Case Report

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Paediatrics Section

Complete Mouth Rehabilitation in a Patient with Fanconi Anaemia: A Rare Genetic Disorder

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ABSTRACT

Fanconi anaemia is a rare, progressive congenital bone marrow failure syndrome characterised by autosomal recessive inheritance and clinical manifestations, including growth retardation, bone marrow failure leading to pancytopenia, an elevated risk of malignancy, skin pigmentation, and skeletal malformations. Patients who have undergone bone marrow transplantation can be treated comprehensively in the dental clinic like other routine patients, provided their blood indices are within normal limits. Its complications include aplastic anaemia, malignancies like acute myeloid leukaemia, liver tumours, and squamous cell carcinoma, often developing post-bone marrow transplantation. Dental anomalies, altered salivary flow, and increased susceptibility to cancer necessitate early diagnosis and specialised dental care. Hereby, the authors present a case of 15-year-old male with Fanconi Anaemia who had undergone bone marrow transplantation and was treated comprehensively in a hospital setting. The present case report aimed to equip healthcare practitioners, especially dentists, with the knowledge needed for accurate diagnosis and effective management.

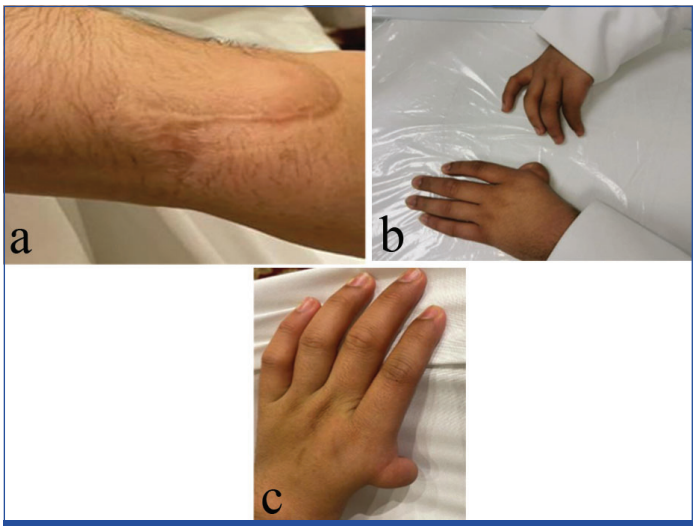
Keywords: Autosomal recessive, Bone marrow transplantation, Hypoplasia, Periodontitis

CASE REPORT

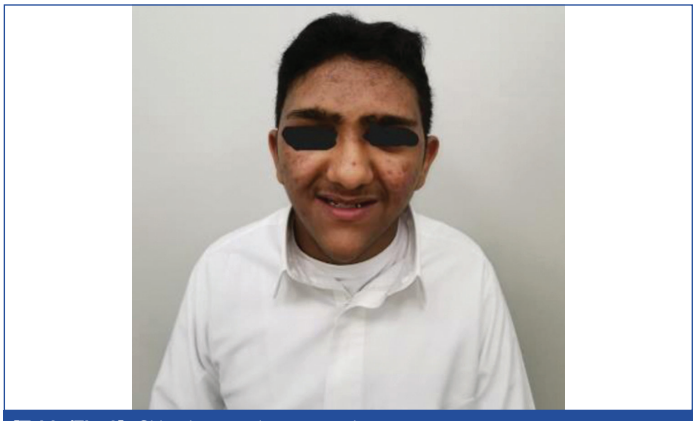
A 15-year-old male patient presented to the Oral Medicine Department at the College of Dentistry, Imam Abdulrahman bin Faisal University, Dammam, Eastern Province, Saudi Arabia, for a routine dental check-up. He was diagnosed with Fanconi Anaemia (FA) at the age of two and underwent cord blood transplantation in 2008.

Physical examination revealed a short height of 150 cm [Table/Fig-1] and a weight of 40 kg, suggestive of a stunted growth pattern. The flexor surface of the right hand [Table/Fig-2a] showed scarring, likely due to healed lesions of neutrophilic dermatosis, also known as Sweet syndrome. Both the right and left thumbs displayed abnormal shortening compared to the other digits [Table/Fig-2b,c]. The patient has a tendency to keep his digits inwards to hide the malformed thumbs. Additionally, midface hypoplasia was evident, characterised by the absence of cheekbone prominence along with skin pigmentation over the malar area [Table/Fig-3].

[Table/Fig-1]: Short height of the patient with inward stance of digits.



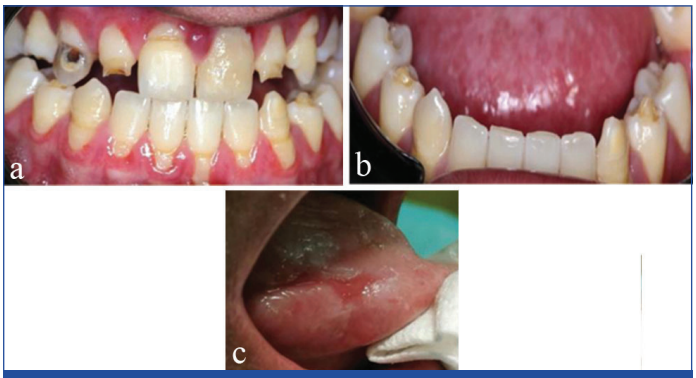
[Table/Fig-2]: a) Scarring on flexor surface of right hand; b) Abnormal shortening of thumbs of both hands; and c) Close up view of left hand.



[Table/Fig-3]: Skin pigmentation over malar area.

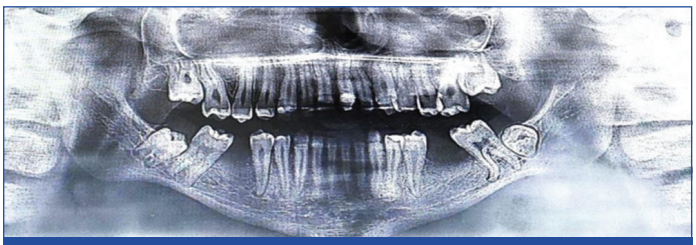
Intraoral examination revealed poor oral hygiene, generalised gingivitis, and gingival recession. Arrested caries was seen on the labial surface of the maxillary right canine (tooth # 13) [Table/Fig-4a], along with congenitally missing first permanent molars. Erosion was noted on the palatal surface of tooth # 21, which was non tender to percussion. Enamel hypoplasia with a fracture up to the cervical 1/3rd was seen in the maxillary right and left lateral incisors (Teeth # 12, 22). Dens evaginatus was seen on the mandibular right and left premolars (teeth # 34, 44 [Table/Fig-4b]). The dorsal surface of the

tongue exhibited a non specific ulceration [Table/Fig-4c] with a size of 3×2 cm, erythematous in appearance with ill-defined margins and no signs of tenderness upon scraping. The lesion has been present for one month. The patient was unable to move the tongue freely, with a restriction of lateral movements. Food debris was seen on the tongue.



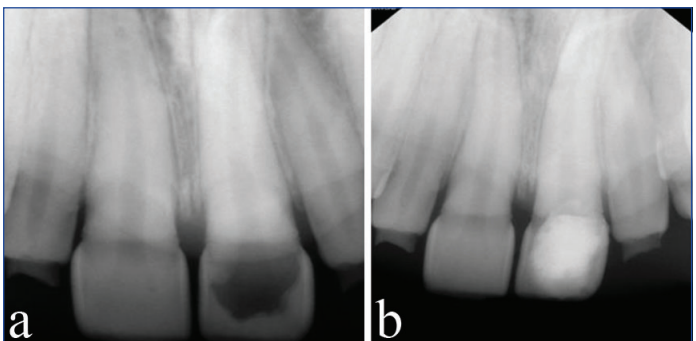
[Table/Fig-4]: a) Arrested caries on labial surface of tooth # 13 (b) Dens evaginatus over teeth# 34,44 (c) Ulcer on dorsal surface of tongue.

A panoramic radiograph [Table/Fig-5] confirmed the presence of dilaceration in the mesial root of tooth # 37, along with congenital absence of teeth # 16, 26, 36, and 46. Radiolucency involving the crown of tooth # 21 was observed. Reduced jaw height suggestive of jaw hypoplasia was apparent. The radiographic contrast between enamel and dentin was decreased, suggestive of hypoplasia. Furthermore, mandibular angle hypoplasia was evident on the radiographs.



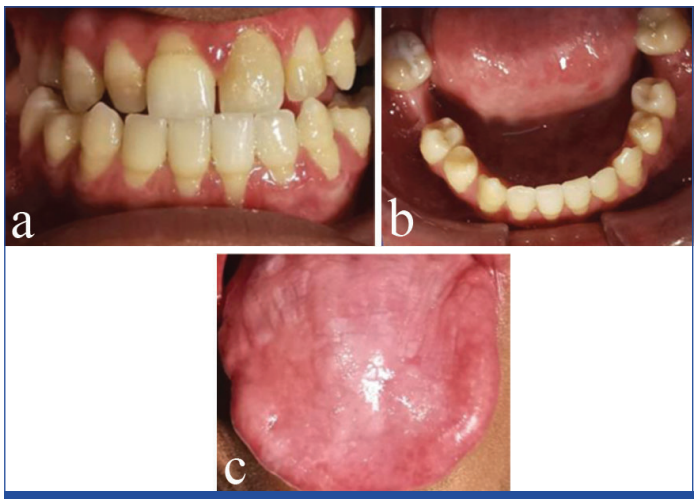
[Table/Fig-5]: In-treatment panoramic radiograph showing dilaceration in tooth # 37, missing tooth # 16,26,36,46.

Complete Blood Count (CBC) showed normal values for haemoglobin, Red Blood Cells (RBC), White Blood Cells (WBC), and platelet count, which can be attributed to the bone marrow transplant the patient underwent early in his childhood. Since, the patient's blood values were within normal limits, a comprehensive treatment plan was formulated for the patient. This included scaling and root planning, fissure sealants in the disease control phase. Pulp extirpation was performed, and temporary restorations were placed in tooth # 21 [Table/Fig-6a,b]. Composite build-up was performed at a later visit for teeth # 13, 12, 22, and composite build-up for teeth # 34, 44 was done in the restorative phase to enhance his smile line [Table/Fig-7a,b]. Candid mouth paint was prescribed for the tongue lesion, and there was complete resolution of the ulcer after two weeks



[Table/Fig-6]: Intraoral Periapical Radiograph (IOPAR) of tooth # 21 showing carious lesion on the palatal surface of tooth and placement of temporary restoration after pulp extirpation and canal cleaning and shaping.

[Table/Fig-7c]. The patient was recalled for completion of endodontic treatment of tooth # 21 and replacement of missing teeth but was unable to follow-up.



[Table/Fig-7]: (a,b) Post treatment image showing restored carious teeth and healthy gingiva (c) Complete healing of ulcer on dorsal aspect of tongue.

DISCUSSION

The FA, an autosomal recessive genetic disorder, presents a multitude of challenges in dental management. The present case report highlights the significant dental and oral health issues faced by a 15-year-old male patient with FA. The management of dental care for such patients requires a comprehensive, multidisciplinary approach that takes into account the patient's underlying condition, oral manifestations, and the potential complications associated with FA [1].

First described by Dr. Guido Fanconi in 1927, this genetic disorder has been the subject of extensive research and understanding over the years. FA is characterised by chromosomal instability and affects proteins responsible for Deoxyribonucleic acid (DNA) repair and cell cycle regulation [2-5]. It primarily presents as progressive bone marrow failure, growth retardation, and an increased susceptibility to malignancies [5]. Early diagnosis is essential, as the clinical presentation can vary significantly among affected individuals.

Dental anomalies are common in FA patients and encompass a wide range of issues, including gingivitis, periodontitis, dental caries, dental anomalies, and soft tissue lesions [1]. Poor oral hygiene, plaque accumulation, and hyposalivation contribute to dental caries, which affects a significant portion of these patients. Additionally, dental anomalies such as microdontia, tooth agenesis, abnormal tooth shape, and micrognathia are frequently observed [1].

One of the most critical challenges in managing dental care for FA patients is their elevated risk of developing head and neck Squamous Cell Carcinoma (SCC) [4,5]. A recent study reported a lower lip SCC in FA and its management [4]. This underscores the importance of regular oral examinations and early cancer detection, making dental professionals vital contributors to the overall healthcare team [1]. Another recent study by Gaitán-Fonseca C et al., stressed the preventive and restorative approach for a FA patient and reported a significant reduction in the recurrence of infections after a follow-up of more than12 months [5].

In present specific case, the patient presented with poor oral hygiene, gingivitis, gingival recession, severe caries, congenitally missing first permanent molars, enamel hypoplasia, and tooth malformation. A notable tongue lesion further complicated the picture, affecting the patient's ability to move the tongue freely. The comprehensive treatment plan included various dental interventions to address these issues, with a focus on oral infection removal and the restoration of function and aesthetics. The patient's case reinforces the need for a multidisciplinary approach to the management of FA. This approach involves collaboration with Haematologists, Paediatricians,

Endocrinologists, and other specialists to provide comprehensive care that addresses both systemic and oral health issues.

Effectively managing a patient with FA in the dental office requires a thorough approach. Collaborate closely with the patient's medical team to gather a comprehensive medical history and understand potential dental procedure risks. Emphasise minimally invasive techniques to minimise the risk of bleeding and infection, and consider antibiotic prophylaxis based on medical team recommendations.

Schedule regular follow-ups to stress the importance of oral hygiene. Educate the patient on proper oral care practices, limit radiation exposure, and create a comprehensive treatment plan addressing both preventive and restorative aspects. Use sedation judiciously and be prepared for emergencies. Maintain open communication with the patient, caregivers, and the medical team for a collaborative and well-informed dental care approach, ensuring optimal oral health while considering the distinctive challenges of FA.

CONCLUSION(S)

In conclusion, the present case underscores the importance of early diagnosis, specialised dental care, and a multidisciplinary approach in managing FA.

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