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Syndromal diagnostic program “SynDiag”

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The program *SynDiag* allows to make preliminary syndrome diagnosis on the basis of the phenotypic description of a patient selected from sign library. *SynDiag* is the diagnostic program, working under MS Windows NT/2000/XP/7/8/10 operating systems. *SynDiag* and intend for diagnosing MCA/MR syndromes of different etiology (monogenic, chromosomal, teratogenic, etc.), ectodermal and skeletal dysplasias, some systemic complexes. The program can also be used as a textbook on syndromology and is intended for every physician, who contacts with malformed patients in their practice. The program has three main libraries-

1. Sign library is organised in a three-step manner: a) System, b) Organ, c) Sign.

The library includes the list of major and minor malformations and dysplasias, functional, laboratory, roentgenologic and some other data (near 1700 signs). All signs have an explanation of their meaning and many of them have photoillustrations.

2. Syndrome pattern library includes 3013 nosologic forms. Each pattern gives the full image of the syndrome clinical picture and also includes- a) The name and main synonyms, b) The amount of patients, being included in the description, c) The amount of signs, d) The amount of screens with photos, e) The population frequency, f) Literature, g) Complete syndrome description in which the frequency (in %), diagnostic value of every sign and mode of inheritance. Diagnostic value subdivides all signs of a syndrome pattern in three groups: Diagnostic, helpful and auxiliary.
3. Photolibrary includes near 2751 screens with 1 to 20 color or gray photos of our patients with different syndromes. There are two ways of diagnosis search in the program. One is based on originally modified pseudo- Bayes algorithm, which operates with all parameters of syndrome patterns and suggests syndromes with probability of at least 10%. The testing of this variant showed the 94% diagnostic effectivity. The other way gives all syndromes containing from 1 to all selected signs without probability evaluation. *SynDiag* is actively using program in many Genetic Centers of the former USSR.

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