

Third French National Plan for Rare Diseases: establishment of a diagnosis observatory



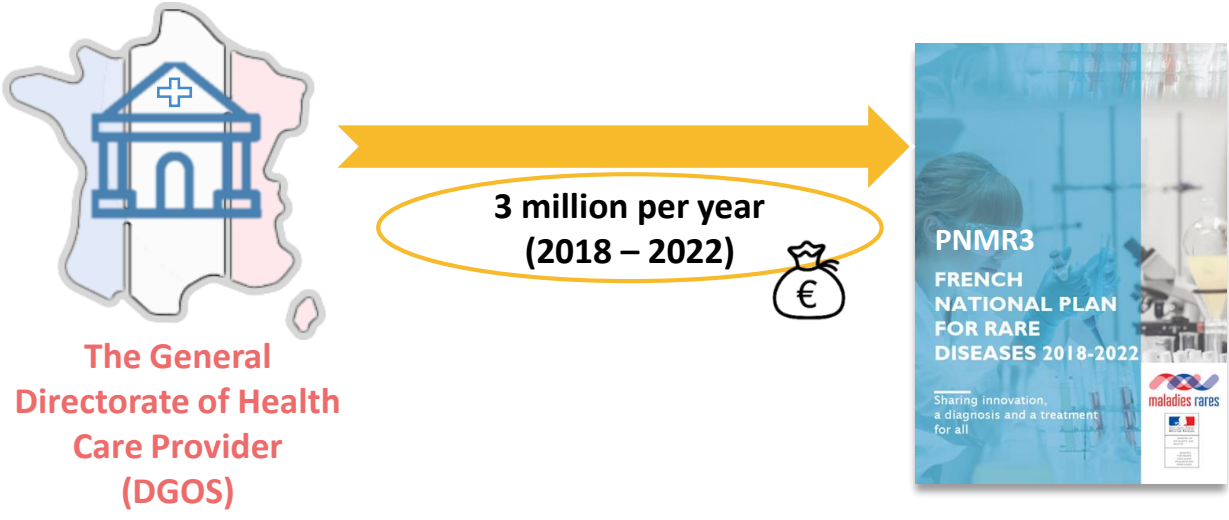
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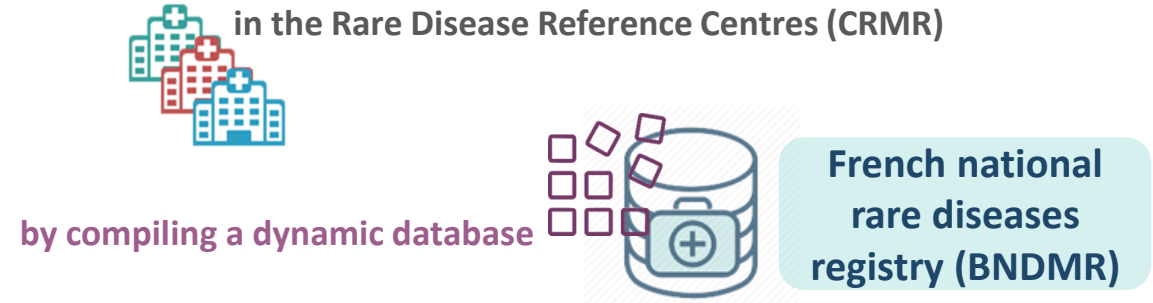
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The French National Plan for Rare Diseases

The strategy



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



Action 1.7 of the PNMR3

Reduced diagnostic delays and undiagnosed diseases

with the support of the 23 (FSMR)

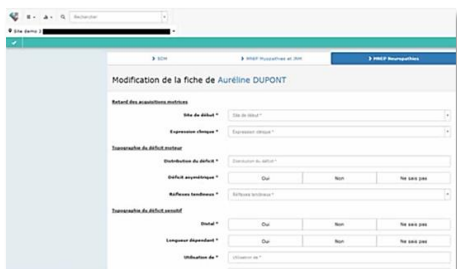




Minimal Data Set (MDS) : www.bndmr.fr/publications/le-set-de-donnees-minimal

The FSMRs :

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



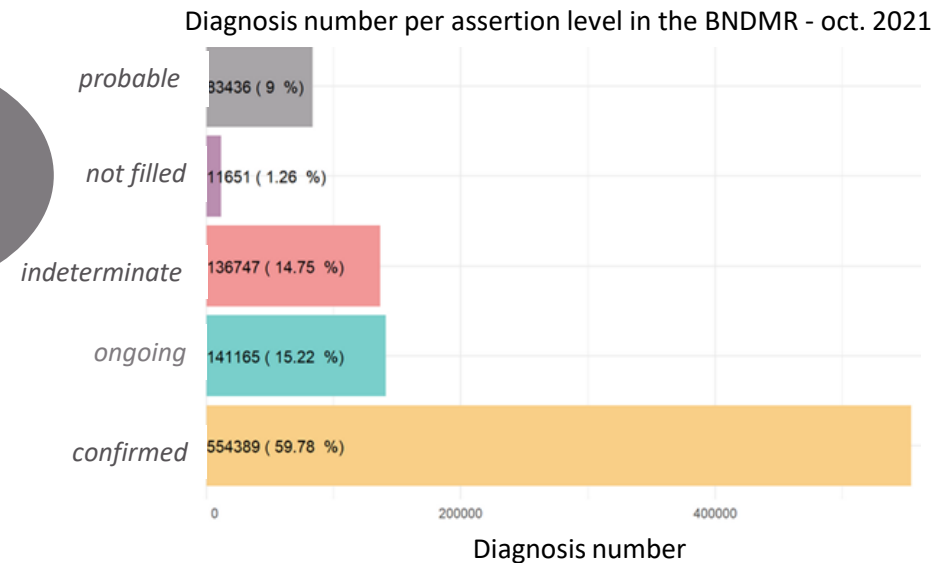
More than 50 people (especially clinical research associates and technicians) were recruited



- 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, distribution of the diagnosis confirmation levels



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



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