OPEN ACCESS

Saudi Journal of Medicine

Abbreviated Key Title: Saudi J Med
ISSN 2518-3389 (Print) | ISSN 2518-3397 (Online)
Scholars Middle East Publishers, Dubai, United Arab Emirates
Journal homepage: https://saudijournals.com

Case Report

Dental Management of Fanconi Anemia: Two Case Reports

Jazi Imen[1*], Nait Malek M[2], Zaroui J[3], Chalbi M[4], Essari A[5], Chemli MA[6]

[1]Assistant Professor, Pediatric Dentistry Department, Hospital La Rabta, Tunisia
[2]Resident, Pediatric Dentistry Department, Hospital La Rabta, Tunisia
[3]Associate Professor Pediatric Dentistry Department, Hospital La Rabta, Tunisia
[4]Assitant Professor Pediatric Dentistry Department, Hospital La Rabta, Tunisia
[5]Assistant Professor Pediatric Dentistry Department, Hospital La Rabta, Tunisia
[6]Professor Pediatric Dentistry Department, Hospital La Rabta, Tunisia

DOI: 10.36348/sjm.2022.v07i07.006 | **Received:** 06.05.2022 | **Accepted:** 10.06.2022 | **Published:** 16.07.2022

***Corresponding Author:** Jazi Imen
Assistant Professor, Pediatric Dentistry Department, Hospital La Rabta, Tunisia

Abstract

Fanconi Anemia (FA) is an extremely rare autosomal recessive disorder characterized by chromosomal break up that induces congenital abnormalities. FA results from a mutation in one of the 15 genes involved in the DNA repair pathway that is essential for the proper development of white blood cells, red blood cells, and platelets (Dental Perspective of Rare Disease of Fanconi Anemia (2). Some signs made up of a short stature, hyperpigmentation and bone marrow failure should suggest the diagnosis. In this paper we report two case reports of FA of in different ages who were followed in our Paediatric Dentistry Department in la Rabta Hospital and in which various classical signs were present. Comparing the different symptoms, we noticed that developmental and physical abnormalities are in common such as hyperpigmentation, short stature, skeletal abnormalities and some oral manifestations such as microdontia, periodontitis, and dental caries. Due to an increased risk of malignancies in this population, we have given emphasis on oral manifestations and the role of pediatric dentist in making early diagnosis and ensuring the maintenance of oral health for these patients.

Keywords: Fanconi anemia; rare genetic disease; malignancies; oral manifestations, dental management.

I. INTRODUCTION

Fanconi Anemia (FA) is a rare genetic autosomal disease occurring in 1-2 per 100000 births [1] and which is characterized by a chromosomal break up and DNA damage.

This syndrome is caused by a mutation in one of the 15 genes involved in the DNA repair pathway that is essential for the development of blood cells and platelets [2].

The most frequent hematological complications of FA are pancytopenia, anemia, thrombocytopenia, and leukopenia. Because of these complications, patients affected by FA may develop severe bleeding as well as recurrent infections [4]. It is usually diagnosed at early age, presents a high risk of malignancies which complicate the prognostic [3].

It is an extremely rare disease with a prevalence of 1:350,000 births [2]. Patients have a 500-to 700-fold higher incidence of head and neck squamous cell carcinoma (SCC) than the general population and a 14% cumulative incidence of head and neck SCC by the age of 40 years [6].

Clinically, FA presents multiple congenital abnormalities which may be alarming such as short stature, abnormal skin pigmentation, thumb anomalies, gastrointestinal alteration and microcephaly that are recognizable at birth or during early childhood [4].

The oral manifestation may be present beginning from poor oral hygiene, gingival bleeding and dental caries to supernumerary teeth, agenesis and cross-bite.

These oral symptoms have rarely been reported in the literature and there are only few reports with regard to the oral manifestations of FA [4].

The purpose of this paper was to report two cases of FA highlighting the oral repercussions of this disorder and discussing the multidisciplinary management in which the pediatric dentist should be a part.

II. CASES' PRESENTATION

Case1

A 12 years old boy was referred to the Department of Pediatric Dentistry and Prevention in the Dental Clinic of Monastir, Tunisia for dental caries.

Medical history revealed that he had been diagnosed with FA few months after birth by a cytogenetic test (figure 3).

Physical examination showed a various manifestations such as Height-weight delay, thin features and a triangular face, low ears implanted, facial dystrophy, slanting eyes (Figure 1; Figure 2) and "café au lait" spots scattered all over the body (Figure 4; Figure 5).

Fig-1: Patient face having a triangular shape

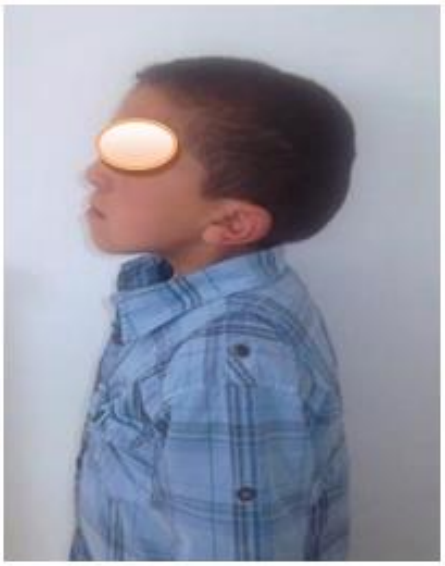

Fig-2: Physical examination showing low ears implanted

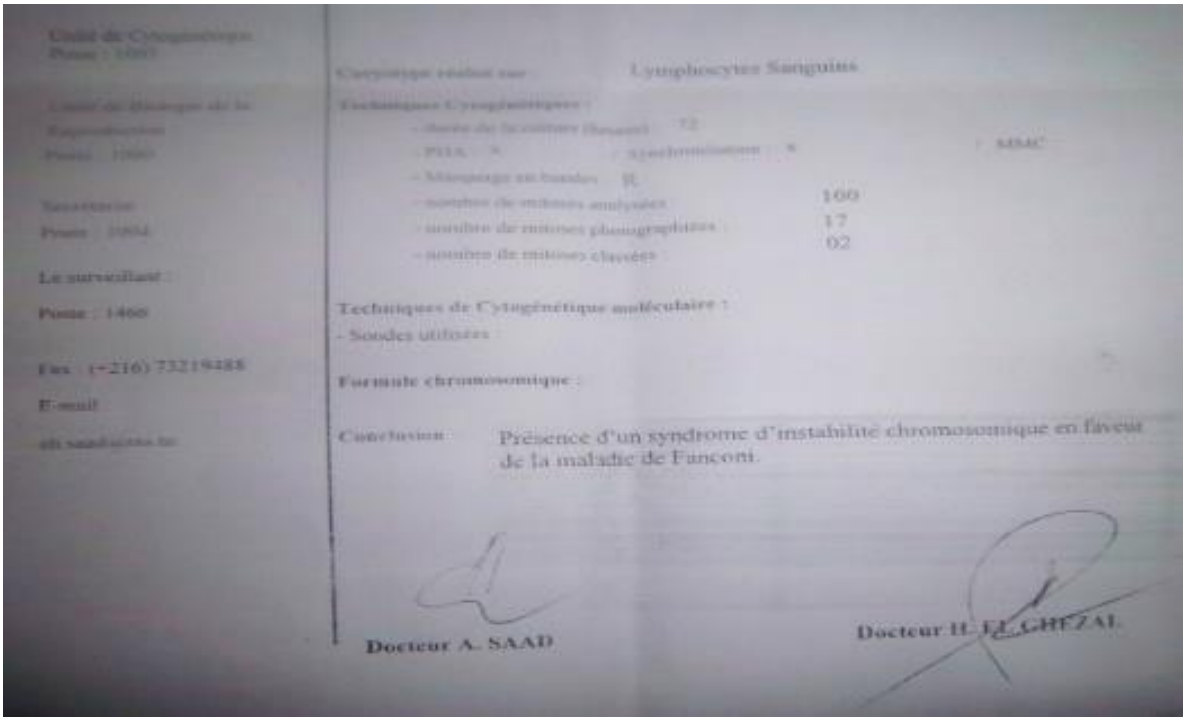

Fig-3: confirmation by a cytogenetic test

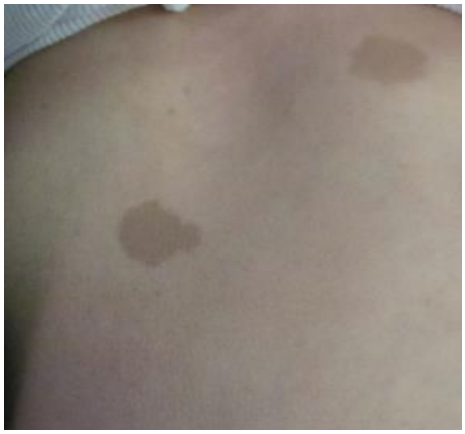

Fig-4: "café au lait "spots scattered all over the body sca

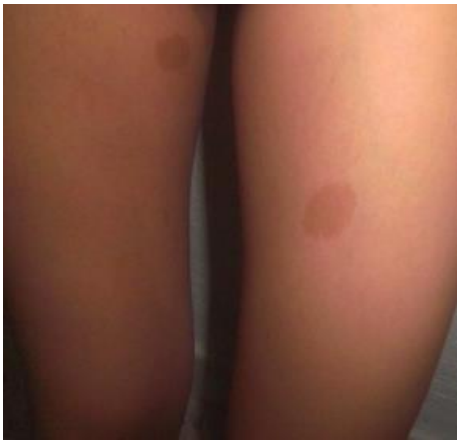

Fig-5: "café au lait "spots scattered all over the body

Patient présent Short stocky fingers and low thumb implantation (Figure 6; Figure7)

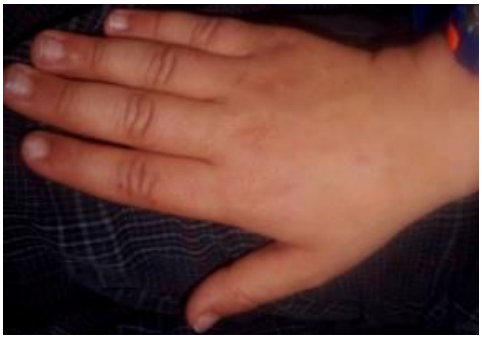

Fig-6: Short socky fingers

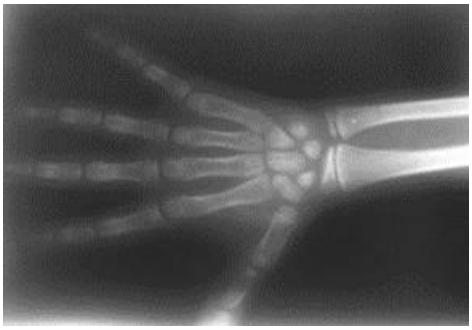

Fig-7: Low thumb implantation

Intra-Oral examination revealed a narrow palate (Figure 8; Figure9), inverted anterior joint, deviation of the inter-incisor midpoint (figure 10; Figure11) and multiple dental caries diagnosed in panoramic x-ray (figure11).

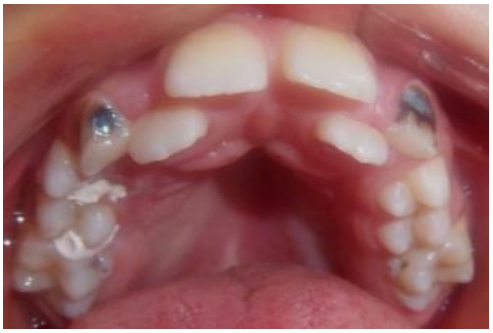

Fig-8

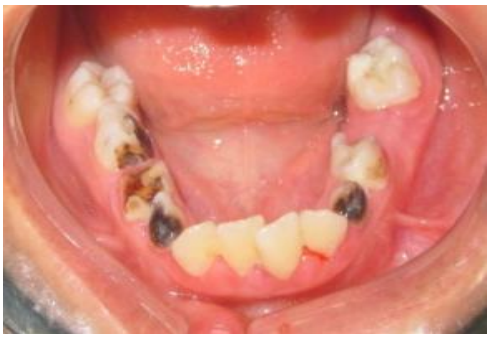

Fig-9
Poor hygiene and presence of multiple dental caries

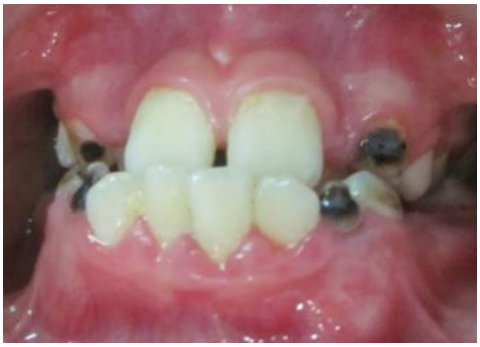

Fig-10: Inverted anterior joint

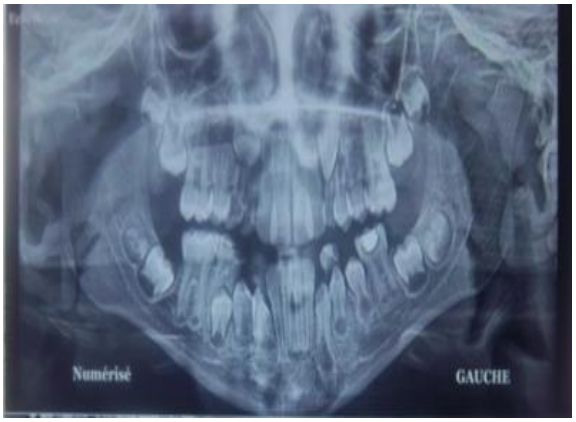

Fig-11: Panoramic X-ray showing multiple caries

Therapeutic approach consists in oral rehabilitation (coronal restauration and extraction of decapitated teeth) (Figure 12).

These dental therapeutics are carried out under antibiotic prophylaxis and good compression using haemostatic agent. Regular follow up was recommended in order to insure a good oral health.

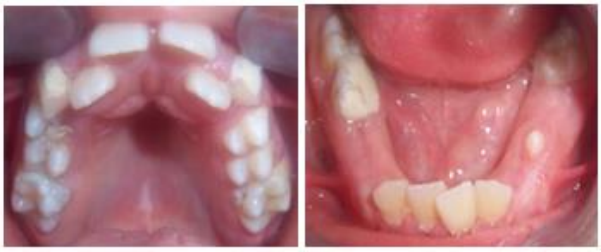

Fig-12: Intra oral view after oral conditioning

Case 2

A 5 year old boy was referred to the Pediatric Dentistry Department at La Rabta hospital of Tunis from the transplant center in order to eliminate any infectious focus of dental origin before the bone marrow transplant scheduled in few days.

He had been diagnosed with FA at the age of 3 after repeated unexplained first-line epistaxies. The patient's family history was significant as he was the second child of a consanguineous marriage.

Medical anamnesis revealed a myelo-suppression that compromises any bleeding act. Physical examination showed some characteristics beginning from growth retardation, thin features, slanting eyes (Figure 13) to the most common manifestation which is "café au lait" spots and pale skin (Figure 14; 15).

Fig-13: Physical examination showing the short stature of the patient

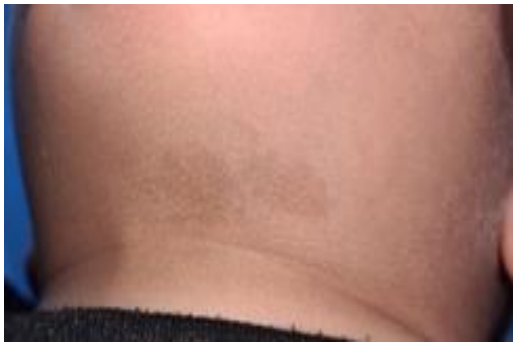

Fig-14

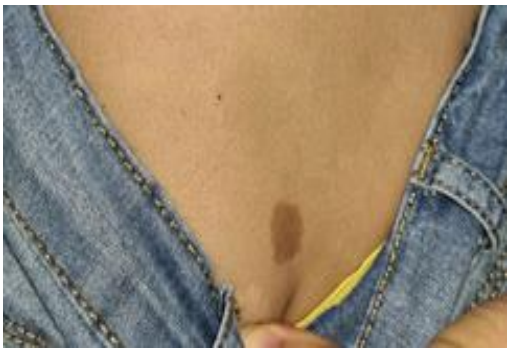

Fig-15
« Café au lait » spots on the back and neck

Intra-oral examination revealed teeth decay that affected all primary molars which is confirmed by the radiological exploration (figure16) and oozing blood from the gum which was more evident in posterior region (Figure 17; 18).

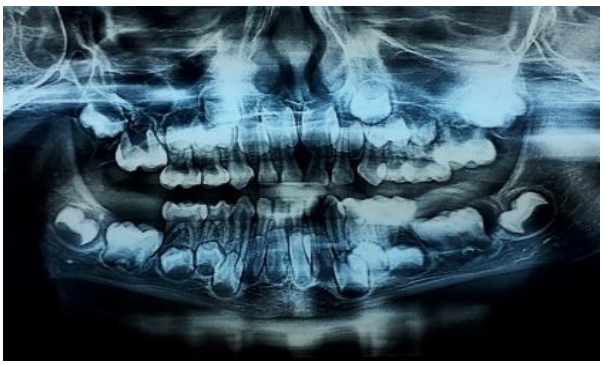

Fig-16: Radiological examination showing dental caries

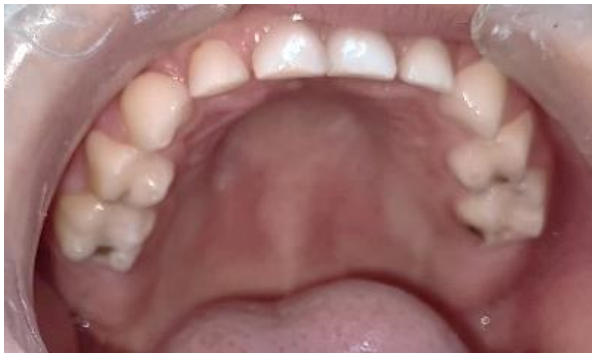

Fig-17: Oozing blood from the gum

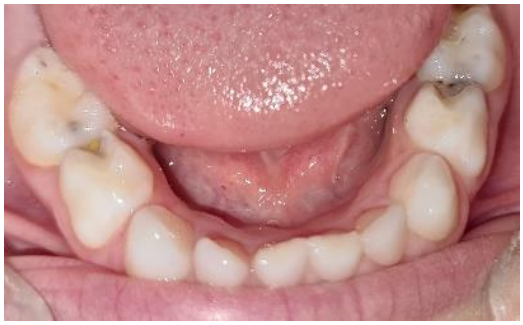

Fig-18: Multiple tooth decay

Our treatment plan has begun with contacting the haematologist in order to inquire about the possibility of insuring non-invasive dental care temporarily until the improvement of the patient's health after the bone marrow transplant (Figure 19; 20).

The gingival bleeding was controlled by local compression using haemostatic agent and prescription of a mouthwash.

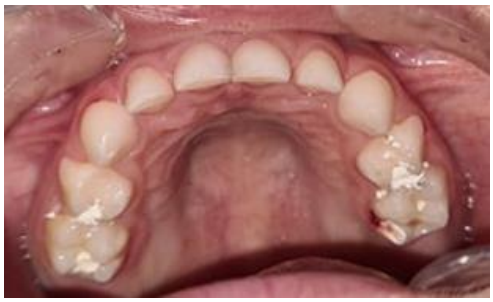

Fig-19

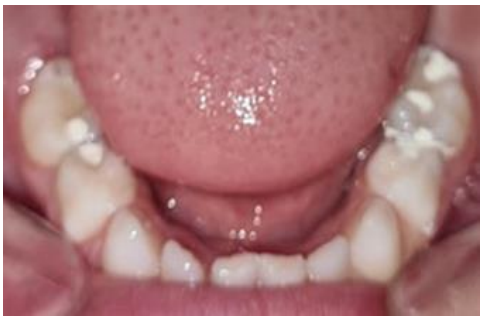

Fig-20

Superficial caries curettage and temporary filling before the bone marrow transplant

III. DISCUSSION

FA is a syndrome characterized by bone marrow failure, congenital malformations and a high risk to develop squamous cell carcinomas. Thus patients may develop head and neck cancers at young age [1, 4].

Clinically, we can have a wide array of manifestations and abnormalities such as bifid thumbs, malformations of hands and fingers, skin pigmentation in the form of "café au lait" spots [8] and growth retardation [2]. These were seen in our present cases but we should notice that some patients may not have any physical anomalies and skin lesions.

In the international Fanconi Anemia Registry (IFAR) a third of patients with FA were reported as not having any congenital malformations [3, 6].

In our cases, short stature and growth deficiency were also present which affects 81% of FA individuals as reported in the literature. Height-weight delay and developmental disorders such as microcephaly are also common [2].

The most frequent hematological complications are anemia, thrombocytopenia and leucopenia [7] thus patients affected by FA may develop severe bleeding and recurrent infections and this was seen in our cases.

Less common abnormalities in this syndrome include gastrointestinal defects, cardiac anomalies and central nervous system alteration [7] that was not seen in our cases.

The high risk of developing carcinoma is due not only to the syndrome but also to the treatment administrated (immunosuppressive, treatment of GVHD) [11]. Two-thirds of the head and neck squamous cell carcinoma in FA patients are located within the oral cavity, most frequently at the tongue margins and gingival areas [5].

The Oral manifestations are mentioned in the clinical description of our cases but their prevalence has not been well illustrated.

As reported in the literature, the most frequent oral abnormality associated with FA is gingivitis and periodontitis (50%) and this may be related not only to the disease but also to the medications applied during the immunosuppressive treatment and to the poor oral hygiene during long hospitalisation periods [4]. Spontaneous gingival bleeding is also common and was seen in our cases due to the decreased platelet level.

Few studies have mentioned the prevalence of early childhood caries in this population and reported that 35%_66% of FA patients develop dental decay. Caries are associated with poor oral hygiene and accumulation of plaque and with our cases this has been explained by the attention being focused on systemic alteration and not on the oral health [2, 4].

Dental anomalies such as microdentia, tooth agenesis, rotation, supernumerary teeth was mentioned in the literature but was not seen in our cases. However oral malformations such as micrognathia, narrow palate, crossbite was diagnosed in our first case and which is an extremely rare occurrence [9].

Dental treatment in this population should be specific and limited to prevention and some non-invasive procedures.

Two major risks we may face with FA patients which are infectious and haemorrhagic risk thus a close cooperation between the haematologist and the paediatric dentist is necessary. Treatments in our cases were done under antibiotic prophylaxis and with a good compression using haemostatic agent. A platelet count and hospitalisation are important to ensure safe care.

Because of the high risk of developing oral cancers, an early and frequent examination of the mucosal tissues is very important. Instructions to maintain excellent oral hygiene and parents' information of this risk are also primordial in this population [4, 10].

IV. CONCLUSION

Fanconi anemia is a rare genetic disorder whose symptoms start at an early age. Paediatric dentist should not only ensure oral treatments but also identify early changes and lesions associated with FA in the oral cavity. Thus Knowledge of the various oral manifestations and being aware of the risk of developing cancers is vital.

Pediatric dentists are in a unique position to identify early changes or lesions associated with FA in the oral cavity. They must collaborate with the child's hematologist before any invasive procedure is undertaken [2].

We should conclude that regular follow-ups and prevention are the best way to reduce the incidence of caries and gingivitis.

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