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KINDLER SYNDROME- A RARE CASE REPORT

Dermatology

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ABSTRACT

Kindler syndrome is a rare autosomal recessive disorder characterized by acral blistering, skin fragility and progressive poikiloderma which is usually present at birth. The genetic defect is caused by the loss of function mutations in the KIND1 gene which encodes kindlin-1 protein. We report a case of Kindler syndrome in a 14 years old patient who came with complaints of recurrent blisters over the palms and soles since childhood, photosensitivity and generalized xerosis.

KEYWORDS

Kindler syndrome, KIND1 gene, Poikiloderma

INTRODUCTION:

Kindler syndrome was first described in the year 1954 by Theresa Kindler in a 14 year girl with features of poikiloderma congenitale and epidermolysis bullosa [1]. Clinically, it presents with four major features such as acral blistering in infancy and childhood, skin atrophy, progressive poikiloderma and abnormal photosensitivity [2][3].

CASE REPORT:

A 14 years old boy presented to our OPD with complaints of recurrent blisters over the dorsal aspect of both palms and soles since childhood. History from his mother revealed that he was born out of third degree consanguineous marriage. Antenatal and postnatal period was uneventful. He was delivered by normal vaginal delivery and birth weight was 2.5 kg. There was no developmental delay. The patient started developing blister from the 10th day of neonatal period and persisted till date. The blisters occurs spontaneously or following trauma. The blisters rupture within 2-3 days and leave behind erosions which heals with atrophy and hypopigmentation or develops following trauma. He started developing dark colored skin lesions over the neck at the age of 5. It was initially small in size which later progressed to attain the present size and later he started developing multiple dark colored lesions over the forearms, trunk, back, and both the legs. History of photosensitivity was present for the past 5 years. Bowel and bladder habits were normal. Mental and motor development was normal. There was no history of similar complaints in the family members.

Dermatological examination revealed diffuse hyper and hypopigmentation present over the neck **[Figure 1]**. Multiple irregular, hyperpigmented macules of varying sizes 2-5mm present over the trunk, back and both the legs **[FIGURE 2]**. Cutaneous atrophy and hypopigmentation present over the dorsal aspect of both hands and feet **[FIGURE 3]**. Generalized xerosis present all over the body. Blepharitis was present. Hypopigmentation was present over the palmar aspects of both the hands with keratoderma. Nail discoloration and ridging was present. Scalp hair was normal. Teeth, oral and genital mucosa were normal. Systemic examination done was normal. Skin biopsy done showed features of hyperkeratosis, sub epidermal bulla with eosinophils and neutrophils. The dermis shows dermal melanophages with lymphocytes. **[FIGURE 4]**.

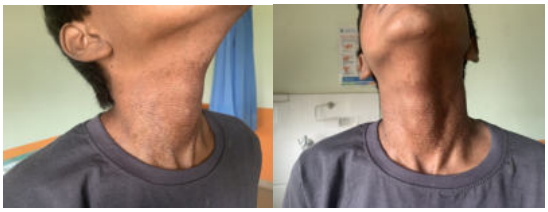

Figure 1: Diffuse Hyper And Hypopigmentation(poikiloderma) Present Over The Neck

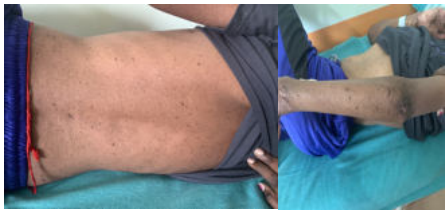

Figure 2: Multiple Irregular, Hyperpigmented Macules Of Varying Sizes 2-5mm Present Over The Trunk, Back And Forearms

Figure 3: Cutaneous Atrophy And Hypopigmentation Noted Over The Dorsal Aspect Of Both Hands And Feet

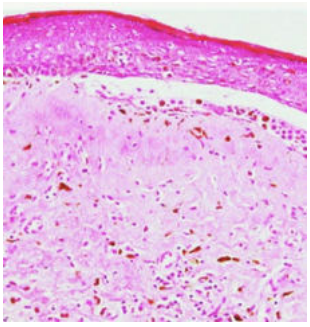

Figure 4: Histopathology Showed Features Of Hyperkeratosis, Sub Epidermal Bulla With Eosinophils And Neutrophils. The Dermis Shows Dermal Melanophages With Lymphocytes.

DISCUSSION:

Kindler syndrome is a very rare genodermatosis caused due to the mutation of KIND1 gene which encodes kindlin-1, which is a membrane-associated signaling protein. Loss of this protein results in abnormal skin fragility with defects in actin-extracellular matrix linkage [4] [5]. Recently, novel mutation in FERMT1 gene has been discovered [6].

The disease usually presents at birth with blistering of skin induced by trauma, which is more prominently seen over the limbs and regress with age. The blisters heals with minimal scarring. Photosensitivity begins in infancy or early childhood and improve significantly with age. Diffuse poikiloderma appears gradually over the sun exposed areas (localized to the face and neck) and becomes more predominant later in life to involve non exposed areas also. Atrophic changes (cigarette paper wrinkled appearance) of the skin are more prominent in the sun exposed areas such as dorsal aspect of the hands and feet and it becomes generalized during adolescence stage. The atrophy and poikiloderma are persistent whereas the appearance of blisters and photosensitivity tends to reduce with age.

Esophageal strictures, rectal bleeding, anal stenosis, meatal stenosis, urethral bleeding has been reported. The other common associated feature includes ophthalmic (ectropion, keratoconjunctivitis, conjunctival scarring, severe corneal ectasia), mucosal changes (chronic gingivitis, dental caries, periodontitis, leukokeratosis of buccal, urethral, esophageal and anal mucosa), dental abnormalities, palmoplantar keratoderma, ichthyosis, milia, blond hair, pseudoainhum, webbing of fingers and toes and rarely nail changes (long and thick cuticles of nail, nail dystrophy [7][8]) and increased susceptibility to squamous cell carcinoma [9][10].

The diagnosis is clinical. The histopathology from the atrophic skin lesions shows epidermal atrophy, vacuolar degeneration of basal layer, edema at the dermo-epidermal junction, pigment incontinence and dilatation of blood vessels in the papillary dermis. Biopsy from the bulla by immunofluorescence antigen mapping and electron microscopy shows single or multiple cleavage planes at the level of basement membrane zone.

Treatment is mainly symptomatic with proper counseling. Patients should be advised to avoid trauma and photoprotection to prevent or slow the progression of poikiloderma [11]. Topical and systemic antibiotics are given for infected bullous lesions.

This case is because of the rarity and its association with mottled hyperpigmentation and ichthyosis.

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