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

Jannik Schaaf[1], Martin Boeker[2], Thomas Ganslandt[3], Christian Haverkamp[4], Tim Hermann[5], Dennis Kadioglu[1], Hans-Ulrich Prokosch[6], Thomas O.F. Wagner[7], Michael von Wagner[8], Johanna Schaefer[1], Martin Sedlmayr[9], Holger Storf[1]

[1] Medical Informatics Group (MIG), University Hospital Frankfurt, Frankfurt am Main, Germany

[2] Institute of Medical Biometry and Statistics, Medical Faculty and Medical Center— University of Freiburg, Freiburg, Germany

[3] Departement of Biomedical Informatics, University Medicine Mannheim, Ruprech-Karls-University Heidelberg, Mannheim, Germany

[4] IT-Department, University Hospital Freiburg, Freiburg, Germany

[5] Institute for Biometry and Medical Informatics, Otto-von-Guericke-University Magdeburg, Magdeburg, Germany

[6] Chair of Medical Informatics, Departement of Medical Informatics, Biometrics and Epidemiology, Friedrich-Alexander University Erlangen-Nürnberg, Erlangen, Germany

[7] Frankfurt Reference Center for Rare Diseases, University Hospital Frankfurt, Frankfurt am Main, Germany

[8] Executive Departement of Medical IT-Systems and Digitalization, University Hospital Frankfurt, Frankfurt am Main, Germany

[9] Institute for Medical Informatics and Biometry, Carl Gustav Carus Faculty of Medicine Technische Universität Dresden, Dresden, Germany

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 the 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.

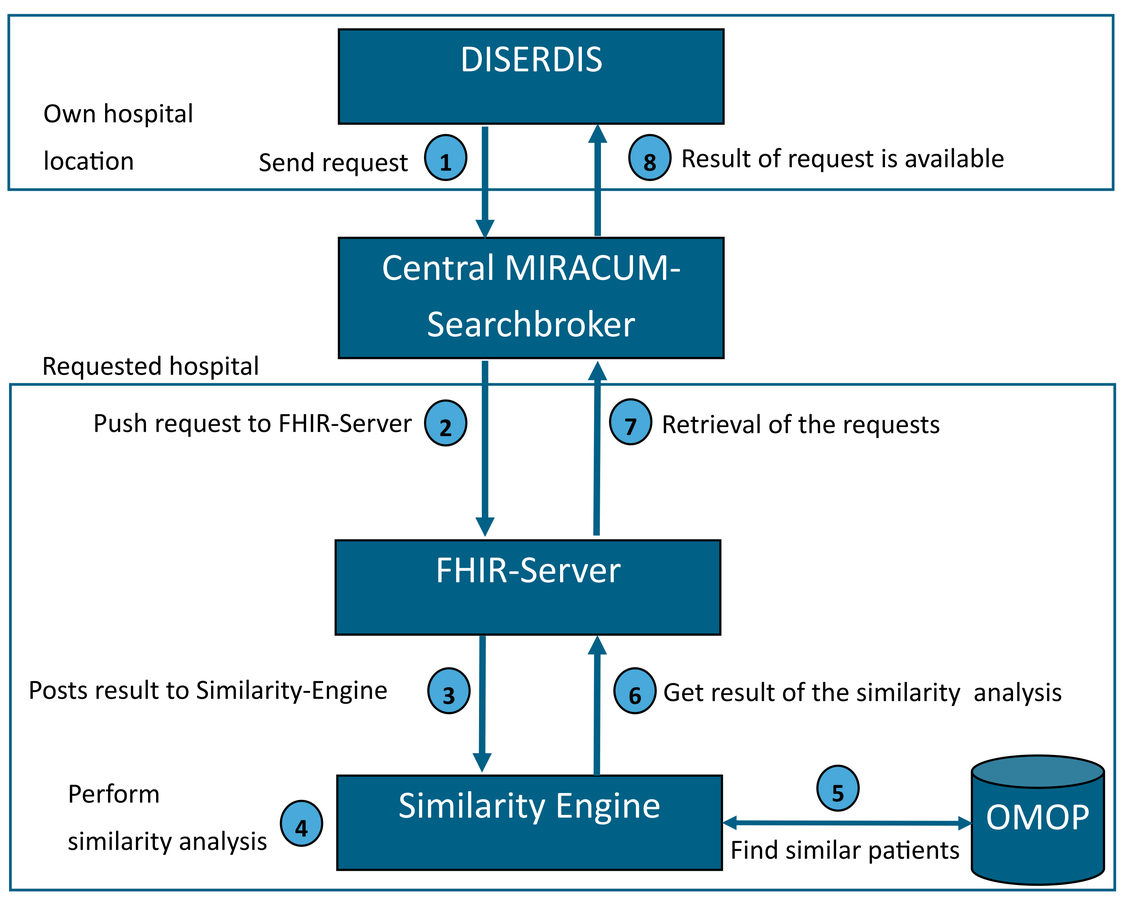

Figure 1: Detailed architecture for diagnosis support

CONCLUSION

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

CONTACT INFORMATION

Jannik Schaaf

schaaf@med.uni-frankfurt.de

Medical Informatics Group (MIG), Universitätsklinikum Frankfurt

www.mig-frankfurt.de

ACKNOWLEDGEMENTS

MIRACUM is funded by the German Ministry for Education and Research (BMBF), FKZ 01ZZ1801C.

REFERENCES

[1] R. Dragusin, C. Petu, C. Lioma, H. Jorgensen, I. Cox, L. Hansen, P. Ingwersen, and O. Winther, FindZebra: a search engine for rare diseases, Int J Med Inform. 82 (2013) 528–538.

[2] H. Prokosch, T. Acker, J. Bernarding, H. Binder, M. Boeker, M. Borries, P. Daumke, T. Ganslandt, J. Hesser, G. Höning, M. Neumaier, M. Marquardt, H. Lenz, H. Rothkötter, C. Schade-Brittinger, M. Sedlmayr, K. Sohrabi, and H. Storf, MIRACUM: Medical Informatics in Research and Care in University Medicine - A Large Data Sharing Network to Enhance Translational Research and Medical Care, Methods Inf Med. 57 (2018) 82–91.

[3] ISO/IEC 11179-1:2015, (2015). https://www.iso.org/standard/61932.html.

[4] D. Kadioglu, B. Breil, M. Lablans, S. Mate, D. Schlue, H. Serve, H. Storf, F. Ückert, T. Wagner, P. Weingardt, and H. Prokosch, Samply MDR - A Metadata Repository and Its Application in Various Research Net-works, Stud Health Technol Inform. (2018) 50–54.

[5] P. Stang, P. Ryan, J. Racoosin, J. Overhage, A. Hartzema, and C. Reich, Advancing the science for active surveillance: rationale and design for the observational medical outcomes partnership, Ann Intern Med. 153 (2010) 600–606.

[6] C. Mandel, D. Kreda, K. Mandl, I. Kohane, and R. Ra-moni, SMART on FHIR: a standards-based, interoperable apps plattform for electronic health records, J. Am. Med. Inform. Assoc. 23 (2016) 899–908