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# Context-Sensitive Common Data Models for Genetic Rare Diseases – A Concept

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**Abstract.** Current challenges of rare diseases need to involve patients, physicians, and the research community to generate new insights on comprehensive patient cohorts. Interestingly, the integration of patient context has been insufficiently considered, but might tremendously improve the accuracy of predictive models for individual patients. Here, we conceptualized an extension of the European Platform for Rare Disease Registration data model with contextual factors. This extended model can serve as an enhanced baseline and is well-suited for analyses using artificial intelligence models for improved predictions. The study is an initial result that will develop context-sensitive common data models for genetic rare diseases.

**Keywords.** Common Data Model, Rare Disease, Context-Sensitive

## 1. Introduction

Rare Diseases (RD) pose a tridimensional challenge involving patients, physicians, and the research community. Physicians have dispersed encounters with RD patients, which together with lack of diagnosis/therapy procedure and medications lead to patients' often struggle to find a specialist (1), or researchers a reasonable cohort for a study (2). Here, Common Data Models (CDMs) facilitated data standardization and integration for both rare and common diseases. Prominent examples are the Observational Medical Outcomes Partnership (OMOP) CDM and the European Platform for Rare Disease Registration Common Dataset (ERDRI CDS) (3) that introduced Common Data Elements (CDE). We propose a concept to extend the existing ERDRI CDS with Contextual factors (CFs), which include patient, physician, and environment-related parameters that can greatly impact the decision-making process (4). This article is part of an interdisciplinary project that develops a generalizable and context-sensitive CDM for genetic rare diseases (GRDs) that is also suitable for Artificial Intelligence (AI) analyses. In brief, current methods for developing a CDM in the health domain were recently reviewed (5) and applicability of OMOP CDMs hosting genomic data and performing analytics on the data was shown (6).

## 2. Method

The CFs especially for RDs were extracted from the literature indexed in PubMed in a scoping review method. A detailed description of the study components and extracted elements can be obtained from our OSF protocol (7). The elements are aggregated into groups and discussed in an interdisciplinary team until consensus was reached and then compared to the ERDRI CDS. As shown in Figure 1A, a CDM including CFs will be developed as part of the SATURN project (https://www.saturn-projekt.de/).

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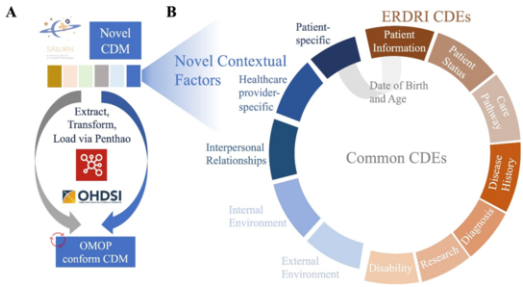


Figure 1: (A) Dissertation method overview, (B) comparison of CFs with the elements in ERDRI. Only two patient-specific CFs (date of birth and age) are part of ERDRI CDS.

## 3. Results

We identified three groups of CFs (Figure 1) in the literature: A) Person-related factors: patient-specific and healthcare provider specific. B) Factors of the immediate environment: interpersonal relationships. C) Organization-related factors: internal environment and external environment. The comparison results between the CFs and the ERDRI CDS are summarized in Figure 1B. A full list of the CFs can be found in (7).

## 4. Discussion

Despite advancements in the field of RDs, patients still go through an odyssey before getting diagnosed. It is the large number of RDs that pose a huge challenge. The inclusion of patient context through the CDM might increase the accuracy of the decision support models. Therefore, the extended ERDRI CDS for registries and points of care can be a basis and motivation for structured documentation of these contextual elements.

## 5. Conclusion

We have conceptualized an extension of ERDRI CDS with our CFs that can be used on the registry level to collect patient data. We will develop a context-sensitive RD-CDM model that might increase the documentation of meta and genomic data for GRDs, which can be used for conducting subsequent analyses via AI approaches.

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