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Case Report

Beta Thalassemia Major -A Case Report

Sahana K[1]*, Raghavendra Kini[2], Shweta Kajjari[3], Prasanna Kumar Rao J[4] and Vathsala Heggade KN[5]

[1]*Postgraduate Student, Department of Oral Medicine and Radiology, AJ Institute of Dental Sciences, Mangalore, Karnataka, India*

[2]*Professor and Principal, Department of Oral Medicine and Radiology, AJ Institute of Dental Sciences, Mangalore, Karnataka, India*

[3]*Senior lecturer, Department of Pedodontics and Preventive Dentistry, Subbaiah Institute of Dental Sciences, Shimoga, Karnataka, India*

[4]*Professor and Head, Department of Oral Medicine and Radiology, AJ Institute of Dental Sciences, Mangalore, Karnataka, India*

[5]*Postgraduate Student, Department of Conservative dentistry, AJ Institute of Dental Sciences, Mangalore, Karnataka, India*

***Corresponding Author:** Sahana K, Postgraduate Student, Department of Oral Medicine and Radiology, AJ Institute of Dental Sciences, Mangalore, Karnataka, India.

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Abstract

Thalassemia, the epitomy of haemolytic anemia is the most widely distributed genetic disorder. It occurs due to defect in the globin chain of haemoglobin. The more severe form, Beta Thalassemia major, presents with a diverse spectrum of clinical features caused by anaemia, bone marrow hyperplasia and blood transfusions. Here is a rare case presentation of beta thalassemia major with its clinical intraoral, extra oral features and radiographic appearances with the review of literature.

Keywords: Thalassemia Major; Chip Munk Facies; Hair on End Appearance; Rodent Facies

Introduction

Haemoglobinopathies are most frequently encountered disorders in South East Asia [1]. Thalassemia, which is one among them, is due to defect in the globin chain of haemoglobin. Based on the involvement of the globin chain, it is further subdivided into alpha thalassemia and beta thalassemia [2].

Thalassemic individual a present with the diverse clinical manifestations. It can be asymptomatic in heterozygotic individuals can present as thalassemia minor and sickle cell trait [3].

Beta Thalassemia major is the homozygous state affecting the beta genes presents as severe anaemia, expansion of skull bones with typical clinical and radiological manifestations [4].

Case Report

A 14 yrs. old male patient visited department of oral medicine and radiology with the chief complaint of decayed upper left back tooth for 6 months. It was not associated with the pain or swelling.

Patient was diagnosed with the thalassemia major 5 yrs. back and is undergoing blood transfusion every month since then. Patient also gives the history of splenectomy and cholecystectomy four years back. Family history revealed presence of thalassemia in one of his family members. Personal history was non-contributory. Patient was poorly built and nourished with short stature. Extra-oral examination of the patient revealed frontal bossing, maxillary protrusion, malar prominence, saddle nose and frontal bossing giving an appearance of 'chip-munk facies' or rodent facies' (Figure 1A) Patient exhibited asthenic body with weakness of scapular girdle muscles (Figure 1B and C) and grade 3 systolic murur. Patient's finger nails and skin extremities exhibited yellowish tinge and sclera showed icterus (Figure 1D, E, F) There was a presence surgical scar in the left hypochondrium which was matched with history of splenectomy. (Figure 1B). Intraoral examination demonstrated proclined upper anterior teeth, generalised yellowish discolouration teeth, mild generalised diastema, grossly decayed left maxillary second molar, enamel hypoplasia with left lower second premolar, localised periodontitis with lower anteriors. (Figure 2A, 2B).

The patient was further subjected to haemotogic and radiologic investigation. Further, there was a mucosal pallor with blackish pigmentation of the buccal mucosa present. Also, yellowish tinge

at the junction of hard and soft palate was noted. Hematologic investigation revealed microcytic hypochromic anaemia with anis poikilocytosis, nucleated RBC'S along with thrombocytosis which was consistent with the thalassemia.

Later Hb electrophoresis was done which confirmed the diagnosis of beta thalassemia. Further, Lateral skull view of the patient showed widened diploe space. (Figure 2C) Orthopantomogram did not show any changes.

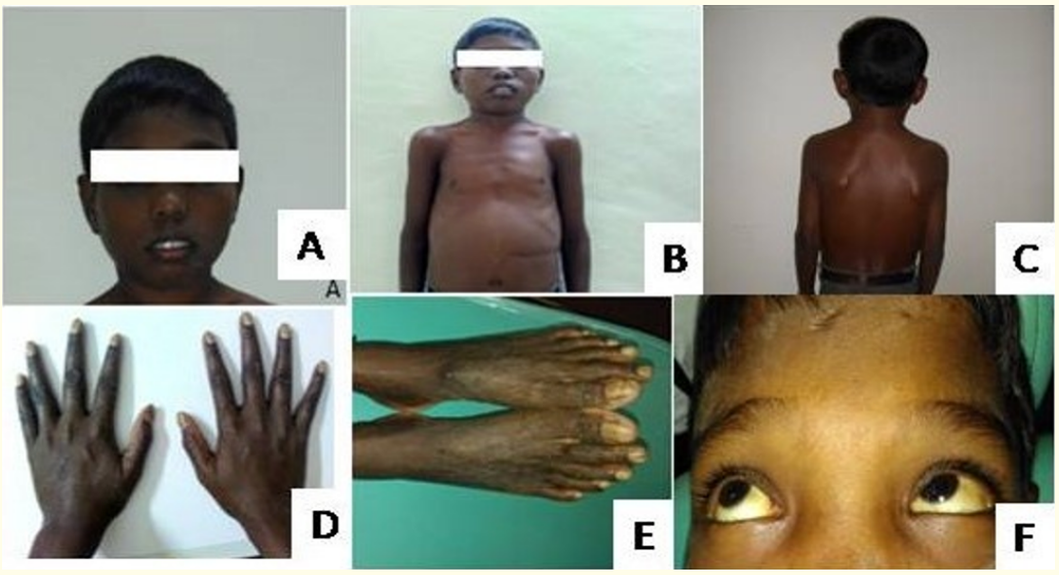

Figure 1

Figure 1A- frontal bossing, maxillary protrusion, malar prominence, saddle nose and frontal bossing giving an appearance of 'chip-munk facies' or rodent facies'

Figure 1B- Showing asthenic body with weakness of scapular girdle muscles

Figure 1C- Showing asthenic body with weakness of scapular girdle muscles.

Figure 1D- showing fingernails with yellowish tinge.

Figure 1E-Showing skin extremities showing yellowish tinge

Figure 1F- Showing scleral icterus

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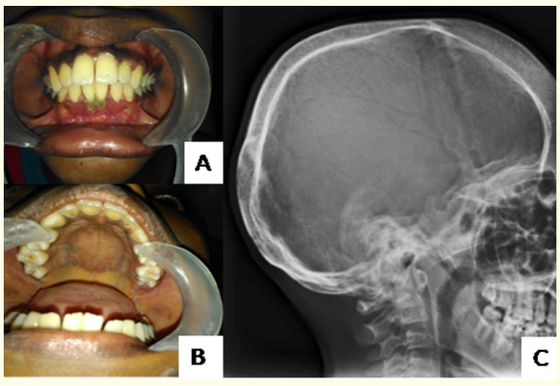

Figure 2

Figure 2A- Showing proclined upper anterior teeth, generalised yellowish discolouration teeth, mild generalised diastema.

Figure 2B- Showing a mucosal pallor with blackish pigmentation of the buccal mucosa and yellowish tinge at the junction of hard and soft palate.

Figure 2C- Lateral skull view of the patient showed widened diploes.

Discussion

The word thalassemia was derived from greek thalassa," meaning sea, as the condition is more frequent in the region of the Mediterranean Sea. The condition was explained by Cooley and Lee, in 1925 as anemia that associated with bone changes and splenomegaly [5]. Later on, thalassemia was found to be genetic disorder, which presents as a homozygous or heterozygous state. Homozygous form of thalassemia considered as thalassemia major and later one is considered as thalassmia minor. β-thalassemia major which is a subtype of thalassemia occurs when beta globin production hampered by defect in the both the genes.

The disease, also called as a Cooley's anemia, presents with characteristic features as depicted in the present case [6]. Thalassemia, one of the hemoglobinopathies often presents with diverse clinical picture which may cause diagnostic dilemma. Bone marrow hyperplasia and enlargement contributes to peculiar orodental manifestations [7]. Fragile RBC's which are produced in thalassemia further results in increased haematopiosis causing

bone marrow hyperplasia. Bone marrow hyperplasia or enlargement plays a main role in causing peculiar 'chip –munk facies' as in our case, which is characterised by maxillary protrusion, frontal bossing, malar prominence [8]. Further, thalassemic patients often present longipin or asthenic body type which is similar to present case [9,10].

Anaemia is the consistent feature of thalassemia, results from the synthesis of abnormal haemoglobin. Microcytic hypochromic anaemia subtype is more frequently observed in thalassemia major patients as early as 1ST year of life as the fetal haemoglobin formation stops after 3 - 4 months [12]. Same sub type of anemia was observed in our case. Fragile RBC's which are produced in thalassemia further results in increased haematopiosis causing bone marrow hyperplasia. Bone marrow hyperplasia or enlargement plays a main role in causing peculiar 'chip - munk facies' as in our case, which is characterised by maxillary protrusion, frontal bossing, malar prominence [8]. Further, thalassemic patients often present longilin or asthenic body type which is similar to present case [9,10].

The main therapeutic approach of thalassemia major is by transfusion of the blood every 2-3 weeks. Frequent transfusions results in iron overload which further causes secondary haemochromatosis, with involvement of heart, liver and endocrine glands [10].

Cardiac manifestations of thalassemia such as congestive cardiac failure, systolic murmur was reported in the literature. Above patient presented with the systolic murmur [11,13].

Scleral icterus, yellowish tinge of fingernails, skin extremities and oral mucosa associated with thalassemia, caused by bilirubin formed from degradation of RBCs, which was also eminent in our case [14].

Mucosal pallor and atrophic glossitis are also evident due to anaemia, as in our case anemia was evident. Hyposalivation and inflammation of salivary gland also found in thalassemic individual due to iron deposition, which was not evident in the above case.

Further, blackish pigmentation of the buccal mucosa due to increased ferritin in the blood, which was seen in the above case [15].

Bone marrow hyperplasia caused due to chronic anaemia gives rise peculiar radiographic feature of thalassemia. Radiographically thalassemia characterised by diploe space widening and outer table thinning was seen in above case. Diploe trabeculae aligned perpendicular to outer table producing typical hair on end appearance which was not evident in the above case [16].

Conclusion

Thalassemia is characterised by diverse oral and dental features, a dentist should have complete knowledge of the thalassemia and its complications to treat the dental conditions effectively.

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