Personal Genomics

DESIGNING
THE FUTURE
OF GENOMICS

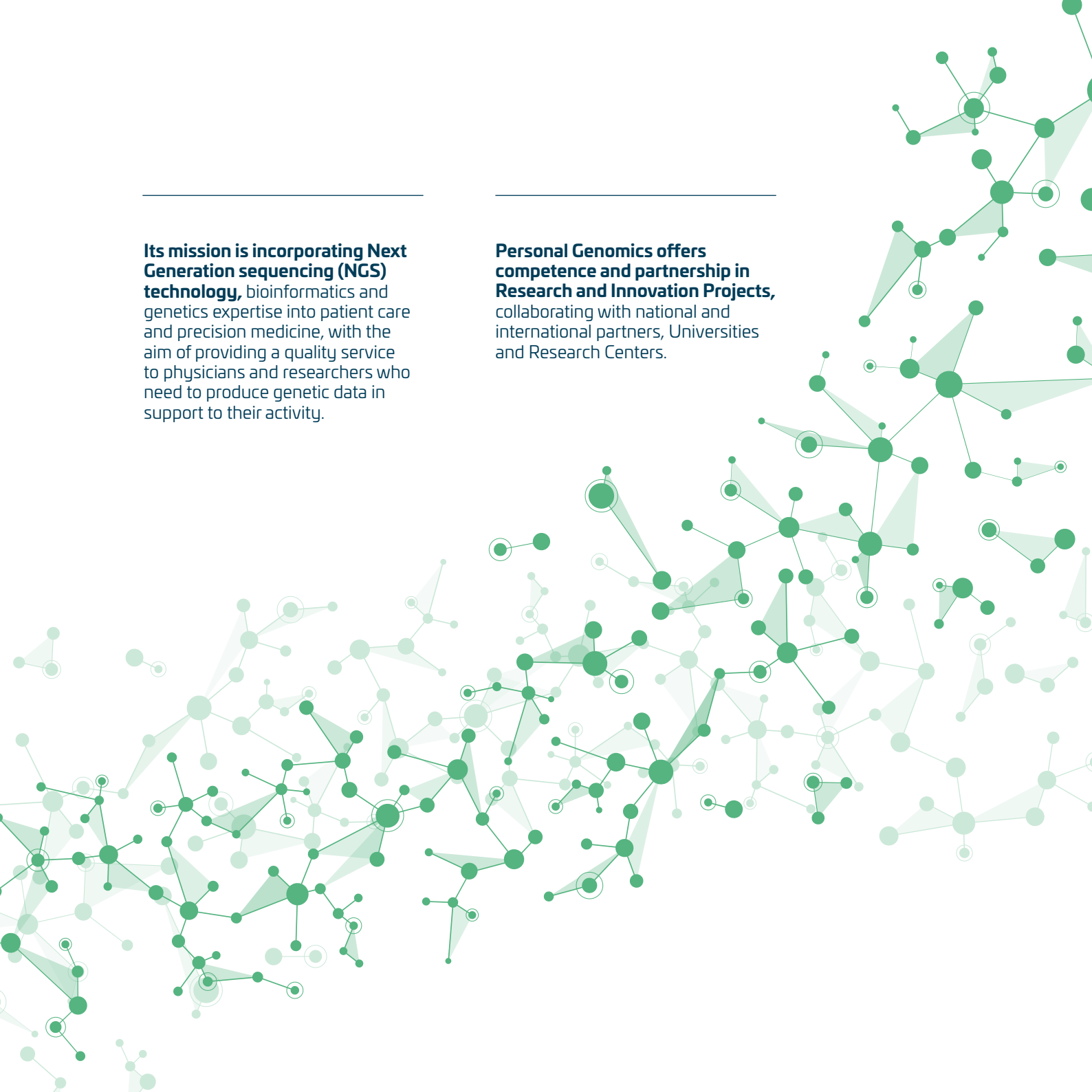
Personal Genomics provides genetic tests and services, ensuring high quality standards in each stage of the production process and in the interpretation of genetic data.

Personal Genomics is a provider of genomics services, born in 2011 as a spin-off of the University of Verona.

Personal Genomics is certified ISO 9001 and Sigucert ®.

Its mission is incorporating Next Generation sequencing (NGS) technology, bioinformatics and genetics expertise into patient care and precision medicine, with the aim of providing a quality service to physicians and researchers who need to produce genetic data in support to their activity.

Personal Genomics offers competence and partnership in Research and Innovation Projects, collaborating with national and international partners, Universities and Research Centers.



1

DNA EXTRACTION

- Extraction from different samples with optimized kit for NGS analysis

DNA/RNA

SALIVA
BLOOD
PARAFFIN
PLASMA

2

LIBRARIES PREPARATION

- Libraries preparation
- Support in the choice of the enrichment kit in order to obtain the best performance
- Personalized panels design

GENOME

EXOME

GENETIC PANELS

RNA LIBRARIES

3

SEQUENCING

- Sequencing services based on different technologies available
- Support in the choice of which technologies is the most performant according to the goal

NGS SERVICES

4

DATA ANALYSIS

- Production of BAM and VCF file
- Production of gVCF file that allow us to identify genotypable regions

ALIGMENT AND VARIANTS CALLING

Annotation of variants with:

- Public DB
- Commercial DB

ANNOTATION

- Prioritization of annotated variants for considering clinical significance, frequency in population and functional annotation
- Prioritization of not annotated variants, distinguishing between coding and non-coding regions, considering frequency in population, frequency in our private database PGVD and functional annotation of the variant

PRIORITIZATION AND INTERPRETATION

5

PERSONALIZED REPORT

- Production of a report for clinical interpretation of germinal and somatic variants

NGS Services

EXPERIMENTAL AND PANEL DESIGN

- Support in the experimental design of NGS projects, together with data production and data analysis.
- Support for the selection of the best enrichment kit based on desired genes list.

SAMPLE PREPARATION AND LIBRARY CONSTRUCTION

- Extraction (DNA/RNA) from different biological sources (saliva, blood, plasma, fresh or fixed tissues) with optimized kit for NGS.
- Library preparation.

SEQUENCING

- Sequencing services based on different platforms (e.g., Illumina, PacBio, Oxford Nanopore).

GENOME

- Whole Genome Sequencing.
- Bioinformatic analysis (using public, commercial and private DBs).
- Structural Variants identification.

EXOME AND GENETIC PANELS

- Exome sequencing.
- Panel design and sequencing (commercial and custom).
- Bioinformatic analysis (using public, commercial and private DBs).

TRANSCRIPTOMICS

- Transcriptome sequencing (polyA or rRNA depletion).
- Bioinformatic analysis (gene-level, isoform-level).

METAGENOMICS

- 16S/ITS amplicon sequencing.
- Shotgun genome/transcriptome sequencing.
- Bioinformatics analysis (standard or custom pipeline).

DATA ANALYSIS

- Standard products including validated pipelines for NGS data analysis.
- Support for custom analysis and pipeline development.

VARIANT ANNOTATION

- Annotation of variants with: Public DBs and Commercial DBs.
- Prioritization of annotated variants considering the clinical significance, the frequency in the population and the functional annotation.
- Prioritization of not-annotated variants, distinguishing between coding and non-coding regions, considering the frequency in the population, the frequency in our proprietary database PGVD and the functional annotation.

Diagnostics

PANORAMA TEST - NIPT

Panorama® is an NGS-based test that analyses cell-free fetal DNA to determine the risk that the fetus will be born with certain chromosomal aneuploidies and/or microdeletions.

FETAL SEX DETERMINATION

Non-invasive prenatal screening test performed in the first weeks of pregnancy using qPCR technique, useful in case of pregnancies with an increased risk for X-linked genetic disease.

FETAL RH GENOTYPING

Non-invasive prenatal screening test performed in the first weeks of pregnancy using qPCR technique, to prevent unnecessary immunization in RhD-negative women.

X FRAGILE SYNDROME TEST

This test aims to quantify the number of CGG triplets in the FMR1 gene using TP-PCR technique, to determine the association with FMR1-related disorders.

SPINAL MUSCULAR ATROPHY TEST

This test screens for the deletion of exons 7 and/or 8 of SMN1 gene using the MLPA technique, either to identify carriers or confirm a clinical diagnosis of SMA.

ALPHA-THALASSEMIA TEST

This test screens for deletions in HBA1 and/or HBA2 genes using the MLPA technique, either to identify carriers or confirm a clinical diagnosis of alpha-thalassemia.

THROMBOPHILIA TEST

Sanger sequencing is used to assess whether the patient is carrier of one or more variants associated with hereditary thrombophilia to eventually set up a preventive antithrombotic therapy.

CARRIER TEST

This NGS-based test aims to investigate the presence of variations in genes causative of recessive genetic diseases with highest prevalence in Europe: Cystic fibrosis, SMA, Alpha- and Beta-thalassemia, Phenylketonuria, Non syndromic hearing loss, Duchenne and Becker muscular dystrophy (only for females).

CYSTIC FIBROSIS TEST

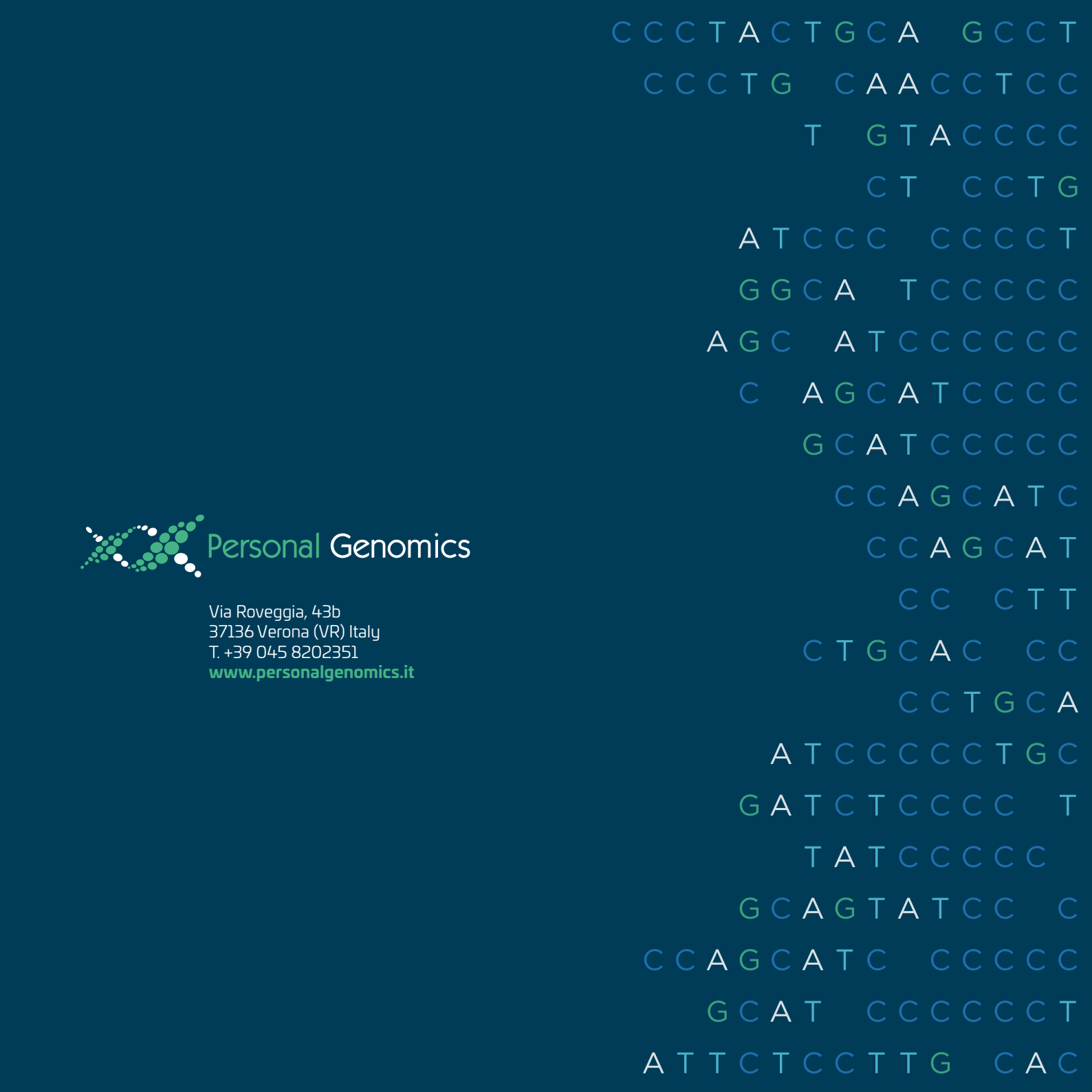
This NGS-based test investigates the presence of pathogenetic variants (both point and copy-number) in the CFTR gene, either to identify carriers or confirm a clinical diagnosis of Cystic Fibrosis.

BRCA TEST

This NGS-based test aims to identify pathogenetic variants in the BRCA1 and BRCA2 genes. Variants located in these genes are related to a higher risk of developing breast and/or ovarian/tubal cancer.

AUTISM SPECTRUM DISORDERS TEST

Whole Genome Sequencing enables the detection of not only small variations, but also large chromosomal rearrangements associated with autism spectrum disorders, to understand the molecular basis of the disease.



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