

# An unusual complication after craniofacial surgery for Apert syndrome

Abhay A. Lune<sup>1,3</sup>, Bharat B. Dogra<sup>2</sup>, Sonali A. Lune<sup>3</sup>

Departments of <sup>1</sup>Ophthalmology and <sup>2</sup>Surgery, Padmashree Dr D Y Patil Medical College, Hospital and Research Centre, Dr D Y Patil Vidyapeeth, Pimpri, Pune, <sup>3</sup>Lune Eye Clinic, Pune, Maharashtra, India

## ABSTRACT

Apert syndrome is a rare genetic disorder, characterized by premature fusion of skull sutures, mid-face hypoplasia and syndactyly of the hands and feet. It is inherited as autosomal dominant or sporadic and is associated with increased paternal age. It arises from mutations in the fibroblast growth factor receptor 2 gene on chromosome 10q26.

A case of Apert syndrome who had undergone craniofacial surgery elsewhere 4 years back presented to us with purulent discharge near the lateral orbital margin of right orbit, watering and redness of the right eye. He had telltale signs of this syndrome in the form of skull deformities such as brachycephaly, frontal bony prominence, mid-face hypoplasia, proptosis and syndactyly of both hands and feet. There was a surgical scar of previous craniofacial surgery over the bi-coronal region. He had a discharging granuloma over the lateral orbital margin and the adjacent lower eyelid had developed cicatricial ectropion. X-ray and computed tomography scan orbit confirmed the clinical suspicion of osteomyelitis of the underlying zygomatic bone at the site of miniplate and screw fixation of the earlier surgery. He was treated with excision of granuloma and extraction of loose screw and infected miniplate while ectropion was corrected by rotation advancement of temporal skin flap.

**Keywords:** Apert syndrome, brachycephaly, craniofacial surgery, ectropion, syndactyly

## Introduction

Apert syndrome is a rare genetic disorder, characterized by premature closure of skull sutures, commonly the coronal sutures, maxillary hypoplasia and syndactyly of the hands and feet.<sup>[1]</sup> The prevalence is 1 in 65,000.<sup>[2]</sup> The early bony fusion affects the shape and size of the orbit, head and face causing proptosis and mental retardation. Ectropion, which is an eversion of the lid margin, is not a feature of this syndrome. It occurred as a complication of previous craniofacial surgery.

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## Case Report

A 21-year-old mentally-retarded male patient presented with the complaints of a tender swelling with purulent discharge near the right lateral orbital margin since 1 month along with eversion of the right lower eyelid since 15 days. He had undergone craniofacial surgery in the form of the orbital rim advancement for his skull deformity 4 years back elsewhere.

On examination, he had the classical features of Apert syndrome, namely, brachycephaly, bird-beak appearance of nose, mid-face hypoplasia and syndactyly of hands and feet [Figures 1-3]. Detailed ocular examination revealed a cycloplegic refraction of +3.00 Dsph OD and +4.00 Dcyl × 180 OS, with best-corrected vision of 20/80 in both eyes. He had an inter-pupillary distance of 80 mm and inter-medial canthal distance of 40 mm. Both eyes had proptosis. The left eye had hypotropia of 10 prism diopters with exotropia of 36 prism diopters. Ocular movements showed poor left superior rectus action and right inferior oblique overaction. The anterior segment findings were unremarkable except for a nebular corneal opacity in the inferior cornea of the right eye. Fundus examination of both eyes revealed tortuous blood vessels with normal discs and fovea.

Eversion of the lateral one-third of the right lower lid margin was noted with a 2 cm × 1 cm firm, tender,

### Address for correspondence:

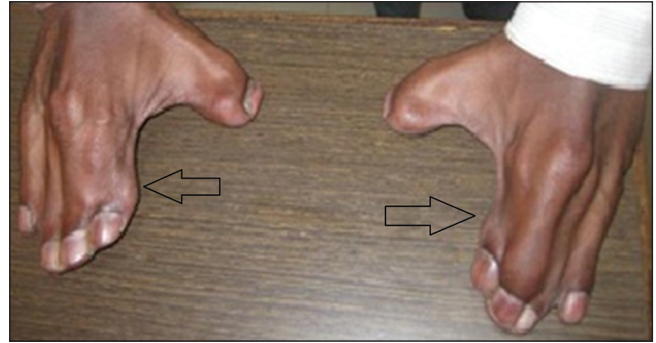
Dr. Abhay A. Lune, Lune Eye Clinic, 1641, Madhav Heritage, Tilak Road, Pune - 411 030, Maharashtra, India.  
E-mail: [abhaylune@gmail.com](mailto:abhaylune@gmail.com)

granulomatous mass with purulent discharge fixed to the underlying zygomatic bone near the right lateral orbital margin [Figure 4].



**Figure 1:** Full image of the face frontal view showing bird-beak nose, brachycephaly and exotropia with hypotropia of the left eye. A granuloma near right lateral canthus (arrow) and lower lid ectropion

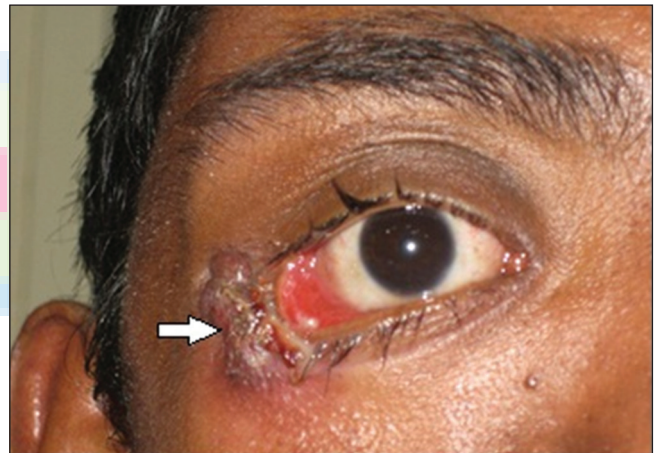
Skull radiographs revealed shallow orbits, hypertelorism and fixation plates and screws of the osteotomy in both the orbital margins and mid-face. A lytic lesion in the right zygomatic bone at the site of the granuloma was noted near the right orbital margin [Figure 5]. Computed tomography orbit confirmed a hyperdense lesion at the right lateral orbital margin with erosion of the underlying zygomatic bone, suggestive of osteomyelitis [Figure 6]. Brain



**Figure 2:** Syndactyly of hands



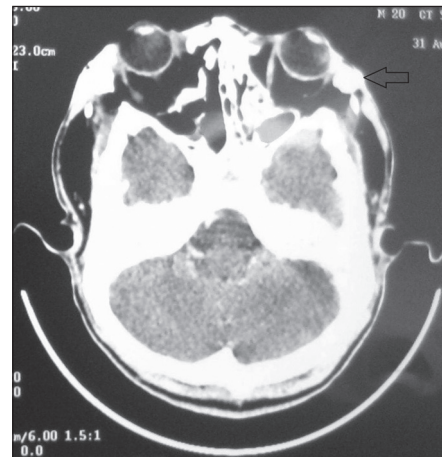
**Figure 3:** Syndactyly of feet



**Figure 4:** Close-up view of granuloma near right lateral canthus (arrow) and lower lid ectropion



**Figure 5:** X-ray skull, depicting hypodense cavity with loosened screw (arrow) and fixation nails and plates in place following previous surgery



**Figure 6:** Computed tomography scan of skull showing hyperdense lesion at the right orbital margin (arrow)

parenchyma and ventricles were normal. Radiographs of the cervical spine showed fusion of the lower cervical vertebrae [Figure 7] and those of hands and feet showed syndactyly [Figure 8].

Excision of the granuloma with debridement of the wound and removal of the loosened fixation screw and part of the plate was done. The cicatricial ectropion was released and a temporal skin flap was rotated and advanced to cover the resultant soft tissue defect. At 15<sup>th</sup> post-operative day the temporal flap and sutures were in place with mild residual ectropion [Figure 9]. Surgical treatment of syndactyly of hands and feet was also advised, which he wanted to undergo later.

## Discussion

Apert syndrome was first described by Eugene Apert in 1906.<sup>[3]</sup> It is inherited as autosomal dominant trait or is sporadic with mutations in fibroblast growth factor receptor 2 gene on chromosome 10q26.<sup>[4]</sup> It is usually associated with increased paternal age.<sup>[5]</sup>

Cicatricial ectropion of the lower eyelid is caused by a shortening of the anterior lamella, with resultant eversion of the eyelid away from the globe and occurs secondary to mechanical, thermal, chemical or surgical trauma.<sup>[6]</sup> In our case, osteomyelitis of the underlying zygomatic bone resulted in granuloma formation and the resultant skin scarring caused eversion of the lateral part of the lower lid.

Shallow orbits in Apert syndrome caused proptosis, luxation of the globes, exposure keratitis, exotropia and the hypertelorism as is shown by the increased inter-pupillary and inter-medial canthal distances. The nebular opacity in the right inferior cornea was due to the exposure keratitis following proptosis in the past. Other ocular features of hyperopia, high astigmatism and strabismic amblyopia were also noted in our case. The hypotropia noted may be due to structural alterations of the extraocular muscles, suggesting that ocular motility disturbances may not be caused solely by mechanical factors.<sup>[7]</sup> Absence of the superior rectus is also known to occur.<sup>[8]</sup>

Antimongoloid palpebral fissures, break in the continuity of eyebrows, horizontal groove above supraorbital ridge, keratoconus, ectopia lentis, corectopia, nystagmus, congenital glaucoma, albinoid alterations of the fundus with occasional papilledema and optic atrophy may also occur.<sup>[8]</sup>

The retarded skull growth affects the brain development, which may result in mental retardation as in our case.<sup>[9]</sup> Dental deformities like teeth malposition occur due to mid-face hypoplasia for which he was treated.

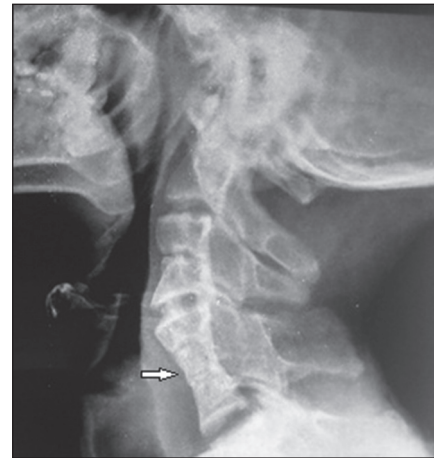


Figure 7: X-ray cervical spine showing fusion of cervical vertebrae (arrow)



Figure 8: X-ray hand showing syndactyly (arrows)



Figure 9: Post-operative photograph showing temporal skin flap and sutures in place with mild residual ectropion

Treatment of Apert syndrome involves a multi-disciplinary approach by Ophthalmologist, plastic surgeon, neurosurgeon and orthodontist. Le Fort craniofacial osteotomy involves disjunction of coronal sutures and fronto-orbital advancement to reduce the proptosis and the dysmorphic features.<sup>[10]</sup>

During infancy, the nasofrontal skeleton is modeled and advanced in order to protect the proptotic globes and to expand the anterior cranial fossa. This procedure also sets the stage for midfacial advancement, done in early or late childhood. Different secondary surgical procedures such as midfacial advancement, facial bipartition and monobloc osteotomy for mid-face hypoplasia and hypertelorism and orthognathic surgery to correct any dentofacial deformities can be done. After completion of skeletal growth, adjustments of maxilla and mandible and procedures to refine the forehead, chin, eyelids and nose can be done. Surgical separation of digits is done for syndactyly.

In conclusion, this case report emphasizes that while ocular and craniofacial manifestations of Apert syndrome are well-documented, post-surgical osteomyelitis of the underlying bone can occur, which is not reported in the literature studied. Thus, Apert syndrome patients should have regular ophthalmic, orthopedic and craniofacial reviews every year. To the best of our knowledge, we have not come across a similar case in the literature studied.

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