

# Visualization techniques in a Clinical Decision Support System for Rare Diseases

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## Introduction

Clinical Decision Support Systems (CDSS) can assist clinicians in the diagnosis of rare diseases (RD). In Germany, the MIRACUM consortia (Medical Informatics in Research and Care in University Medicine), which comprises ten university hospitals, developed a CDSS for RD. MIRACUM is funded by the German Ministry of Education and Research (BMBF) in context of the Medical Informatics Initiative (MI-I). The aim is to make data interoperable for research and patient care by creating data integration centres (DICs) at each hospital [1, 2]. The diagnosis support system identifies similar patient cases in the DICs and can support the clinicians in finding diagnosis for a RD. The aim of this study was to identify visual techniques, which show results of similar patients, as well as to evaluate these techniques in a clinical prototype.

## Methods

A workshop with representatives of a focus-group was held in March 2019 to gather requirements of visualization techniques. The identified visualisation techniques were implemented in a web-based prototype. To check the relevance of the prototype in the clinical practice, we conducted a qualitative study with a “Thinking-Aloud Test” (TA-Test) using a test-patient scenario between September and December 2019. We involved clinicians of rare diseases centres, with the focus in diagnosis and treatment of RD.

## Results

A total of 17 clinicians and computer-scientists participated in the focus group. In the discussion, the participants identified e.g. scatter-plots and patient-timelines as relevant for the visualization. Figure 1 shows an overview of similar patients in a scatter plot. The scatter plot sorts the similarity of patients from 0 to 100 %. With a click on one data point, various data can be viewed, e.g. diagnosis, location and age of the patient.

Figure 2 shows a patient-timeline of an identified similar patient. The diagnoses and symptoms of the patient are arranged horizontally in time. With a click on a diagnosis, diagnosis code and the diagnosis date are shown.

The results of the TA-Test with eight clinicians show a fundamental acceptance to the prototype. However, the study participants noted a more comprehensive transparency in the presentation of similar patients.

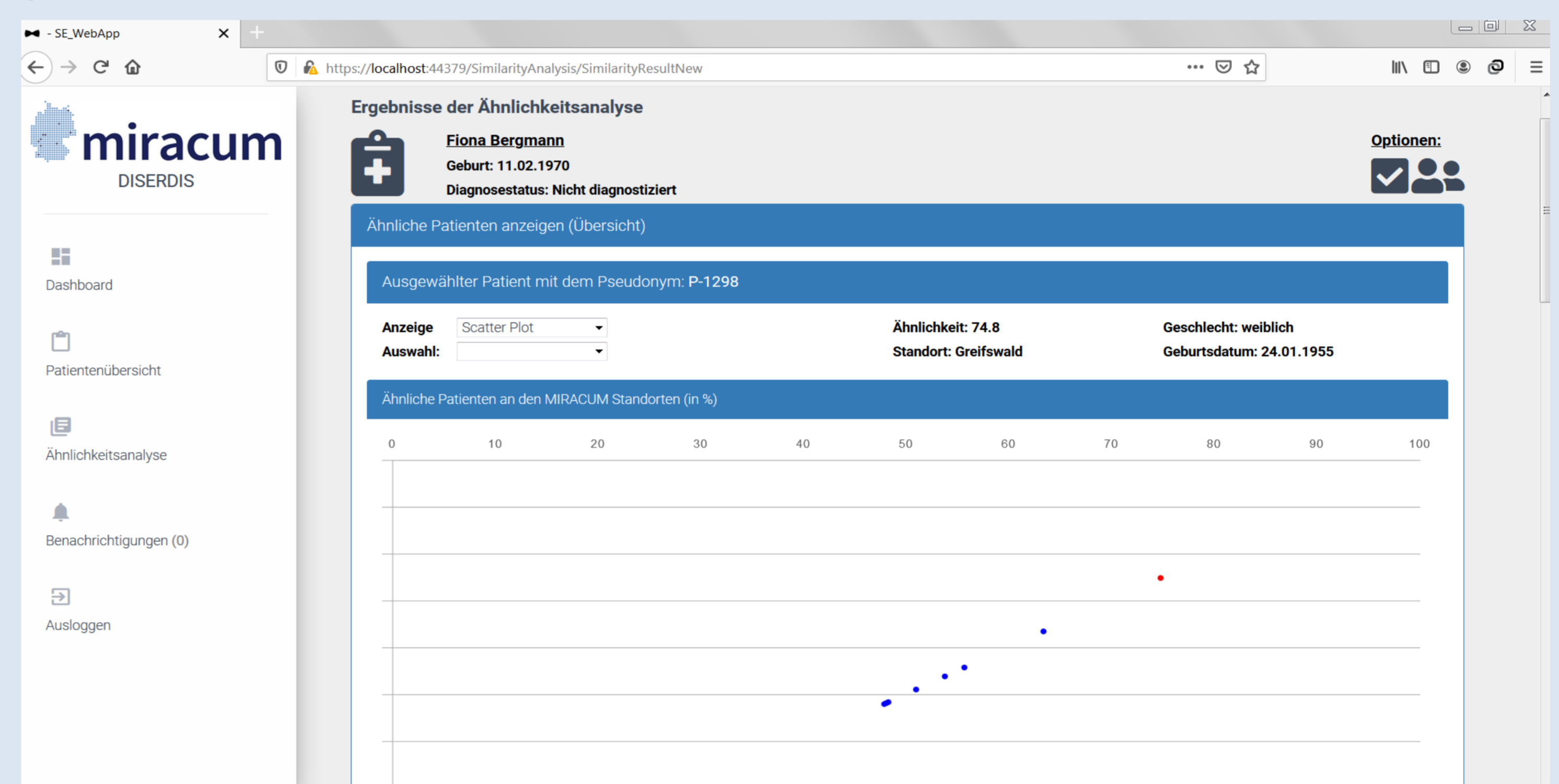


Figure 1: Scatterplot of similar patients

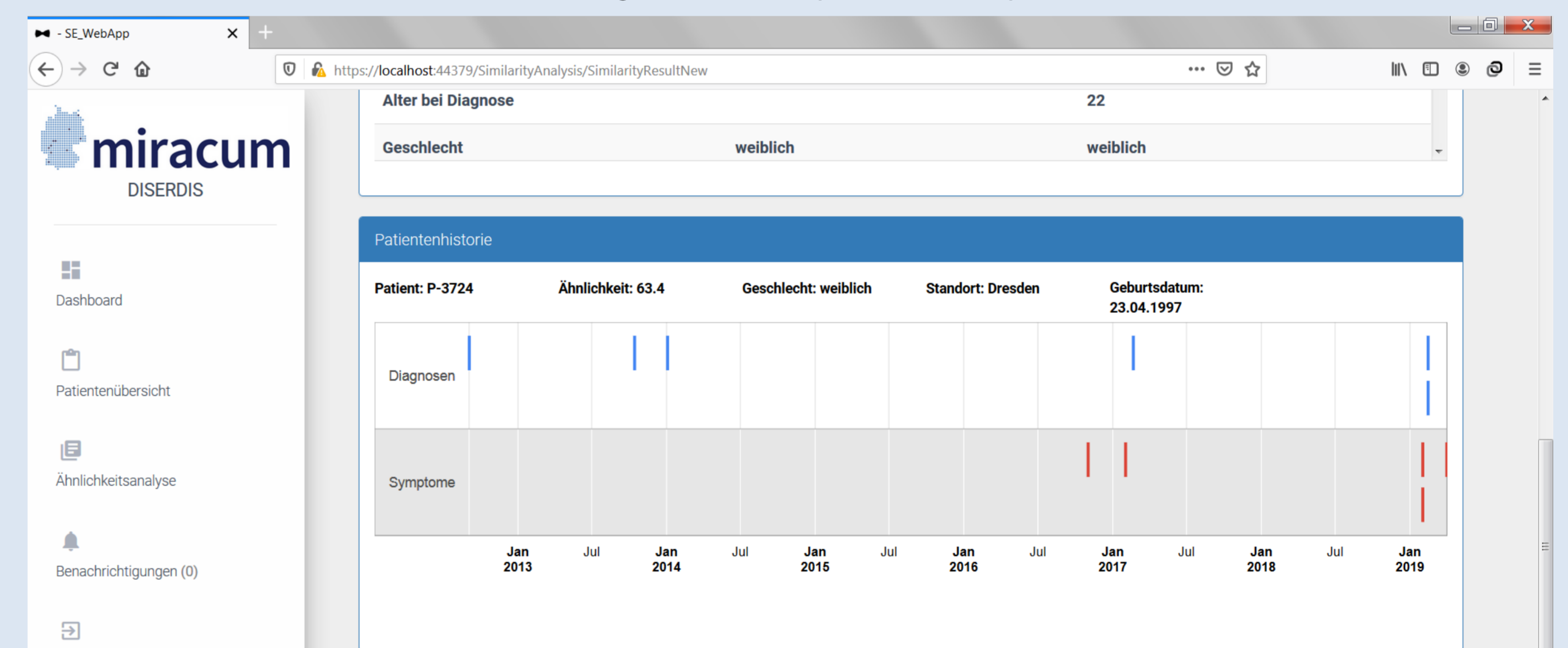


Figure 2: Patient timeline of a similar patient

## Conclusion

The developed clinical prototype enables the identification of similar patient cases and can be a solution to support the clinician finding a correct diagnosis on the long-term view.

## Acknowledgements

MIRACUM is funded by the German Federal Ministry of Education and Research (BMBF) within the “Medical Informatics Funding Scheme” (FKZ 01ZZ1801A, 01ZZ1801B, 01ZZ1801C, 01ZZ1801L).

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