FINDING THE NEEDLE IN THE HAY STACK: AN OPEN ARCHITECTURE TO SUPPORT DIAGNOSIS OF UNDIAGNOSED PATIENTS

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INTRODUCTION

A big challenge with rare diseases (RD) is to find the correct diagnosis for a patient. A study by the EU showed that 25% of the patients waited between 5 and 30 years for a correct diagnosis. In Europe, a disease is declared as “rare” if less than 5 out of 10,000 people are affected [1]. To tackle the problem of undiagnosed patients with RD, Clinical Decision Support Systems (CDSS) are promising. This work focuses on a software-based architecture for a CDSS for RD, which uses distributed clinical data from ten university hospitals in Germany. The concept and the development of this system are part of the MIRACUM consortium (Medical Informatics for Research and Care for University Medicine), which is funded within the Medical Informatics Funding Scheme by the German Federal Ministry of Education and Research (BMBF) [2]. Within MIRACUM, the university hospitals will establish Data Integration Centers (DIC) with the goal to improve collaborative research as well as clinical processes. A DIC will be established in the IT-infrastructure of each hospital which enables to exchange data among the partners in MIRACUM based on the principle of data federation. The CDSS for RD will perform a similarity analysis of an undiagnosed patient at the hospitals DIC to give a physician a hint to a possible diagnosis.

METHODS

To perform a data analysis at each MIRACUM site, the data of each site needs to be harmonized. To formally describe all data elements used for a similarity analysis, a Metadata Repository (MDR) is used, which is based on the international meta data standard ISO 11179 [3]. All data in the DIC will be described at each site with their MDR. Used data elements will be mapped to a common dataset, which is available in the local databases [4]. The common dataset will be based on Common Data Models (CDM) such as the OMOP CDM, which includes a standard representation of common vocabularies for coding of clinical concepts (e.g. SNOMED-CT) [5]. For data exchange between sites, HL7-FHIR is used [6].

RESULTS

A physician formulates a query for similarity analysis to find similar patients. The web application DISERDIS (Diagnosis Support in Rare Diseases) will be available at each site. The patient data is stored in a OMOP database which is periodically updated with data from the respective hospital information system of the site via ETL processes. When a similarity analysis is triggered at one site, a request is sent to the central MIRACUM searchbroker which provides the request to the respective local FHIR-Server. The clinical hospital IT networks usually block external access to the data. Therefore, the searchbroker is designed as a central request point allowing to forward the search request to the FHIR-Server which manages access to the data. The FHIR-Server is a REST API that can retrieve requests via corresponding FHIR resources. It retrieves the query from the searchbroker and submits it to the similarity engine which performs the similarity analysis on the database. The data is available in the OMOP-CDM and must be transformed to the FHIR resources. The result is returned to the FHIR-Server which sends the result of the similarity analysis to the searchbroker. The architecture is displayed in Figure 1.

CONCLUSION

This paper demonstrates a concept of a system-architecture establishing a diagnosis support system for RD based on distributed clinical data.

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REFERENCES