

5th International Conference on

Big Data Analysis and Data Mining

June 20-21, 2018 | Rome, Italy

Diagnosis support for patients with rare diseases based on cross-institutional clinical-data

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The Federal Ministry of Education and Research in Germany (BMBF) funds four consortia for establishing data integration centers (DIC) in University Hospitals, one of which is MIRACUM (Medical Informatics in Research and Care in University Medicine), with the goal of fostering linkage and exchange of data between the hospitals. One target is to support the diagnosis of patients without diagnosis or patients with rare diseases (RD). It is estimated that there are between 5,000 and 8,000 different RD. A challenge for patients with an RD is to be diagnosed by their general practitioner in a timely and correct way. Affected patients are often on a psychologically stressful journey from doctor to doctor with many suspected diagnoses and contradicting statements. A study conducted by the European Organization for RD (EURORDIS) showed that 40% of RD patients were wrongly diagnosed before the correct diagnosis was given. A variety of databases, web resources and clinical decision support systems have been developed providing information on RD. While those systems search the internet or dedicated disease databases, an alternative approach is to use a continuously updated database of earlier patient cases in order to find similar cases. The MIRACUM partner site Frankfurt will develop and validate a machine learning approach to find similar patients based on the clinical data of a patient stored within the DIC of all MIRACUM sites. These data include inter alia the patient history, diagnoses, lab results and genetic testing.

Biography

Jannik Schaaf is a PhD student at the Goethe University, Frankfurt Big Data Lab. He is employed at the Medical Informatics Group at the University Hospital Frankfurt and works on different projects in Data Integration and Machine Learning.

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