



# Tracking auto-immune diseases in electronic health record.

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## 1 - Motivations

The collection of biomedical data requires a multitude of software tools for purposes ranging from websites for patients' follow-ups to alarm algorithm for medical monitoring by physicians or large scale collection of data for epidemiological analyses. Yet many more opportunities remain to leverage technology to better patients' care. Vast amount of human and technical efforts have already been spent on genetic disorders and the emergence of deep-sequencing analyses for patients' samples has led to significant progress. On the opposite, little or no attention has been paid to autoimmune diseases in patients' medical records.

## 3 - Proposed solution

The aim is to develop a comprehensive thesaurus of all autoantibodies found in patients. To date, more than 200 different autoantibodies has been identified by the 23 FSMR (Filières de Santé Maladies Rares) and one ERN (European Reference Network) who contributed to the development of this list. All these items are then implemented in the HPO (human phenotype ontology) database. A unique code will then be assigned which can be used to complement the patient's diagnosis. For example, in the National Data Bank for Rare Diseases, these codes can be used in the clinical description field.

## 4 - Dataset Description

The dataset is very straightforward and contains all terms/names of all autoantibodies reported by the physicians of the expert filières (and a data curation on Pubmed), i.e. more than 200 terms, the associated HPO code, the synonyms (based on literature, Pubmed), and the ORPHA code of the associated disease if known.

## 2 - Problem statement

(Rare) auto-immune diseases, which can affect multiple organs simultaneously, have yet to be thoroughly investigated and effectively reported in the electronic health record.

In the last years, MDs reported the lack of available tools to properly declare the autoimmune diagnosis of patients in health care databases. An accurate tool will allow better information into the health databases and help in the follow-up of patients and would allow better epidemiological monitoring.

## 5 - Related Work

The objective is to model knowledge on auto-antibodies using different sources of data that will be combined into knowledge graphs. Different approaches based on knowledge graphs models will be used and compared (different types of distance, different treatments of multimodal information). The different sources of data that will be used as knowledge source will be: The French BNDMR, ORDO (Orphanet data), Pubmed and the EDS (French entrepôt de données de santé)

#RAREDISEASES

#AUTOIMMUNITY

#healthrecord

## Results

Altogether, those data will shed light on auto-immune diseases and their respective epidemiology. This project is the result of a global collaboration of the 23 FSMR, Orphanet and the HPO database and will shortly result in a publication. This work will drive the attention of all physicians and health care providers to increase knowledge on the association between phenotypes and auto-antibodies.

