

The possibility of simplifying the diagnostics and therapy of rare diseases in dentistry with the help of a database-based expert system in 2D and 3D imaging

Abstract

The present dissertation describes the methodology of creating a database of patients with rare diseases in the orofacial area. Clear and structured data are the basis for working with knowledge-based systems also in clinical practice. The use of database data is described for two specific diseases, namely Ectodermal Dysplasia and Zimmermann-Laband Syndrome.

The methodology for working with the database consists of the steps of identifying a patient, adding a new card to the database, adding patient records and creating layouts. In the case of ectodermal dysplasia, a set of 13 Czech paediatric patients with ectodermal dysplasia presenting in the oral cavity was processed. The patients underwent genetic testing of candidate genes EDA, EDAR, EDARADD, TP63 and WNT10A. Three-dimensional facial scan images were taken of the patients and compared with facial scans of a healthy control group. The dental treatment of the patients was described using three of these patients as examples. Two patients with suspected Zimmermann-Laband syndrome were examined in both the genetic and dental departments despite not very pronounced manifestations of the syndrome.

The ERN Cranio database at Motol University Hospital was established as a centralized database of patients with rare disease. At the time of writing, it has 113 patients with rare diseases and contains information on 30 rare diseases. Both text and image documents are collected in the database. Among the image documents we find photographs, orthopantomograms, telereöntgenes, CBCT and facial scans. Patients with ectodermal dysplasia were all found to have pathogenic or likely pathogenic variants of one of the above genes. By comparing the facial scans, we verified dysmorphic features in the lower 1/3 of the face in patients with ED. After the removable denture was made, the height of the lower 1/3 of the face approached the average values of non-syndromic subjects. We find clinically surprisingly mild manifestations in patients with Zimmermann-Laband syndrome. No gingival fibromatosis is present in the oral cavity. Nevertheless, genetic testing revealed a pathogenic variant in the c.1606G>A p gene (Ala536Thr).

Keywords

Database, knowledge system, rare diseases in dentistry, ectodermal dysplasia, Zimmermann-Laband syndrome