

The French national registry for rare diseases: feedback from a FAIRification process



BNDMR

Banque Nationale de Données Maladies Rares

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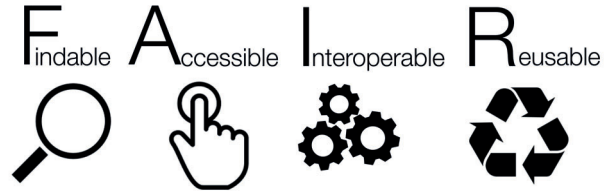
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► What is the F.A.I.R. process?

It's to construct, store, present or publish the data to enable them to be « Findable, Accessible, Interoperable and Reusable ».

It is a process of sharing and opening up data, promoted within the framework of open science.

The F.A.I.R. principles were described in the following originator publication: Wilkinson MD et al., **The FAIR Guiding Principles for scientific data management and stewardship**. *Sci Data*. 2016 Mar 15;3:160018. doi: 10.1038/sdata.2016.18. || *Addendum in: Sci Data*. 2019 Mar 19;6(1):6.



More information on the FAIR data principles: <https://www.go-fair.org/fair-principles/>

► The BNDMR FAIRification process

Description of the types of data and their collection format (metadata) of the Rare Disease Minimum Data Set (MDS)

- BNDMR website : <http://www.bndmr.fr>
- Interoperability framework of Health Information Systems Rare diseases component designed by the French Agency for Digital Health <https://esante.gouv.fr/volet-sdm-mr-set-de-donnees-maladies-rares>
- Metadata made public on the Central Metadata Repository (ERDRI.mdr) website <https://eu-rd-platform.jrc.ec.europa.eu/mdr/>



« Metadata and data should be easy to find for both humans and computers. Machine-readable metadata are essential for automatic discovery of datasets and services.»

Findable

Interoperable

«The data usually need to be integrated with other data. In addition, the data need to interoperate with applications or workflows for analysis, storage, and processing.»

- Use of international standards and controlled vocabularies for coding information and exchanging data :
=> medical repositories : HPO, Orphanet, HGNC, orphans drugs...
=> countries, city codes, hospitals, rare diseases sites repositories...
- 1 data element = 1 unique identifier / pseudonymized data with the IdMR*
- Data transmitted from hospitals to the BNDMR in a HL7 promoted format in accordance with several ISO standards: CDA (R2 level 3)
- Implementation of the common OMOP data model (used by many data warehouses) in the BNDMR for combined data processing



Standardized procedures for accessing BNDMR data

- => Procedures defined by the BNDMR Scientific Committee
- => Standard documents to be completed and sent to analyse.bndmr@aphp.fr
- => Application assessment by the BNDMR team and Scientific Committee

« Once the user finds the required data, she/he needs to know how can they be accessed, possibly including authentication and authorisation.»

Accessible

Reusable

«The ultimate goal of FAIR is to optimise the reuse of data. To achieve this, metadata and data should be well-described so that they can be replicated and/or combined in different settings.»

- Metadata and data precise description in the MDS and in the Interoperability framework of Health Information Systems Rare diseases component
- Each nomenclature license described in the Interoperability framework of Health Information Systems Rare diseases component designed by the French Agency for Digital Health (ASIP-Santé)
- MDS distributed under Creative Commons license

► Conclusion

No database is completely F.A.I.R. However, assessing the level of maturity, on the principles mentioned above, makes it possible to identify the missing bricks. This to allow a wider sharing of knowledge, in the strictest respect for the rights of patients and the general data protection regulations (GDPR).

The objectives for the project are the improvement of its FAIRification, a better readability of the data by machines, a transparent communication on the authentication and authorization policy, but also a better documentation of its sources.

The BNDMR is part of the current European dynamic, promoted by the European Joint Programm on Rare Diseases (EJP-RD) and the European Commission (JRC), alongside with the ERN registers.

Consult the rare disease Minimum Data Set on:

<http://www.bndmr.fr/le-set-de-donnees-minimal/>

*IdMR: rare diseases identifier

Methodolgy published in: M.Maaroufi et al., **Federating patients identities: the case of rare diseases**. *Orphanet J Rare Dis*. 2018 Nov 12;13(1):199. doi: 10.1186/s13023-018-0948-6



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