Third French National Plan for Rare Diseases: a diagnosis observatory overview

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The French National Plan for Rare Diseases

The General Directorate of Health Care Provider (DGOS)

MINISTÈRE DU TRAVAIL DE LA SANTÉ ET DES SOLIDARITÉS

3 million per year (2018 – 2022)

Action 1.7 of the PNMR3

Reduced diagnostic delays and undiagnosed diseases

The strategy

Reassess the cases of currently undiagnosed patients in order to obtain a diagnosis in light of scientific advances

in the Rare Disease Reference Centres (CRMR)

by compiling a dynamic database

French national rare diseases registry (BNDMR)

with the support of the 23 (FSMR)

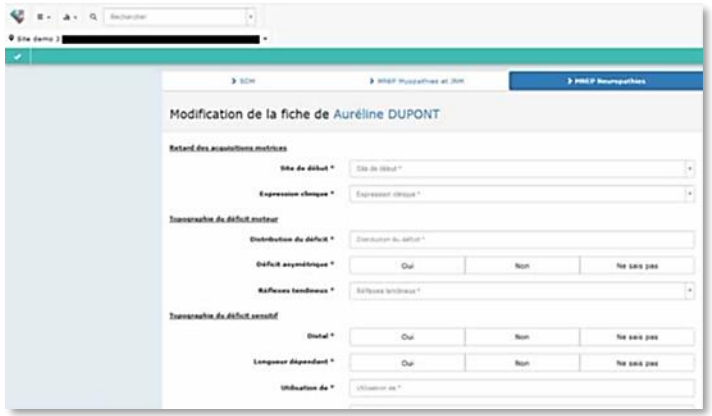
French Rare Diseases Networks

The BNDMR - created during the PNMR 2

Minimal Data Set (MDS) : www.bndmr.fr/publications/le-set-de-donnees-minimal

The FSMRs :

- are committed to entering this MDS in all the CRMRs
- support them towards better data quality /completion
- some collect additional data in BaMaRa for patients without a diagnosis

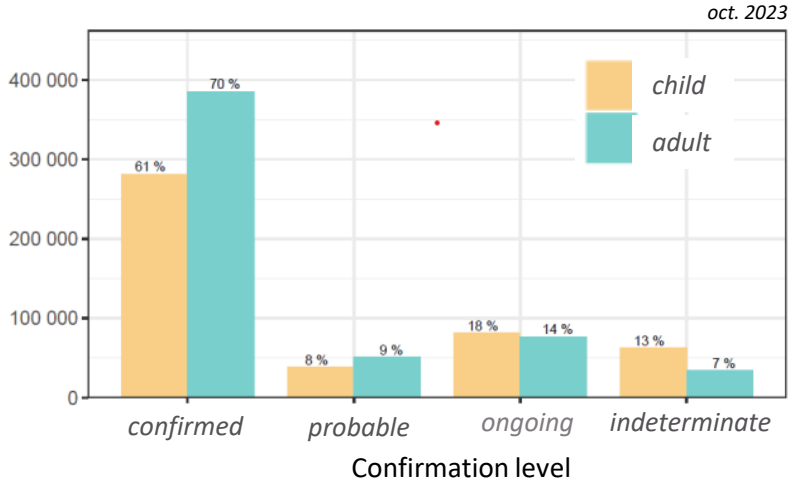


The diagnosis observatory

- to follow the diagnostic delay and undiagnosed diseases situations
- to assess the impact of the measures taken

⇨ based on annual indicators from the database

For example, number of patients per diagnosis assertion level



to ensure consistency of practices and the incorporation of recent discoveries in medical care

The overwiew

https://solidarites-sante.gouv.fr/soins-et-maladies/prises-en-charge-specialisees/maladies-rares/article/l-observatoire-du-diagnostic

More than 100,000 patient records were reviewed

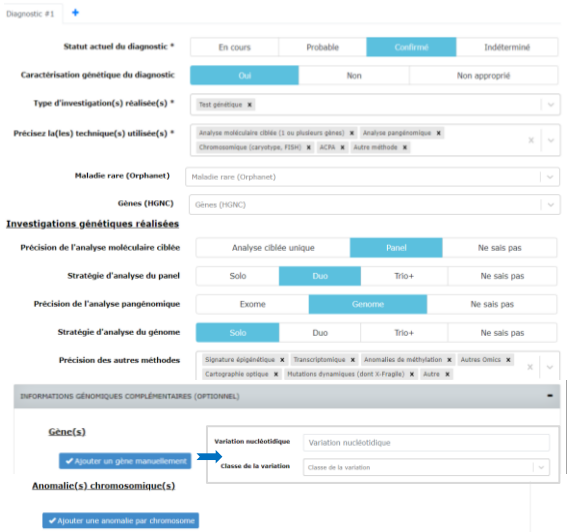
More than 300 people (especially clinical research associates and technicians) were recruited

More than 1400 people trained

More than 200 documents (various coding guides, diseases or Orpha codes list,..) created and 141 revised

5 dedicated data (and 1 to come) collections for patient without a diagnosis

MDS expansion with genomic items: distinction between clinical and genetic diagnosis, deeper genetic tests precisions and genomic anomalies description



www.bndmr.fr/les-donnees-collectees/le-sdm-g-genomique/

Numerous research projects and with patients associations

This work will be amplified during the next French National Rare Disease Plan 4

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