Rare Diseases in the German Medical Informatics Initiative and Network University Medicine

Strengthening potentials for digital transformation in healthcare is realized by two large German initiatives: The Medical Informatics Initiative (MII) aims to transform routine patient data into research-ready resources[1] and the Network University Medicine (NUM) aims to establish a nationwide study and data environment to advance research and strengthen pandemic preparedness. Rare diseases (RD) are emerging as a focus of both research frameworks, as the majority of RD patients are receiving care in university hospitals and the lack of available clinical data impacts clinical care and research. Optimized access to standardized longitudinal clinical data would allow identifying patients for clinical trials and benchmarking of therapies, registry development and facilitate research. The RDs identified within the national newborn screening program could serve as a blueprint for other RD.

We propose a concept for a newborn screening registry that leverages infrastructures from MII and NUM to expand the national data space with additional RD data. A gradual implementation of the concept could begin with 2-3 newborn screening centers at German university hospitals. The knowledge gained from the pilot phase will be used to expand the concept to additional sites.