SAVE - AN ONLINE TOOL TO IMPROVE THE DIAGNOSIS AND THERAPY OF PATIENTS WITH CYSTINOSIS AS AN EXEMPLARY APPROACH FOR RARE DISEASES

Vasseur J¹, Hohenfellner K³, Göbel J², Froshauer S², Boulesteix A-L², Erler J³, Herzog N³, Holla I³, Knerr C³, Koeppel C³, Passow M³, Priglinger C³, Steidl G², Treikauskas J³, Wolff S², Weitzel D², Viß K⁴, Storf H¹
¹ Medical Informatics Group, University Hospital Frankfurt, Germany
² Cystinosis Foundation, Munich, Germany
³ Department of Medical Informatics Biometry and Epidemiology, LMU Munich, Germany
⁴ Interdisciplinary Cystinosis Clinic, iMed-Hospital, Rosenheim, Germany

BACKGROUND
Cystinosis is a rare multi-systemic disease with a prevalence of around 1:150,000. To meet its requirement for interdisciplinary patient care, the Cystinosis Clinic for paediatric and adult patients was established in 2012 following the concept of a „one-stop clinic“ [1]. As of today, 98 of the approximately 130 cystinosis patients in Germany regularly visit the Cystinosis Clinic at least once a year to consult clinicians from up to 12 specialties within one day. Like many other rare diseases, the care of cystinosis patients is facing a number of challenges, including the lack of standardised data collection and little communication between the different healthcare professionals.

OBJECTIVES
We decided to develop a web-based tool specifically for cystinosis in order to support the complex management of this disease with the following objectives:

- To ensure the structured and standardised documentation of medical data.
- To facilitate the sharing of data between other specialists and local doctors.
- To create a knowledge base of prospective data for research purposes.
- To enable exchange of data with international registries.

METHODS
1. Development of documentation forms
   Relevant findings to document the course of the disease were compiled by the various specialists involved in the Cystinosis Clinic.
   The resulting requirements for registry forms were defined in Excel sheets.

2. Creation of a patient registry
   Using OSSE [2,3], a flexible open-source software for patient registries (Figure 1), data elements and forms were created based on the templates previously prepared in Excel.
   The registry forms were revised as required in consultation with the specialists.
   OSSE was extended with several novel features to support everyday use.

REFERENCES

RESULTS
As of now, the SAVE patient registry comprises 38 standardised data entries containing nearly 2,000 data elements (Figure 2). These allow the comprehensive collection and evaluation of clinical, imaging and laboratory data from cystinosis patients. In accordance with data protection rules, all data in the registry is pseudonymised, ensuring a strict separation of medical and personal data, and patient consent is required for data processing.
SAVE is available online and allows - with the permission of the patient - all participating doctors to access an individual patient’s data in the registry. This is of central importance to ensure communication within the multi-disciplinary team and with local family doctors, especially for patients during the transition stages from paediatric to adult care.

SUMMARY
In SAVE, we established a comprehensive web-based electronic record for the standardised documentation of the medical history of patients with cystinosis. The use of this patient registry promises benefits for both health care providers and patients and will hopefully help to improve the diagnosis and therapy of cystinosis patients. This approach could also serve as an example for other rare diseases facing similar challenges regarding patient care.