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A rare case of Proteus syndrome

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Abstract

Introduction: We report a rare case of Proteus syndrome which presented with dysphagia due to unilateral tonsillar hypertrophy.

Case report: Our case had involvement of multiple organs in addition to bony deformities, namely, enlarged right parotid gland and right testis, verrucous naevus, varicose veins and abdominal lipoma. The features unique to this case were a larger right optic disc and expansion of the inner table of the skull.

Discussion: A literature review revealed less than 100 bona fide, published cases which fit the revised diagnostic criteria proposed by Turner.

Summary: The ENT manifestations of Proteus syndrome are high arched palate, gingival hypertrophy, malocclusion and overcrowding of teeth, hyperostosis of the external auditory meatus, and low nasal bridge. Our patient had a unilateral enlargement of the tonsil causing dysphagia, which was relieved by tonsillectomy.

Key words: Dysphagia; Hypertrophy; Exostosis; Nevus; Proteus Syndrome

Introduction

Proteus syndrome is a rare, congenital, hamartomatous disorder affecting all three germ cell layers and potentially all bodily tissues. It is characterised by disproportionate, asymmetrical overgrowth and skeletal abnormalities which are distorting and relentlessly progressive. The patients typically develop asymmetrical enlargement of the hands and feet, varicosities, verrucous epidermal naevi, haemangiomas, and lymphangiomas. Overgrowth of long and cranial bones is also an associated feature. Diagnosis is made according to Turner's criteria.

Our patient presented with dysphagia, exostosis, hemihypertrophy and other features. Tonsillectomy was performed without complication.

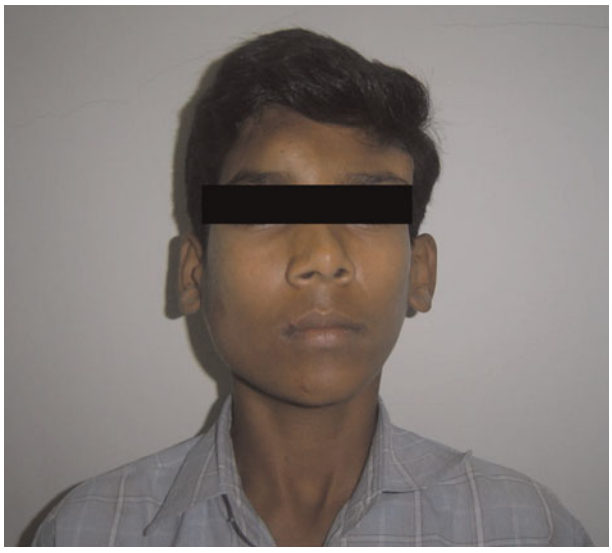

Fig. 1
Enlargement of right half of lower jaw and right parotid gland.

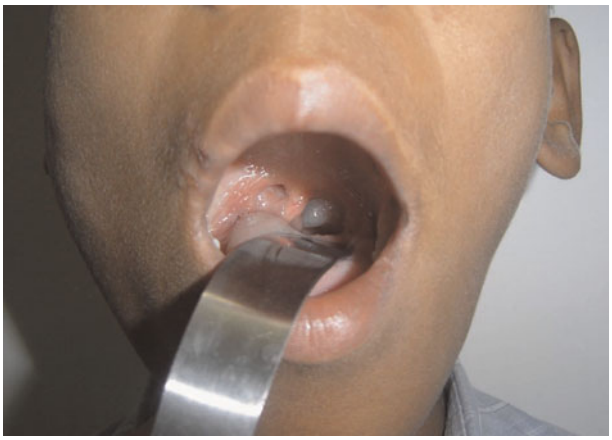

Fig. 2
Enlarged right tonsil.

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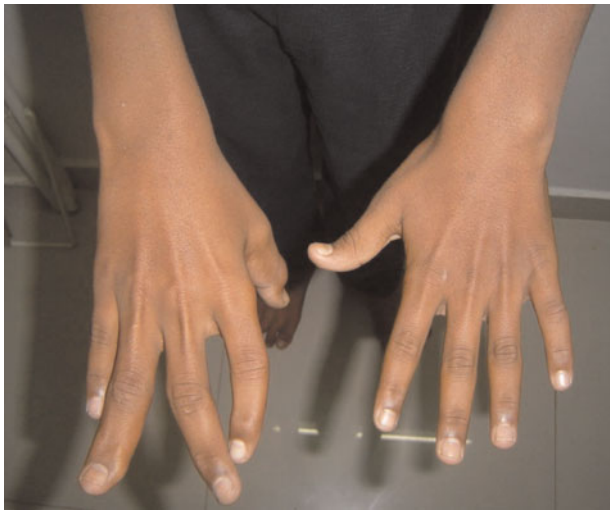

FIG. 3
Enlarged right hand showing longer fingers; middle & ring fingers show convergent tips middle & ring fingers show divergent tips.

Case report

A 15-year-old boy presented to the ENT department of the Karnataka Lingayat Education Society's Hospital and Medical Research Centre, Belgaum, Karnataka, India, with dysphagia of six months' duration. There was no history of pain in the throat. The patient also complained of a swelling on the right side of the forehead above the nasal bridge, a swelling over the left half of the forehead lateral to the eyebrow, and swellings on the vertex, left temporal and occipital regions. Facial asymmetry was present due to enlargement of the right half of the lower jaw (Figure 1). In addition, the patient had noted

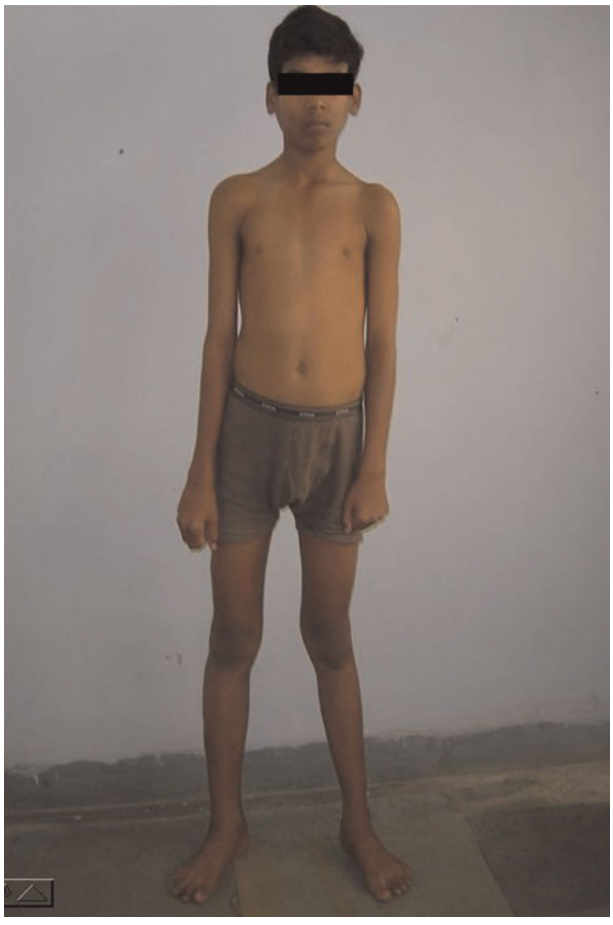

FIG. 4
Longer & bulkier right upper & lower limbs.

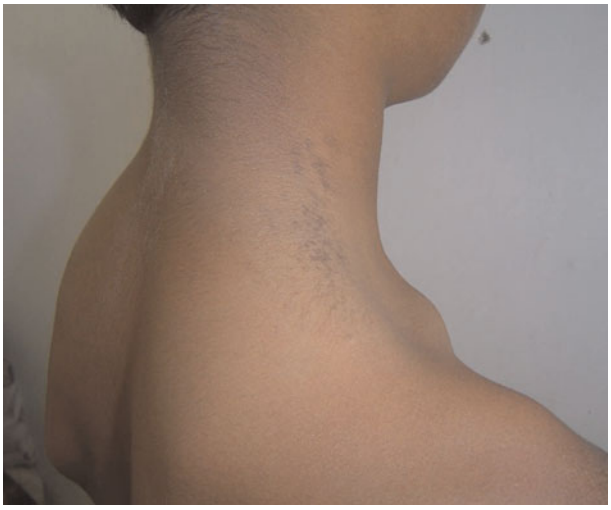

FIG. 5
Linear, epidermal, verrucous naevus on the right of the neck.

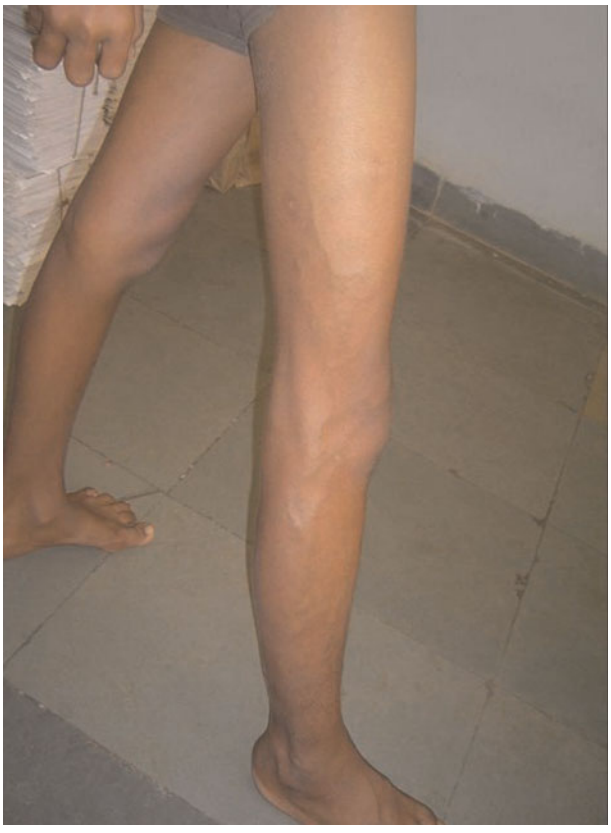

FIG. 6
Varicosity of velns on the lateral aspect of right thigh & leg.

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excess growth of the right upper and lower limbs, with finger deformities.

The boy had been born via a full term, vaginal delivery, from a nonconsanguineous marriage.

On examination, the oropharynx showed the right tonsil to be enlarged and crossing the midline, narrowing the oropharyngeal isthmus (Figure 2).

Hemispherical, bony swellings were observed over the forehead and the scalp, varying in size from 3 to 5 cm. The right half of the mandible and the right parotid gland were enlarged. The right upper limb was longer than the left by 3.3 cm, and the circumference of the right arm exceeded that of the left by 1 cm. The fingers of the right hand were longer, with the middle and index fingers showing convergent tips and the middle and ring fingers showing divergent tips (Figure 3).

The right lower limb was longer than the left by 2 cm, and the circumference of the right thigh exceeded that of the left by 3 cm (Figure 4).

The right testis was enlarged. A subcutaneous, anterior abdominal wall lipoma measuring 5 × 5 cm was observed in the right hypochondriac region. Dermatological evaluation revealed a single, linear, epidermal, verrucous naevus on the right side of the neck (Figure 5).

Varicosity of the veins on the lateral aspect of the right leg and thigh was observed (Figure 6). Fundoscopy showed the right optic disc to be larger than the left by approximately 30 per cent. In view of the mosaic pattern, sporadic occurrence and progressive course of the lesions, a diagnosis of Proteus syndrome was made.

Computed tomography (CT) of the brain revealed diffuse calvarial thickening involving the frontoparietal area bilaterally. Using the bone window CT setting, multiple bony outgrowths were noted in the occipital, left frontal and both parietal bones. Expansion of the outer skull table was noted at the left frontal, left parietal and right high parietal areas,

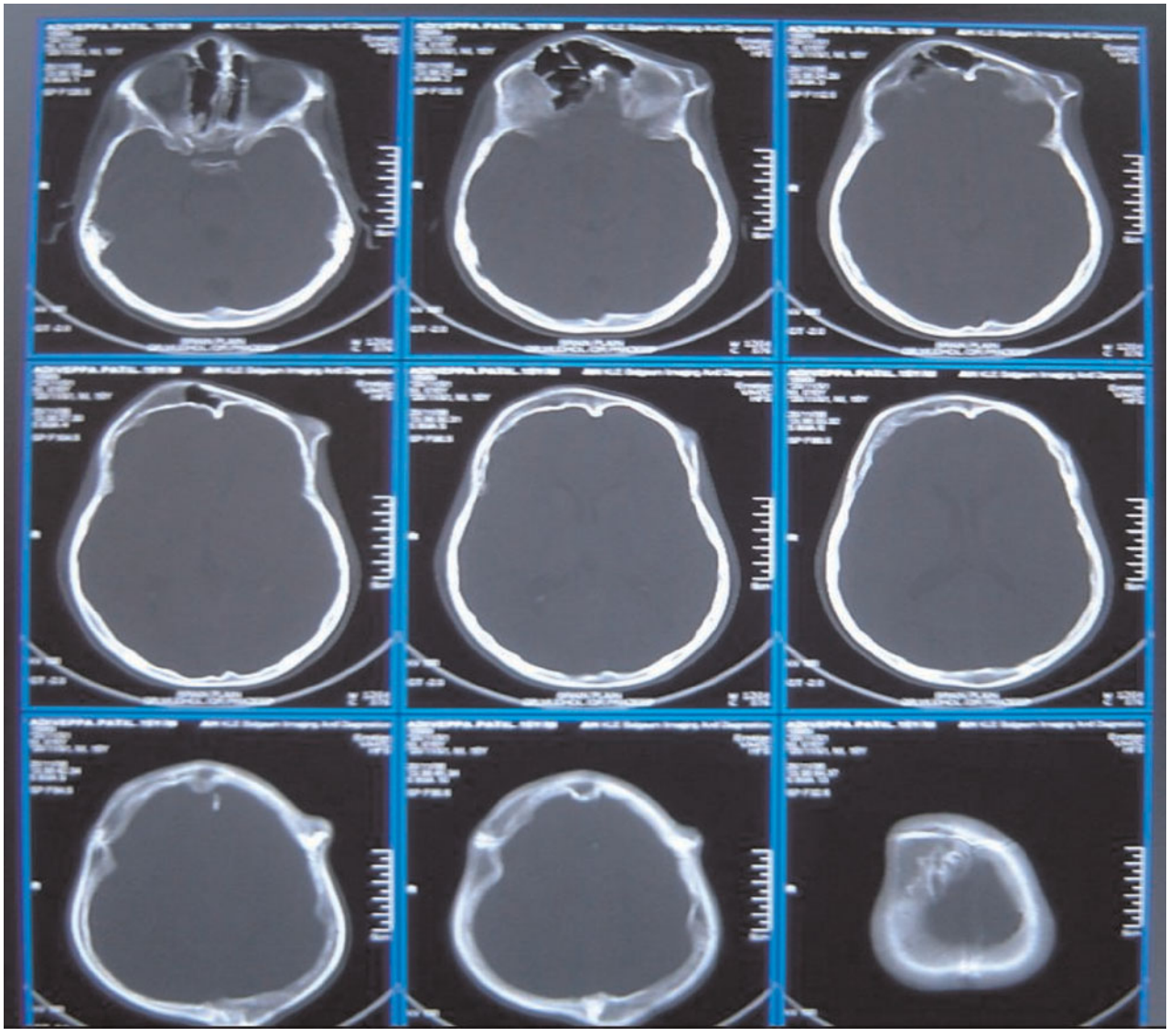

FIG. 7

Bone window CT showing expansion of outer skull table at left frontal, left parietal & right high parietal areas with expansion of inner skull table at mid frontal area.

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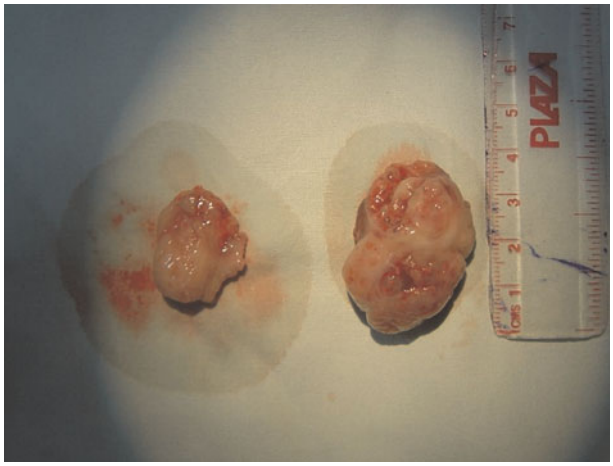

FIG. 8
Enlarged right tonsil.

and expansion of the inner skull table at the mid-frontal area (Figure 7).

A CT scan of the hands revealed macrodactyly and clinodactyly of the right second, third and fourth fingers. A CT scan of the feet showed overgrowth of the right calcaneum. Computed tomography scans of the thorax were normal, with no evidence of pulmonary cystic lesions. Fine needle aspiration cytology of the abdominal mass revealed a lipoma, and skin biopsies of the neck lesions confirmed an epidermal naevus. Echocardiography revealed a grade one mitral valve prolapse (i.e. an anterior mitral leaflet and posterior mitral leaflet prolapse with no mitral regurgitation) and normal pulmonary artery pressure.

A tonsillectomy was performed under general anaesthesia, under 'grave risk' consent, since the patient had varicosities of the veins of the right lower limb. The right tonsil measured 4 cm in length, compared with a normal left tonsil of 2 cm (Figure 8).

The patient made an uneventful recovery and was discharged three days after surgery. Tonsillar biopsy was suggestive of chronic, non-specific tonsillitis with increased fibrosis.

Discussion

The first case of Proteus syndrome was reported in 1979,[1] but it was not until 1983 that Widemann *et al.* conferred the name 'Proteus syndrome'.[2] 'Proteus' refers to the Greek god of the sea, who had the ability to change his shape at will in order to avoid capture. The first case was reported from India in 1990. Less than 100 bona fide cases of this syndrome have been reported in the world literature.[3]

Proteus syndrome is a heterogeneous disorder that can affect any body tissue, resulting in a wide variety of deformities in a mosaic pattern. Although rare, the disease is said to be caused by a somatic alteration in a gene. No specific mutation has yet been identified.[4]

Therefore, the diagnosis and management of Proteus syndrome depends on clinical evaluation and imaging. The fundamental manifestation of this disease is dysplasia. The features of the disease are not seen at birth but become apparent from one year of age. The disease involves multiple systems, with disordered skeletal growth being the obvious feature. Patients typically develop asymmetrical enlargement of the hands and feet, varicosities, verrucous epidermal naevi, haemangiomas and lymphangiomas. The cerebriform connective tissue naevus is specific, showing raised and rugose lesions composed of collagenised connective tissue. These lesions may occur on the plantar surface of the foot, palmar surface of the hand, lateral and dorsal surfaces of the fingers, the nose, and, rarely, on the chest, abdomen and back.

Patients carry a social stigma due to the disfiguring effect of the syndrome. Evaluation includes skeletal survey with CT and magnetic resonance imaging in order to rule out any central nervous system manifestations.[5]

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Dr N D Zingade takes responsibility for the integrity of the content of the paper.
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