CASE REPORT

Axenfeld-Rieger syndrome associated with severe maxillofacial and skeletal anomalies

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Received: 01-05-2014
Accepted: 21-03-2015

ABSTRACT

This article reports an unusual case of Axenfeld-Rieger Syndrome (ARS) associated with severe maxillofacial and skeletal anomalies.A 55-year-old man with ARS revealed interesting and unexpected radiological findings. Severe maxillofacial and skeletal anomalies, in addition to the well-recognized extraocular findings, that occurs in patients with ARS is reported and our case highlights this possible rare association between ARS and maxillofacial and skeletal anomalies.

Key words: Axenfeld-rieger syndrome, corectopia, dextrocardia, maxillary aplasia, pseudopolycoria

INTRODUCTION

Axenfeld–Rieger Syndrome (ARS) is an autosomal dominant genetic disorder and the incidence of ARS is estimated at 1 in 200,000.[1] Although cases with ARS show a wide spectrum of developmental defects, including ocular, umbilical, auricular, dental and craniofacial abnormalities; the ocular and craniofacial features are primary presentations of this syndrome.[2] Cardiovascular and endocrinological anomalies may also be seen.

ARS manifests with varying degrees of ocular anterior segment dysgenesis including heterochromia, aniridia, coloboma of the iris and persistent papillary membrane, corneal opacities.[1,3] Craniofacial malformations include midface hypoplasia, hypertelorism, telecanthus, mandibular prognatism, hypodontia and microdontia. Cleft palate may also be found.[1] In this case report, a brief description of ARS with severe maxillofacial and skeletal anomalies which has not been reported previously was presented.

CASE REPORT

A 55-year-old man presented with gradual painless vision loss in both eyes since his childhood. Ophthalmic evaluation showed visual acuity of light perception bilaterally. Ocular manifestations are iris hypoplasia and atrophy, corectopia (refers to displacement of the pupil), pseudopolycoria (refers to multiple colobomata in the iris body that do not have associated pupillary musculature), corneal vascularization resulting from extensive peripheral anterior synechiae (refers to adherence of iris to the cornea) and acute corneal hydrops (refers to leakage of aqueous through a tear in descemet membrane) resulting from rupture of the Descemet's membrane and Dua's layer [Figure 1]. Ophthalmic ultrasonography showed no posterior segment abnormality. Intraocular pressures taken by tonopen were 28 mmHg in the right eye and 27 mmHg in left eye. Neurological examination was normal. The radiological investigation revealed dextrocardia, aplasia of nasal bone, upper alveolar process of maxilla and displasia of both upper and lower extremities [Figure 2]. Transthoracic echocardiography revealed no abnormality. Thoraco-abdominal and cerebral computed tomographies (CT) showed normal anatomy. A diagnosis of Axenfeld-Rieger Syndrome (ARS) was made. Intraocular pressure was successfully controlled with the dorzolamide 2% and timolol 0.5% fixed combination (Cosopt®, Merck and Co., Inc., Whitehouse Station, NJ, USA).

COMMENT

ARS is one of the ocular anterior segmentdysgenesis syndromes.[4-6] The pathogenesis is believed to be the impaired neural crest cell and ectodermal migration and differentiation during embryonic development.[1,7] The ocular and systemic features of ARS including craniofacial, dental, abdominal abnormalities are well-described in literature and multiple additional abnormalities have been reported in association

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Website: www.jomfp.in
DOI: 10.4103/0973-029X.157219

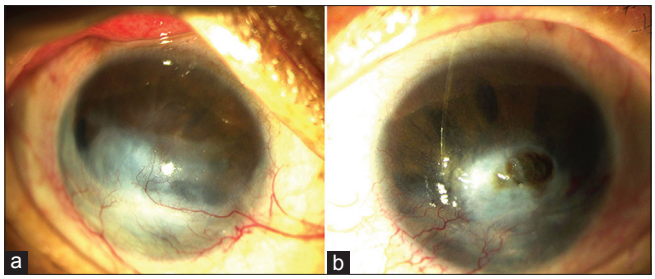

Figure 1: Ocular manifestations like iris hypoplasia and atrophy, corectopia, pseudopolycoria and corneal vascularization resulting from extensive peripheral anterior synechiae were noted in (a) Right eye; (b) Left eye (Acute corneal hydrops resulting from rupture of the Descemet's membrane and Dua's layer occured at follow-up)

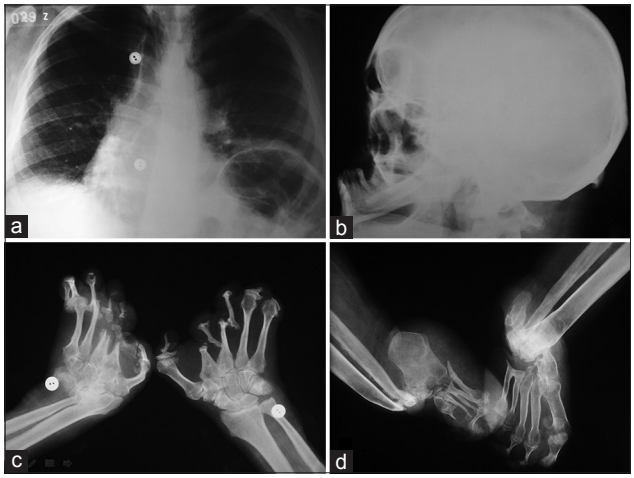

Figure 2: Radiological findings (a) Dextrocardia; (b) Aplasia of nasal bone, upper alveolar process of maxilla; (c) Ulnar deviation of the hands, displasia of second and third metacarpal bones in both hands, displasia of the second and third proximal phalanx in both hands, hypoplasia and aplasia of the all middle and distal phalangeal bones in both hands except the left hand the fifth proximal, middle and distal phalanges and the forth proximal phalanx; (d) Aplasia of lateral and medial malleoles in right and left ankle and displasia of the distal ends of tibia and fibula, hypoplasia and displasia of right calcaneus, hypoplasia of talus in both sides, aplasia of tarsal bones in both sides except hypoplasic navicular bones, displasia of tarsal bones with the fourth and fifth tarsal bones hypoplasia and hypoplasia of proximal phalanges and aplasia of middle and distal phalanges in both feet are seen

with ARS, including sensorineural hearing loss and cardiac abnormalities[6] however the association of ARS with severe maxillofacial and skeletal anomalies has not been reported previously.[5] These anomalies, in addition to the well-recognized extraocular findings, may occur in patients with ARS and our case highlights this possible rare association by this photo assay. As visual function can be preserved with early diagnosis, the knowledge of this presentation is important for the ARS patients. ARS is a multisystem disorder and a team approach including specialists in ophthalmology, cardiology and medical genetics is necessary. Further clinical reports accompanied by molecular diagnoses will elucidate this co-occurence in detail in the future.

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How to cite this article: Gokce G, Oren NC, Ozgonul C. Axenfeld-Rieger syndrome associated with severe maxillofacial and skeletal anomalies. J Oral Maxillofac Pathol 2015;19:109.

Source of Support: Nil. **Conflict of Interest:** None declared.