From the French national rare disease registry (BNDMR) towards ITHACA ERN registry (ILIAD):

Data reusability to ease the burden of data entry

Arnaud Sandrin[1], Céline Angin[1], Morris Swertz[2], Fernanda De Andrade[2], Klea Vyshka[3,4], Alain Verloes[3,5]

contact.bndmr@aphp.fr

[1] French National Rare Disease Registry (BNDMR) - Assistance Publique-Hôpitaux de Paris, Paris, France | [2] University Medical Center Groningen, Dept. of Genetics, Genomics Coordination Center, Groningen, Netherlands | [3] Assistance Publique-Hôpitaux de Paris - Université de Paris, Department of Genetics, Paris, France | [4] CERCRID, UMR 5137, "Centre de Recherches Critiques en Droit", Université de Lyon, Lyon, France | [5] INSERM UMR 1141 "NeuroDiderot", Hôpital R DEBRE, Paris, France

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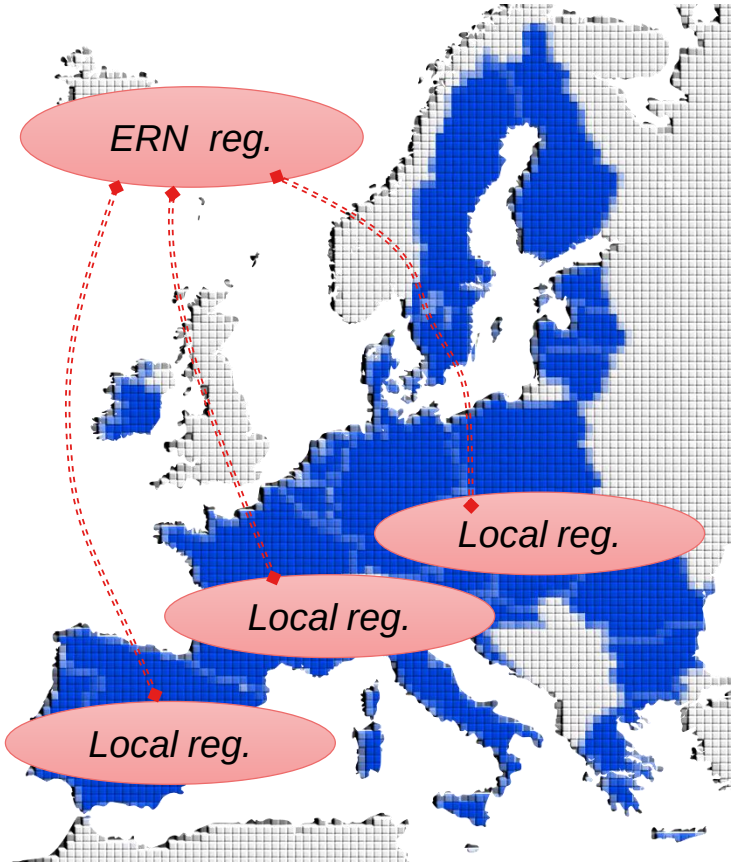
All European Reference Networks (ERN) are currently developing registries in order to collect or make Findable, Accessible, Reusable and Interoperable (FAIR) the **common data elements (CDE)** required by the European Joint Research Consortium (JRC) are



Connecting such a large number of local registries altogether is an unprecedented interoperability effort. It faces significant challenges of various sorts :

- **Regulatory** : patient re-information *and* inter-HCPs contracts requiring law and data protection regulation expertise
- **IT :** compliancy with interoperability international standards requires highly technical IT development and infrastructure expertise
- **Data :** managing inconsistencies, data duplication, data source tracing is mandatory in a fully automated system

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Data reusability to avoid double entry of the same data for the same patients

To mimic double entry of hundreds of patients data : a short-run pragmatic alternative to longer-run fully-fledged FAIR system

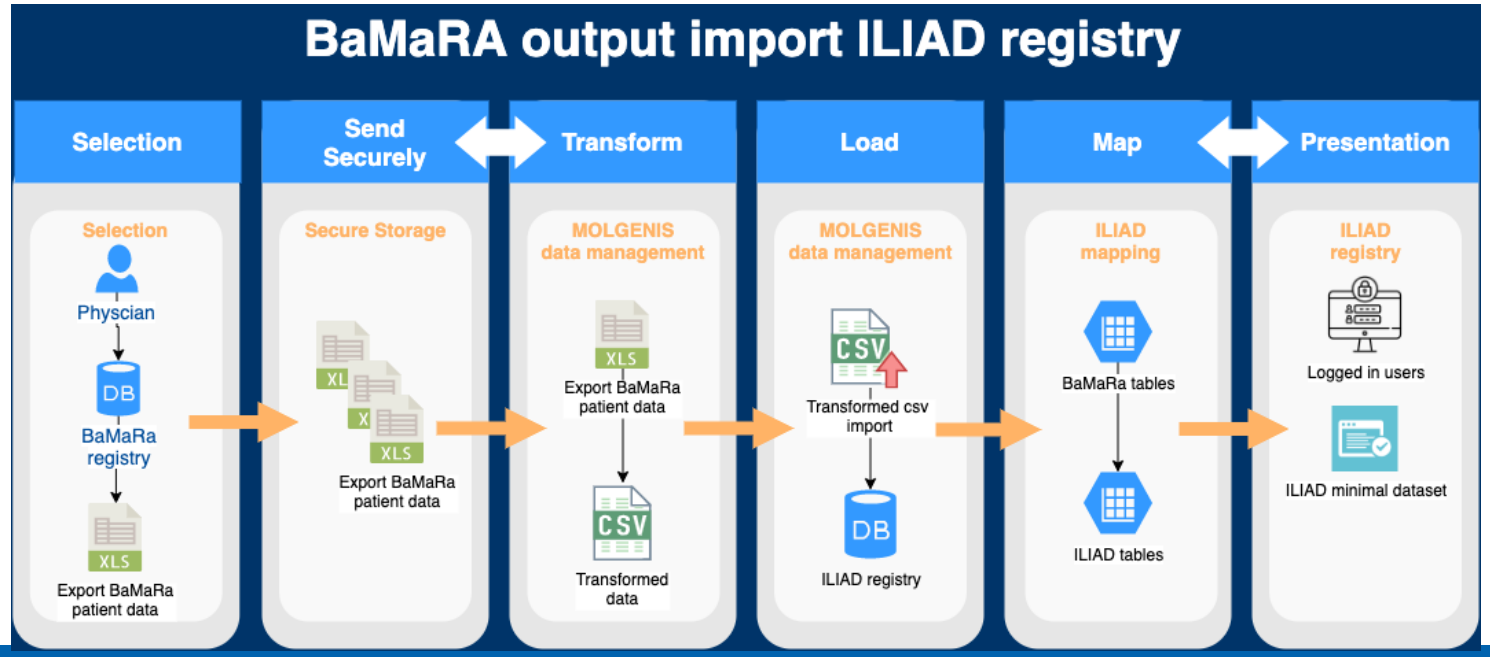
ERN ITHACA is developing a “meta-registry” called ILIAD, connecting 71 HCPs, databases, and biobanks across the EU for patients with dysmorphic/MCA syndromes and/or intellectual disability. The registry is built on MOLGENIS open-source software, providing flexible rich data structures, user-friendly data import and querying, and FAIR interfaces for programmatic data exchange. **Collected data include the CDE.**

In France, all Rare Disease (RD) expert centers patients are registered in the National RD Registry (BNDMR). Data is collected through HCP local IT systems and/or via a web application called BaMaRa (interoperability between HCPs and BaMaRa is based on HL7 CDA standards). The system is developed at AP-HP where data is accessible in a national data warehouse. **Collected data also include the CDE.**

In the BNDMR collection tool (BaMaRa), patient data can be downloaded in an Excel file, that is uploaded securely on the Molgenis platform where it is transformed (including translation, format reconciliation and pseudonimisation). The new file is then loaded in the ILIAD registry and mapped with available items so data can be displayed for users.

→ **No data transfer *per se***

→ **Same and simple and safe as double entrying, but for hundreds of patients in only few clicks**



Perspectives about patient information, benefits of the approach

Patients information (RGPD) : what about retrospectively included patients ?

Many patients are already included in local registries and are not likely to make another visit at the expert center which included them. Therefore, they have not received information about ERN registries.

Having informed that data feeds into BaMaRa does *not* mean the data can then feed into ILIAD. Indeed : patients *must* be informed individually by the clinicians about the ILIAD registry and a consent will be needed to allow the reuse of data in ILIAD.

GDPR article 14(5)(a) and (b) allows alleviating the information process in some cases, e.g. if “the provision of such information proves impossible or would involve a disproportionate effort”. Discussion with local authorities should be undertaken, no conclusion is yet available.

A pragmatic approach : cutting double-entry burden, enhancing epidemiology opportunities in the short-run, while preserving high security standards

No double-entry, a clinician can download local registry data and upload it within ERN registry at a taylor-made frequency (every 6 months, e.g.), depending on ERN research topics.

Data is effectively accessible at an ERN registry in the short-run, research project can be launched at European level on rare diseases epidemiology.

The process is secure : there is no digital interface between the two systems (BNDMR and ILIAD), which minimizes cyber-risks.

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