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A Case Study of Dyskeratosis Congenita: Clinical Manifestations and Diagnostic Challenges

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Abstract: ***Dyskeratosis congenita (DC), is a rare hereditary disorder predominantly affecting males and characterized by reticular skin pigmentation, nail dystrophy, and oral leukoplakia. This case study presents a 17 - year - old male diagnosed with DC, with classical triad of dystrophy of the nails, reticular skin pigmentation and oral leukoplakia along with additional complications including dyspnea, bilateral pedal edema, and multiple pathological fractures. Despite an early diagnosis in 2018, the patient received no treatment until recent symptoms prompted further medical intervention. This case underscores the importance of early diagnosis and ongoing management to mitigate the severe complications associated with DC.***

Keywords: dyskeratosis congenita, anemia, bone marrow failure, pathological fractures

1. Introduction

Dyskeratosis congenita (DC), or Zinsser - Cole - Engman syndrome, is a rare hereditary disease primarily affecting males, typically between 5 to 12 years old (1). It is characterized by a triad of reticular skin pigmentation, nail dystrophy, and oral leukoplakia (2). DC is a fatal condition often leading to aplastic anemia and cancer due to shortened telomeres and mutations in telomere biology (3, 4). Additional symptoms include epiphora, blepharitis, alopecia, esophageal and urethral stenosis, pulmonary fibrosis, avascular necrosis, epithelial cancers, myelodysplastic syndrome (MDS), and leukemia. This case study involves a young male with the classic triad, dyspnea, bilateral pedal edema, and multiple pathological fractures.

2. Case Report

A 17 - year - old male presented with dyspnea on exertion and bilateral pedal edema over the past month. He denied yellowish discoloration, hematuria, melena, chest pain, palpitations, or decreased urine output. Additionally, he had spooning and delayed nail growth, with brownish skin pigmentation over his chest, trunk, arms, and face since 2018. He underwent surgery for bilateral lacrimal punctal stenosis and phimosis the same year, complicated by post - operative bleeding. Further examination and investigations in 2018 diagnosed him with dyskeratosis congenita, though he received no treatment. In 2020, he developed a pathological fracture of the left femur, managed conservatively with a Thomas splint and traction due to surgical ineligibility (Image 7).

During the physical examination, he appeared as a young man of average build with a body mass index of 19.53 kg/m² (weight 50 kg, height 161 cm). Pallor was present. Mucocutaneous examination revealed fine, reticulate, grey - brown pigmentation on the chest, abdomen, arms, thighs, and back (Image 1, 2, 3). Leukoplakia was observed in the oral cavity, characterized by irregular blackish and whitish patches on the buccal mucosa and tongue (Image 4). Dental caries was evident, and dystrophy was present in all the nails (Image 5, 6).

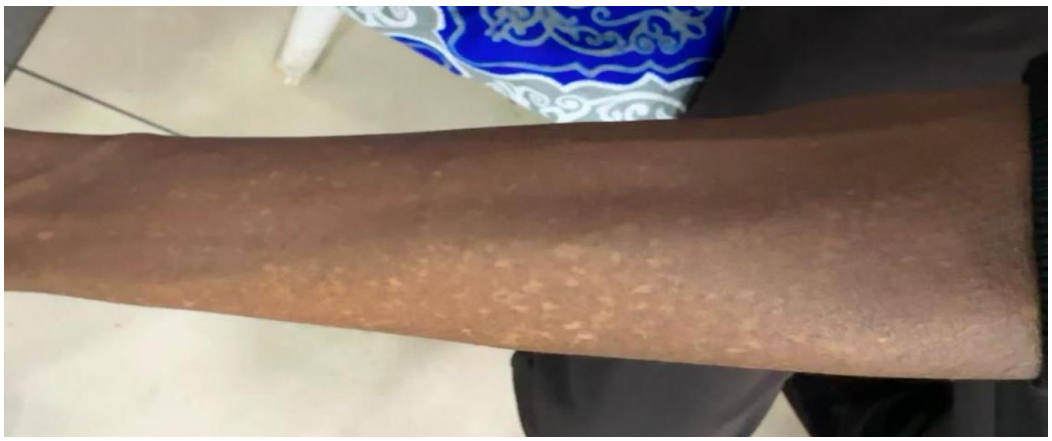

Image 1: Reticulate hyperpigmentation on forearm

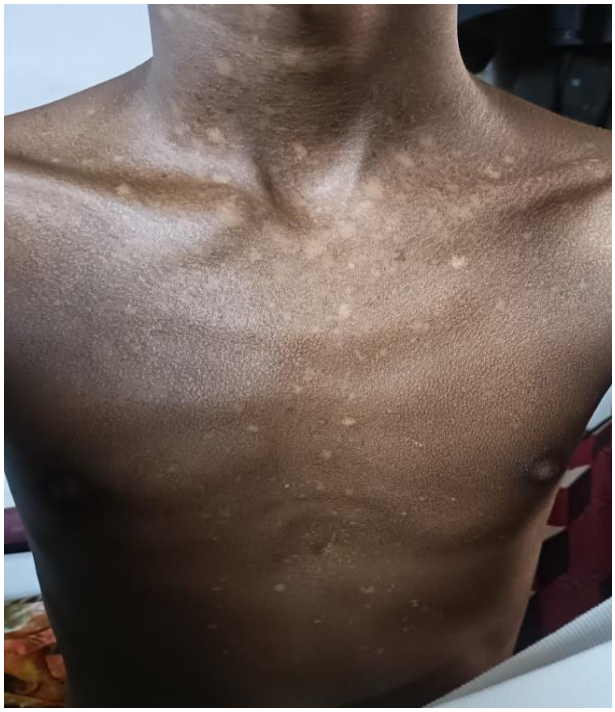

Image 2: Reticulate hyperpigmentation on chest

Image 3: Hyperpigmentation on face

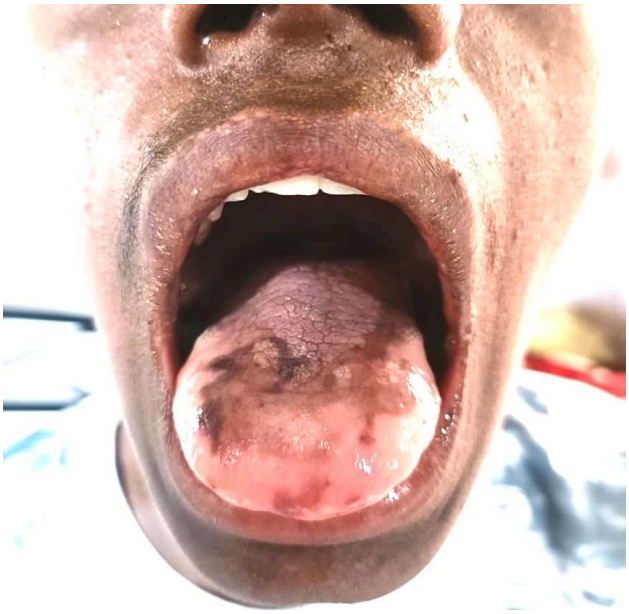

Image 4: Leukoplakia on tongue

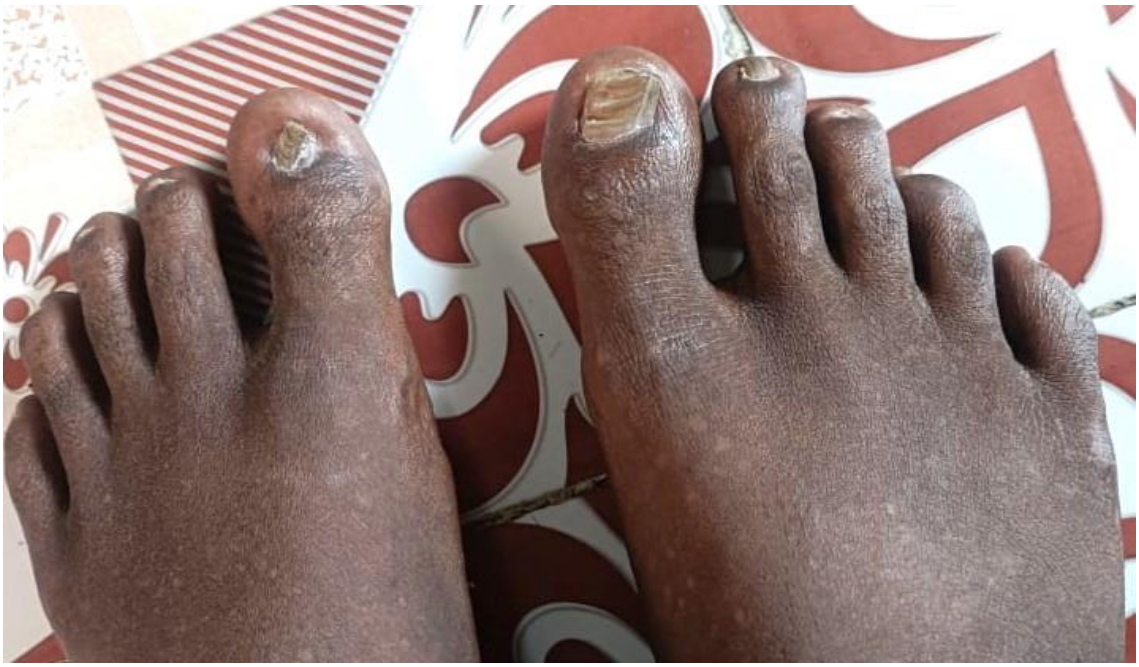

Image 5: Nail dysplasia of toe nails

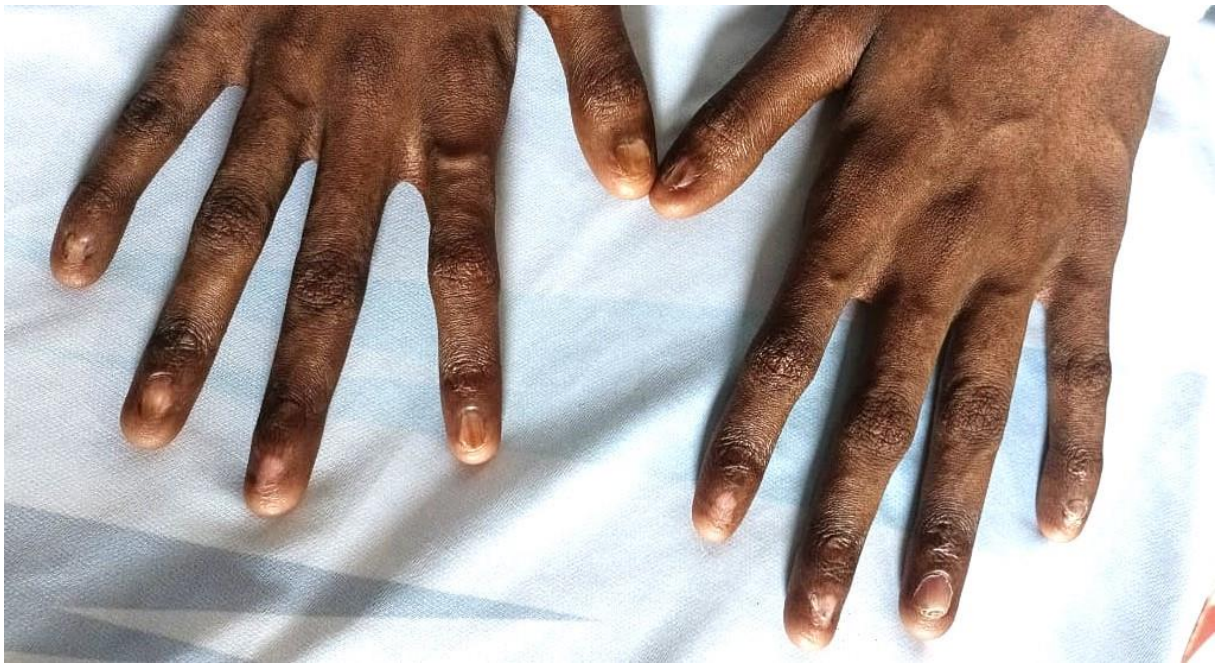

Image 6: Nail dysplaisa of finger nails

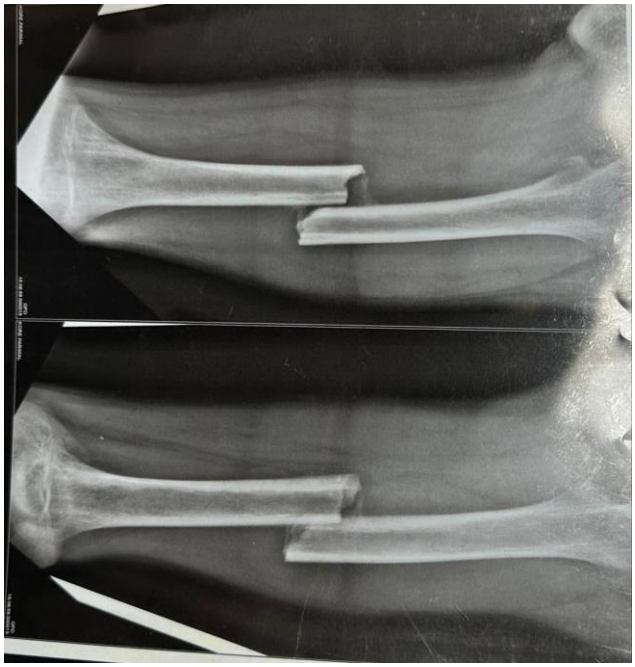

Image 7: Fracture of shaft of right femur

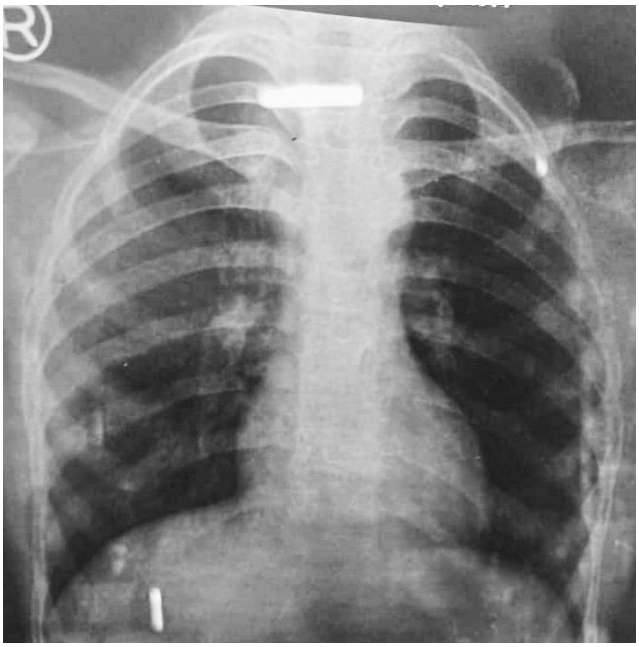

Image 8: Chest Xray

3. Investigation

Routine hematological tests revealed the following results: Hemoglobin (Hb) 3.2 g/dL, Total leukocyte count (TLC) 1600/µL with differential count (Neutrophils 63%, Lymphocytes 30%, Monocytes 6%, Basophils 1%), and platelet count 5000/µL. Red blood cell (RBC) count was 1.03 million/µL, hematocrit (Hct) 10.9%, Mean Corpuscular Volume (MCV) 105.6 fL, Mean Corpuscular Hemoglobin (MCH) 30.6 pg, Mean Corpuscular Hemoglobin Concentration (MCHC) 29 g/dL, and Red Cell Distribution Width (RDW) 15.4%. Peripheral blood smear revealed anisopoikilocytosis, macrocytosis, hypochromasia, thrombocytopenia and leukopenia.

Bone marrow aspiration done in 2020 showed reduced erythrocyte precursors, thrombocytopenia, and leukopenia. Bone marrow biopsy showed markedly reduced hemopoietic marrow with predominant fatty marrow, sparse erythroid and myeloid cells, markedly reduced megakaryocytes, suggestive of hypoplastic marrow.

Other investigations showed negative results for ICT, DCT, and sickling test. Serum levels of LDH, liver function tests (LFT), renal function tests were within normal limits.

Additional findings includes 2D echocardiogram showing preserved left ventricular systolic function with a 60% ejection fraction and a thin layer of pericardial effusion. Abdominal ultrasound revealed mild splenomegaly, and chest X - ray findings were normal (Image 8).

Treatment

The patient at present recieves danazol, calcium and vitamin d supplementation, ferrous sulphate and folic acid supplimentaion, and multivitamins, along with high protein diet.

4. Discussion

Dyskeratosis congenita (DKC) manifests typically with a triad of symptoms, reticulated skin hyperpigmentation, nail dystrophy, and oral leukoplakia (7). However, milder forms may not exhibit the complete triad. Nail changes usually appear first, followed by skin pigmentation and then oral leukoplakia, beginning by 10 years of age (7). Diagnosis requires at least two major features (abnormal pigmentation, nail dystrophy, leukoplakia, bone marrow failure) and additional somatic features seen in DKC (8).

The hyperpigmentation appears in a reticulated pattern due to epidermal atrophy, capillary hyperplasia, and melanin deposition near blood vessels in sun - exposed areas. Ectodermal abnormalities include scalp, eyebrow, and eyelash alopecia, premature graying, and palmoplantar hyperkeratosis. Nail dystrophy starts with ridging and can progress to absent nails, typically affecting fingernails before toenails. Leukoplakia affects oral mucosa, with potential constriction and stenosis in other areas like the esophagus or genitals (9).

Approximately 90% of patients develop peripheral cytopenia, often by age 20, with bone marrow failure a major cause of mortality due to bleeding and infections (10). Malignancy is another significant cause of death, usually emerging in the third decade of life (11). Early diagnosis and vigilant monitoring are crucial for managing dyskeratosis congenita effectively.

5. Conclusion

This case highlights the classic presentation of Dyskeratosis Congenita, demonstrating the triad of symptoms alongside features such as epiphora, phimosis, and multiple pathological fractures. The patients extensive cutaneous involvement and systemic manifestations emphasize the need for early diagnosis and comprehensive management to improve outcomes and quality of life for individuals affected by this rare condition.

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