

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

Vasseur J^{*1}, Hohenfellner K⁴, Göbel J¹, Froschauer S², Boulesteix A-L³, Erler J⁴, Herzig N⁴, Holla H⁴, Knerr C⁴, Koepl C⁴, Passow M⁴, Priglinger C⁵, Steidle G⁴, Treikauskas U⁴, Wolff S⁴, Weitzel D⁴, Vill K⁶, 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, RoMed Hospital, Rosenheim, Germany

⁵ Department of Ophthalmology, LMU Munich, Germany

⁶ Department of Neurology and Developmental Medicine, Dr. von Hauner'sche Kinderklinik Munich, 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 communication with the specialists.
- OSSE was extended with several novel features to support everyday use.

RESULTS

As of now, the SAVE patient registry comprises 30 standardised data entry forms 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 enable communication within the multi-disciplinary team and with local family doctors, especially for patients during the transition stage from paediatric to adult care.

Over the course of the project, we developed and implemented several new functionalities for OSSE, including advanced plausibility checks or comparisons to reference values. Users will be able to quickly generate a comprehensive report based on the registry data to use, for instance, in a doctor's letter. Further features such as automatic import of laboratory data or a visual display of data are currently in development.

Following an ongoing test phase, SAVE will be used to document all patients consulting the Cystinosis Clinic from spring 2020. While currently only available in German, we are planning to establish an English version of SAVE to make it accessible on an international platform.

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.

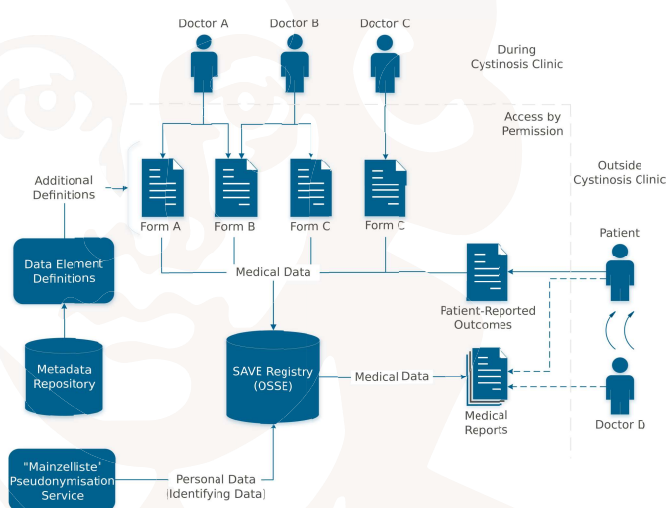


Figure 1: Interaction between the individual participants and software components.

Figure 2: Example data entry form from the SAVE patient registry

REFERENCES

[1] Hohenfellner K, Harms E (2015) Seltene Erkrankungen: Spezielle Cystinosesprechstunde. Dtsch Arztebl 2015; 112(33-34): A-1378 / B-1160 / C-1132

[2] Muscholl M, Lablans M, Wagner TOF, Ückert F. OSSE – open source registry software solution. Orphanet Journal of Rare Diseases. 2014 Sep (1)

[3] Storf, H., Schaaf, J., Kadioglu, D., Göbel J, Wagner T, Ückert F (2017) Register für seltene Erkrankungen. OSSE – ein Open-Source-Framework für die technische Umsetzung. Bundesgesundheitsbl. doi:10.1007/s00103-017-2536-7

THE PROJECT WAS SUPPORTED BY

Bayerisches Staatsministerium für
Gesundheit und Pflege

