

Development of a prototype system to support decision-making for recommendations for genetic testing in monogenic syndromes with Orofacial clefts

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Orofacial cleft is one of the most common heterogeneous etiologies of diseases encountered in the practice of any dentist and periodontist with a specific weight among all congenital malformations, more than 87%. Optimization of genetic testing requires the development and use of specialized computer programs that simplify the confirmation of diagnosis by recommending the selection of genes for testing, taking into account the clinical picture of the patient. The study was performed within the framework of research works on creation of decision support systems in xGen Cybernetics.

The aim of the study was to create a prototype system to support decision-making for recommendations on genetic testing and the knowledge base in monogenic syndromes with Orofacial clefts.

Available public INTERNET sources were used to fill the knowledge base of the decision support system (DSS). For the formation of the directory monogenic forms used information from the databases OMIM and 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.

Developed a prototype system to support decision-making for recommendations for genetic testing of Orofacial clefts. 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 551 genes that lead to the development of this group of diseases. Knowledge base of diseases with Orofacial clefts includes 811 nosological forms. Of the presented forms, 811 syndromes are characterized by anomalies of the head and neck. Of the 811 forms for 636 units characteristic eye abnormalities, 690 forms the characteristic disorders of the nervous system, 591 - with hearing problems and abnormalities of the ears, 387 - anomalies of the muscles, 384 - anomalies are bronchopulmonary system, 464 - gastrointestinal tract, 535 - with manifestations in the urogenital system (including 418 with lesions of the male genital system, and 264 female) with 313 syndromes described lesions of kidney, 286 syndromes with anomalies of the endocrine system, 497 - with anomalies of the cardiovascular system, abnormalities of growth described at 533 syndromes, anomalies of the blood and blood-forming tissues 107, lymphatic system with 43 syndromes, immunology system at 167 syndromes, defects of the connective tissue at 321, and anomalies of the integument at 518 syndromes.

According to the types of inheritance monogenic forms of this group of diseases are distributed as follows: 278 with autosomal dominant inheritance type, 345-with autosomal recessive inheritance type, 17 forms - with X-linked inheritance type, 46 forms - with X-linked recessive inheritance type, 19 forms - with X-linked dominant inheritance type and 4 - with mitochondrial inheritance type.

The functionality of the prototype DSS provides the following functions: 1) entering data on the patient (phenotype, family history, environmental conditions of residence and negative professional factors) in the form of formalized information; 2) the formation of the ranked list of genes; 3) conclusion of conclusion with recommendations for genetic testing. The structure of the conclusion includes a list of entered parameters for the patient and recommendations for genetic testing with a list of genes.

Created a prototype automatiseret recommendations for genetic testing in Orofacial clefts. Completion of a prototype will allow you to optimize the work of the doctor, simplifying the differential diagnosis.