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Letter to Editor

Kindler Syndrome: A Close Mimic of Dyskeratosis Congenita and the Need to Distinguish the Two Clinical Entities

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To the Editor,

I read with great interest the recent article by Sinha et al.[1] Interestingly, one rare condition that often mimics dyskeratosis congenita, and that needs to be distinguished from it, is Kindler Syndrome (KS).

KS primarily occurs secondary to "loss of function" mutations in the FERMT1 gene. Mode of inheritance is autosomal recessive. Interestingly, FERMT1 gene mutations are absent in 30% of cases. The characteristic histological feature of KS is attenuated keratinocyte proliferation. Piccinni et al. in a recent study have shown that KS related deficiency of Kindlin-1 results in the premature senescence of the keratinocytes.[2] Dermal as well as intra-epidermal cleavage is seen on ultra- structural examination of the skin.

The disease phenotype is characterized by an increased incidence of trauma induced acral blisters. The blisters typically start appearing during infancy. Premature aging and progressive bullous poikiloderma of the skin is another characteristic feature of KS.[3] Hyperpigmentation as well as hypopigmentation may occur. Accompanying telangiectasiasis is seen. In addition, "cigarette paper" like atrophy is seen in most patients with KS.[4] Increased photosensitivity is another typical feature of the syndrome. Nofal et al. have recently also reported webbing of the fingers in 60% of cases.[5] Associated nail dystrophy is seen in three out of five patients with KS.

"Owl-like" facies have been reported in 60% of cases. Gingival hypertrophy and ankyloglossia may also be seen. Leuko-keratosis of the lips is an additional common feature. Rarely, ocular lesions may be seen. For instance, Cheour et al. recently reported corneal deformities with accompanying bilateral ectropion and symblepharon in a 57-year-old patient with KS.[6] In addition, deaf mutism is seen in 25% of the cases. The gastrointestinal tract may also be involved resulting in esophageal strictures and consequent dysphagia (40%).[7] Similarly, colitis with accompanying bloody diarrhea may occur. Anal stenosis (40%), as well as uretheral stenosis (20%) have also been reported in KS. An increased incidence of cutaneous squamous cell carcinomas is seen. Mizutani et al. have also reported laryngeal carcinomas in patients with KS.[7, 8]

The management of KS is symptomatic, and no specific therapy is available yet.[9] Squamous cell carcinomas require local resection of the tumor. Severe esophageal stenosis occurs in some patients necessitating the need for esophageal dilatation.[10] Parental consanguinity is seen in almost all cases. Hence, genetic counseling is of paramount importance. As is obvious from the above discussion, the clinical picture in patients with KS may mimic dyskeratosis congenita. A close dermatological and systemic assessment can help to distinguish between the two disorders.

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Sincerely,

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