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EVALUATION OF THE “FACE2GENE” PROGRAM AS A TOOL FOR PHENOTYPICAL IDENTIFICATION OF GENETIC PATHOLOGY

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Introduction. Approximately 4% of the world's population suffers from orphan diseases, the majority of them has a genetic origin. Syndromological analysis is crucial in studying the semiotics of hereditary pathology, but it requires substantial experience and can be subjective. Additionally, there is a shortage of trained clinical geneticists, leading to long waiting times for appointments. To address these challenges, computer programs have been developed with utilizing artificial intelligence to detect rare hereditary disorders by analysing facial images. The "Face2Gene" application generates a list of ten genetic pathologies that the patient may have, aides geneticists in more effective screening of genetic syndromes.

Objective. 1) Determine the indicators on which the "Face2Gene" application creates a sample of ten pathologies and understand the analysis of morphological changes in the face. 2) Apply the program in practical settings.

Materials and Methods. In this study, we analysed 46 scientific publications that describe algorithms for recognizing and analysing morphological changes of the face, forming the basis of the "Face2Gene" application. We examined patients who sought medical and genetic counselling due to suspected genetic diseases, considering their clinical and laboratory examination results.

Results. The "Face2Gene" application employs deep learning technology to learn face representation using large-scale recognition. This is followed by a fine-tuning phase, where the learned data is transferred to the genetic syndrome domain.

During our examination of patients, automated anthropometric measurements revealed several characteristic features of specific genetic syndromes. For instance, brachycephaly, hypertelorism, sinophrysis, long philtrum (long distance between nose and upper lip), and thin upper lip were observed in one of the examined patients with KBG syndrome. It's important to note that this syndrome exhibits wide phenotypic variability, which can pose challenges in its initial diagnosis. The application also showed positive results in diagnosing EEC1 syndrome, characterized by ectrodactyly, cleft lip, abnormal hair, glands, and eyes, even without specific characteristic facial changes.

Conclusion. DeepGestalt technology, along with the "Face2Gene" application, demonstrates high accuracy in analysing craniofacial dysmorphism in patients with genetic pathology. However, it should not be considered a substitute for clinicians' knowledge of phenotypes. Further research should focus on acquiring and interpreting DNA analysis of patients.

References:

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