CZU: 616-056.7-07

USE OF AI-ASSISTED TOOLS IN HEREDITARY DISEASE DIAGNOSTIC

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One of the most difficult parts of medical genetics is analysis of a huge amount of patient's data and presumption of diagnostic. It can be even harder if we take in account multiplicity of hereditary diseases, presense of many phenocopies at some of them and purely human factors as tiredness or some form of unconscious prejudices, all of them having possibility to interfere and impair ability to put correct diagnosis, what definitively is not beneficient for patient.

This situation began to change in 1980's, when first expert systems in domain of human genetics appeared. But those systems included only limited amount of data, were statical, not able to self-improve and were composed by humans, so being influenced by all human errors.

Major changes in this field began in 2011, when FDNA company was founded and began to develop Face2Gene program, based on next-generation phenotyping and artificial intelligence technologies. This program compares patient's face photo and clinical signs with a database of previously diagnosed patients using AI and proposes a list of possible diagnoses with ranking for similarity with photo and with clinical signs.

This software helped us to diagnose such diseases as Coffin-Siris, Bloom and Myhre syndromes and much more. In this publication we summarize our experience of Face2Gene software and next-generation phenotyping use.

Keywords: AI, next-generation phenotyping, medical genetics.