

FRANCE MEDECINE
GENOMIQUE 2025



PFMG2025

INTEGRATING GENOMIC MEDICINE INTO THE NATIONAL HEALTHCARE SYSTEM IN FRANCE

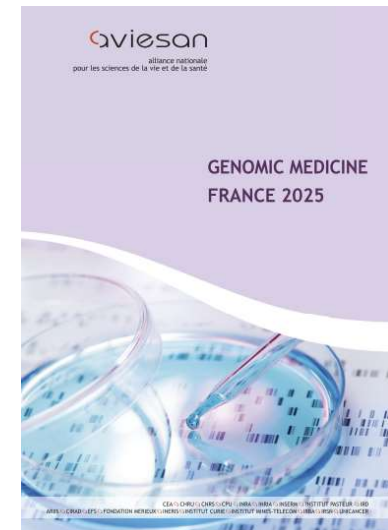
Frédérique Nowak, coordinator of the 2025 French Genomic Medicine Initiative
ICPerMed Webinar of the Internationalisation Working group, February 26, 2026



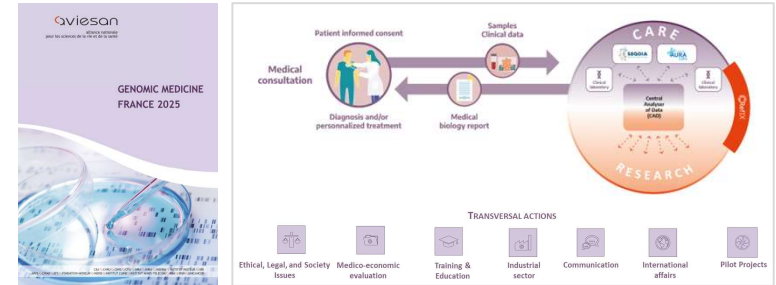
2025 FRENCH GENOMIC MEDICINE INITIATIVE AMBITION

**Launch of the initiative in 2016 upon Prime Minister request,
with the ambition of:**

- ⇒ Integrating genomic medicine into the patient's care pathway
- ⇒ Allowing equal access to clinical whole genome sequencing, first for rare diseases and cancers and then for common diseases, as new therapeutic indications are validated
- ⇒ Developing a genomic medicine sector



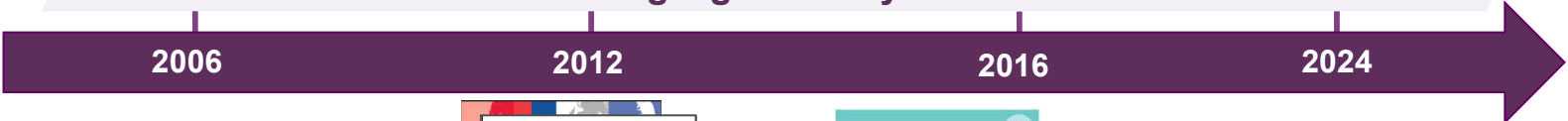
THE FRAMEWORK FOR GENOMIC MEDICINE IN FRANCE



Genome sequencing

Genes panel sequencing

Single gene analysis



- ✓ A network of tumour genetics laboratories
- ✓ A network for cancer genetics predisposition
- ✓ A network of early phases clinical trials centres



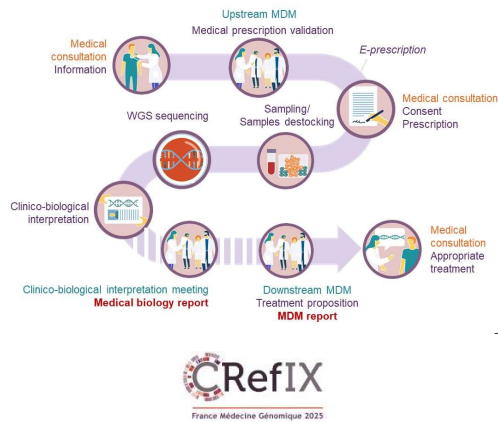
- ✓ Regional diagnostic laboratories
- ✓ Centres of Reference
- ✓ Thematic rare diseases health networks
- ✓ A national database



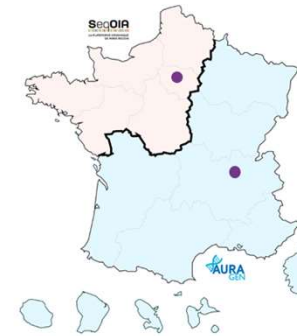
INTEGRATION INTO THE HEALTHCARE SYSTEM



✓ A genomic healthcare pathway



✓ 2 clinical WGS laboratories



✓ 82 clinical preindications

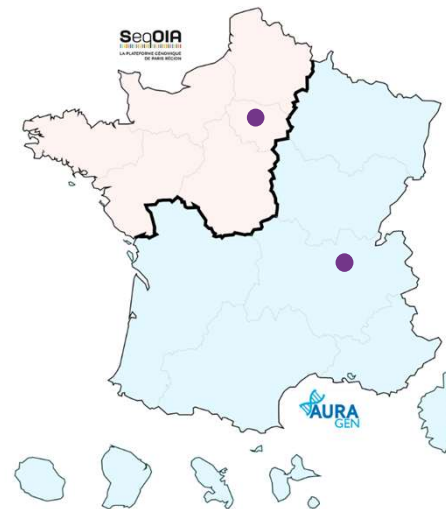
✓ Data reuse for research



Central Data Analyzer (CAD)

THE FIRST 2 CLINICAL SEQUENCING LABORATORIES

- ⇒ Selected through a call for proposals launched by the Ministry of Health
- ⇒ Generic activity: rare diseases and cancers
- ⇒ Clinical laboratories
- ⇒ Access for all patients in France
- ⇒ Integration into the patient's healthcare pathway and the existing healthcare networks



Rare diseases: WGS of trios

Cancers : tumour WGS, WES
and RNAseq + germline WGS

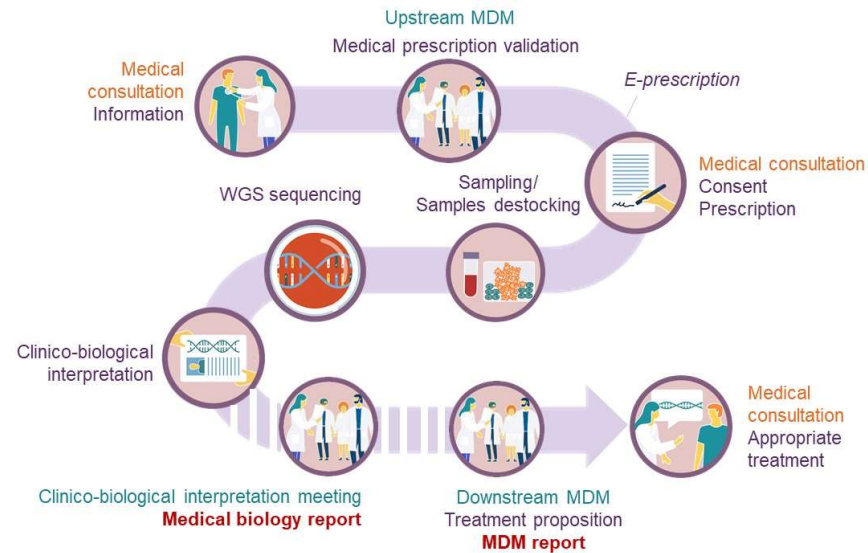


CLINICAL PREINDICATIONS



- Clinical WGS prescription is possible for well defined preindications
 - 5 rounds of selection have been organised to date
 - **82 preindications:**
 - ✓ 67 for rare diseases
 - ✓ 4 for cancer genetic predisposition
 - ✓ 11 for cancers
 - At term, these preindications will be evaluated by the French HTA agency (HAS) for long-term reimbursement.
 - Seqogen: national medico-economic evaluation project funded by MoH
-

MEDICAL PATHWAY FOR PREINDICATIONS



For each preindication:

- Structuration and setting up of Multi-Disciplinary Meetings
- Organisation of samples circuits
- Definition of the diagnostic strategy
- Vetting of prescribers
- Gestion of clinical biologists

CONTINUOUS IMPROVEMENT OF THE GENOMIC PATHWAY

Several simplified versions of information leaflets

Creation of local non-thematic MDM for rare diseases

Upstream MDM

Medical prescription validation

Medical consultation
Information



WGS sequencing



Sampling/
Samples destocking

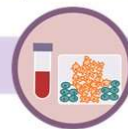
E-prescription



Medical consultation
Consent
Prescription

Pilot study for FFPE samples for cancer patients

Clinico-biological
interpretation



Clinical interpretation meeting
Medical biology report



Downstream MDM
Treatment proposition
MDM report

Medical consultation
Appropriate
treatment



Interpretation can be carried out by clinical biologists from any hospital

Structuration
of a network
of 51
genomic
pathway
managers



Several versions of information notices:

- ✓ for tumor genetics and germline genetics
- ✓ for adults and children
- ✓ suitable for different levels of understanding
- ✓ translated into several languages for non-native French speakers

FRANCE MÉDECINE GÉNOMIQUE 2025
avieson

Notice d'information pour les examens de génétique constitutionnelle

Version mineurs
Janvier 2023

6 - Qu'est-ce que le séquençage du génome ?

L'examen génétique qui vous est proposé consiste à séquencer l'ensemble de votre ADN (génome), c'est-à-dire le lire. L'objectif est de repérer les variants génétiques pouvant expliquer votre maladie, trouble ou handicap.

Bien que la lecture de l'ADN soit aujourd'hui possible, son interprétation reste difficile et limitée aux connaissances actuelles.

Les nouvelles technologies de séquençage produisent de grandes quantités de données, analysées grâce à des outils informatiques puissants combinés à l'expertise de différents spécialistes.

Étant donné le grand nombre de variants dans l'ADN de chaque individu, le défi consiste à identifier le variant en cause dans la maladie, le trouble ou le handicap. Cela revient à chercher une aiguille dans une botte de foin.

Afin de faciliter cette interprétation, il est souvent nécessaire d'étudier l'ADN des parents, et plus rarement celui d'autres membres de la famille.

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FRANCE MÉDECINE GÉNOMIQUE 2025
avieson

Notice d'information pour les examens de génétique constitutionnelle

Mineurs
version illustrée
Janvier 2023

Voici tes résultats

Le docteur va peut-être proposer à tes parents de faire le test génétique.

Lors d'une prochaine consultation, le docteur te dira si on peut expliquer tes difficultés ou pas.

9

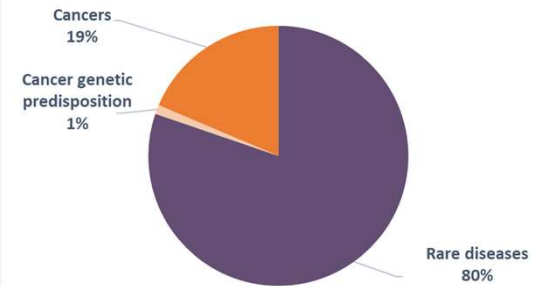


ACTIVITY REPORT AS OF 31 DECEMBER 2025

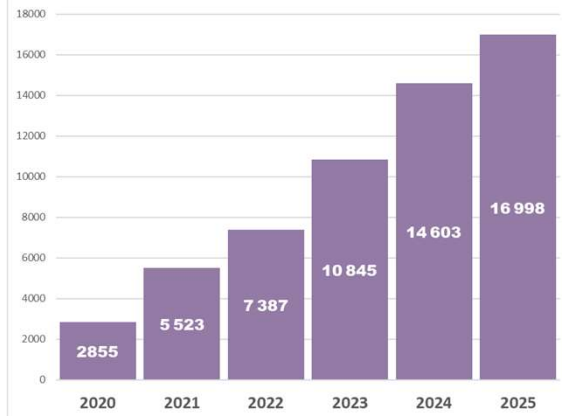
Since 2019 :

- 58,200 requests
- 44,200 clinical reports
- > 115,000 individuals

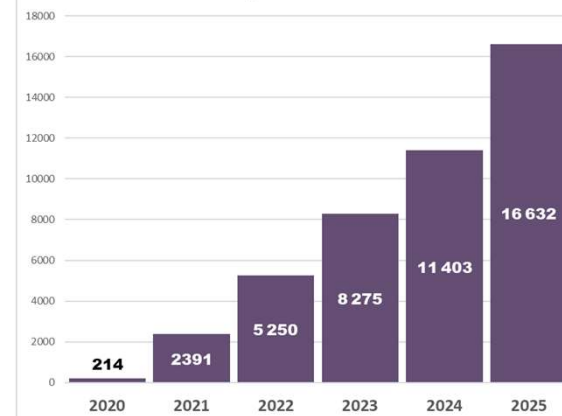
Distribution of medical prescriptions



Number of prescriptions
All preindications



Number of medical reports
All preindications

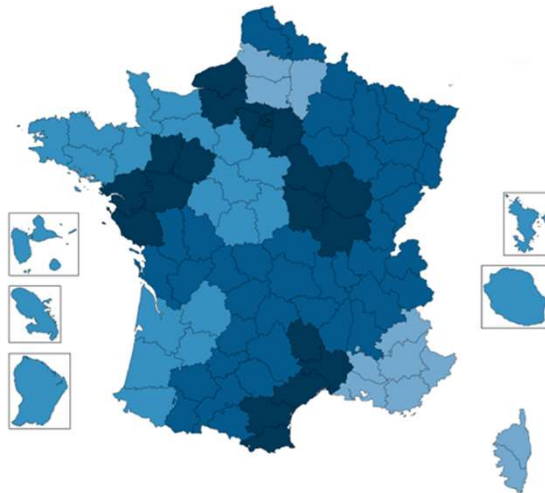




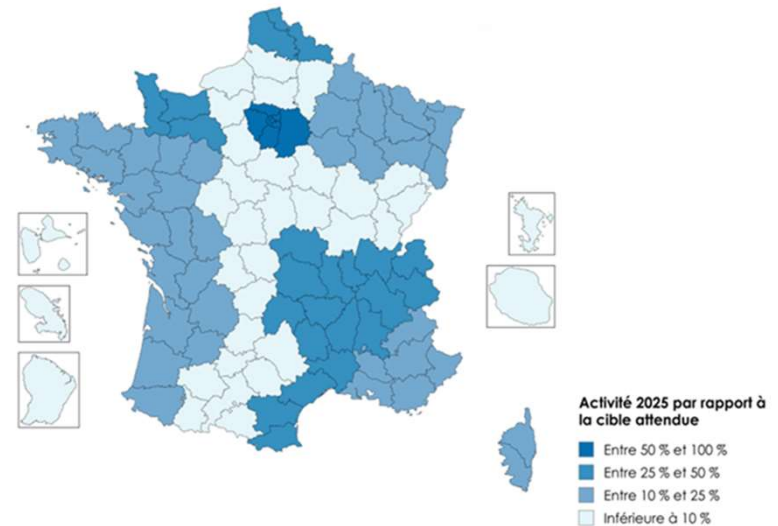
GEOGRAPHICAL DISTRIBUTION OF PRESCRIPTIONS



Rare diseases – 2025



Cancers – 2025





SECONDARY USE OF GENOMIC DATA

Dynamic of open science: wide opening of data, while ensuring their security and respecting scientific and ethical criteria





PROGRESS OF DATA REUSE FOR RESEARCH



2 projects have access to PFMG2025 data and have begun their first reanalyses.



■ **3 projects :**

- ✓ Data transferred to CAD
- ✓ Bioinformatic tools are being prepared for use in a secure space



INTERNATIONAL COLLABORATIONS

Participation in the European 1+ Million Genomes Initiative

Objective : Enabling secured access to genomic data and the associated clinical data in Europe for research, healthcare and public health.

- ✓ Genomic Data Infrastructure



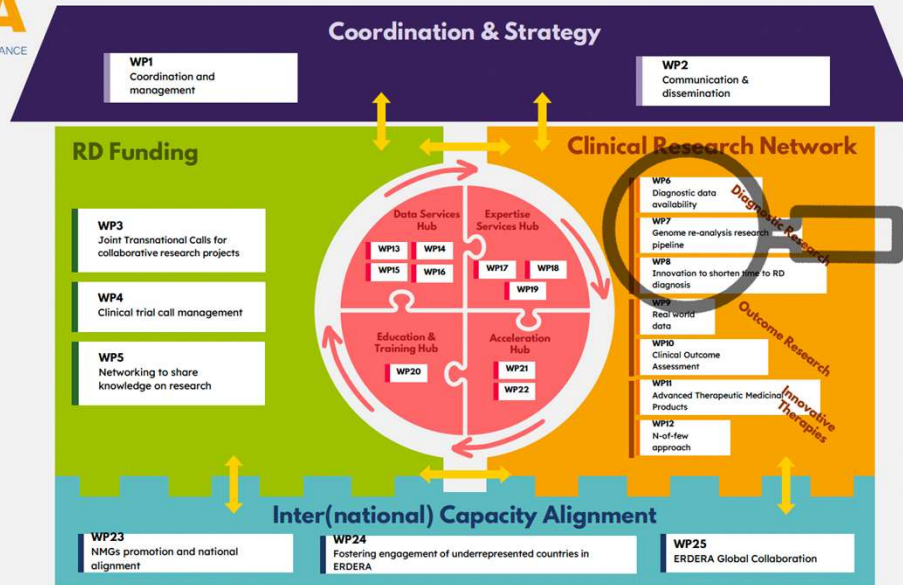
- ✓ Genome of Europe



- ✓ Co-pilot with Portugal of 1+MG WG7 : Implementation of genomics in healthcare



EUROPEAN RARE DISEASES RESEARCH ALLIANCE



Objective:

Genomic data reanalyses for patients with a negative result, as an extension of the Solve-RD initiative.

For PFMG2025: patients with a negative WGS result

WP13 Rare Diseases-Virtual Platform (RD-VP): Finding and accessing the data ecosystem	WP16 Knowledge bases and ontologies for RD research	WP19 Methodological Support
WP14 Data readiness services	WP17 Mentoring and consultancy	WP20 Education and training in rare diseases research
WP15 Data sharing and analysis services	WP18 Regulatory support service	WP21 Technology accelerator
		WP22 Public-Private Collaboration Accelerator

COLLABORATION WITH OTHER EUROPEAN GENOMIC MEDICINE INITIATIVES

Objective: Sharing experiences and best practices

- **Collaboration agreements :**
 - ✓ 2018: Genomics England
 - ✓ 2021 & 2024: Genomic Medicine Sweden
 - ✓ 2023: Danish National Genome Center
 - ✓ 2025: The Hartwig Medical Foundation (Netherlands)
- **A collaboration agreement in preparation with:**
 - ✓ The NHS Genomic Medicine Service (UK)





CONCLUSIONS



Ten years after its launch, *PFMG2025* has successfully integrated genome sequencing into the French healthcare system for rare diseases and cancers



The Lancet Regional Health - Europe
Available online 6 January 2025, 101183
In Press, Corrected Proof [What's this?](#)

ELSEVIER

Health Policy

PFMG2025–integrating genomic medicine into the national healthcare system in France

PFMG2025 contributors^a

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Remaining challenges:

- Low number of prescriptions for cancer patients
- Clinical biological interpretation and turnaround time
- Complexity of the genomic medical pathway
- Involvement of patients and citizens
- Data sharing & data access for research purposes



Towards the implementation of a “2030 or 2035 French Genomic medicine initiative”

- ⇒ Evaluation of the 2025 French Genomic Medicine Initiative
 - ⇒ Setting up of working groups to draft the second phase of the initiative
-

