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Thesis Abstract

Ectodermal dysplasias (group A): elaboration of a computerized database to help with genetic-clinical diagnosis (Displasias ectodérmicas do grupo A: elaboração de um banco de dados informatizado para auxílio ao diagnóstico genético-clínico)

Toni Lisboa Costa

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Ectodermal dysplasias are conditions that involve structures derived from the ectoderm, such as the epidermis and its enclosures, constituting a large and complex nosologic group, composed of rare entities with genetic etiology (in their majority). Freire-Maia, in 1971 and 1977, proposed a clinical definition and a clinical-mnemonic classification of the ectodermal dysplasias, which are used today by researchers and clinicians throughout the world. This classification divides them into two main groups (A and B), based on classic signs in structures related to hair, teeth, fingernails and sweat glands. Our main objectives were: 1) an analysis of the literature concerning genetic and clinical aspects of ectodermal dysplasias, 2) an update of the ectodermal dysplasias in Freire-Maia's classification (1971, 1977), reviewed by Pinheiro and Freire-Maia (1994), making the necessary alterations in group A (joining conditions previously described as different subgroups, and inclusion of new conditions that have been described), and 3) creation of a computerized control system (available in a web page on the Internet), with a database that contains the information obtained, as well as a database of selected images of the known ectodermal dysplasias. There is also a search function, where the information supplied by the user (according to the patient's clinical signs) is compared with the database, indicating the possible ectodermal dysplasias. The database is available at http://displasias.bio.ufpr.br. Each condition was analyzed individually, a "file card" was elaborated for each of them, with the following data: general information, genetic aspects, "classic" clinical signs, other frequent signs, commentaries

and bibliographical references. Images and search systems (based on key words and clinical criteria) are also available. The present study enlarged the number of known ectodermal dysplasias to 192 (group A). The subgroups with the largest number of conditions are the so-called 1-2-3 and 1-2-3-4 groups, with 48 and 40 dysplasias, respectively. They are followed by subgroup 1-2 with 34 conditions, subgroup 1-3 with 25 and subgroup 2-3 with 15 dysplasias. The other subgroups have less than 10 dysplasias. Most of the ectodermal dysplasias have autosomal recessive inheritance (38%), followed by those that possess autosomal dominant inheritance (28%). The X-linked dysplasias constitute 5% and the remaining 29% include syndromes whose etiology is not yet known.