Секция «Клиническая медицина»

Development of a prototype information and diagnostic program for hereditary diseases accompanied by periodontitis

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Periodontitis is one of the most common diseases encountered in the practice of any dentist and periodontist. To date, there are 33 known monogenic syndrome, accompanied by periodontitis. Domestic periodontics and medical genetics are almost completely divided with significant progress in the study of etiology of a large number of diseases of this group. Inaccurate diagnosis entails errors in the conduct of medical and genetic counseling in burdened families. One way to address this gap may be the development and introduction of periodontists automated diagnostic programs. The study was performed within the framework of research works on creation of systems of support of decision-making in xGen Cybernetics.

Purpose: to create a prototype of information and diagnostic system for hereditary syndromes accompanied by periodontitis.

Materials and methods: available public sources of the INTERNET are used to fill the knowledge base of the decision support system (DSS). For the formation of the directory syndromes used information from the databases OMIM, ORPHANET. Description of the clinical picture according to the literature data was carried out on the basis of modified and extended ontology HPO. The program was implemented on the basis of the platform for development of XGEN IDS diagnostic programs and xGenCloud service.

As the main algorithm of differential diagnostics, implemented in the DSS of the algorithm of automated diagnosis search, is known in the theory of decision-making method of deterministic classification with linear decisive function.

Results: we have developed an information and diagnostic system of hereditary syndromes accompanied by periodontitis. In the process of systematization of knowledge on monogenic diseases the basis of syndrome forms described in the international catalogues OMIM and ORPHANET. The knowledge base includes 33 of nosological forms. According to the literature for 29 nosological forms genes are mapped and pathogenic mutations are determined, leading to the development of hereditary syndromes, accompanied by periodontitis manifestations for most of the presented forms of this group of diseases, a specific combination of it with the features of the development and functioning of other organs and systems is described.

Functional prototype of DSS provides the following functions: 1) input data on the subject (phenotype, family history, living environment and negative occupational factors) in the form of formalized information; 2) the formation of a ranked list of diseases; 3) dialogue mode for differential diagnosis.

Conclusions: the prototype automates differential diagnosis in monogenic syndromes accompanied by periodontitis.