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Zinsser-Engman-Cole syndrome: A Rarity

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Abstract

Dyskeratosis congenita (DC) also known as Zinsser-Engman-Cole syndrome is very uncommon. The primary clinical features include oral leukoplakia, nail dystrophy, and reticular skin pigmentations. Hoyeraal–Hreidarsson syndrome (HH) is a variant of DC and has a multi system involvement. It commonly affects males and presents with aplastic anemia, multiple infections, cerebellar-hypoplasia and growth retardation. The etiology is unknown.

Introduction

Dyskeratosis congenita (DC) and HH has inherited disorders. DC was first studied by Zinsser in the year 1906 **[1]** and Engman recognized it as a clinical condition in the year 1926 **[2]** and Cole in 1930 **[3]**. DC has a classical triad including reticular skin pigmentation, nail dystrophy and oral leukoplakia. Bone marrow involvement being the most common cause of death **[4]**. The age of onset is 5 - 50 years. The prevalence of the disease is 1 in 1 million individuals **[5,6]**.

Case presentation

A 10 years of age female patient from Pakistan presented to the pediatrics outpatient department with painful mouth ulcers, dryness of skin, hair loss and drying of the scalp for the past 3 years. She was born at full-term and her parents had a non-consanguineous marriage. She was the only child of a 28-year-old mother. At birth, her head circumference was 27 cm. The patient had delayed milestones. Laboratory investigations are given below in **table 1**. Physical examination showed oral white patches showing oral leukoplakia and acute necrotizing ulcerative gingivitis (ANUG) with focal acute pseudomembranous candidiasis with dental caries as shown in **figure 1**. She also had full body xerosis. She was micro cephalic, with sparse eyebrows and eye lashes, dystrophic fingernails as shown in **figure 2**. The skin of soles was hyperkeratotic. Rest of the examination was unremarkable. Her weight, height and head circumference were under 3rd centile. Her Echocardiogram showed hypertrophied left ventricle with evidence of aortic stenosis (13.2 mm of hg) and moderate regurgitation of aortic valve with trace of mitral regurgitation as shown in **figure 3**. Rest of the examination was unremarkable. Bone marrow aspiration study showed aplastic anemia. Computerized tomography (CT) scan showed cerebellar hypoplasia as shown in **figure 4**. The patient was started on oral retinoid treatment for the oral lesions. Topical steroid ointment was prescribed for external use for the oral lesions. Patient was regularly monitored for any dermatological, hematological, and malignant changes.

Table 1: Laboratory investigations

Complete Blood Count (CBC)	
Red blood cells (RBCs)	1.42 × 10^12/L
White blood cells (WBCs)	9.45 × 10^9/L
Platlets	55 × 10^9/L
Hemoglobin (Hb)	2.4 g/dl
Neutrophils	4.69 × 10^9/L
Lymphocytes	4.25 × 10^9/L
Eosinophils	0.00 × 10^9/L
Basophils	0.10 × 10^9/L

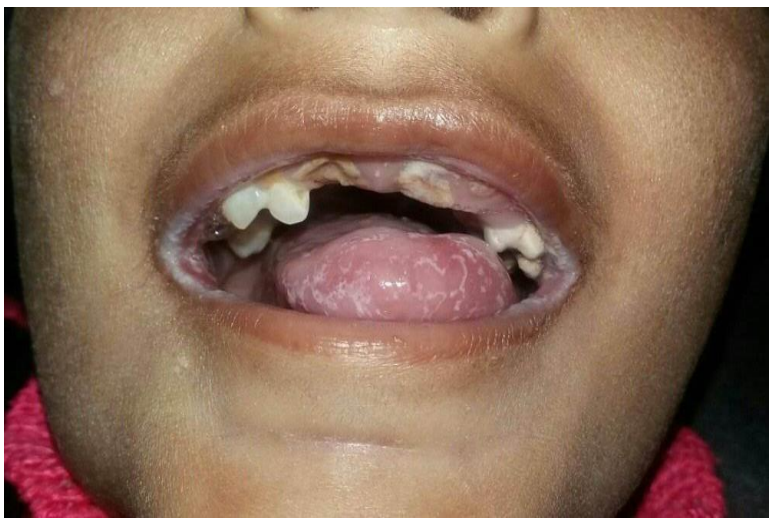

Figure 1: Oral leukoplakia and acute necrotizing ulcerative gingivitis

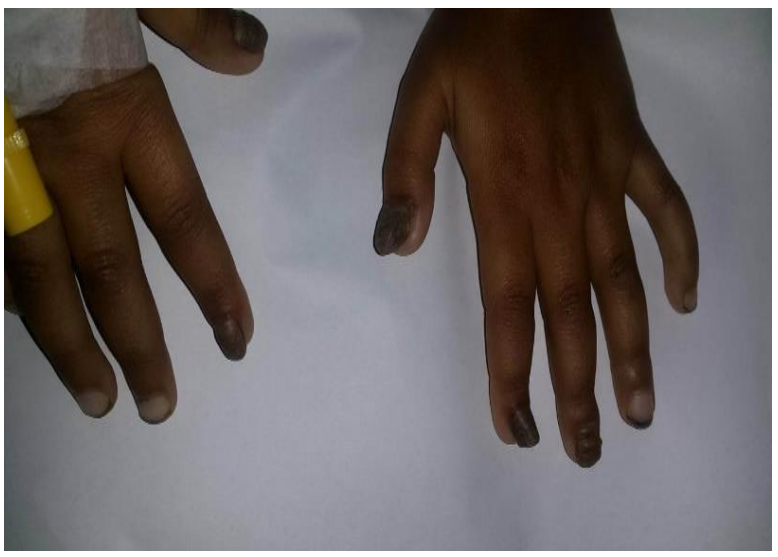

Figure 2: Dystrophic fingernails

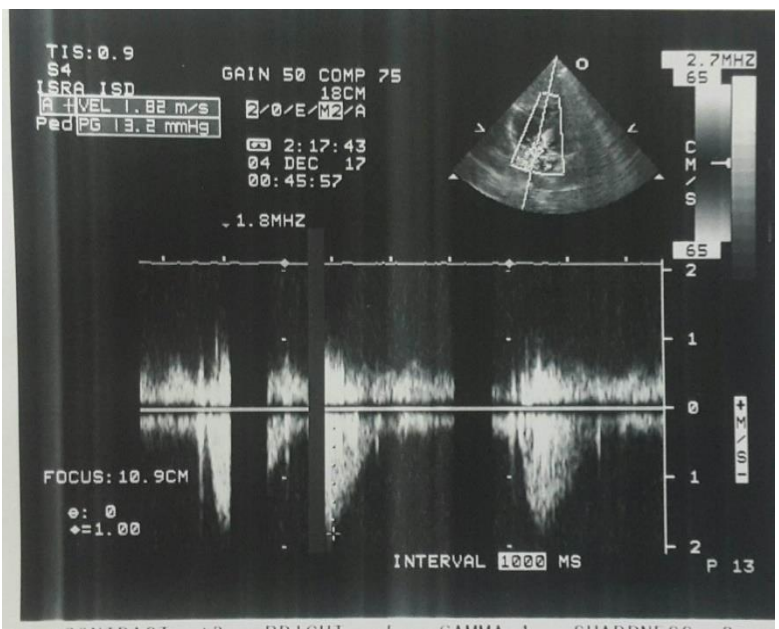

Figure 3: Echocardiography report showing aortic stenosis

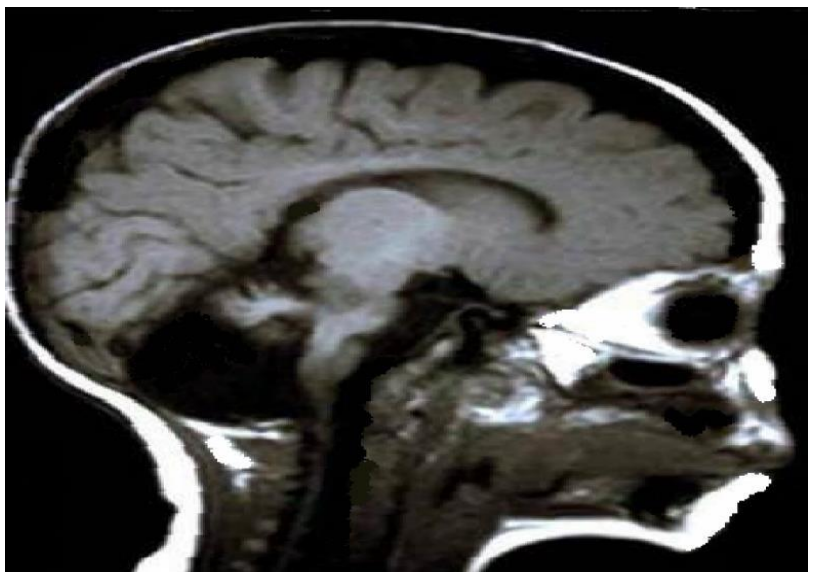

Figure 4: Cerebellar hypoplasia

Discussion

DC has characteristic features such as nail dystrophy, hyper pigmentation of skin and oral leukoplakia **[7]**. The disease is very rare and in Pakistan this is the first case reported for DC and HH. The age of onset of this disease is 5-15 years **[8]**. Our patient was ten years old. ANUG is rare in DC, our patient had ANUG with oral leukoplakia. Bone marrow failure is a very late complication of DC **[9]**. Our patient had deteriorating bone marrow function. Hoyeraal–Hreidarsson (HH) syndrome is a very uncommon type of dyskeratosis congenita (DC). The patients have growth retardation, microcephaly, cerebellar hypoplasia, aplastic anemia and bone marrow failure **[10]**. Our patient had growth retardation microcephaly and cerebellar hypoplasia.

Conclusion

Pancytopenia and other complications as a result of DC can lead to immunological disturbances making the person more prone to infection and death. The etiology of the disease and the treatment is still not known. More studies should be done on this clincial manisfestation to help decrease morbidity and mortality.

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