The French national registry for rare diseases: A whole rare disease registry at national level meeting challenges at EU level

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Context and objectives
The French national registry for rare diseases (BNDMR) was created as part of the second National Plan for Rare Diseases. Its aim is to collect from all expert centers on rare diseases in France a Minimal Data Set (MDS), either from a web app (BaMaRa) or from the electronic health record (EHR). In the end, these data are sent to the BNDMR data warehouse to allow national analyses.

The BNDMR team is also committed to directly make possible the upload of its data to the forthcoming ERN registries and CPMS platform to avoid multiple inputs for the same data for professionals.

Results (March 2020)

The RD French minimum data set (MDS)
The French MDS contains about 60 items, including:
- Patient identification
- Vital status
- Care pathway & activities
- Ante and neonatal data
- Treatment (orphan drugs)
- Diagnosis (ORPHAcodes)
- Phenotype (HPO / ICD-10)
- Genotype (HGNC / HGVS)

It was the source of the Common data elements of the European Commission constituting the core of ERN registries, therefore it natively overlaps with it.

Study requests made on the BNDMR data
30 applications received in 2019
- 12 during the first trimester 2020

Various stakeholders ask for BNDMR data

Granularity level of the data requests

The participating network
2 700 professionals involves in the data collection
(physician or paramedical staff, secretory, clinical research associate...)
67 hospitals including overseas
820 expert centres actively collecting data

The database content
700 000 patients (3.2% declared deceased)
22 700 healthy carriers
37 500 foetuses
33% deceased
37 500 foetuses

The rare diseases diagnosis
4 640 diagnostic terms (orphanet acronym)
More than half of the diagnoses concern less than 10 cases

Distribution of diagnoses
by number of cases (in classes)

Level of diagnostic confirmation

Confirmed diagnosis in only 1 out of 2 cases

*requires a unique national identifier (INS)