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A Case Report on: Dyskeratosis Congenital

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ABSTRACT: -

Bone marrow failure, an 11-fold increase in cancer risk relative to the general population, ectodermal dysplasia (nail dystrophy, mouth leukoplakia, and aberrant skin pigmentation), and other somatic abnormalities are the hallmarks of the rare genetic condition dyskeratosis congenita (DC). A male, age 22, had symptoms of chills, fever, and sore throat. His tongue showed signs of leukoplakia, and some of his fingers and toe nails showed obvious dystrophies. He appeared to have pigmentation spots on his neck and front chest. Leukaemia and "blood disease" were mentioned by the patient's family history. In order to treat his neutropenic fever (102.9 °F, WBC: 940, ANC: 404, platelets: 21,000, and Hb: 9.2), he was admitted and started on wide spectrum antibiotics. The results of a bone marrow biopsy showed mild dyserythropoiesis and normocellular marrow with an erythroid predominance.There were no structural or numerical chromosomal abnormalities found by chromosomal analysis. No assay-specific anomalies were found in the fluorescence in situ hybridization report. Studies on autoimmune antibodies and the hepatitis virus marker produced disappointing findings. Splenomigly was revealed on a CT scan. With a follow-up appointment at the haematology clinic, the patient was released once he had no fever or symptoms. As in this example, leukoplakia, dystrophic nails, and skin pigmentation are the most prevalent presentations for people with DC. Although our patient's genetic anomaly was not validated, it is hypothesised that the X-linked recessive pattern, which is more common in men than in women (10:1), would be relevant.Our patient is classified in the most common class of mild aplastic anaemia based on the distribution of blood counts and a bone marrow biopsy. It's critical that medical professionals rule out any underlying causes of fever in young kids whose bone marrow is declining. Even though DC is an uncommon disease, it can be identified with just a cursory examination of the mucocutaneous anomalies. DC is a serious multisystem illness linked to early morbidity and death. Reporting this occurrence, in our opinion, would contribute further details to the body of current knowledge.

KEYWORDS: Congenital dyskeratosis, hereditary illness, failing bone marrow, nail deterioration Phallosalpage, colouring of the skin, Tetronomere, uncommon illness

INTRODUCTION:-

The three main symptoms of the uncommon genetic disease dyskeratosis congenita (DC) are nail dystrophy, oral leukoplakia, and aberrant skin pigmentation [1]. Additional manifestations consist of bone marrow insufficiency, an inclination towards cancer, and lethal respiratory issues [2]. Bone marrow failure accounts for 80% of observed mortality in DC patients and is the primary cause of death in approximately 85% of cases [3]. DC was initially believed to be a skin condition that also affected the mouth and nails when it was first mentioned in medical literature in 1906. It was discovered that patients with these skin abnormalities experience bone marrow failure only later in the 1960s. Classic DC affects about 1 in every 100,000 people; 200 cases were reported in the literature.As researchers gain more knowledge, the diagnosis and treatment of DC by medical professionals are rapidly evolving. Nevertheless, immunodeficiency is a major cause of premature mortality in DC, and despite this, the immunologic features of DC remain underdiagnosed and undertreated [4]. One significant and significant side effect of DC is thought to be bone marrow failure. In an effort to raise awareness of this uncommon disease, we report on a case of a young man with DC who also had neutropenic fever.

PATIENT INFORMATION

Patient-Specific Information:- In A. V. B. R. H., a 22-year-old guy was admitted. On the 20th of June, 21st, with the chief complaint of fever, chills, a sore throat, fatigue, weakness (especially in the lower extremities), difficulty with gait, impaired poise and synchronization, fever, headache, chills, and arthralgias, the doctor diagnosed a case of Dyskeratosiscongenita after a physical examination and investigation.

MEDICAL HISTORY:-

A patient was admitted to the A. V. B. R. hospital on the 20th of June, 2021. The patient has complaints of being unwell. He had been sick for a few days with a fever, chills, and a sore throat that was becoming worse. And it's been three years since I've consumed alcohol or smoked. Five days before admission, he had a fever, headache, chills, and myalgia, and he finished illicit substances on a daily basis.The patient's tongue, as well as a few of his finger and toe nails, were noticeably dystrophic after his fever and headache subsided on the fourth day of symptoms. The patient had a history of Dyskeratosiscongenita**.**

CLINICAL FINDING:-

PHYSICAL EXAMINATION:-

Reticulated darkening of the skin, nail dystrophy, and leukoplakia are all symptoms of Dyskeratosiscongenita (D.K.C.). Affected patients may have a number of other clinical signs in addition to the diagnostic mucocutaneous features and bone marrow loss, such as fever, chills, and a sore throat that is worsening. Temperature 98.6 degrees Fahrenheit, pulse 72 beats per minute, respiration 20 breaths per minute, and blood pressure 120/80 mm of mercury

IMPORTANT CLINICAL FINDINGS:-WBC: 5000 cells/mm3 (Blood Investigation) IgM and IgG tests: Positive, RBC: 20,000-40,000 (cells/mcL), IgM and IgG test: Positive, 12,000 cells/mm3 platelet count

THERAPEUTIC INTERVENTIONS:-Antipyretics were administered in the present case of Dyskeratosiscongenita. Inj. Ceftriaxone 1gm IV (BD), Inj. Pantoprazole 40 Mg iv (OD), Inj. Ondansetron 4MG iv (TDS).

NURSING PERSPECTIVES:-Fluid substitution (DNS and R.L.) is administered, and vital signs are monitored hourly. 2 hourly vital charts were meticulously kept, as was the intake output chart. Tab. Antibiotics with paracetamol were prescribed by the doctor.

DISCUSSION:-

The three main characteristics of DC are leukoplakia, skin pigmentation, and dystrophied nails. It may be connected to pancytopenia, or bone marrow failure, as demonstrated in this instance. Bone marrow failure typically develops before the age of 20, as was characteristically observed in our patient [1]. Skin pigmentation and nail alterations typically manifest first, before the age of 10. Although the presence of X-linked recessive gene was not confirmed in our patient, its higher frequency in men than in women (ratio of 10:1) suggests that this gene would be significant [2,3]. Over the past ten years, research has shown that misplaced telomere maintenance is the primary cause of DC. Patients with FC have extremely short telomeres, and genetically defined cases of DC have mutations in various genes encoding telomerase complex components [4]. Thus far, mutations in CTC1, DKC1, TERC, TERT, TINF2, NHP2, NOP10, and WRAP53 have been identified as the genes responsible for DC and extremely short telomeres. About half of the people who fit the clinical diagnostic criteria for DC have mutations in one of these eight genes [5].Excessive telomere attrition resulting from telomerase mutations causes premature cell death and chromosome instability, which ultimately depletes the stem cell reserve and causes clinical features like BM failure [6]. The blood count distributions in our case place the patient in the moderate aplastic anaemia class; these results are indicative of the most common pattern. Malignancy, which mostly affects mucosal surfaces exhibiting leukopenia, is another important cause of death. The elevated risk of upper aero-digestive tract cancers in these patients (11-fold increase compared to general population) must thus be acknowledged [7, 8].It's critical for medical professionals to rule out any underlying causes of fever in young patients whose bone marrow is failing. Although DC is a rare disease, it can be identified with a straightforward examination of the mucocutaneous abnormalities. The classic triad of oral leukoplakia, reticular pigmentation of the neck and upper chest, and abnormal fingernails and toenails serves as a diagnostic tool [9]. Individuals who fit the clinical diagnostic requirements ought to be the subject of more research. Consequently, physicians should be informed that DC may contribute to bone marrow failure in paediatric patients, which can result in life-threatening consequences like bleeding and infection.

CONCLUSION:-

The patient was brought to A.V.B.R.H. with a fever, chills, sore throat, increased exhaustion, limitation (especially in the lower limbs), walking difficulties, impaired equilibrium and coordination, and other symptoms. The patient with dyskeratosis congenita received the appropriate care and medication. The patient's condition has improved.

CONCLUSION:-

DC is a serious multisystem illness linked to early death, typically as a result of bone marrow failure. We saw a patient with moderate aplastic anaemia complicated by neutropenic fever, who was identified as DC. We think that sharing the patient's presentation will broaden the body of knowledge already in existence. DC is currently the subject of ever-intense research, and this case study can help both medical professionals and patients who are afflicted with this uncommon illness.

REFERENCE :

1. Dyskeratosiscongenita in all its manifestations (Dokal I). Br J Haematol, vol. 110, no. 4, pp. 768–779. http://dx.doi.org/10.1046/j.1365-2141.2000.02109.x [PubMed] [CrossRef] [Source: Google Scholar] https://bmchematol.biomedcentral.com/articles/10.1186/1471-2326-11-3

2. Stem cells, and telomeres. 2. Kirwan M, Dokal I. BiochimBiophysActa, vol. 1792, no. 4, pp. 371–379. http://dx.doi.org/10.1016/j.bbadis.2009.01.010. [Free article from PMC] [PubMed] [CrossRef] [Source: Google Scholar]

3. Carola Duran-Mckinster, MarimarSaez-De-Ocariz, Luz Orozco-Covarrubias pp. 661–668 in Dyskeratosis Congenital: Neurocutaneous Disorders, Phakomatoses, and Hamartoneoplastic Syndromes. [Source: Google Scholar]

4. S.Jyonouchi, L. Forbes, E. Ruchelli, and K. E. Sullivan. A single-centre paediatric experience with dyskeratosis congenital, a combined immunodeficiency with a broad clinical range. 10.1111/j.1399-3038.2010.01136.x. Pediatr Allergy Immunol. 2011;22(3):313–319. [PubMed] [CrossRef] [Source: Google Scholar]

5. Advances in the knowledge of dyskeratosis congenital. Walne AJ, Dokal I. 10.1111/j.1365-2141.2009.07598.x.Br J Haematol. 2009;145(2):164–172. DOI: 10.1111/j.1365-2141.2009.07598.x. [Free article from PMC] [PubMed] [CrossRef] [Source: Google Scholar]

6. Sharon is a ruthless woman. Congenital Dyskeratosis Gene's Opinions. NBK22301; NBK22301; NBK22301; NBK22301; NBK22301; NBK [Source: Google Scholar]

7. Dyskeratosiscongenital ,Dokal I. 2011;480–486 in Hematology Am SocHematolEduc Program. [PubMed] [Source: Google Scholar]

8. B.P. Alter, N. Giri, S.A. Savage, and S.A. Rosenberg PS. Dyskeratosiscongenita cancer. Blood, 113(26), 6549–6557, 2009. Blood, 2008-12-192880, DOI: 10.1182/blood-2008-12-192880. [Free article from PMC] [PubMed] [CrossRef] [Source: Google Scholar]

9. Ballew, B.J., and Savage, S.A. Updates on dyskeratosis congenital and associated telomere biology diseases, including biology and treatment. Expert Rev Hematol, 6(3), 327–337, 2013. ehm.13.23 (DOI: 10.1586/ehm.13.23). [PubMed] [CrossRef] [According to Google Scholar]