LETTER TO THE EDITOR

Crouzon syndrome: Ophthalmologic complications in an untreated adult patient

Syndrome de Crouzon : complications ophtalmologiques chez un patient adulte non traité

Premature craniosynostosis, midfacial hypoplasia, and exophthalmos form the triad known as Crouzon syndrome [1]. These malformations are caused by premature cranial suture closure (craniosynostosis). Facial abnormalities include proptosis due to shallow orbits, divergent strabismus or exotropia, ocular hypertelorism and hypoplastic maxilla [1].

Early surgical craniofacial treatment is the standard procedure to avoid further complications; hence, the vast majority of publications are case reports that describe children’s reconstructive surgery results. However, there is scarce information published about ophthalmologic complications in adult Crouzon syndrome patients, which have not been treated in their early years of life. We report a case of an adult patient with Crouzon syndrome who never underwent surgical treatment.

A 69-year-old woman diagnosed from Crouzon syndrome attended our clinic due to ocular pain and irritation after dislocation of her left eye, which she had repositioned by herself. She denied recent vision loss. The woman referred repeated episodes of eye dislocation of both eyes (OU), approximately once per month. No surgery had been performed in this patient and she followed no regular ophthalmologic treatment. The patient previously had consulted to several ophthalmologists because of similar episodes. She referred occasional headaches, and neurological examination only evidenced mild intellectual disability, and optical nerve damage. She didn’t have any personal or family relevant medical history and denied allergies.

External examination revealed exophthalmos in OU, hypertelorism and ptosis of the upper eyelid of OU, mainly in the left eye (OS) (Fig. 1). The upper eyelid of the OS was the one keeping the globe inside the orbit and when it was lifted dislocation of the eyeball occurred.

Non-corrected visual acuities showed amaurosis in the right eye (OD) and 0.2 in the OS. Intraocular pressure was 10 mmHg in OU. She presented 4 mm of lagophthalmos with preserved Bell phenomenon and only the lower conjunctiva was exposed in OU. Slit lamp examination showed intense conjunctival hyperemia, and severe keratitis with epithelial defects in the OD. Nuclear cataracts were described in OU.

Figure 1. Anterior view of the patient where intense ocular proptosis, hypertelorism and ptosis of upper eyelids are observed.

Non-dilated fundus examination found pale papillae with diffuse retinal retinochoroidopathy in OU. In the primary position of gaze, 20° exotropia, 15–20° hypertropia OD, due to obtuse angle of the orbits, with left eye dominance.

Head computed tomography (CT) and magnetic resonance (MRI) (Figs. 2 and 3) showed typical signs of Crouzon syndrome, such as marked ocular hypertelorism, bilateral...
proptosis, orbital deformity, craniosynostosis with important acrocephaly along with marked hydrocephalus.

Lubrication and topical antibiotic treatment was prescribed and she was then referred to the neuro-ophthalmology department for follow-up. The patient had always refused any surgical treatment.

In 1912, the French surgeon Crouzon, described this hereditary craniofacial syndrome named after him [2]. He described the triad as skull deformities, facial anomalies and exophthalmos [1]. It is caused by premature craniosynostosis, which induces abnormal skull growth and affects the orbital and maxillary complex development.

Crouzon syndrome occurs in approximately 1 in 25,000 births worldwide and makes up approximately 4.8% of all cases of craniosynostosis [1]. It is caused by a mutation in the fibroblast growth factor receptor-2 (FGFR2) [3]. It is an autosomal dominant genetic disorder with complete penetrance and variable expressivity [1]. Risk factors include patients with parents who manifest the syndrome or relatives known to be gene carriers and those with advanced paternal age fathers at the time of conception [4]. Diagnosis is based in most cases on clinical findings and radiological examinations.

Crouzon’s syndrome universal sign is proptosis caused by retraction of the lateral and inferior orbital margins with a very short orbital floor [5]. Corneal exposure and dislocation of the eyeballs are secondary to proptosis. Hypertelorism is also common, possibly due to a decrease in the growth of sphenozygomatic and sphenotemporal sutures [4]. However, there are isolated cases described of dislocation of eyeballs and cataracts in these patients. Other ophthalmologic complications due to neurological disorders include optic neuropathy and papilledema from raised intracranial pressure, being the optic atrophy the most common structural cause of reduced visual acuity in these patients [6].

Timothy et al. [6] describe amblyopia as the main cause of reduced visual acuity in Crouzon syndrome. Strabismus is the most common condition leading to amblyopia, they often show a V-Pattern exotropia. Early strabismus surgery in these cases may improve binocular vision and reduce amblyopia, and therefore improve the visual prognosis of these patients.

We describe a patient who developed many of the reported complications, presumably because of the long course of this disorder. This case illustrates how non-interventional attitudes in Crouzon syndrome patients evolve into major irreversible ophthalmologic problems.

Disclosure of interest

The authors declare that they have no conflict of interest concerning this article.

References


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