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Clinical Record

Proteus syndrome with huge tonsillar mass causing dysphagia: a rare case

J K Sahni, MS, MNAMS, S Kumar, MS, DNB, V Wadhwa, MS, G Kathuria, MS, DNB

Abstract
Proteus syndrome is a rare and highly variable hamartomatous syndrome that can affect multiple organ systems. It is characterized by hyperplasia of connective tissue, vascular malformations, epidermal naevi and hyperostosis. Most of the cases present to plastic and orthopaedic surgeons. Otolaryngologic presentations are minimal. We report such a rare case, which presented with a huge unilateral tonsillar mass causing dysphagia.

Key words: Proteus Syndrome; Tonsil Hyperplasia; Tonsillectomy

Introduction

Proteus syndrome is a rare, recently described, congenital hamartomatous syndrome involving malformations of multiple organ systems.[1] The diagnostic criteria for the disease are yet to be established properly, and it may be confused with similar syndromes such as neurofibromatosis, Klippel–Trenaunay–Weber syndrome and Mafucci syndrome. We report a case of Proteus syndrome that presented with huge tonsillar enlargement causing dysphagia. The tonsillar mass was surgically removed without any complication. The case is reported on account of its rarity.

Case report

A five-year-old boy was referred to the ENT out-patient department of the Kalawati Saran Children's Hospital with complaints of increasing difficulty in swallowing of one year's duration and also congenital morphological abnormalities. The child had been born at full term to a prima gravida mother by normal vaginal, hospital delivery, from a non-consanguineous marriage. There was no family history of similar abnormalities. The ante- and peri-natal history was insignificant. The birth weight had been 2.7 kg and the infant had shown mild right hemifacial hypertrophy, linear epidermal naevi on the right half of the body, and mild hypertrophy of the right upper and lower limbs. Hypertrophy of the left ring finger had been noted. The parents advised that all these features had become more obvious with the growth of the child. The motor milestones had also been delayed.

On examination, the child had a 'hot potato' voice. The above-mentioned skeletal findings were confirmed. The child had mild developmental delay, with frontal bossing, scoliosis, epidermal naevi, palmar and plantar hypertrophy, venous varicosities, and left genu valgum (Figure 1). Oropharyngeal examination revealed gingival hypertrophy, malocclusion and overcrowding of teeth, and a huge right tonsillar mass that was almost touching the opposite side (Figure 2). The left tonsil was normal in size. The rest of the ENT examination was unremarkable. Ophthalmological examination showed bilateral choroid sclerosis.

Routine investigations, including serum calcium, phosphorus and alkaline phosphatase levels, were reported to be within the normal range. The soft tissue lateral X-ray of the neck showed no adenoid hypertrophy. Thyroid function tests were also normal. An abdominal ultrasound was normal except for the absence of the left testis. Whole body scans showed skeletal hypertrophy of the right upper and lower limbs and scoliosis. Fine needle aspiration cytology of the tonsillar mass showed lymphoid hyperplasia. Karyotyping from a peripheral leucocyte culture revealed a normal 46XY pattern.

Because of complaints of increasing dysphagia, tonsillectomy was conducted under general anaesthesia. The tonsillar mass was easily dissected out in toto, following a routine tonsillectomy procedure (Figure 3). The post-operative period was uneventful and the child was discharged after three days. Histopathological examination of the tonsillar mass revealed normal tonsillar tissue. Six months after the operation, the child was asymptomatic, with no oropharyngeal complaints and no signs of recurrence.

Discussion

Cohen and Hayden first reported a case of Proteus syndrome in 1979,[2] but it was not until 1983 that Widemann *et al.* conferred the name 'Proteus syndrome'.[1] The first case in India was only reported in 1990.[3] Proteus was the Greek god who could change shape to avoid capture. On the basis of around 200 cases reported in the world literature, Proteus syndrome has been found to have a sporadic occurrence, with an equal male to female ratio.[4]

Apart from some cutaneous manifestations, features of this disease are usually not marked at birth but start to become apparent after one year of age.[4] The disease involves multiple systems, with skeletal overgrowth being the most apparent feature. The characteristic overgrowth features are: hemifacial hyperplasia; hypertrophy of part or whole of one or both upper and lower limbs or the

From the Department of ENT and Head & Neck Surgery, Lady Hardinge Medical College, New Delhi, India.
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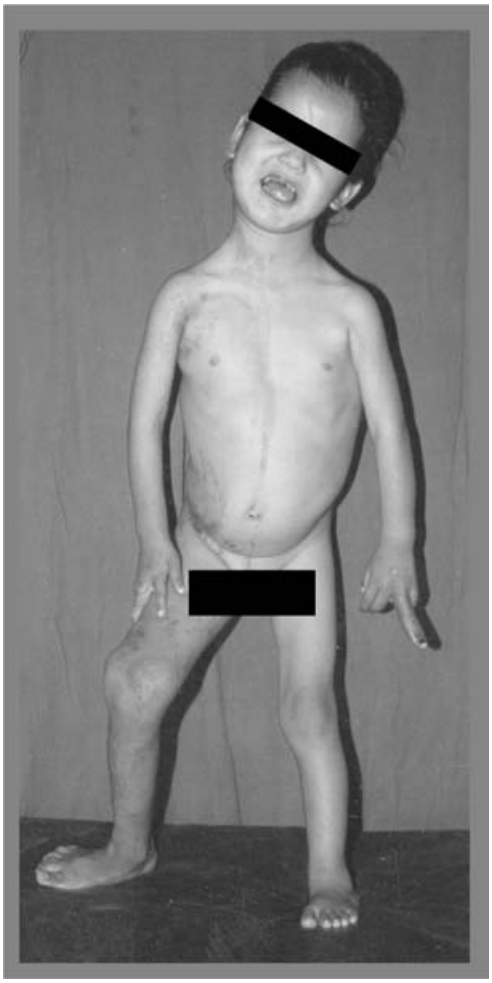

FIG. 1
Child showing right hemihypertrophy of the body, with left ring finger macrodactyly.

trunk; or any combination of these (including hemihypertrophy of one side of the body). Macrodactyly has been regarded as a characteristic feature but not diagnostic.[4] Other common features included are verrucous epidermal naevi, plantar and palmar gyriform masses, ambiguous genitalia,[5] infantile haemangiomas and lipoma-like subcutaneous hamartomas. Among these, the palmar and plantar gyriform mass may be considered a pathognomonic sign.[5] Developmental delay also occurs in about 50 per cent of cases.[4]

The majority of Proteus syndrome cases present to plastic and orthopaedic surgeons. Very few cases directly present to the otolaryngologist because ENT manifestations are less obvious and seldom require prompt treatment. However, the otolaryngological problems associated with this disease which have been reported are high arched palate, gingival hypertrophy, malocclusion and overcrowding of teeth.[6] Because of the known association of Proteus syndrome with vascular abnormalities, it was feared that our patient's tonsillar mass might bleed during the tonsillectomy; however, no abnormal bleeding was observed and the tonsillar mass was easily dissected out in toto.

Our patient was a classical case of Proteus syndrome, showing typical features such as macrodactyly, plantar and palmar gyriform masses, ambiguous genitalia and epidermal naevi. These features helped us to differentiate the syndrome from neurofibromatosis, Klippel–Trenaunay–Weber syndrome and Mafucci syndrome, which all may

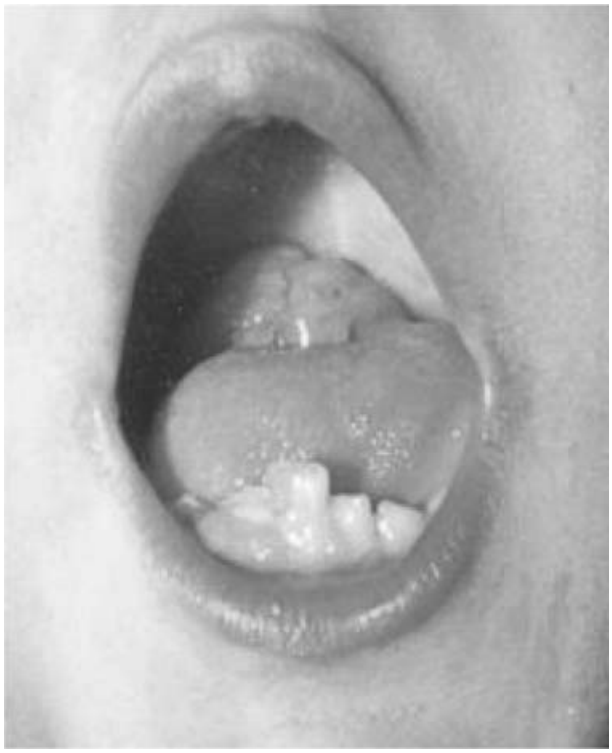

FIG. 2
Huge right tonsillar mass causing almost total oropharyngeal obstruction.

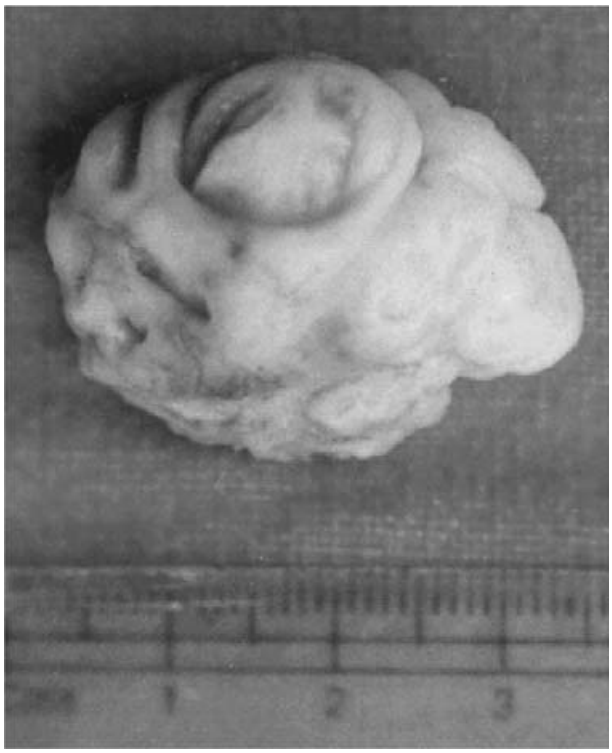

FIG. 3
Dissected tonsillar mass.

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contain similar hypertrophy features.[1] Our patient had a huge right tonsillar enlargement causing dysphagia. Unilateral enlargement of the tonsil was due to this syndrome's tendency to prefer organs of one half of the body. However, such symptomatic tonsillar enlargement, which required tonsillectomy, has never before been reported in Proteus syndrome.

The objective of this article is to educate otolaryngologists about this rare syndrome, which may cause significant tonsillar enlargement. Although plastic and orthopaedic surgeons would normally manage Proteus syndrome cases, otolaryngological surgeons can certainly improve the quality of life of those patients with obvious ENT complaints.

- **This report describes the otolaryngological features in a child with Proteus syndrome, a rare congenital disorder involving multiple organs with hamartomatous malformations**
- **The primary feature of this case was of extreme, unilateral tonsillar enlargement, managed successfully by tonsillectomy**

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Address for correspondence:
Dr Vikram Wadhwa,
B-7, Pandara Road, New Delhi 110003, India.

E-mail: drvikramwadhwa@yahoo.com

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