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A CASE REPORT ON A RARE DISEASE: ZINSSER-ENGMAN-COLE SYNDROME (A.K.A DYSKERATOSIS CONGENITA)

Pathology

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ABSTRACT

Zinsser-Engman-Cole syndrome is a rare but complex multi-system disorder. The clinical diagnosis of classical Dyskeratosis Congenita requires at least two features of the triad of dysplastic nails, lacy reticular pigmentation of the upper chest and/ or neck and oral leucoplakia. A 16-year-old boy presented to our hospital with complaints of swelling on the right side of neck and pain on swallowing solids, liquids and saliva. Leucoplakia was noted on his tongue and all of his fingers and toe nails were markedly dysplastic. His skin was spotted with reticular pigmentation on the upper trunk and back. Also, a swelling of size 6 cm in diameter was noted on the right side of his neck. FNAC was done from the right neck swelling which showed metastatic Squamous Cell Carcinoma. It was followed up with a biopsy from laryngeal wall which showed Well Differentiated Squamous Cell Carcinoma. Individuals with DC most commonly present with skin pigmentation, dystrophic nails and leucoplakia, as presented in this case. Associated epithelial malignancies are common. DC is a severe multisystem disorder associated with premature morbidity and mortality. We believe that reporting this case would add more information to the existing fund of knowledge.

KEYWORDS

Dyskeratosis congenita, Skin pigmentation, leukoplakia, Dysplastic nails, Epithelial malignancy.

INTRODUCTION

Zinsser-Engman-Cole syndrome is a rare but complex multi-system disorder [1]. The clinical diagnosis of classical Dyskeratosis Congenita requires at least two features of the triad of dysplastic nails, lacy reticular pigmentation of the upper chest and/ or neck and oral leucoplakia.[2] Additional features include bone marrow failure, epithelial cancers, myelodysplastic syndrome, leukemia, epiphora, blepharitis, prematurely gray hair, alopecia, developmental delay, short stature, cerebellar hypoplasia, microcephaly, esophageal stenosis, urethral stenosis, pulmonary fibrosis, liver disease and avascular necrosis of hips or shoulders.[3]

Initially believed to be a skin condition that also affects the mouth and nails when it was first mentioned in the medical literature in 1906, DC has now been more widely recognised.[4] Patients with these skin abnormalities eventually develop bone marrow failure, but this wasn't discovered until later in the 1960s. 200 cases have been documented in the literature, and the incidence of classic DC is 1/1,000,000 people.[5]

Case Report

A 16-years old male presented to our hospital with complaints of swelling on the right side of neck and pain on swallowing solids, liquids and saliva. He had fever in the last couple of days and complained of feeling easily exhausted but his weight or appetite was not changed. He had no cough, bowel or urinary compliant. He had no travel history or similar symptoms in the family. He was never exposed to poisonous gas or radiation. He had not taken any medications for his illness or other reasons. He did not smoke, drink alcohol or use illicit drugs.

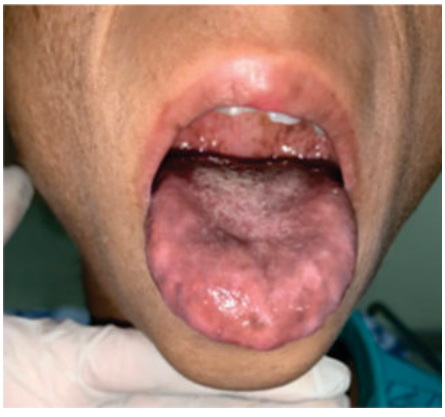
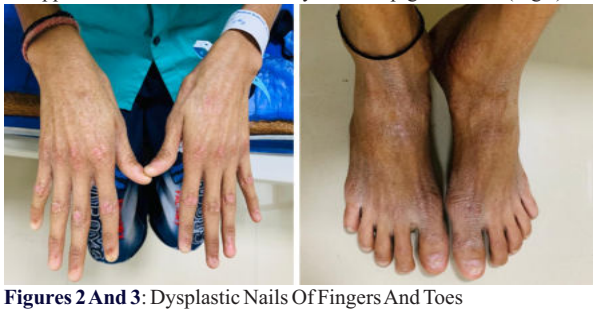


Figure 1: Oral Leukoplakia

On clinical examination of this patient, a swelling of size approximately 6 centimeters in diameter was noted on the right side of the neck. Leukoplakia (Fig 1) was noted on his tongue and his entire fingers and toenails were dysplastic (Fig 2 and Fig 3). Also, the skin on his upper trunk and back showed lacy reticular pigmentation (Fig 4).



Figures 2 And 3: Dysplastic Nails Of Fingers And Toes

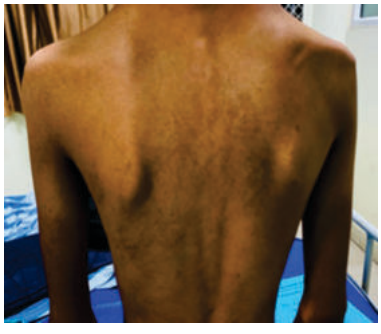
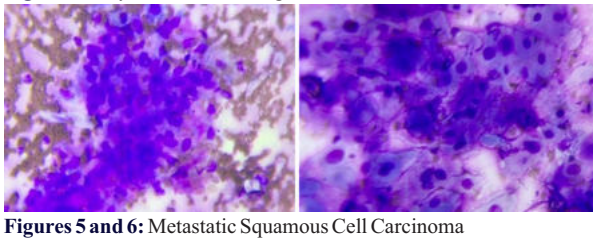


Figure 4: Lacy Reticular Skin Pigmentation



Figures 5 and 6: Metastatic Squamous Cell Carcinoma

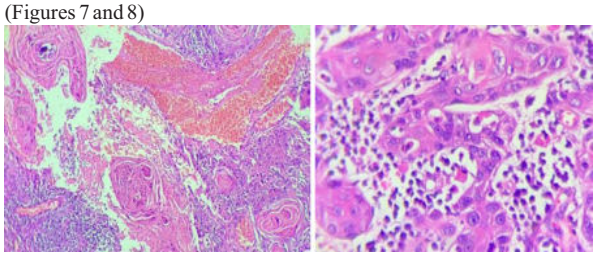
Fine Needle Aspiration Cytology Was Performed From The Right Neck Swelling

FNAC showed malignant cells arranged in sheets, clusters and singly

dispersed. Tumor cells are highly pleomorphic with round to oval angulated nuclei and abundant glassy blue cytoplasm. Areas of necrosis, anucleate squamous and keratin pearls are also seen. (Figures 5 and 6).

Bronchoscopic biopsy was performed the day after the FNAC from the laryngeal wall

Biopsy showed tumor tissue arranged in trabeculae, sheets and nests. The tumor cells have pleomorphic vesicular nuclei, conspicuous nucleoli, dense eosinophilic cytoplasm. Individual cell keratinization and abundant keratin pearls seen. Dense lymphocytic infiltrate noted. (Figures 7 and 8)



Figures 7 and 8: Squamous Cell Carcinoma: Well-differentiated

Final diagnosis given was Squamous cell carcinoma of the larynx with metastasis to right cervical lymph node.

The patient was then referred to the department of oncology for further treatment and management.

DISCUSSION

DC is characterized by a triad of dysplastic nails, skin pigmentation, and leukoplakia. The Dyskeratosis Congenita Registry (DCR)[6] requires the presence of either two of the three features of the triad or one of the triads with at least two minor features. It can be associated with epithelial malignancies as presented in this case. The classical age group is 15 years(range 0-75 years)[7] as presented in this case.

Studies over the last 10 years have demonstrated that DC is principally a disease of defective telomere maintenance. DC patients have very short telomeres and the genetically characterized cases of DC have mutations in different genes which encode components of the telomerase complex.[8]

Major cause of death is malignancy occurring mainly on mucosal surfaces.[9] Therefore, it is vital to recognize the increased risk of upper aero-digestive tract cancers in these patients (11-fold increase compared to general population) as seen in our case with the presence of laryngeal squamous cell carcinoma.

Hence we would like to emphasize the importance of early diagnosis, prompt actions, the importance of follow-up and the inter-disciplinary approach to tackle this disease.

CONCLUSION

DC is a severe multisystem disorder associated with premature mortality. We presented a patient who was diagnosed as DC with laryngeal squamous cell carcinoma. We believe that by reporting this presentation of the patient would add more information to the existing fund of knowledge. DC is being now studied ever more intensively and this case report can be beneficial in early diagnosis of underlying associated malignancies and to save the lives of those suffering from it.

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