International Journal of Research in Medical Sciences
Narendra S et al. Int J Res Med Sci. 2015 Nov;3(11):3402-3405
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: http://dx.doi.org/10.18203/2320-6012.ijrms20150930

Case Report

Changing spectrum of Zimmerman-Laband syndrome: a six years follow up case report of a family

Suryakanta Narendra[1]*, Surendra nath Senapaty[2], Chaitali Bose[2], Deepti Samanta[2], Dhirendranath Moharana[3]

[1]Department of Periodontology, SCB medical/dental College, Cuttack, Odisha, India
[2]Department of Radiotherapy, Acharya Harihar Regional Cancer Centre, Cuttack, Odisha, India
[3]Department of Medicine, SCB medical/dental College, Cuttack, Odisha, India

Received: 14 September 2015
Accepted: 07 October 2015

***Correspondence:**
Dr. Suryakanta Narendra,
E-mail: suryakantanarendra@yahoo.com

ABSTRACT

The Zimmermann-Laband syndrome (ZLS) is a rare genetic disorder inherited as an autosomal dominant fashion, with clinical characters like, gingival fibromatosis, bulbous soft nose, thick floppy ears, nail dysplasia, joint hyperextensibility, hepatosplenomegaly, skeletal anomalies and occasional mental retardation. Idiopathic gingival enlargement is a hereditary condition; it can be expressed as autosomal dominant inheritance. Here association of Idiopathic gingival enlargement has been reported in a family, with an autosomal dominant inheritance diagnosed to be ZLS. Other clinical features associated are, hypertrichosis, bulbous soft nose, thick floppy ears, nail dysplasia, joint hyper extensibility, bimaxillary protrusion and enlarged palm and fingers with flat feet. The unusual clinical presentations of massive gingival fibromatosis, unusual length of upper limbs and bimaxillary protrusion supported the variable spectrum of phenotype expression of the ZLS. The Biopsy report confirmed the diagnosis of gingival fibromatosis. Gingivectomy was carried out in all four quadrants for exposing the natural teeth and to bring back the original contour of the gingiva. But there was recurrence of the enlargement found to be associated during 6 years follow up.

Keywords: Autosomal dominant inheritance, Idiopathic gingival Enlargement, Zimmerman-Laband syndrome

INTRODUCTION

Idiopathic gingival enlargement associated with Zimmermann-Laband syndrome (ZLS) is a hereditary disorder, regarding which data has been already reported several times.[1] It has been reported that it can be expressed as one autosomal dominant inheritance.[2] It may appears as isolated entity or it may be a part one of the several syndrome.[3] The ZLS is a rare disorder inherited as an autosomal dominant fashion, clinically characterized by gingival fibromatosis, bulbous soft nose, thick floppy ears, nail dysplasia, joint hyperextensibility, hepatosplenomegaly, skeletal anomalies and occasional mental retardation.[3] Very often idiopathic gingival enlargement is associated with hypertrichosis as a central clinical feature, usually become evident after eruption of the permanent teeth.[2,3] Early detection and timely recognition of the syndrome to allow adequate dental care, ophthalmic screening at periodic intervals is merited to improve the overall quality of life for these patients.[4] This case report is about the diagnosis and treatment of a case of idiopathic gingival enlargement in a 14-year-old female and her family. The patient presented with massive, generalized, firm and fibrous gingival enlargement along with severe skeletal bimaxillary protrusion. The patient is subjected to esthetic and functional disability because of this enlargement since childhood. In addition to this, characteristic atypical hair distribution over face, bimaxillary protrusion with

disproportionately long arms, forearms and fingers stretches the phenotyping characters of ZLS.

CASE REPORT

A fourteen year old female presented with signs of inability of chewing and closing the mouth, enlarged gums, and protrusion of both upper jaw and lower jaw. She has been extremely concerned because of her unsociable appearance. The growth of maxilla and mandible was noticeable since she was three years of age as reported by her parent. The massive abnormal growth of gingiva leads to difficulty in chewing, swallowing and speaking Figure 2. Her lip is incompetent and whole body and face is covered with hair Figure 1. At the age of 14 years she reported at the Dental Wing, SCB Medical College and referred to the department of Periodontology for need full action. Her father suffering from the same idiopathic gingival enlargement shown in Figure 6. One of her brother was also suffering from the similar condition and died because of nutrition problem as reported by his parents. Her father is also suffering from the same idiopathic gingival enlargement but degree of expression is comparatively less and hair distribution over his face is not similar to that of his daughter. Her mother on the other hand carries no such clinical features and they are absolutely normal. Her psychological health is absolutely normal with normal IQ showing Figure 4. Her menstrual cycle is also normal. On general examination it was found that, there is massive hair distribution all over the body and face with normal body build and other normal vital signs Figure 4. Systemic evaluation suggests no internal organ anomaly and no cardio respiratory anomalies.

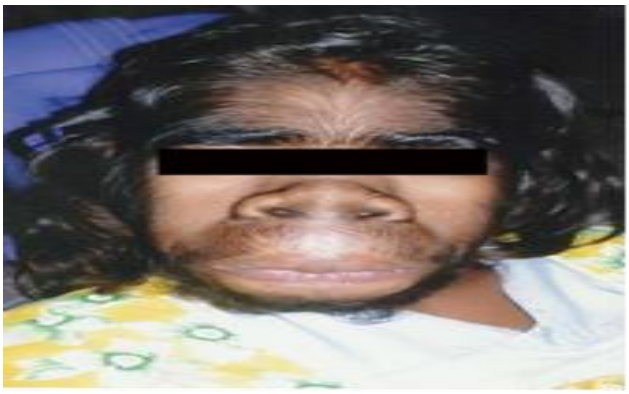

Figure 1: Patients suffering from idiopathic gingival enlargement.

On intraoral and extra oral examination it was found that, Incompetent lip is due to protrusion of the enlarged gingiva and bimaxillary protrusion. All the teeth were covered and deeply embedded under massive tissue folds. The gingival growth is firm, non-tender & free from inflammation. Attached gingiva so massively over grown which covered the whole of the jaw. It is also pink, firm & leathery in consistency with lobulated and pebbled surface. The jaw is distorted due to the bulbous enlargement. The tongue is slightly enlarged. Cephalometric dimensional analysis of face confirmed the skeletal problem of bimaxillary protrusion. On radiological examination, all the permanent teeth and some retained deciduous teeth are found to be completely covered by the enlarged mass of the gingiva.

Figure 2: Massive enlargement causing problems during chewing.

Diagnosis

The diagnosis of ZLS was performed by a pediatric geneticist based on his medical history and the presence of characteristic manifestations. The diagnosis of gingival fibromatosis was given after histopathological examination of the excised gingival mass. The histopathological features are; hyperplastic, orthokeratinised stratified squamous epithelium with elongated rete ridges. The underlying connective tissue showed densely packed collagen bundle, few blood vessels and few lymphocytes.

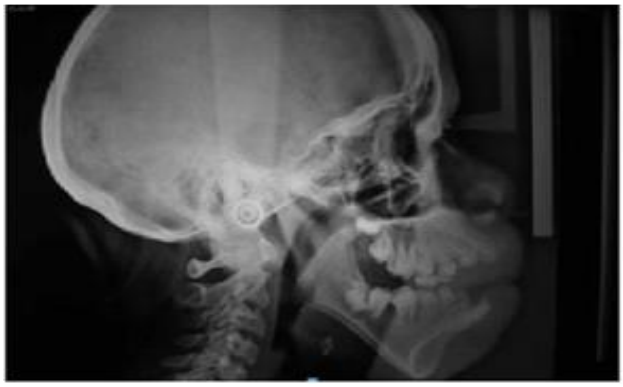

Figure 3: Lateral cephalogram showing bimaxillary protrusion with over lying soft tissue.

Treatment and management of gingival enlargement

The rejection of massive gingival tissue is done in phase wise manner in order to avoid post-operative complications & pain that could arise because of the development of anticipated raw area. The massiveness of

the enlargement and the risk of bleeding from the vessels like incisive vessel and Grater palatine vessel are the 2 main reasons for completing the case in phase wise manner. The gingivoplasty was finally done with an objective of restoring normal contour of gingiva, exposure of the teeth and bringing back the normal facial profile. Recurrence of the problem of gingival fibromatosis became evident during the six years follow up of this case.

Figure 4: Patient showing normal IQ and massive hair distribution over hands.

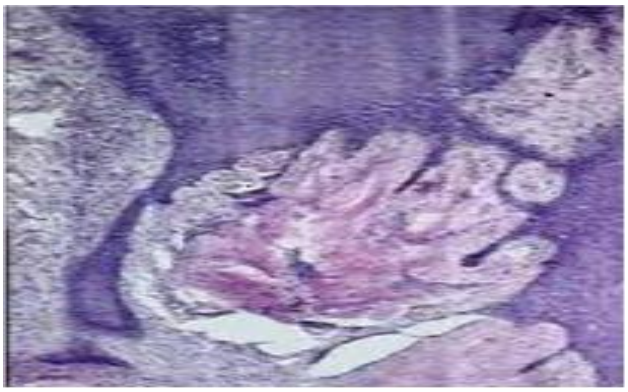

Figure 5: Histopathological slide showing long slender tube shaped rete pegs with densely arranged collagen bundles.

DISCUSSION

The presence of physical characters like gingival fibromatosis, Hypertrichosis, aplastic or hypoplastic distal phalanges with absent nails, and enlargement of soft tissues of the face, moderate learning disability and mild hearing loss is recognised as ZLS.[1] The syndromic features of ZLS are highly variable and can overlap with other entities featuring gingival fibrosis, colpocephaly, hemivertebra, polydactyly, hyperpigmentation, and hemihyperplasia.[3] These cases are discussed in the light of recognizing different characteristic features from time to time in different literatures. ZLS can be part a contiguous genes syndrome or be consequence of a gene mutation with wide variable expression.[1] Hereditary gingival fibromatosis may exhibit autosomal dominant mode of inheritance .[5] Genetic loci for autosomal dominant modes of gingival fibromatosis have been localized to chromosome 2p21p22 (HGF-1) and chromosome 5q12-q22 (HGF-2).[6] It has been discovered that a mutation in the son of sevenless-1 (SOS-1) gene is responsible for gingival fibromatosis.[7] Hypoplasia of the nails and hyperflexibility of the joints are also reported to be associated with hereditary gingival fibromatosis.[8] Gingival fibromatosis combined with cherubism was reported as Ramon syndrome.[9] Hepatomegaly was also reported to be associated with gingival fibromatosis.[10] Gingival fibromatosis and its related syndromes are mainly inherited in an autosomal-dominant manner, but autosomal-recessive inheritance has also been reported.[11] Other Clinical syndromic presentation with phenotypic overlap, includes Cowden syndrome, Cross syndrome, Göhlich-Ratmann syndrome, Avani syndrome, and I-cell disease.[11] The present report supports that ZLS has a wide clinical spectrum because of the presence of massive idiopathic gingival enlargement, hypertrichosis, hyperextensibility of joints and some additional clinical features. With the notice of additional clinical features of bimaxillary protrusion with disproportionately long arms, forearms and fingers, phenotyping characters of ZLS could be stretched and resumed to be variable. Thus, the present report expands the phenotypic spectrum of this uncommon syndrome. This case is clearly a case of autosomal dominant inheritance, as in this family, out of three children from a parent constituting one affected phenotypic male and a normal unaffected phenotypic female, one male child and another female child manifested the disease and third male child is absolutely normal without any clinical feature like her mother. So the 2:1 ratio of affected children going absolutely in favour of autosomal dominant inheritance. Bimaxillary protrusion is evident from lateral cephalogram. Early detection and timely recognition of the syndrome to allow adequate dental care, ophthalmic screening at periodic intervals is merited to improve the overall quality of life for this patients.[2-4] The histopathological features are going in favour of Idiopathic gingival enlargement.

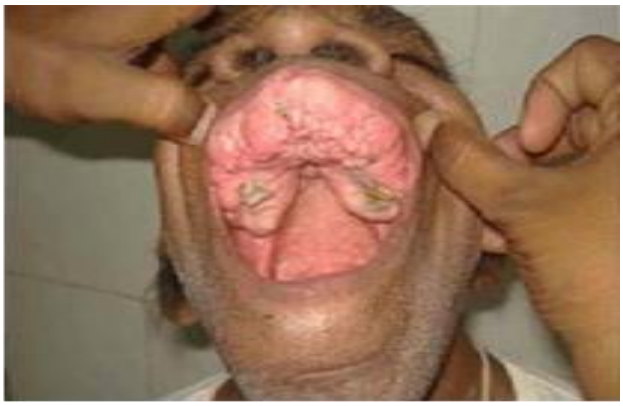

Figure 6: Patient father showing protrusion of face.

CONCLUSION

This case report of idiopathic gingival enlargement associated with ZLS is attributed to a hereditary disorder transmitted as autosomal dominant inheritance. In this case report, ZLS being characterized by presence of the idiopathic gingival enlargement, skeletal bimaxillary protrusion, massive hair distribution over the entire body with classic hair distribution over the face, comparatively enlarged palm, disproportionately long arms, forearms and fingers, waddling front bending gait, is reflecting a change in the spectrum of phenotype characters of this syndrome. The esthetic and functional need dictates the need of surgical intervention. The gingival enlargement cannot be cured but may be controlled with varying degree of success for improving the normal functions. This case of idiopathic gingival enlargement will be in need of the detection of risk allele responsible for this phenotype expression in future research. The future research could be directed towards the therapeutic field that could change dramatically by one of the recombinant DNA and monoclonal antibody technology for prevention of these syndromes.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Davalos IPL, Brambila-Tapia AJ, Dávalos NO, Durán-González J, et al. Wide clinical spectrum in Zimmermann-Laband syndrome, Genet Couns. 2011;22(1):1-10.
2. Laband PF, Habi BG, Humphrey GS. Hereditary gingival fibromatosis. Report of an affected family with associated splenomegaly, skeletal and soft-tissue abnormalities. Oral Surg. 1964;31:691-5.
3. Chacon-Camacho, Vázquez J, Zenteno JC. Expanding the phenotype of gingival fibromatosis-mental retardation-hypertrichosis (Zimmermann-Laband) syndrome. Am J Med Genet A. 2011;155A(7):1716-20.
4. Shah N1, Gupta YK, Ghose S. Zimmermann-Laband syndrome with bilateral developmental cataract - a new association? Int J Paediatr Dent. 2004;14(1):78-85.
5. Bazopoulou-Kyrkanidou EL, Papagianoulis L, Papanicolaou S, Mavrou A. Laband syndrome: a case report.J Oral Pathol Med. 1990;19(8):385-7.
6. Hart TC, Pallos D, Bozzo L, et al . Evidence of genetic heterogeneity of hereditary gingival finromatosis . J Dent Res. 2000;79:1758-64.
7. Hart TC, Zhang Y, Gorry MC, et al. A mutation in the SOS1 gene causes hereditary gingival fibromatosis type 1. Am J Hum Genet. 2002;70:943-54.
8. Haytac MC1, Ozcelik O. The phenotypic overlap of syndromes associated with hereditary gingival fibromatosis: follow-up of a family for five years. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 2007;103(4):521-7.
9. Ramon Y, Berman W, Bubis JJ. Gingival Fibromatosis combined with cherubism, Oral Surgery, Oral Medicine, Oral pathology. 1967;24:435-48.
10. Alavandar G. Elephantiasis: Report of an affected family with associated hepatomegaly , soft tissue abnormalities. Oral Surg Oral Med Oral Pathol. 1964;17:339-51.
11. Poulopoulos A1, Kittas D, Sarigelou A. Current concepts on gingival fibromatosis-related syndromes. J Investig Clin Dent. 2011;2(3):156-61.

Cite this article as: Narendra S, Senapaty SN, Bose C, Samanta D, Moharana D. Changing spectrum of Zimmerman-Laband syndrome: a six years follow up case report of a family. Int J Res Med Sci 2015;3:3402-5.