OROFACIAL MANIFESTATIONS OF COFFIN-LOWRY SYNDROME – A CASE REPORT AND REVIEW

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Abstract

Coffin-Lowry syndrome (OMIM ID 303600) is a rare X-linked dominant genetic disorder and is associated with mutation of ribosomal S6 kinase 2gene, which is located on the short arm of the X chromosome and is designated as Xp 22. This syndrome has a distinctive facial appearance, with developmental delay, slower physical growth pattern, puffy and tapered fingers, along with few skeletal changes and often muscular hypotonia. Besides these, the orofacial findings like thick prominent lips, high arched palate, midline lingual furrow, microdontia, hypodontia, early tooth loss, and delayed eruption are quiet characteristic in this syndrome. In 1966, this condition was first demonstrated by Coffin and later in 1971, Lowry also presented such a similar report. In this case report and review, we demonstrate the orofacial findings in a 9-year-old female patient suffering from Coffin-Lowry syndrome.

Key Words Coffin-Lowry, X-linked dominant

INTRODUCTION

Coffin-Lowry syndrome (OMIM ID 303600) is a X-linked dominant genetic disorder which is quite rare and is associated with distinctive facial appearance, delayed developmental milestones, slower physical growth pattern, tapering fingers, together with few skeletal changes and often muscular hypotonia [1]. Besides these, the orofacial findings like thick prominent lips, high palate, midline lingual furrow, microdontia, hypodontia, early tooth loss, and delayed eruption are characteristic in this syndrome. This syndrome is more common in males than in females. The aim of this presentation is to help the dental surgeons to diagnose such cases, though rare, and provide necessary guidance for their well being and appropriate treatment

CASE REPORT

A 9-year-old female patient reported to the Department of Oral Pathology, Dr. R. Ahmed Dental College & Hospital, Kolkata with a complain of multiple carious teeth associated with pain and difficulty on chewing food.

Physical examination revealed puffy hands with tapered fingers, short stature and straight coarse hairs on the scalp. Extraoral examination revealed coarse facies, large forehead and prominent chin. Hyperteloric eyes with wide eyebrows. Ears were prominent with mild degree of hearing loss. Nose was fleshy and broad with thick alae nasi. Lower lip was also thick and everted.

Intraoral examination revealed multiple carious teeth, retained maxillary deciduous laterals and delayed

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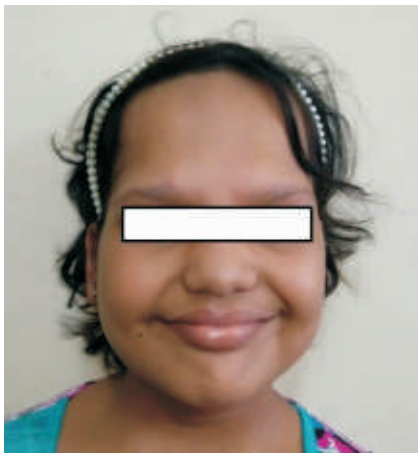

Figure 1. Frontal profile of the patient showing prominent forehead, fleshy nose, thickened lower lip and prominent chin.

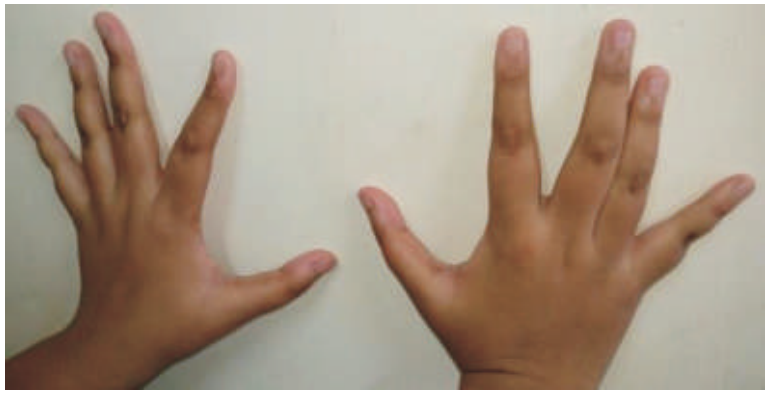

Figure 2. Tapering fingers

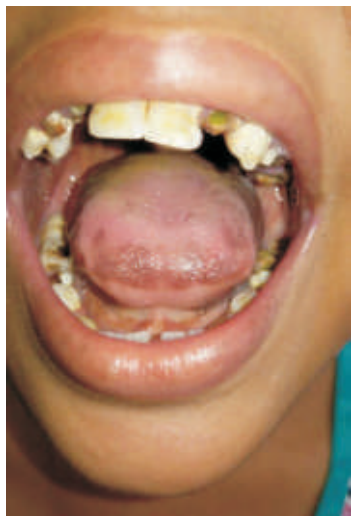

Figure 3. Retained roots of deciduous maxillary laterals and multiple carious teeth

eruption of maxillary permanent laterals. Palate was high arched. Malocclusion noted due to multiple carious teeth and delayed eruption of permanent teeth.

After consideration of the intraoral, extraoral and overall physical findings, patient was provisionally diagnosed to be a case of Coffin-Lowry syndrome. Patient was referred to the Department of Oral Surgery & Conservative Dentistry for management of retained root stumps and carious teeth respectively.

DISCUSSION

Though a rare disease, it has a distinct clinical manifestations. It is more common in males and cannot be readily identified at birth.

In 1966, this condition was first demonstrated by Coffin in two separate families [2]. Five years later i.e. in 1971, Lowry also presented such a similar report [3].

Prevalence of this syndrome is 1 in 50,000 to 1 in 1,00,000 male infants as reported by Stevenson[4].

Patients usually present with mental retardation and typical characteristic facial features. These facial features include a prominent forehead, hyperteloric eyes, a fleshy broad nose along with mid face hypoplasia. Such typical facial appearance starts evolving after two years of birth, which progresses slowly throughout childhood. A further consistent physical finding that is found in both sexes is soft, stubby, tapered fingers [5]. Other abnormalities may include mitral insufficiency, pectus carinatum, bifid sternum, rectal prolapse, uterine prolapse, inguinal hernia, thick calvarium, scoliosis, coxa vulga.

The differential diagnosis for infants with oro-facial appearance suggestive of Coffin-Lowry syndrome include Soto's syndrome, idiopathic hypercalcemia, phenytoin embryopathy and fragile X syndrome[6].

The gene responsible for Coffin-Lowry syndrome, ribosomal S6 kinase 2, is present on the short arm of the X chromosome and is designated as Xp 22. Mutation of this gene ribosomal S6 kinase 2 leads to the disease in males as only single X chromosome is present. In case of females, where

two X chromosomes are present, the genetic expression is much more variable[7]. Diagnostic confirmation by genetic analysis is presently available in some research laboratories now-a-days.

CONCLUSION

The clinical oro-dental findings along with the other typical characteristics, especially fingers and facial findings, finally led to the provisional diagnosis of this Coffin-Lowry syndrome. This has to be confirmed by genetic analysis. The dental surgeon has a vital role in diagnosing such cases. Moreover during providing dental treatment, one should take into account of the complications associated with this syndrome such as mitral insufficiency, mental retaradation, delayed growth.

REFERENCES

1. Stevenson RE. Coffin-Lowry syndrome. In: Nairns B. The Gale encyclopedia of genetic disorders. 2nd ed. Vol 1. Detroit: Gale; 2005: 266–7.

2. Coffin GS, Siris E, Wegenkia LC. Mental retardation with osteocartilaginous anomalies. Am J Dis Child 1966;112:205–13.

3. Lowry B, Miller JR, Fraser FC. A new dominant gene mental retardation syndrome. Am J Dis Child 1971;121:496–500.

4. Stevenson RE. Coffin-Lowry syndrome. In: Nairns B. The Gale encyclopedia of genetic disorders. 2nd ed . Vol 1. Detroit: Gale; 2005: 266–7.

5. Temtamy SA, Miller JD, Hussels-Maumenee I. The Coffin-Lowry syndrome: an inherited faciodigital mental retardation syndrome. J Pediatr 1975;86:724–31.

6. Young ID. The Coffin-Lowry syndrome. J Med Genet 1988;25:344–8.

7. Ian R. Lange, Peter Stone, Salim Aftimos: The Coffin-Lowry Syndrome: A Case Report and Review of the Literature: J Obstet Gynaecol Can 2010;32(7):691

