Letters to the Editor

A CLINICAL REAPPRAISAL OF A NEWLY RECOGNIZED ECTODERMAL DYSPLASIA

To the Editor: Two patients showing a newly recognized form of ectodermal dysplasia [1-6] (termed odontotrichomelic hypohidrotic dysplasia [7]) were re-examined 2 years after the first investigation. Some signs not seen initially have now been verified and some others previously described have not been confirmed. After this clinical reappraisal, the signs may be listed as follows: (1) hypotrichosis; (2) abnormal dentition; (3) onychodysplasia (hypoplastic nails; this trait is present only in the boy—the girl has no fingers or toes); (4) hypohidrosis (as determined by pilocarpine iontophoresis; a full analysis of this problem is presented elsewhere [8]); (5) thin, dry, shiny skin (unusual wrinkles are formed when the patients smile or grimace); (6) hypoplastic nipples and areolae; (7) extensive tetramelic reductions with dermatoglyphic abnormalities; (8) large, thin, protruding and deformed ears; (9) protruding lips; (10) enlarged nose at the distal two-thirds; (11) possible abnormalities of tyrosine and/or tryptophane metabolism; (12) cleft lip (unilateral incomplete; only in the boy); (13) growth retardation (only in the boy); (14) EEG abnormalities (only in the boy); and (15) ECG abnormalities (only in the girl).

Odontotrichomelic hypohidrotic dysplasia is an ectodermal dysplasia of the tricho-odonto-onycho-dyshidrotic subgroup (signs 1-4 in the above list) according to Freire-Maia's classification [7, 9]. Some of the above-mentioned defects may not be components of this syndrome, but only coincidental findings. Observations on other patients are necessary for a better delineation of the syndrome.

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REFERENCES

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