedule of Services (Annex A), respectively.

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4.5. Technical notes on performance

- NIPT (Non Invasive Prenatal Test): a non-invasive prenatal screening test consisting of the analysis of free foetal DNA circulating in maternal blood by means of Next Generation Sequencing (NGS) techniques and sophisticated bioinformatic analysis. Analytical sensitivity 74.1 - 95.5% and analytical specificity >99%.
- NGS (Next Generation Sequencing): set of massively parallel sequencing technologies that have the capacity to sequence millions of nucleic acid fragments simultaneously. Analytical sensitivity >99%, with a minimum coverage of 10X, and analytical specificity >99%.
- Real Time qPCR: technology used to quantify nucleic acids by measuring the fluorescence emitted by a fluorophore. Analytical sensitivity and specificity >99%.
- Amplification and separation of the amplified products by capillary electrophoresis: a methodology that makes it possible to determine the number of consecutive repeats, in variable numbers and therefore polymorphic, of identical sequences. Analytical sensitivity >99% and analytical specificity 98.4%.
- MLPA® (Multiplex Ligation-dependent Probe Amplification): a technique for detecting copy number alterations of certain genomic regions and is therefore able to identify deletions and duplications of coding regions (exons). Analytical sensitivity and specificity >99%.
- Sanger direct sequencing: reference technique for molecular genetics that allows the sequencing of small gene portions. Analytical sensitivity and specificity >99%.

4.6. Storage times: data and biological material

The minimum guaranteed deadlines for the storage of biological and documentary material relating to the various types of genetic tests are based on the document drafted by the SIGU-Health Working Group 'Guidelines on the Conservation of Biological and Documentary Material relating to Genetic Testing" dated 19/01/2021 and are as follows:

SUBJECT	**STORAGE TIME** (from the date of issue of the report)
Patient-Laboratory User Card	20 years (from compilation)
Informed consent	20 years (from compilation)
Report	Unlimited
Files/pictures/photos	20 years
NGS data: bam or fastq files	2 years
NGS data: vcf files	20 years
In prenatal care	
Residual biological material	6 months after the expected date of delivery (10 years if anomalies are not fully characterised)
In the postnatal environment	
Residual biological material (cytogenetic tests)	15 days (10 years if anomalies are not fully characterised)
Residual biological material (molecular tests including those performed by NGS methodology)	15 days if the diagnostic question has been resolved (10 years if unresolved case, which may require further examinations)
In oncology	
DNA/biological material processed if cases do not fully defined	10 years

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5. Annex A: Catalogue and schedule of services.

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