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Case Report

Sporadic dyskeratosis congenita in a male – A case report

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ABSTRACT

Dyskeratosis congenita was first described in 1960 as Zinsser-Engman-Cole syndrome. It is a rare inherited condition with a progressive nature and a tendency to involve multiple systems like pulmonary, gastrointestinal, genitourinary, cerebral, and dental. It has an X-linked recessive (most common) or Autosomal dominant or recessive inheritance with a high male preponderance. The genetic defect lies in the DKC1 gene which encodes for Dyskerin protein. Dyskeratosis congenita patients are at a higher risk of development of malignancies, pulmonary fibrosis and eventually aplastic anemia and bone marrow failure which may be the cause of death. This report details a case of Dyskeratosis congenita affecting a 21 year old male patient with the most benign presentation.

1. Introduction

Dyskeratosis congenita (DC), also called as Zinsser-Engman-Cole syndrome is a rare inherited, progressive multisystem syndrome characterized by a classic triad of reticular skin pigmentation, dystrophic nails and oral leukoplakia. Here in, we report a sporadic case of DC without similar clinical presentation in the first degree and second degree relatives. [1,2]

2. Case Report

A 21 year old male patient born of second degree consanguineous marriage presented with the complaints of dark pigmentation of skin over face, neck, arm pits, hands, legs, groins, palms, soles and mouth; occasional burning sensation in mouth; and excessive sweating of palms and soles for the past 15 years. There was no significant family history. On examination: diffuse, lacy, reticular gray brown pigmentation of the skin of face, neck, axillae, cubital fossa, hands, legs, groin, palms and soles was noted. Oral examination revealed leukoplakia with black pigmentation of the tongue and mucous membranes and dental caries. Nails showed dystrophy of all the fingers and toes, and reverse pterygium of all the finger nails. Premature greying of hair and adermatoglyphia were noticed. Routine laboratory investigations were normal. Histopathology revealed keratinized stratified squamous epithelium with basal vacuolar degeneration, pigment incontinence in upper dermis. A diagnosis of Dyskeratosis congenita was made based on the history, clinical examination and biopsy findings.

3. Discussion

Dyskeratosis congenita is a fatal condition with multisystem involvement which has an X-linked recessive (most common) or Autosomal dominant or recessive inheritance with a high male preponderance. It is characterized by severe shortening of telomeres (hence considered as

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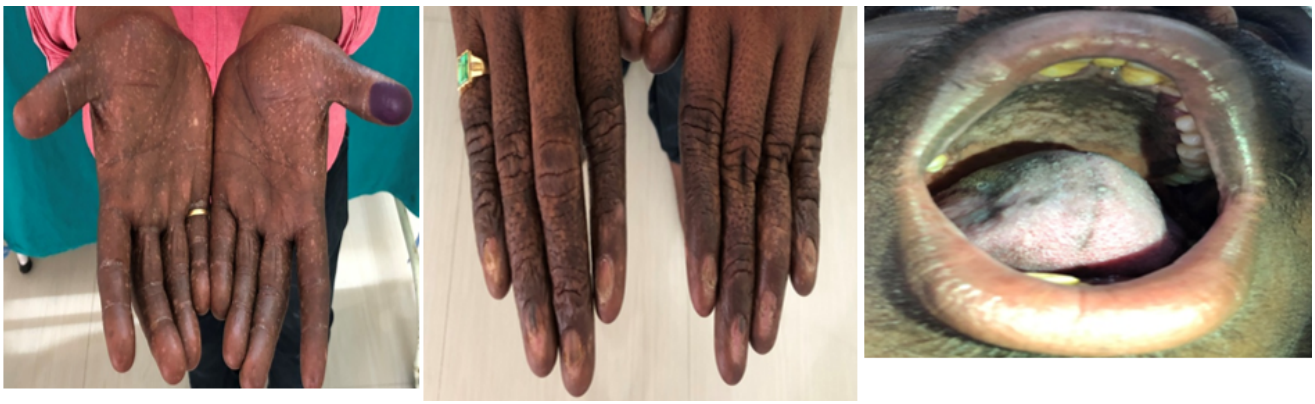

Fig. 1: Pictures of the patient showing reticulate pigmentation of the palms, dystrophy and reverse pterygium of all the nails and oral leukoplakia.

Telomeropathy) leading to arrest of cell proliferation and senescence. Extreme telomere shortening causes the clinical features of this disease. Hypo – or pigmented macules and patches of tan-to-gray color in a mottled or reticulate pattern in the sun-exposed areas including the upper trunk, neck, and face are the primary diagnostic features. Poikilodermatous changes with atrophy and telangiectasia may be common. Ectodermal abnormalities such as alopecia of the scalp, eyebrows, and eyelashes; hyperhidrosis; premature graying of the hair; hyperkeratosis of the palms and soles; and adermatoglyphia (loss of dermal ridges on fingers and toes) can also be noticed. Nail dystrophy is seen in approximately 90% of patients which begins with ridging and longitudinal splitting and progresses resulting in small, rudimentary, or absent nails. Mucosal leukoplakia involving the buccal mucosa, tongue, and oropharynx occurring in approximately 80% of patients is the pathognomonic feature. Diagnosis is based on clinical evaluation and identification of characteristic physical findings. Histopathology of the skin is non specific and shows epidermal atrophy, increased vascularity of the dermis with a sparse lymphocytic infiltrate, and pigment-laden macrophages. Patients have a high propensity of development of malignancies, pulmonary fibrosis and aplastic anemia which may be the cause of death.[3,4]

4. Conclusion

Dyskeratosis congenita is a rare genodermatosis with an incidence of 1 in 10,00,000. However, given the fatal consequences of development of malignancies or hematological abnormalities, we hereby report a case of dyskeratosis congenita to maintain a high index of suspicion and recommend an early prompt diagnosis and long term care to improve the quality of life. We would thus recommend a close multidisciplinary follow – up of such cases for progression to malignancies of blood through regular blood count, and skin and bone marrow biopsies as necessary.

5. Conflict of Interest

The authors declare they have no conflict of interest.

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