Kindler syndrome with facial telangiectatic hyperpigmentation: need for modification of diagnostic criteria.

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Sir,

Our interest was aroused by your recent publication (1) on Kindler syndrome (KS) . The rarity of the syndrome and the observations that its major clinical features (photosensitivity and blistering) improve after childhood, contribute to the fact that many cases remain undiagnosed. Symptoms appearing later in life may differ from the classical presentation.

We report on a 44-year-old Pakistani of non-consanguineous parents with a long lasting facial hyperpigmentation. The examination disclosed that an extensive facial telangiectasia had been mistaken for black discoloration as it almost merged with his dark complexion. In addition poikiloderma was expressed on the chest. There was no scarring of hands and feet, while the teeth appeared normal. A careful enquiry revealed that he experienced blisters and photosensitivity in early childhood. Two of his younger brothers had similar symptoms, and one of them had died in early childhood. The family members were not available for examination as they live in Pakistan. He had no complaints for many years. Two months earlier he had an injury on one hand that took a month to heal.

Based on our previous study (2) we believe that this patient is suffering from KS. The syndrome comprises seven major characteristics: a positive family history, oral abnormalities, telangiectasia on the face and neck, blistering and photosensitivity in childhood, poikiloderma, mucosal involvement and acral abnormalities. Other potential symptoms in KS include actinic keratoses, anemia, psychological distress and skeletal abnormalities (3). A history of neonatal death may be present. In a large kindred from Panama (4), 26 cases were examined. The major findings were skin fragility with blistering (100%), poikiloderma (96%), photosensitivity (92%), sever cutaneous atrophy (89%), hyperkeratosis of palms and soles (81%), congenital acral blisters (81%), severe periodontal disease (81%), and phimosis in males (80%). We would like to add to the major criteria facial and neck erythema, telangiectasia and the history of an affected child in the family. Our patient had only two out of five major criteria (acral blistering in early life and photosensitivity), the diagnosis remained probable as four major criteria are required for a reliable diagnosis of KS. We consider that our patient was a KS case, since extensive variation in the severity of symptoms can be seen among members of the same family (2). The type of the genetic defect may be involved in the genotype-phenotype relationship. With the mild phenotype reported here it is clear that the syndrome has great heterogeneity. Thus far, 17 different loss-of-function mutations in Kind1 gene in 41 families have been uncovered (5,6).

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