

A Case Report of Fanconi Anemia: A Literature Review and Dental Perspective of Rare Disease

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ABSTRACT

Introduction: Fanconi Anaemia (FA) is a very rare genetic disease characterised by genetic alterations, which causes congenital abnormalities in individuals. This clinical case report of Fanconi anaemia, will review classical signs of the disease in detail. The various aspects of this rare condition are examined, with an emphasis on oral manifestations and their impact on affected patients overall health. Since this group is more likely to acquire cancers, paediatric dentists must be aware of common oral symptoms and possibly malignant lesions in order to make an early diagnosis and provide thorough care and maintenance of oral health to those who are affected.

Case Presentation: A four-year-old female patient was brought by her parents to the Department of Pediatric and Preventive Dentistry, Yenepoya Dental college, with a chief complaint of pain in the upper back tooth region. The child had already been diagnosed with FA.

Conclusion: Patients with FA are more prone to get infections, so proper precautions should be taken to avoid any situation that might put them at risk of infection or bleeding. Patients should be motivated to have regular follow-ups and encourage them to maintain good periodontal health, to help prevent the incidence of caries and to monitor their overall oral wellbeing.

Keywords: Fanconi anaemia, genetic disease, rare disease, Congenital defects

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INTRODUCTION

Fanconi Anemia (FA) is a rare multisystem hereditary illness with symptoms that include fatigue, anaemia, bone marrow failure, somatic dysfunction abnormalities, pancytopenia and risk for cancer. Acute myeloid leukaemia (AML) is the most common type of malignancy seen in this group.¹

It was named after Guido Fanconi, a Swiss doctor who initially identified the condition in 1927. With an incidence of 1:350,000 births, it is an extremely rare condition.²

Congenital defects include short stature; microcephaly; endocrine problems; skin pigmentation in the form of café au lait spots; petechiae and bruises; infections; developmental disabilities; and abnormalities of the skin, arms, head, eyes, kidneys, and ears in up to 60%–75% of patients. Pancytopenia, anaemia, thrombocytopenia, leukopenia, macrocytosis, and fetal-like erythropoiesis are among the haematological consequences (D'Andrea 1997).

There are limited reports of oral manifestations in people with Fanconi Anemia in the literature. In this article, we present a case of FA, as well as a discussion of the most common oral symptoms and their impact on the health of FA patients.

CASE PRESENTATION

A four-year-old female patient was brought by her parents to the Department of Pediatric and Preventive Dentistry, Yenepoya Dental College, with a chief complaint

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of pain in the upper back tooth region. The child had already been diagnosed with FA. A cytogenetic test demonstrated chromosomal breakdown when mitomycin C was administered, confirming the diagnosis of FA. As the child's parents had a consanguineous marriage, the family history was significant. She was the only child and was born through a caesarean operation after 32 weeks of pregnancy. The child had undergone surgery for jejunum atresia after the fourth day of birth. The patient was found to be thrombocytopenic with low haemoglobin.

The child was conscious and cooperative, with well-

developed cognitive. The patient's height and weight were below average that is, 57cm and 5.5kg (Figure 1). Congenital abnormalities like hypoplasia of thumbs (Figure 2 &3) in both hands and deformed forearms were noted. No pigmentation of skin was observed.

The patient was a hyperactive child who couldn't sit still in a dental chair. Oral hygiene maintenance was poor, leading to recurrent carious lesions in the oral cavity.

On intraoral examination, multiple carious lesions were seen, dentoalveolar abscess was noticed on upper right buccal mucosa region (Figure 4 & 5). And soft tissues appeared to be normal.

Radiation exposure raises the likelihood of tumour formation in FA patients, thus radiographs were required but could not be taken in this case. Provisional diagnosis of acute periapical abscess in maxillary second deciduous molar was made and indicated for emergency access opening to relieve pain.

After Physicians consent was obtained emergency access opening was carried out. On subsequent visits cleaning and shaping done, the roof of the pulp chamber was removed using a high-speed handpiece and a no. 330 carbide bur (Dentsply Professional). A slow-speed no. 4 round bur was used to clear the pulp chamber. The canals were enlarged with files up to size #30. Sodiumhypochlorite and Saline was used to irrigate the canals, which were then dried with sterile paper points. Vitapex was used as the root canal filling material. The filling material was injected directly into the canals. And finally the tooth was restored with Glass ionomer cement. The patient was placed on 3 month recall schedule but patient didn't turn up for

recall visit.

DISCUSSION

Fanconi anaemia is a rare autosomal recessive disease marked by a variety of congenital defects. Short height, microcephaly, endocrine disorders, pigmentation of the skin in the form of brown patches, petechiae, and contusions, infections, and developmental difficulties, as well as abnormalities of the skin, arms, head, eyes, kidneys, and ears, affect 60 to 75 percent of FA patients. Increased pancytopenia, anaemia, thrombocytopenia, leukopenia, macrocytosis, and foetal erythropoiesis are among the hematologic problems that develop with time³. Almost 300 people with FA showed thumb anomalies such as a missing thumb or hypoplastic thumb (Figure 2 & 3), floating or bifid thumb, polydactyly, and triphalangeal thumbs, according to data analysed from 700 patients with FA in the literature.⁴

For accurate diagnosis and treatment, it's essential to rule out hereditary illnesses. In the developing world, consanguineous marriages are common and provide as a favourable environment for the spread of various genetic instability syndromes⁵. Parental consanguinity was reported to be as high as 94.2 percent among Fanconi anaemia patients and 57.9% among non-Fanconi anaemia patients.⁶

Oral manifestations such as dental agenesis, supernumerary teeth, small teeth, abnormal roots, dental malformations, positioned teeth, dental caries, gingivitis, periodontal disease, oral mucosal lesions, and dysfunction of the salivary glands, to predisposition to develop oral cancers, such as Squamous Cell Carcinoma.³

The most common oral symptoms in people with FA are



Fig. 1: Patient with history of FA



Fig. 2 and 3: Absence of left and right hand's thumb



Fig. 4 and 5: Presence of multiple carious lesions in maxillary arch

gingivitis and periodontitis.⁷

Chronic anaemia is another prevalent haematological change in people with FA, with pallor of the mucosa and gingiva as the major oral clinical features.⁸ Recurrent aphthous ulcers and eruption cysts are particularly frequent lesions in the oral soft tissues in individuals with FA. These individuals frequently exhibit traumatic non-ulcerated lesions and petechiae, which are usually associated to low platelet counts.³ There are limited studies on the prevalence of caries in this population in the literature. Caries is associated to the accumulation of biofilm in the mouth and poor oral hygiene.^{3,8} Due to endocrine changes, hyposalivation is usually observed in FA patients, which dramatically increases their risk of developing dental caries and also condition xerostomia can be a predisposing factor for dental caries.³ The difference in dental, chronological, and bone ages in patients with FA is significant, because dental and bone ages are lower than chronological ages.⁷ Patient may develop developmental disorders such microcephaly and a deficiency of growth hormones.³ In this reported case, short stature and growth hormone deficiency were also present. These patients' small stature is associated with growth hormone insufficiency, which affects 81% of people with FA.⁹ In order to compare the chronological age (CA), bone age (BA), and dental age (DA) of patients with FA and to ascertain whether the therapy received had any influence on the BA or DA, Koubik et al. conducted their study. They came to the conclusion that the BA and DA are delayed and are not affected by the therapy given. Individuals with FA have a series of changes to their tongue. Papillary atrophy, saburrall tongue, macroglossia, and melanic pigmentation, which can extend up to the floor of the mouth and gingivae, are the most commonly reported.^{10,11,12}

Furthermore, lesions and conditions that have the potential to become cancerous, such as leukoplasias, erythroplasias, lichen planus, and squamous cell carcinoma, must be considered significant changes in the tongue in patients with FA.^{13,14,15,16}

Pediatric dentists are an important part of the patient's care since they can build a preventive strategy to promote oral health which in turn will improve the patient's quality of life. Routine dental examinations ensure that patients are motivated to re-establish and maintain periodontal health, as well as reduce the occurrence of caries and monitor their oral health. Treatment for dental infections should be targeted and limited.¹⁷

For a variety of reasons, providing dental treatment is difficult in this group. The radiosensitivity of FA cells is a lesser-known and yet controversial feature. FA cells have been reported to be highly susceptible to x- or gamma-irradiation. So radiographs are not advisable in FA patients due to induction of tumors, which makes effective identification of oral disorders problematic. Therefore radiographs were not taken during the treatment of the present case.¹⁸

CONCLUSION

Pediatric dentists are crucial members of the treatment team's multidisciplinary approach, and they must understand the pathophysiology of the various oral conditions observed in FA. Patients with FA are more prone to get infections, so proper precautions should be taken to avoid any situation

that might put them at risk of infection or bleeding. Patients should be motivated to have regular follow-ups and encourage them to maintain good periodontal health, to help prevent the incidence of caries and to monitor their overall oral wellbeing.

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