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Nasopalpebral Lipoma *sine* Coloboma Syndrome—First Case Report

Ved Prakash Rao Cheruvu[1] Manal M. Khan[1] Madhuri Arora[1] Vikas Chaudary[1]

[1] Department of Burns and Plastic Surgery, All India Institute of Medical Sciences Bhopal, Bhopal, Madhya Pradesh, India

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Address for correspondence Ved Prakash Rao Cheruvu, MCh, Department of Burns and Plastic Surgery, All India Institute of Medical Sciences Bhopal, Room no. 1017, 1st floor, Academic Block, Saketnagar, Bhopal 462020, Madhya Pradesh, India (e-mail: vedprakash.plasticsurg@aiimsbhopal.edu.in).

Abstract

The nasopalpebral lipoma-coloboma syndrome was described for the first time in 1982. It is an autosomal dominant syndrome with complete penetrance and is characterized by features like congenital symmetric upper eyelid and nasopalpebral lipomas, bilateral symmetric upper and lower eyelid colobomas, broad forehead, widow's peak, abnormal eyebrow pattern, telecanthus, broad nasal bridge, maxillary hypoplasia, and ophthalmological abnormalities. We report a case of a milder variant of the nasopalpebral lipoma-coloboma syndrome that we have termed "nasopalpebral lipoma *sine* coloboma syndrome." Such a milder variant is not reported hitherto in the literature. We also describe the surgical correction of the deformity in a case that presented in adulthood, with a satisfactory and pleasing aesthetic outcome.

Keywords

- abnormalities
- coloboma
- lipoma

Introduction

Nasopalpebral lipoma-coloboma syndrome was described for the first time by Penchaszadeh et al in 1982.[1] Eight cases of this syndrome involving three generations were reported by them in a Venezuelan family. Akarsu and Sayli, in 1990, reported this syndrome in a Turkish family and found a similar pattern.[2] Two reports of solitary cases of this syndrome are also published.[3,4] Tadisina et al published a literature review in 2015 in which they identified 11 cases of this syndrome.[5] This is described as an extremely rare autosomal dominant syndrome with complete penetrance, although the more recently reported cases have been sporadic. This is characterized by symmetric upper eyelid and nasopalpebral lipomas, bilateral symmetric upper and lower eyelid colobomas, broad forehead, "widow's peak," abnormal eyebrow pattern, telecanthus, broad nasal bridge, and maxillary hypoplasia. Ophthalmological features including persistent epiphora, malposition or aplasia of the lacrimal punctae, aberrant eyelashes, conjunctival hyperemia, corneal and lenticular opacities, and a divergent squint have also been described.[1]

We report a case of a milder variant of this syndrome, hitherto not reported in the literature, and also describe the surgical correction of the deformity in an adult female.

Case Report

A 26-year-old female presented to us for the correction of abnormal facial features. She was the eldest of the three issues of her parents. The second issue, a male, 3 years younger than her, had a congenital meningomyelocele and died at the age of 15 days. As per the available history, the second issue did not have any facial deformity. The third issue, a male, 5 years younger than her has a similar deformity but is yet to present to us for evaluation. In our index case, the course during her mother's pregnancy and labor was uneventful. Physical and mental development in

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Fig. 1 Clinical picture at presentation: (**A**) Frontal view, (**B**) right oblique view, and (**C**) left oblique view.

childhood was normal. On examination, the patient had symmetric lipomas involving the nasopalpebral region bilaterally. The medial canthi on both sides were displaced laterally, the nose root was distorted, and telecanthus was present. The forehead was broad, a "widow's peak" was present, and an abnormal eyebrow pattern was observed (►**Fig. 1**). There were no colobomas in the upper and lower eyelids, and epiphora was not present. The rest of the ophthalmological and physical examination was normal. Computed tomography revealed normal bony anatomy (►**Fig. 2**). Magnetic resonance imaging showed bilateral nasopalpebral lipomas (►**Fig. 3**).

We performed the surgical correction of the deformity on both sides as detailed below:

i. Correction of telecanthus using the Mustarde technique (jumping man flap or five flap Z-plasty; ►**Fig. 4**).
ii. The lipomatous masses were identified in the subcutaneous plane and dissected from the underlying tissues. The lacrimal system was protected from damage by cannulating the inferior canaliculus before the excision of the lipoma (►**Fig. 5**). Adequate hemostasis was achieved with prior infiltration of adrenaline-saline.
iii. The medial canthal ligaments were identified and isolated (►**Fig. 6**). They were shortened appropriately and refixation was performed with polypropylene suture.
iv. Following this, the patency of the lacrimal system was confirmed by lacrimal syringing through the cannula, and skin wound closure was performed.

The final appearance after correction is shown in ►**Fig. 7**. A thermoplastic splint was applied over the nose to reduce the dead space and achieve good contour. The splint was continued

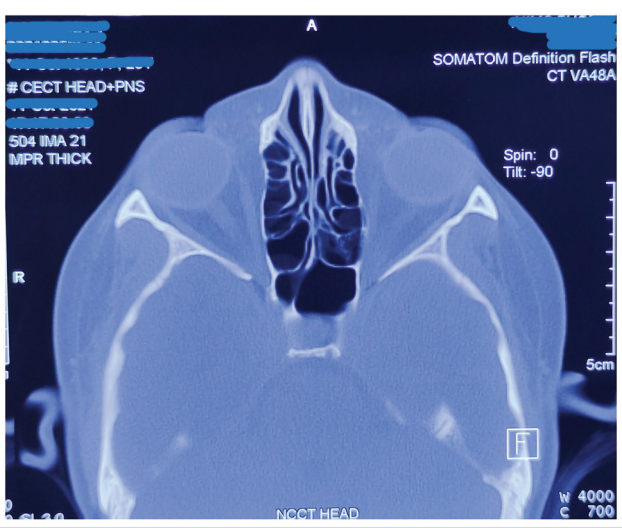

Fig. 2 Computed tomography axial scan showing normal bony anatomy.

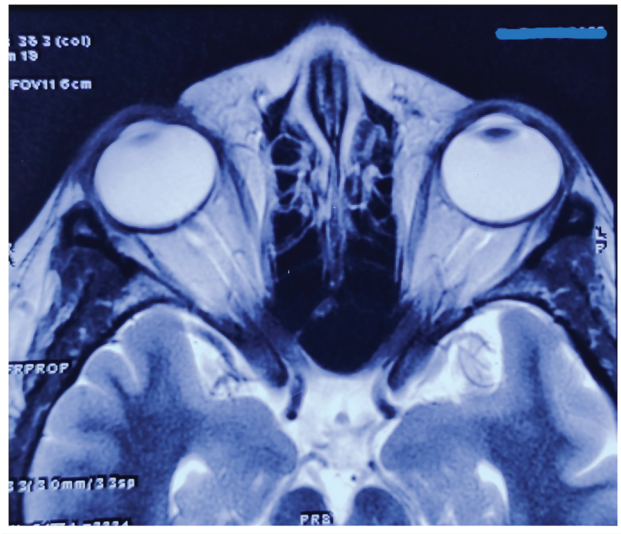

Fig. 3 Magnetic resonance imaging axial scan showing bilateral nasopalpebral lipomas.

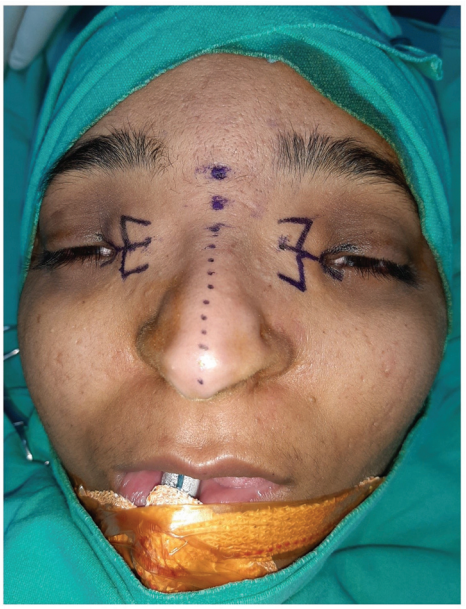

Fig. 4 Skin markings for Mustarde double opposing Z-plasty flaps.

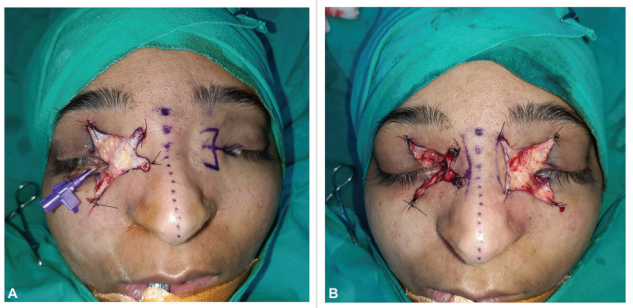

Fig. 5 Subcutaneous lipomatous masses: (**A**) right side and (B) left side.

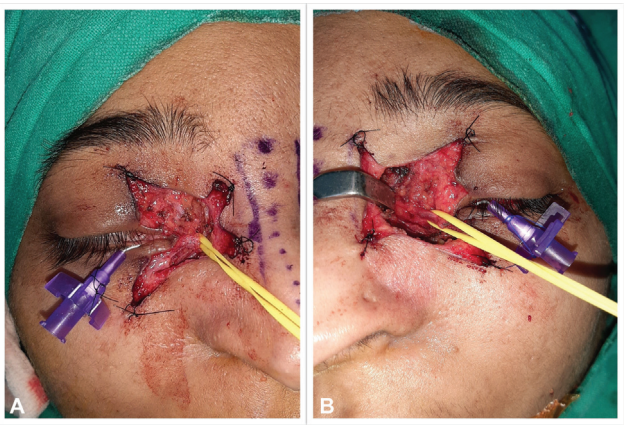

Fig. 6 Stretched medial canthal ligaments: (**A**) right side and (**B**) left side.

Fig. 7 Final appearance after skin closure.

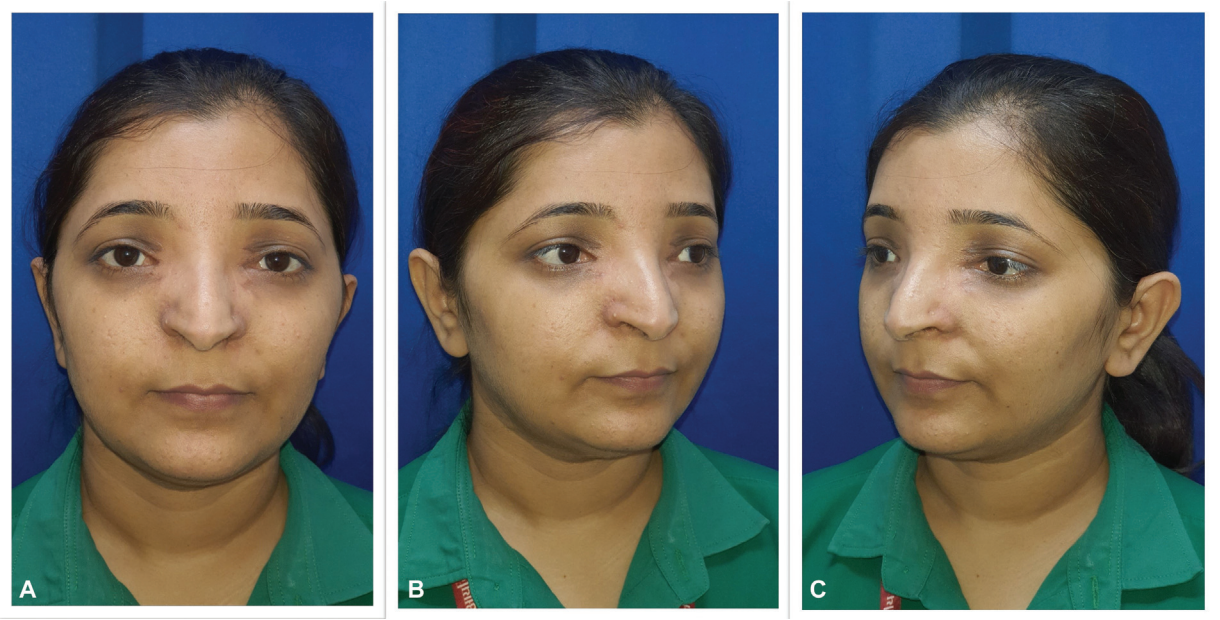

Fig. 8 Picture at 3-month follow-up: (**A**) frontal view, (**B**) right oblique view, and (**C**) left oblique view.

for 2 weeks. The postoperative period was uneventful. Three months follow-up pictures showed complete correction of telecanthus and the medial canthi were positioned normally with nicely shaped palpebral fissures on both sides (►**Fig. 8**).

Discussion

The physical findings in our patient were consistent with the descriptions of the nasopalpebral lipoma-coloboma syndrome, except that there were no colobomas, the lacrimal system was normal, and there were no other ocular anomalies. The severity of telecanthus was also less compared with the cases reported in the literature previously. Therefore, we believe that it should be termed "nasopalpebral lipoma *sine* coloboma syndrome" and should be regarded as a milder variant of the nasopalpebral lipoma-coloboma syndrome. Such a milder variant of the syndrome has not been described in the literature previously.

The pathogenesis of the nasopalpebral lipoma-coloboma syndrome remains unknown; it might be a genetically determined single developmental field complex anomaly.[6] Bock-Kunz et al reported a case of nasopalpebral lipoma-coloboma syndrome that suggested a new mutation in 2000.[3] Chacon-Camacho et al reported a case in which they described nanophthalmos and the histopathological analysis revealed a hamartoma of smooth muscle and lipomatous tissue.[7] Babu et al reported a case with unilateral lipoma and coloboma associated with a limbal dermoid.[8] Moreira Gonzalez and Jackson recommended a coronal approach for surgical correction. They also recommended that surgery should be performed early to avoid secondary ophthalmic problems that can compromise vision.[9]

Our patient is the first affected case in the family, with a similar affliction in her younger male sibling. Her parents are normal. Except for the appearance, the patient was a completely normal child. Imaging showed the lipomatous mass occupying the nasopalpebral area, but hypertelorism was absent. The deformity was milder when compared with the classical nasopalpebral lipoma-coloboma syndrome. We were able to excise the lipomatous mass through the palpebral approach avoiding additional scars. Prior infiltration with adrenaline-saline provided good visualization and adequate hemostasis was secured carefully. In the medial canthal region, meticulous dissection was performed. After isolation of the medial canthal ligament and adjusting the appropriate length, reattachment was performed with some overcorrection. Excision of the skin or skeletal correction was not required in our case. In the postoperative follow-up, the patient was very satisfied with the aesthetic result.

Conflict of Interest
None declared.

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