Press release

Berlin Institute of Health (BIH)
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02/27/2020
http://idw-online.de/en/news733842
Cooperation agreements, Transfer of Science or Research
Information technology, Medicine
transregional, national

Digital collaboration on rare diseases

Twenty university hospitals and other partner institutions have joined forces across Germany under the CORD-MI (Collaboration on Rare Diseases) project to improve patient care and research in the field of rare diseases. CORD-MI has been funded by the German Federal Ministry of Education and Research (BMBF) since February – to the tune of almost six million euros over two years. The project leverages the infrastructure of the BMBF's Medical Informatics Initiative (MII). Its aim is to facilitate the privacy-compliant sharing of information on rare diseases that arises from diagnosis, treatment and research throughout Germany. The project is led by the Berlin Institute of Health (BIH).

Rare diseases are not necessarily that rare: They affect some four million people in Germany alone. But of the estimated 8,000 different diseases that fall into this category, there are usually only a few recorded cases of each one. As a result, it can take years for those affected to receive the correct diagnosis, and in most cases there is a lack of effective treatment options and barely any research.

Many university hospitals already include centers for rare diseases that provide help for these patients. But some rare diseases are so rare that even centers such as these have only ever experienced a few cases. It is therefore all the more important that efficient use is made of the limited data that is available on such diseases. This is exactly where CORD-MI comes in. “We make sure that the BMBF's Medical Informatics Initiative also benefits the centers for rare diseases at university hospitals,” says Dr. Josef Schepers, deputy head of the BIH Core Unit eHealth and Interoperability. He is the spokesperson for CORD, the exemplary “use case” of rare diseases in the BMBF's Medical Informatics Funding Scheme. On February 1, 2020, the BMBF began funding CORD-MI with almost six million euros over two years. “Digital networking can be extremely helpful,” explains Schepers. “Especially for medical diagnoses that occur perhaps just 100 times throughout Germany.”

Shining a light on orphan diseases

“You can only improve what you can measure, and it is here that research into these so-called ‘orphan diseases’ already gets stuck, because they cannot be counted accurately using the usual diagnosis codes,” explains Schepers. “We urgently need rare diseases to be documented and classified using Orpha numbers in regular clinical care, so that the patient data can be used across various locations and in accordance with data protection regulations,” he continues. “The appropriate documentation and digital networking of the existing centers for rare diseases should improve the visibility of these orphan diseases and thus help to accelerate the diagnosis of those affected, develop adequate treatments, and promote research on rare diseases.”

CORD-MI focuses on a selection of rare diseases, including cystic fibrosis. “Cystic fibrosis is one of the more common rare diseases, with a correspondingly large data base that has been growing over a long period of time ,” explains Prof. Helge Hebestreit, a participating clinician from the University Hospital of Würzburg. “It therefore provides a perfect basis for demonstrating the added value of innovative data analyses across different locations.” Prof. Reinhard Berner from Dresden's Carl Gustav Carus University Hospital adds: “We want to demonstrate the added value that medical informatics also represents for people with particularly rare diseases. This means that we are also focusing on diseases that potentially affect only ten people in Germany.”

Uniform documentation required

The BIH Core Unit eHealth and Interoperability is involved in developing concepts that aim to ensure uniform documentation methods are used at as many university hospitals as possible. “Under the Medical Informatics Initiative, almost all German university hospitals are setting up data integration centers tasked with jointly developing concepts for documenting and sharing data in compliance with strict data protection rules. This opportunity should also be used for people with rare diseases,” says Schepers, outlining the task ahead. To achieve this, the parties involved will introduce the FAIR principles, which state that scientific data should be findable, accessible, interoperable and reusable. This way, CORD-MI is also contributing to the overall outcome of the Medical Informatics Initiative – for example, by testing innovative and privacy-compliant approaches to linking and analyzing data.

European-wide collaboration, too

Germany is, of course, not the only place where data on rare diseases is poorly managed. Rare diseases are a perfect example of an area of research that would greatly benefit from the networking of patients, researchers and physicians at the European and international level. The European Commission is therefore supporting the establishment of European Reference Networks (ERNs) for complex and rare diseases as part of the EU’s Horizon 2020 research and innovation program. “We are naturally making efforts through CORD-MI to also achieve the interoperability of patient data across Europe. In the case of many rare diseases, we can only understand what factors contribute to the disease and how it progresses by collaborating across borders. Many people working together can produce results that a few people working alone never could,” says Schepers.

About the Berlin Institute of Health (BIH)
The Berlin Institute of Health (BIH) is a biomedical research institution focusing on translational research and precision medicine. The BIH is dedicated to improving the prediction in progressive diseases and developing advanced therapies for unmet medical needs in order to improve patients’ health and quality of life. The Institute is committed to providing research solutions and innovation enabling value-based, personalized healthcare. The BIH is funded 90% by the Federal Ministry of Education and Research (BMBF) and 10% by the State of Berlin. The two founding institutions, Charité – Universitätsmedizin Berlin and Max Delbrück Center for Molecular Medicine in the Helmholtz Association (MDC), are independent, member entities within the BIH.

About the Medical Informatics Initiative (MII)
The aim of the Medical Informatics Initiative (MII) is to improve the possibilities for medical research and patient care through innovative IT solutions. These will enable data from healthcare contexts and from clinical and biomedical research to be used and exchanged across multiple entities and sites – transcending the boundaries of individual institutions and geographical locations. The Federal Ministry of Education and Research (BMBF) is providing a total of around €160 million for this initiative through 2021. All of Germany’s university hospitals have joined forces with research institutions, businesses, health insurers, and patient advocacy groups in four consortia – DIFUTURE, HiGHmed, MIRACUM, and SMITH – to create a framework across more than 30 sites that harnesses research findings to the direct benefit of patients. Data protection and data security are given the highest priority. CORD-MI is a use case in the Medical Informatics Initiative.

URL for press release: http://More information on CORD can be found at:

URL for press release: https://www.medizininformatik-initiative.de/de/CORD

URL for press release: http://More information on the BIH Core Unit eHealth and Interoperability can be found at: https://www.bihealth.org/de/forschung/core-facilities/interoperabilitaet/?L=o