A New Syndrome Is Born

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In this issue of *Dermatologica*, Dr. Piergiacomo Calza-vara-Pinton in collaboration with Dr. Anna Carlino and Dr. Anna Benetti and Dr. Giuseppe De Panfilis from the Departments of Dermatology and Anatomopathology (II) of the University of Brescia, Italy, describe nothing less but a new genetic syndrome which is characterized by brittle hair breaking off to leave only stubbles of 1-10 mm in length, dystrophies of the distal nail plate and a distinct facies [1]. Scanning electron microscopy revealed focal flattening of the hair shafts and rotations along the axis of hair shafts. The hairs fracture in these twists. This latter finding may have been the reason why the authors decided to report their new hair shaft breakage syndrome under the designation of 'pili torti and onychodysplasia'. The term 'pili torti', of course, is a very old one and has the advantage of being deeply rooted in the medical literature. To me, the latin term 'pili torti' suggests that one deals with a very specific phenomenon that is pathognomonic for a certain disease as is the case in trichorrhexis invaginata and the Comèl-Netherton syndrome. In contrast, 'pili torti' are rather nonspecific and can be found in quite a number of genetic syndromes affecting the hairs. Therefore, I prefer to speak of 'twisted hairs' instead. Nevertheless, these twisted hairs cause breakage of the hair shafts and result in a clinical picture resembling atrichia in the disorder reported by Dr. Calzavara-Pinton and collaborators.

The authors carefully delineate their new syndrome from atrichia with nail dystrophy, abnormal facies and retarded psychomotor development [2] which, at first glance, shows some resemblance because of marked alopecia and distal nail dystrophies, but has a different histopathology and therefore a different pathomechanism underlying the alopecia. The Beare syndrome [3] featuring

'pili torti' and nail dystrophies, the Clouston type of hidrotic ectodermal dysplasia and the Rapp-Hodgkin/ AEC syndrome, which also features 'pili torti' [4], can be immediately excluded because of autosomal dominant inheritance, whereas the disorder described by Dr. Calzavara-Pinton follows an autosomal recessive inheritance that only mimicks dominant transmission due to consanguineous marriages. A normal banding pattern on polarizing microscopy rules out the heterogeneous group of trichothiodystrophy syndromes all of which are inherited in an autosomal recessive manner [5].

Many colleagues who describe a new syndrome then have the understandable urge to classify 'their' syndrome within the framework of a larger disease group. Dr. Calzavara-Pinton and coworkers could not resist this urge and decided to place their hair shaft breakage/nail dystrophy syndrome within the group of 'ectodermal dysplasia'. Nothing is wrong with this, except that this group is already rather crowded. If one applies the very broad definition of ectodermal dysplasia as conditions featuring one of the following signs: (1) trichodysplasia; (2) dental defect; (3)

onychodysplasia, or (4) dyshidrosis as well as the presence of at least one other feature involving a structure of epidermal origin [6] , one ends up with an enormous number of clinical syndromes ranging from focal dermal hypoplasia to the Tay syndrome, and many ichthyoses would qualify as ectodermal dysplasia. Most of us (and I include myself in this group) immediately surrender when facing such bewildering clinical complexity. I see the danger that when confronted with a patient one will be satisfied with the statement: 'this must be some kind of ectodermal dysplasia'. I do hope this will not be the fate of the syndrome described by Dr. Calzavara-Pinton and coworkers and I shall try to remember their syn-

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drome by putting it in my mind into the drawer of 'syndromes with hair shaft anomalies'. There it will be in the fine company of the Comèl-Netherton syndrome, the Björnstad syndrome, Menkes syndrome, and the tricho-thiodystrophy syndromes, to name but a few. In this drawer I hope to find back from now on the 'Calzavara syndrome'. Proficiat!

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