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RD-MON - Building a Rare Disease Monitor to Enhance Awareness for Patients with Rare Diseases in Intensive Care

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Abstract. Rare diseases are commonly defined by an incidence of less than 5/10000 inhabitants. There are some 8000 different rare diseases known. So even if a single rare disease is seldom, together they pose a relevant problem for diagnosis and treatment. This is especially true if a patient is treated for another common disease. University hospital of Gießen is part of the CORD-MI Project on rare diseases within the German Medical Informatics Initiative (MII) and a member of the MIRACUM consortium within the MII. As part of the ongoing Development for a clinical research study monitor within the use case 1 of MIRACUM, the study monitor has been configured to detect patients with rare diseases during their routine clinical encounters. The goal was to send a documentation request to the corresponding patient chart within the patient data management system for extended disease documentation to enhance clinical awareness for the patients' potential problems.

The project was started in late 2022 and has so far been successfully tuned to detect patients with Mucoviscidosis and place notifications within the patient chart of the patient data management system (PDMS) on intensive care units.

Keywords. Rare diseases, mucoviscidosis, study monitor, patient data management system, FHIR, OMOP

1. Introduction

Rare diseases are commonly defined by an incidence of less than 5 per 10000 inhabitants [1]. Rare diseases may pose problems if they go unnoticed during the treatment of other patient problems e.g., during a necessary surgical treatment with postoperative intensive care therapy or as part of a COVID 19 related intensive care treatment.
The University Hospital of Gießen (UHG) is part of the CORD-MI project on rare diseases in the German Medical Informatics Initiative (MII) [2] and a member of the MIRACUM consortium within the MII. As part of the ongoing Development for a research study monitor within the use case 1 of MIRACUM, the clinical study monitor tool has been configured to detect patients with rare diseases during their routine clinical encounters.

2. Methods and Results

To gain first experience with this concept, the study was limited to the ICD-10 code E84 for mucoviscidosis. Because UHG also operates a large pediatrics department and runs a special center for long term treatment of mucoviscidosis we were also able to compare visit frequencies for both the treatment center and other clinical departments.

To support the process of identifying patients with rare diseases, the MIRACUM clinical trial recruitment support system (CTRSS) [3] has been configured to identify patients with mucoviscidosis once they are readmitted for clinical treatment. The CTRSS is installed within the data context of the data integration center (DIC) at the UHG (

The CTRSS has been enhanced to deliver notifications to the patient data management system (PDMS) ICUdata on the intensive care units and activate a template for documentation of mucoviscidosis symptoms within the chart to enhance situational awareness for the disease symptoms and problems.

3. Discussion and Outlook

It could be shown that the CTRSS is able to detect patients with prior diagnosis of mucoviscidosis (ICD-10 E84) and could also decide whether a patient is treated on an intensive care ward.

Furthermore, it could be proven, that it is possible to place requests for further documentation automatically via HL7-messages from the CTRSS into the patient chart. According to the clinical guidelines for documentation within UHG wards, such requests must be fulfilled by the responsible clinicians within a working shift.

For further data analysis and system improvement a working group within the DIC consisting of a physician and a medical documentary has been established to analyze the resulting documentation and make adaptations to the documentation templates.

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