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Journal of Genetic Syndromes and Gene Therapy

Case Report

# Excyclotropia Leading to Meridoneal Amblyopia in Apert's Syndrome

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## ABSTRACT

Apert's syndrome is a very rare disease. Besides scarce features of acrocraniosynostosis, its ocular features are also predominant. Three cases, which are reported here, had typical features of the syndrome. Additionally one case of eighteen year old girl presented with meridoneal amblyopia due to excyclotropia. To our knowledge this is the first case report where fundus picture confirms presence of excyclotropia in Apert's syndrome. Second case had corneal erosion in proptosed eyes and third case was of a 12 years girl with severe proptosis along with craniosynostosis and syndactyly of all four limbs. The purpose of this report is to show the frequency of this syndrome in Jharkhand and highlighting the orbital and facial deformities and severe syndactyly of patients.

**Keywords:** Syndrome; Ocular features; Craniofacial features

## INTRODUCTION

Apert's syndrome (AS) is a very rare condition (15.5 per million births) characterized by craniosynostosis (premature fusion of cranial sutures), prominent forehead, ocular hypertelorism, proptosis, short and broad nose, pseudoprognathism, dental crowding and ectopia, maxillary hypoplasia, low hairline, webbed neck, pectus excavatum, and severe, bilateral syndactyly of hands and feet [1]. In 1842 Baumgartner and 1894 Wheaton reported this condition for first time, but it was Eugene Apert a French paediatrician who in for the first time described nine people with a similar disorder. Hence the name is attributed to him [2,3]. Ocular findings are proptosis (very frequent); strabismus (frequent).other eye features include corneal erosion, visual impairment and optic atrophy [4]. In our present three cases we found V pattern of eye globe with divergent up gaze and isotropic down gaze.

## CASE REPORT

### Case 1

First case was of 18 years old female who came with chief complaints of dimness of vision and watering of eyes for more than a month. No other significant family or therapeutic history. On examination she was found to have classical features of Apert's syndrome: Tower skull, symmetrical syndactyly of all four limbs. Besides she had high arch palate, class III malocclusion, anterior open bite, posterior crossbite, an excessively large appearing tongue and low hairline too. Ocular findings showed proptosis where eye globe was actually protrudes in relation to the cranial base as was seen in CT scan. The distances from the anterior plane of the cornea to the upper, lateral, and lower orbital margins were significantly increased. Also the distance from the anterior plane of the cornea to the facial plane was significantly reduced (Figures 1-6).

**Figure 1:** Tower skull and low hair line.

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**Received:** January 02, 2020; **Accepted:** January 16, 2020; **Published:** January 23, 2020

**Citation:** Chowdhury S, Srivastava M, Chowdhury N (2020) Excyclotropia Leading to Meridoneal Amblyopia in Apert's Syndrome. J Genet Syndr Gene Ther 11: 326. DOI: 10.4172/2157-7412.19.11.326.

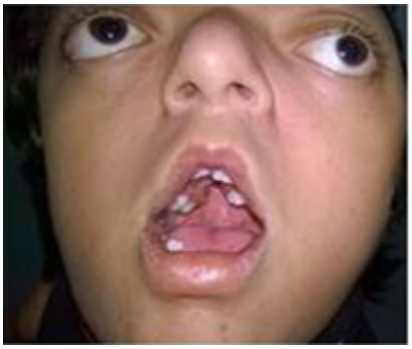

**Figure 2:** Mid face hypoplasia, class III malocclusion, anterior open bite, posterior crossbite, supernumerary teeth, creamy white enamel opacities, large appearing tongue and a v-shaped maxillary arch.

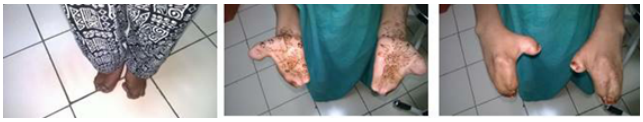

**Figure 3:** Syndactyly of feet.

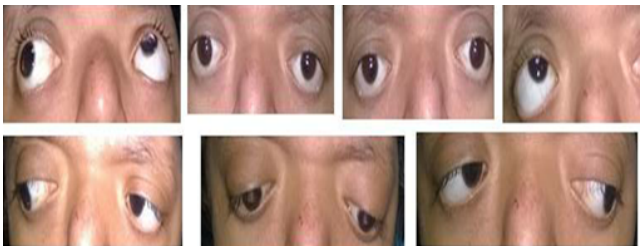

**Figure 4:** Eye positions in different gazes: Excyclotropia.

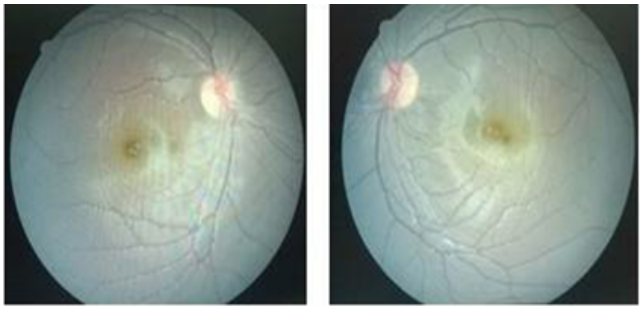

**Figure 5:** Fundus picture confirming excyclotropia.

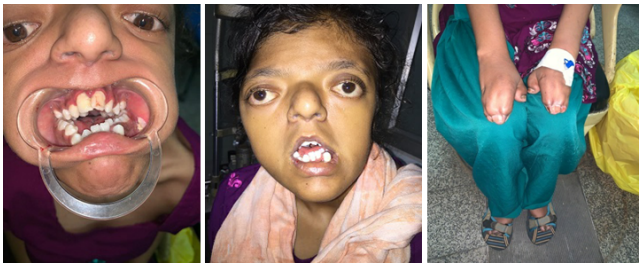

**Figure 6:** Follow up of case 1 after 2 years: Stable Proptosis, No exposure keratitis, no optic atrophy, Visual acuity 6/18.

First case was followed after 2 years, where patient had stable Proptosis, no exposure keratitis, no optic atrophy and visual acuity was 6/18.

## Case 2

Second case was also classical Apert's syndrome where a 9 year old girl presented with severe proptosis and visual impairment due to strabismic meridoneal amblyopia. There was occasional extrusion of globe while coughing. She also had severe syndactyly of upper and lower limbs (Figures 7 and 8).

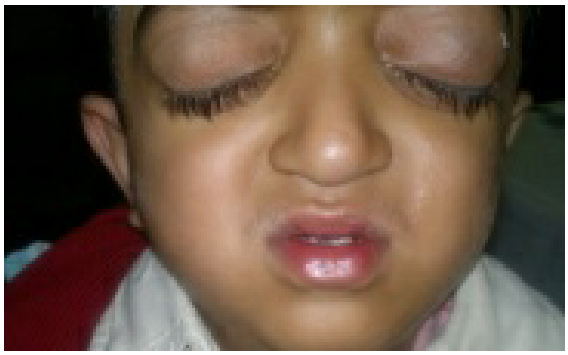

**Figure 7:** Proptosis in Apert's Syndrome.

**Figure 8:** Corneal erosion in Apert's syndrome.

### Case 3

Third reported case is of 7 year old girl who had corneal erosion which is occasional finding in Apert's syndrome.

Treatments in all these cases are mainly symptomatic with lubricants and night patching. Optic atrophy is thought to be due to compression. However none of the three cases had signs of optic atrophy. All are advised to be on follow up. Recurrent lower respiratory tract infection is common due to poor oral hygiene, so patients are advised to maintain it (Figure 9).

**Figure 9:** Supra orbital margin markedly retrude and elevated, both ocular proptosis and down slanting palpebral fissures and syndactyly of all four limbs.

Diagnosis is based on clinical examination. Radiological investigations provide detail of the syndrome. Ocular examination includes best corrected visual acuity, lids, pupillary reaction, and measurement of proptosis by Hertel exophthalmometer. Measurement of strabismus by prism bar funduscopy to rule out optic atrophy, fundus photograph to check excyclotropia. Tear breakup time and fluorescin staining of cornea to rule out ocular surface disorders which are common.

## DISCUSSION

AS is known acrocephalosyndactyly type I which is a form of craniosynostosis. It has an autosomal dominant inheritance, and develops as a mutation of fibroblast growth factor receptor -2 gene (FGFR2) on 10q26 gene locus. AS is especially seen in children whos parents are of advanced age. More than 70 craniosynostosis syndromes are described, which amongst many others, include AS, Crouzon syndrome, Pfeiffer syndrome, and Jackson-Weiss syndrome [5,6]. AS is classified as branchial arch syndrome, affecting the first branchial arch [7,8].

Various etiological hypotheses proposed are as: maternal infection due to virus embryopathy; antenatal drug consumption by mothers; an inflammatory process at the base of the skull and mal development of the skull; high paternal age [9]. Apert fibroblasts synthesized greater quantities of glycosaminoglycans (GAGs). The amount of hyaluronic acid (HA) secreted by Apert fibroblasts was much higher than that secreted by normal fibroblasts, but because the absolute values of heparan sulfate, chondroitin sulfate, and dermatan sulfate also rose in Apert media, the HA-sulfated GAG ratio was similar in the media obtained from both populations [10]. Cohen (2005) states that sutural agenesis in the midline region is characteristic of AS. This midline defect normally obliterates during 2nd-4th year of life [11]. Once a suture becomes fused, growth perpendicular to that suture becomes restricted, and the fused bones act as a single bony structure while compensatory growth occurs at the remaining open sutures to allow continued brain growth. In AS, there is often no suture in the metopic or sagittal regions.

Ocular morbidly can make AS patient's life miserable. Ocular features are studied comprehensively which includes few new findings. This are mentioned here:

Both ocular proptosis and down slanting palpebral fissures are characteristic features, which are often asymmetric were seen in case 1 and 3.

Supra orbital margin was markedly retrude and elevated, most pronounced laterally, especially seen in case 1 and 3.

In the sagittal plane, both the roof and the floor of the orbit were significantly shorter than normal, and the superior and inferior orbital margins were retruded in relation to the cranial base.

Dimunition of vision:due to amblyopia,strabismic or meridoneal

Recurrent corneal erosions were present and pronounced in case 2 and 3 as patients were unable to close their eyes completely.

Absence of exposure keratopathy despite severe proptosis.

Optic atrophy is not seen which probably could be due to presentation at early age.

Diminution of vision is mainly due to strabismic amblyopia

All had astigmatism but compliance with spectacle is very poor.

For medication all has to depend on care giver as they have syndactyly.

But most striking features in all cases were psychological as each patient is considered as differently abled.

Lack of multisystem approach is a hurdle.

Ophthalmic intervention is aimed at prevention of proptosis related problem like corneal erosion and exposure keratopathy. Preservation of vision by preventing optic atrophy and amblyopia should get first priority. Surgeries for ophthalmic features are not reported. Eye care in the form of preventing ocular surface disorders, amblyopia and keeping low intra ocular pressure to protect optic nerve are beneficial [12,13].

## CONCLUSION

Detailed ocular findings are rarely reported in literature. In this study detailed ophthalmic features can explain visual diminution in AS cases. One of the cases had more than a year follow up showing no deterioration.

AS comes under very rare diseases hence genotyping and genetic counseling should be carried out in each case. Prenatal diagnosis and multi-disciplinary intervention by team of neurosurgeon, oral and maxillofacial surgeon, plastic surgeon and ophthalmic

surgeon can improve the quality of life in AS cases. Because of the true proptosis and the major orbital differences, including asymmetry, craniofacial surgery rarely lead to normalization of the relation between the eye and the orbit and between the eye and the facial plane. Indeed some identifiable Apert syndrome characteristics most often remain after craniofacial surgery.

## DECLARATION OF PATIENT CONSENT

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

## FINANCIAL SUPPORT AND SPONSORSHIP

Nil.

## CONFLICTS OF INTEREST

There are no conflicts of interest.

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