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CASE REPORT:

FANCONI'S ANEMIA: A CASE REPORT

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ABSTRACT:

Fanconi's Anemia (FA) is a rare autosomal recessive disorder characterized by thrombocytopenia,diverse congenital malformations which include skeletal malformations, hyperpigmentation, urogenital, renal and cardiac anomalies. We report a case of Fanconi anemia who presented with thrombocytopenia, bilateral hypoplastic thumbs, café-au-lait spots, with severe bleeding from the gingiva managed with platelet transfusion and extraction of the tooth was done

Keywords: Fanconi's anemia (FA), Thrombocytopenia, cafe-au-lait spots, squamous cell carcinoma.
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INTRODUCTION:

Fanconi Anemia (FA) is a rare autosomal recessive disorder (birth incidence of 1 per 350000), first described in 1927 as progressive lethal anaemia associated with brown pigmentation of skin [1, 2]. The disorder includes pancytopenia with hypoplastic bone marrow, skeletal, renal and ophtalmological malformations and chromosomal aberrations [3]. It also involves many organs including skin and genitourinary, musculoskeletal, neurological and cardiovascular systems. The clinical findings in FA patients are Hyperpigmentation on the skin, small reproductive organs in males, kidney problems, abnormalities in the thumbs and arm, skeletal anomalies of hip, spine or ribs, low birth weight, short stature, growth retardation, defects of the tissue separating the heart chambers and mental retardation or learning disability [3, 4]. Most cases of FA manifest anemia symptoms during childhood. However, the symptoms may not become apparent until adulthood [5, 6]. FA patients are at risk for developing secondary malignancies, for example leukaemia, squamous cell carcinoma and hepatocellular carcinoma [7-9]. The risk of squamous cell carcinoma development is high in the anogenital region as well as the head and neck region [10]. There is increased susceptibility of the oral cavity and

anogenital region to local predisposing factors like environmental toxins and viruses [5].

CASE REPORT:

A six year old boy reported to the department of oral medicine and radiology Yenepoya Dental College with the chief complaint of bleeding from the gums since 2 weeks. History revealed similar bleeding from the gums frequently, family history revealed his elder brother with the same problem and died at the age of 3 years. On general physical examination he was conscious, cooperative, poorly built, vital signs were within normal limits. Patient had microcephaly, triangular facies, bilateral anopthalmos, hypertelorism, depressed nasal bridge. High arched palate, low set ears, webbed neck, widely spaced nipples, bilateral undescended testis, radially curved left forearm and hypoplastic biphalangial thumb attached to the palm by thin thread like pedicle. Café au lait spots were noted on the right side of the skin over the neck and shoulder.

Intra oral examination revealed gingiva which was erythematous, soft and edematous. Grade 3 bleeding on probing, chronic pulpitis in relation to 16, 26, 75, 85 and 46. (Figure 1)

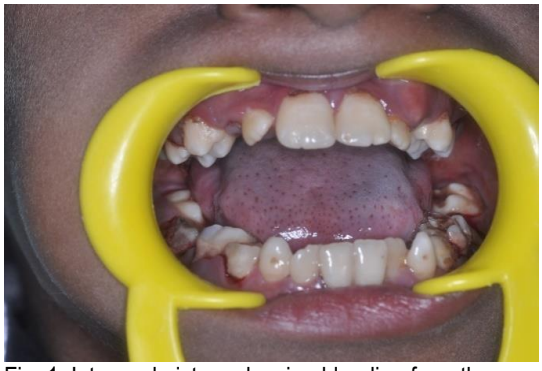

Fig. 1: Intra oral picture showing bleeding from the gingiva, chronic pulpitis 75

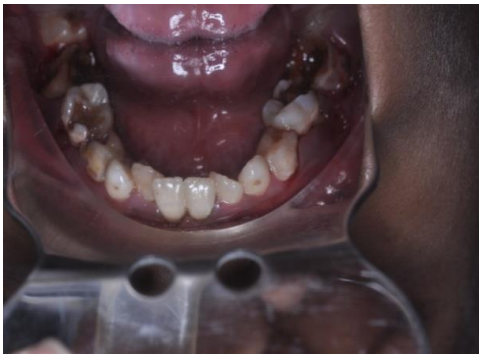

Fig. 2: Postoperative picture after extraction of 75

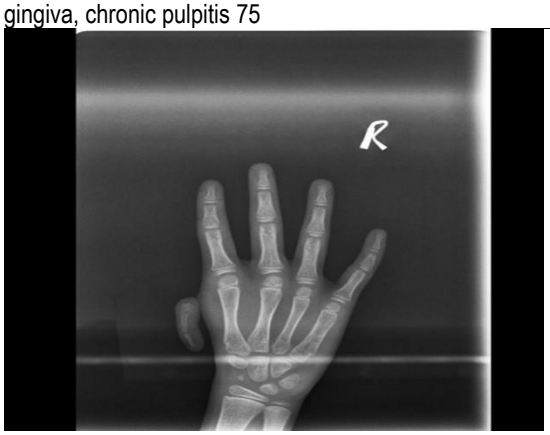

Fig. 3: Right hand wrist radiograph showing hypoplastic thumb, rudimentary 5th Meta carpel bond

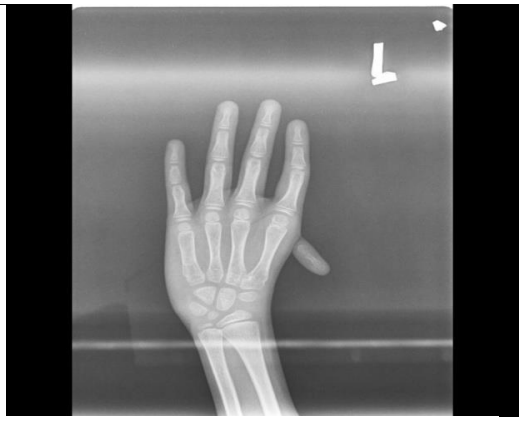

Fig. 4: Left hand wrist radiograph showing hypoplastic thumb

Based on the chief complaint, history given by the parents and clinical evaluation, it was provisionally diagnosed as fanconis anemia, chronic generalized gingivitis and chronic pulpitis 16, 26, 74, 75, 85 and 46 were made.

Hand wrist radiograph revealed hypoplastic thumb, rudimentary 5th Meta carpel on right side (Figure 3). Panaromic radiograph revealed normal anatomical land marks, coronal radiolucency involving enamel dentin and pulp in relation to 16, 26, 74, 75, 85 and 46.

Electro cardiograph showed dilated cardiomyopathy. Hematological investigations show platelet count was 5000/ml, blood slide of peripheral smear showed normochromic blood picture with thrombocytopenia, RBC count was 4.25 million /cu mm, Hb was 12.6/dl, PCV was 34.6%. Based on the radiographic finding and laboratory finding Fanconi anemia was confirmed.

Patient was hospitalized immediately and 4 units of platelet transfusion were given. Post transfusion platelet count was 63000/ml. The next day 2 units of platelet transfusion was done prior to extraction of 75 under local anesthesia with prophylactic antibiotics coverage and 1 unit of platelet transfusion were done immediately after teeth extraction. Patient was fine and there was no report of bleeding. Patient was discharged from the hospital after 2 days.

DISCUSSION:

Fanconi Anemia (FA) is a rare autosomal recessive syndrome. The disease is named after the Swiss paediatrician Guido Fanconi who originally described the disorder in three brothers, in 1927 [1]. Fanconi anemia is remarkable; its phenotype heterogeneity includes bone marrow failure and a variety of congenital malformations. Fanconi anemia has been found in a variety of ethnic groups.

Our present case had growth failure, skeletal malformations, café- au- lait spots, bone marrow failure and absence of left kidney. The hematological disorders resulting from bone marrow dysfunction (thrombocytopenia, leucopenia and anemia) usually appear around a mean age of 7 years, but they can arise very early at birth or even more rarely around 40 years of age [3].

Our case has thrombocytopenia at the age of 5 years. Some patients with FA are prone to develop different types of cancers, commonly leukemia acute myloid leukemia (AML), less commonly liver tumors, cancers of mouth, tongue, throat, genitals and brain tumors. Classic phenotype of FA includes, short stature, abnormality of the thumbs, microcephaly, café au lait and hypopigmented spots with characteristic facial appearance (a broad nasal base, epicanthal folds, and micrognathia) [2]. Our case presented with all these findings. FA newborns commonly have hypogonadism and renal malformations [2,3]. Radial ray abnormality is the most common physical abnormality noted in patients with FA during infancy. Bilateral radial

ray defect is more common (78%) than unilateral (22%) [3]. Hematologic abnormality at birth is very rare [4].

Differential diagnosis considered in the present case includes thrombocytopenia-absent radius (TAR) syndrome, VATER /VACTERAL syndrome and Holt- oram syndrome. TAR syndrome presents at birth with severe thrombocytopenia with bleeding manifestations and radial ray defects [5]. Microcephaly, short webbed neck and skeletal anomalies noted in the present case, though rare, have been reported in patients with TAR syndrome [2,3]. In FA, if the radii are affected, the thumbs are always abnormal (absent /hypoplastic); in TAR radii are absent but the thumbs are always present [2]. The present case had an abnormal thumb, consistent with the diagnosis of Fanconi anemia.

FA has considerable overlap in the physical abnormalities with VATER /VACTERAL syndromes. In a large series of FA patients, 10% patients had three principal clinical features and additional 20% patients had two major defects found in VATER syndrome [6]. Therefore, FA patient can be easily misdiagnosed as VATER/VACTERAL syndrome. The present case had vertebral and limb defects. VATER or VACTERAL syndromes are sporadic whereas FA has 25% chance of recurrence and misdiagnosis has severe consequences for genetic counseling [7]. Therefore it is recommended to rule out FA in patients with suspected VATER / VACTERAL syndromes by chromosomal testing [3,6]. Holt-Oram syndrome has radial ray defects with cardiac defects (100% cases). In the present case there were no cardiac defects [5]. Trisomy 18 can rarely have radial ray defects and eye anomalies. However trisomy 18 has host of different physical abnormalities and a typical facies [2]. Diagnosis of FA requires high index of suspicion as it presents with physical abnormalities involving multiple systems and hematologic abnormalities at birth are extremely rare. Early diagnosis in FA is very important as long term survival depends on the age of onset of hematologic abnormalities or malignancies [8].

If FA is recognize in the preanemic phase, drugs and environmental insults implicated in acquired aplastic anemia or malignancy can be avoided and life span can be prolonged [2]. Early diagnosis also offers options of planning next pregnancy; as the umbilical cord blood can be used for stem cell transplantation. Bone marrow or umbilical cord blood transplantation from identical sibling is now considered the treatment of choice for FA. [3,7]

CONCLUSION:

Fancooni FA patients develop thrombocytopenia at childhood it is recommended to check for thrombocytopenia, requires platelet transfusion for extraction of the tooth. Regular follow up is recommended to screen the developing malignancies. Early diagnosis also offers options of planning next pregnancy as the umbilical cord blood can be used for stem cell transplantation.

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