

Hereditary Gingival Fibromatosis, Hearing Loss, Mental Retardation and Scoliosis: A new syndrome?

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Abstract

Hereditary gingival fibromatosis is a rare genetically inherited fibrous overgrowth of gingiva. It is found to be developed as an isolated disorder but occasionally other abnormalities have also been reported. A case of 17 year old female, presented with localized bilateral gingival overgrowth over posterior palatal slopes in maxillary arch along with classical features of scoliosis of spine which presented familial pattern, hearing loss and mental retardation. Bilateral localized gingival overgrowth was diagnosed to be gingival fibromatosis based on clinical and histological findings. Extensive literature search was carried out and reports of co-existence of localized gingival fibromatosis, scoliosis and hearing loss are very few. Considering positive family history and presence of these findings, a possibility of existence of new syndrome cannot be ruled out.

Keywords: Gingival fibromatosis; Hearing Loss; Mental Retardation; Deviated Nasal Septum; Scoliosis.

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Introduction:

Gingival overgrowth is an abnormal growth of maxillary and mandibular gingival¹. Gingival fibromatosis may be familial or idiopathic affecting both the arches. It normally develops as an isolated finding but can be one of the features of several multisystem syndromes². The present case is associated with scoliosis of spine along with other classical features that occurs in combination gingival fibromatosis. Scoliosis is a term used when a person's spine is curved from side to side, shaped like an "s", and may also be rotated³. The associated syndromes with gingival fibromatosis are Laband, Murray puretic drescher, Rutherford, multiple hamartoma, Cross syndrome, prune belly syndrome^{4,5}. The other disorders seen with Gingival Fibromatosis include hypertrichosis, epilepsy, mental retardation, sensorineural deafness, hypothyroidism, chondrodystrophia & growth hormone deficiency. Autosomal dominant pattern of inheritance is seen in Heredity Gingival Fibromatosis (HGF), although it can be autosomal recessive also, occasionally⁶. The fibrous growth may be generalized or localized to one or more quadrant in either of the jaws. The purpose of this paper is to document a case of localized gingival fibromatosis with mental retardation, deafness, deflected nasal septum and scoliosis of spine which has not been reported yet leading to a possibility of a new syndrome.

Case report

A girl of 17 years of age reported to Department of Oral Medicine and Radiology with a chief complaint of swelling in mouth behind the last tooth on both sides in upper jaw [Fig.1]. History taking revealed that the gingival overgrowth was long standing and was observed by patient's parents almost 6-7 years back. On clinical examination the patient was found to be lean with altered posture. There was a hump on patient's back [Fig.2]. Drooping of eyes and atypical laughing suggested abnormal behavioral response.

On psychiatric examination, GDP interpretation revealed IQ= 56 signifying mental retardation. On ENT examination, there was central perforation of



tympanic membrane of right ear indicating deafness, also deviated nasal septum was diagnosed. Patient was unaware of this and gave no history of injury or infection to the right ear. On examination by Spine specialist, the patient was diagnosed to have idiopathic spine scoliosis [Fig. 2,3].



Fig. 2: Hump on the back of patient.

The intraoral examination revealed bilateral well circumscribed swelling distal to second maxillary molar. Swellings were pale pink in color and covered with smooth surface devoid of stippling. Resorption of root was there with respect to permanent maxillary right first molar. Orthopantomograph was taken to rule out any odontogenic cause or bony lesion.



Fig. 3: Clinical view of the patient showing localized gingival enlargement

Family History revealed the presence of scoliosis, deafness and gingival enlargement in patient's maternal grandmother and also the similar features were seen in patient's aunt (mother's sister). Excisional biopsy was done on both sides over a gap of one week. The excised tissue was of the dimension (1×1.5 cm) right side and (1×2) cm left side. The tissue was subjected to histopathological examination. Based on the histopathological examination in association with the clinical picture it was diagnosed to be Gingival Fibromatosis [Fig.4,5].



Fig. 4: Whole spine Anteroposterior view showing Scoliosis

Discussion

Gingival fibromatosis is a rare condition that occurs as an isolated feature or as a part of multi-system disorder. The syndromes associated are Laband syndrome⁷ (ear, nose, bone and nail defects with hepatosplenomegaly), Rutherford syndrome, Cross Syndrome^{8,9,10} (microphthalmia,

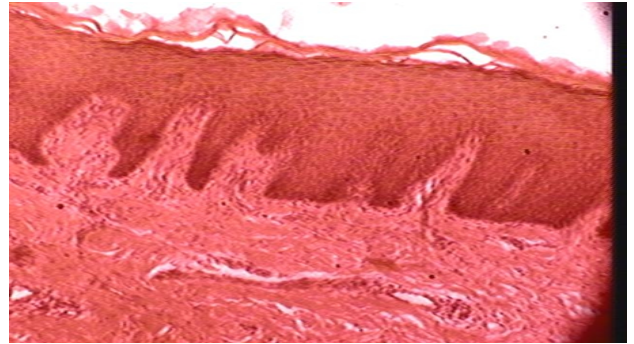


Fig. 5: H & E section showing parakeratinised stratified squamous epithelium and abundant collagen fibres

mental retardation, athetosis and hypopigmentation), new/variant syndrome given by Wynne et al¹¹ (gingival fibromatosis, hearing loss, supernumerary teeth)

In this particular case, the patient is suffering from gingival fibromatosis accompanied by mental retardation, deviated nasal septum, hearing loss and scoliosis of spine. In the literature localized symmetrical form of fibromatosis occurring sporadically without any family history has been reported whereas in contrast in the present case only one of the family members has localized gingival growth whereas the other two had generalized gingival growth. Gingival fibromatosis has been previously associated with hearing loss in three generations in the family along with the supernumerary teeth or sensorineural hearing loss as described in Jones syndrome¹¹. However, in the present case three members in family had hearing loss but no supernumerary teeth were seen. On history analysis, patient's maternal grandmother also had gingival enlargement which extended from permanent maxillary 2nd molar to maxillary first premolar both on right and left side along with deafness and scoliosis. The other member who suffered from similar localized gingival enlargement along with scoliosis and hearing loss is patient's maternal aunt (mother's sister). Scoliosis is a variant feature found in co-existence with other few classical findings. Scoliosis has been mentioned in relation to prune belly syndrome as musculoskeletal abnormalities¹⁰. Observing the presence of these findings in three members of the family suggested it to be an autosomal dominant pattern of inheritance. The

three subjects with gingival fibromatosis had both hearing loss and scoliosis. The presence of these traits along with scoliosis represents variable expression of single condition. Mental retardation which is one of the prime features associated with HGF was seen in the patient but had not manifested in other two members. The biopsy of HGF tissue from the current patient revealed abundant bundles of collagen fibers and sparse vasculature along with inflammatory cell infiltrates (Fig.5). This was incongruent with findings of Johnson et al¹² who found that the fibroblasts from hereditary gingival fibromatosis produced only half of the amount of collagen compared to fibroblasts obtained from normal gingiva.

In the present case, excision of the fibromatosed tissue was done. Previously reported cases have shown no reoccurrence in a period of 14 years follow up in contrast in present case follow up has been done for past 13 months and no reoccurrence has been observed so far.

Conclusion

Gingival fibromatosis is a rare entity presenting a variable expressivity. The previous data and literature when compared to the present report of gingival fibromatosis with hearing loss, deviated nasal septum and scoliosis of spine is sufficiently unique to propose a new syndrome.

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