The 5000genomi@VdA Project: an Italian Whole Genome Sequencing initiative

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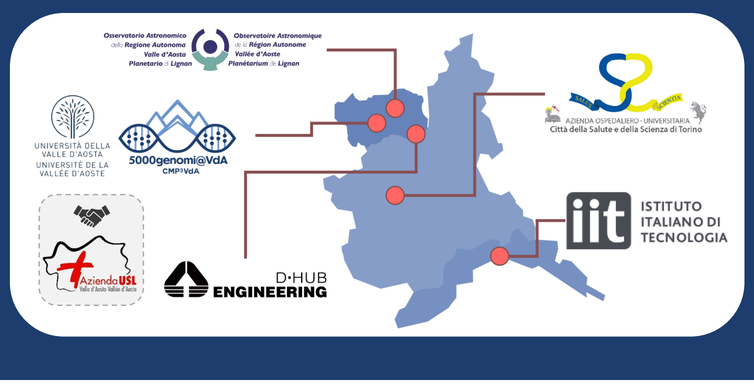
5000genomi@VdA consortium



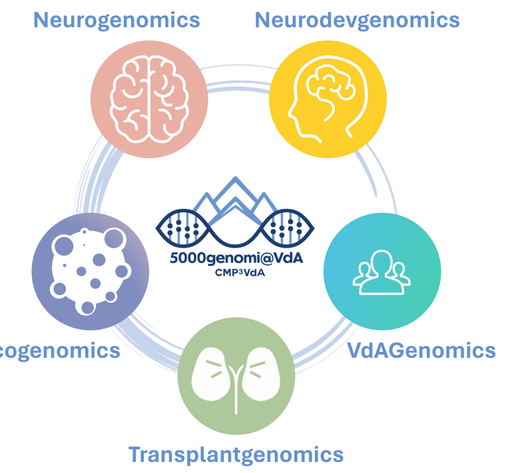
Introduction

The **5000genomi@VdA Project** is a **unique Italian initiative** funded by the Autonomous Region Valle d'Aosta, in the Northwest of Italy:

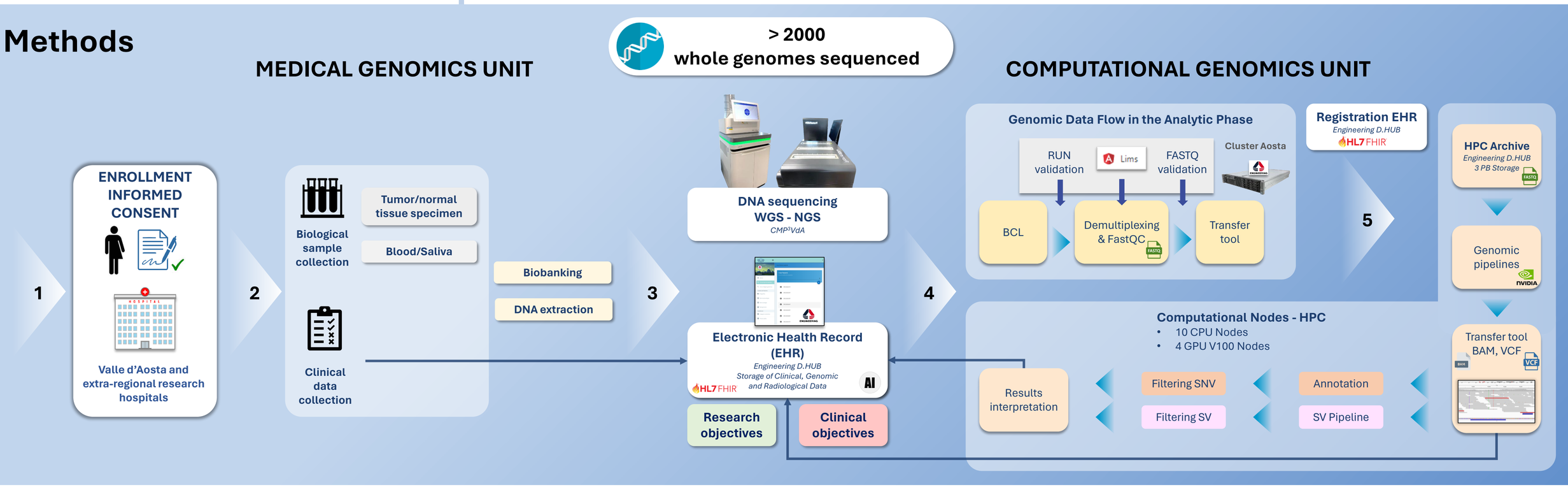
- The project is carried out by a **multidisciplinary consortium** led by the **Italian Institute of Technology - IIT** (Genoa)
- The project takes advantage of collaborating closely with the **Valle d'Aosta healthcare system** and other research centers/hospitals in Italy.

Objectives

The analysis of the entire genome is not a standard practice in clinical genomics studies. In this project, short- and long-read whole genome sequencing (WGS) approaches are employed to study coding and non-coding genomic regions.
The Project aims to sequence **5,000 genomes** of healthy donors (**VdAGenomics**) and patients with neurodevelopmental (**Neurodevgenomics**), neurodegenerative (**Neurogenomics**), oncological (**Oncogenomics**), and organ transplantation (**Transplantgenomics**) diseases.
Through the synergistic interaction between private-public research and healthcare centers, the goal is to study the role our genes play in health and disease conditions, thus promoting the integration of WGS in the clinic.



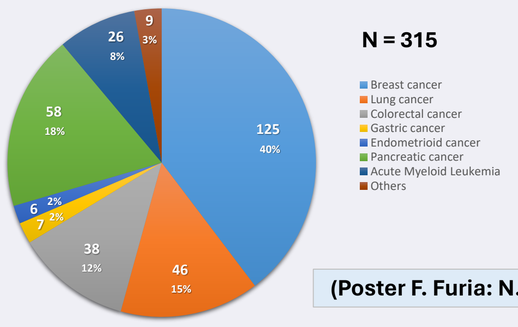
Methods



Results

Oncogenomics

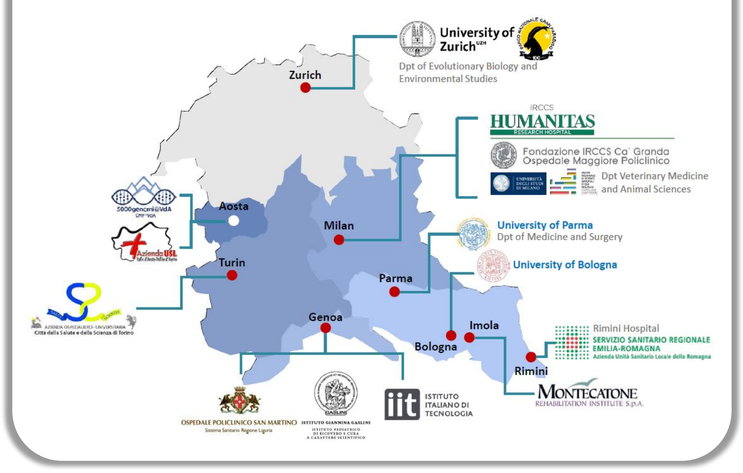
WGS of paired healthy/tumor tissues of patients with different cancer types



N = 315

(Poster F. Furia: N. 144)

The CMP³VdA Network



Neurodevgenomics

Trio-based WGS of families with child affected by autism spectrum disorders (ASD) and intellectual disabilities (ID)

N = 140 families
(459 subjects in total)

Trio with variants of clinical relevance

27%

Trio with variants of uncertain significant of scientific interest

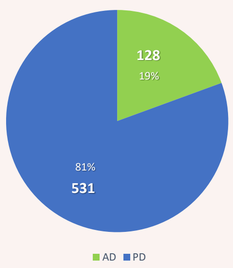
37%

N = 70 (trio analyzed)

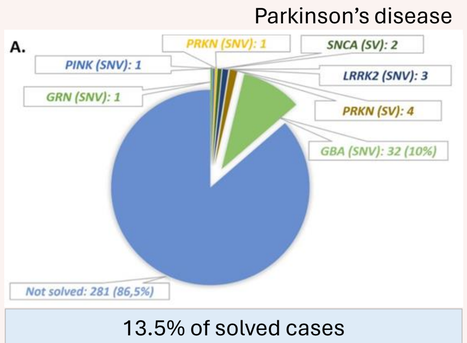
Neurogenomics

WGS of patients with Alzheimer's and Parkinson's disease

N = 659



Parkinson's disease



13.5% of solved cases

(Poster A. Fant: N. 120)

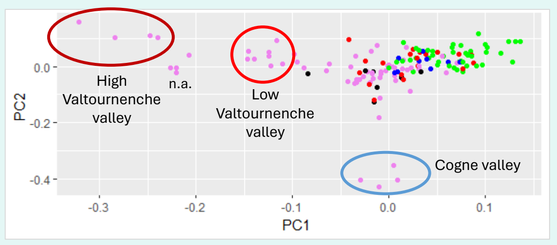
Transplantgenomics

WGS of patients with kidney and/or liver transplantation as well as polycystic kidney disease (PKD)

N = 304

VdAGenomics

WGS of blood donors to study the genomic profile and the genetic characteristics of the modern population of Valle d'Aosta



Stratification of the Aosta Valley population by Principal Component Analysis (PCA)

N = 142

Legend	
Green	0 GP VdA
Blu	1 GP VdA
Red	2 GP VdA
Black	3 GP VdA
Purple	4 GP VdA

7.2M SNPs filtered by:
- MAF > 0.05%
- Genotype > 0.05%

Conclusions and future perspectives

- The consortium sequenced more than 2,000 human genomes.
- This study represents a strategic blueprint healthcare model for implementing WGS into clinical practice in Italy.

References

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GENOMIC INSIGHTS INTO PARKINSON'S DISEASE OF A LARGE ITALIAN COHORT.

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