

An unusual extraocular muscle anomaly in a patient with Crouzon's disease

M. SNIR, E. GILAD, AND I. BEN-SIRA

From the Department of Ophthalmology, Beilinson Medical Center, Tel Aviv University Sackler School of Medicine, Petah-Tikva, Israel

SUMMARY A 29-year-old female suffering from Crouzon's disease was admitted to hospital with retinal detachment in the right eye. At operation agenesis of 4 extraocular muscles (superior and inferior recti and obliquus) was found, together with abnormal insertion of the 2 horizontal muscles. The same extraocular muscular abnormalities were found in the left eye. We suggest here a new surgical treatment in such cases and discuss the reasons for the limitation of ocular motility in such cases.

Crouzon's disease is a rare birth defect^{1,2} which in most instances is inherited as an autosomal dominant trait without complete penetrance.¹⁻³ Craniofacial dysostosis is characterised by bone and eye abnormalities. The bone defects include shallow orbits, protuberant frontal region, hypoplasia of maxilla, mandibular prognathism, enlargement of nasal bone and a beaklike nose, cleft palate, a high-arched palate, and dental malformation.^{1,3-7}

The physical characteristics of Crouzon's disease vary considerably from case to case, but they are essentially the result of premature synostosis of cranial sutures; the sphenozygomatic suture plays an important role in the disease. This premature fusion is responsible for many secondary changes in the basic brain and eye development.^{2,4,8-11}

Ocular manifestations include: exophthalmos, sometimes with luxation of the globe, obliquity of the palpebral fissures, ptosis, hypertelorism, optic atrophy, divergent strabismus, nystagmus, iridic and choroidal colobomas, and congenital cataract.^{1,3-7,11-16} There may be interesting and irregular abnormalities connected with this malformation. These include hypoplasia of orbital margins, anomalous lacrimal passages, and absence of some extraocular muscles.^{1,11,17}

The present report describes a patient with craniofacial disease and bilateral agenesis of 4 extraocular muscles—the superior and inferior recti and obliquus muscles. A special surgical procedure was used to prevent spontaneous luxation of the eyes and also to reduce the proptosis.

Correspondence to Dr M. Snir.

Case report

A 29-year-old female known to be suffering from Crouzon's disease (Fig. 1) was admitted to hospital with retinal tears and subtotal detachment in the right

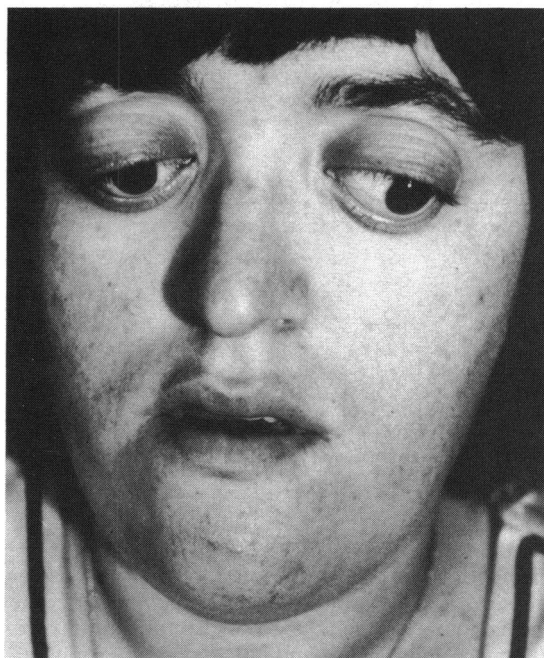


Fig. 1 The typical facial and eye malformations in a patient suffering from Crouzon's disease.



Fig. 2 LE: Severe exophthalmos in primary position in a patient with craniofacial dysostosis. Appearance of the right eye was the same.

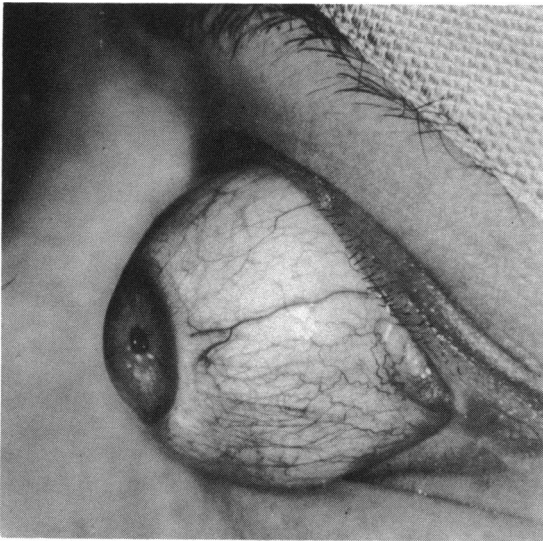


Fig. 3 LE: Extreme spontaneous luxation of the globe. The right eye was the same.

eye. Visual acuity was 6/30, -8.0 D, in the right eye and 6/10, -5.0 D, in the left eye. The intraocular pressure (IOP) and anterior compartment were normal in both eyes. In addition exophthalmos (basis 105 mm, right 31 mm, and left 30 mm on exophthalmometry) (Fig. 2), dislocation of the globes (Fig. 3), and myopic retinal degenerative changes were

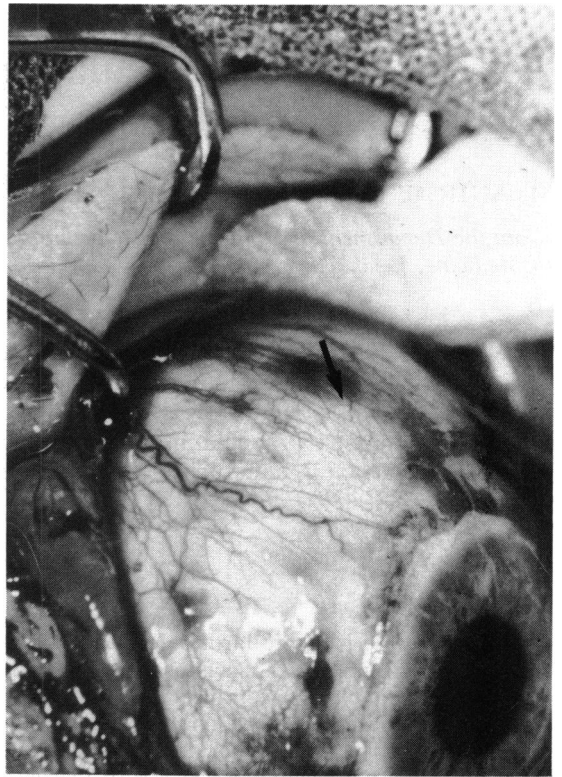


Fig. 4 RE: A close-up photograph of the eye showing the absence of the superior rectus muscle. The same agenesis of this muscle was found in the left eye.

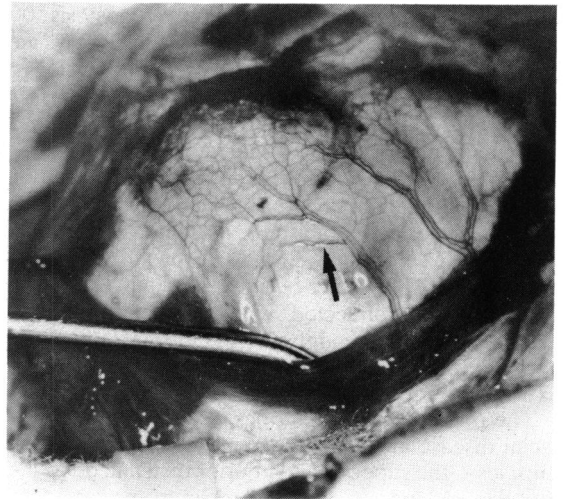


Fig. 5 RE: A close-up photograph of the eye showing the absence of the inferior rectus. There were identical findings in the left eye.

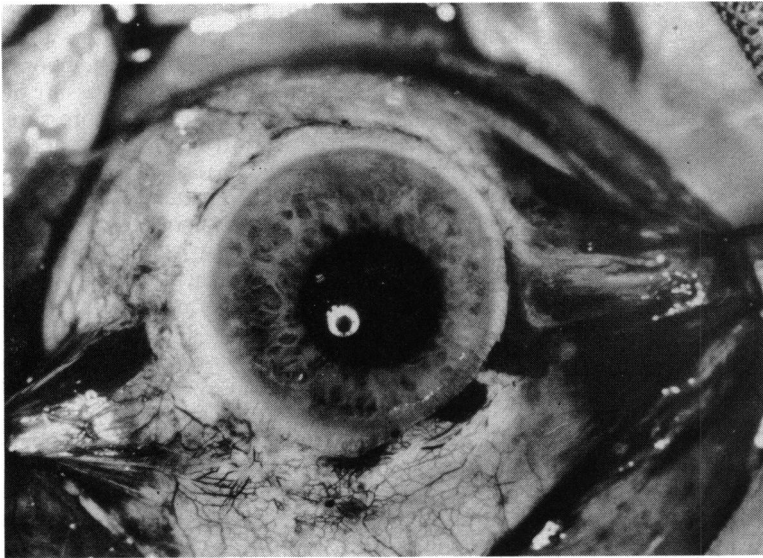


Fig. 6 RE: The only 2 hypertrophic horizontal extraocular muscles, with wide abnormal insertion anterior to normal locations. Findings in the left eye were identical.

observed on both sides. The eyes were in divergent position, and, although horizontal gaze was normal, the vertical movements were limited and greatly deficient.

At operation careful exploration of scleral surfaces disclosed the absence of the superior rectus (Fig. 4) and inferior rectus (Fig. 5) and the obliquus muscles. Moreover, there was hypertrophy of the horizontal muscles, with their wide insertion anterior to the normal location (Fig. 6). A continuous band of tissue

was also noted extending from the lateral rectus to the medial muscle, which looked like an intermuscular septum.

The patient underwent repair of retinal detachment in the right eye. During the operation, in addition to a 2 mm silicone band (for the correction of the detachment), a special procedure was used in an attempt to retract the eye back into the orbit. The Tenon sheath, which was stretched across the 2 horizontal muscles from above and below, was pulled



Fig. 7 RE: A significant reduction of the exophthalmos a few months after the special surgical procedure.

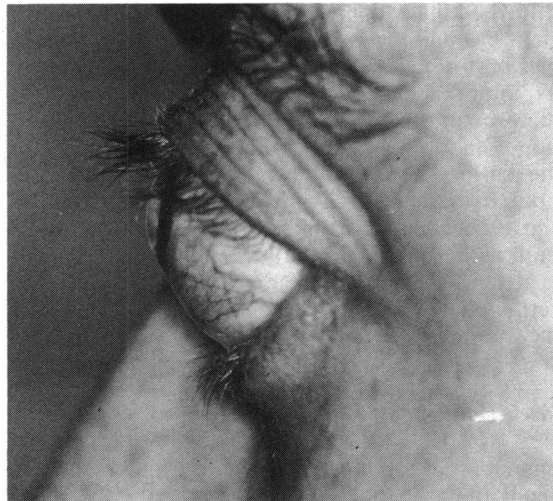


Fig. 8 LE: A significant reduction of the exophthalmic appearance a few months after the surgical treatment.



Fig. 9 Improvement of both eyes, showing retracted positions without luxation 2 years after the operations.

forward and was then sutured to the sclera about 7 mm from the limbus, with 3 7/0 catgut sutures in each half of the globe. This procedure resulted in an immediate retraction of the globe.

After operation the right eye was considerably more retracted than before the operation, with reduction of the exophthalmos (Fig. 7).

Exophthalmometry measurements were now 26 mm in this eye, without any luxation, and the movements were not essentially improved. However, the patient could close her lids without exposure of the globe. The retinal detachment operation was anatomically successful, with visual acuity 6/30, the same as before the surgical treatment (due to amblyopia).

Since the spontaneous luxation of the left eye disturbed the patient and retraction operation on the right eye had proved successful, the same operation was performed on the left eye 4 weeks later. Scleral exploration revealed essentially the same muscular abnormalities in the left eye; the Tenon sheath in the upper and lower globe was therefore pulled forward and sutured anterior to the equator of the eye. Post-operative examination showed a retraction of 5 mm in the left eye (exophthalmometry measurement was 25 mm, with basis of 105 mm) without luxation (Fig. 8).

After 2 years of follow-up the eyes have remained retracted, with the same exophthalmometry measurements and no dislocation of the globes (Fig. 9).

Discussion

Limitation of eye movement in Crouzon's disease is due both to the shallowness of the orbital cavity and to its position in angulation^{2-4 6 8-10 12 13} and to extraocular muscle abnormalities.^{1 11 17}

In craniofacial dysostosis there is stenosis of the cranial sutures and shortening of orbital roof by distinctive bulging of the pterion into the temporal fossa, together with increased intracranial pressure of the growing brain on the orbital roof. This pressure phenomenon leads to downward displacement of the floor of the anterior cranial fossa, combined with vertical displacement of the greater wings of the sphenoid bone. Even the orbital floor is reduced as a result of hypoplasia of the maxilla. All these features lead to restricted functional capacity of the orbits which, in turn causes the abnormal position and motility of the eyes.^{2-4 6 8-10}

Agenesis of extraocular muscles or abnormal insertion of muscle tendons causes misalignment and defective muscular vectors of action. Two cases of absence of the superior recti were reported by Weinstock and Hardesty.¹⁷ Five cases of absence of superior or inferior recti and obliquus muscles (one muscle in each eye) were described by Diamond *et al.*¹¹ In addition, Lyle and McGavic (see Walsh and Hoyt¹) reported on an unusual insertion of the superior obliquus muscle to the eye globe and to the superior rectus muscle.

In the present report the defective position of the eyes, with near total deficiency of vertical and torsional eye motility, was due to the bone malformation and to agenesis of 4 muscles in each eye (the 2 obliquus and vertical recti muscles). The wide insertion of the horizontal muscles in their unusual locations caused the minimal vertical gaze in both eyes.

The reason for the partial underdevelopment of extraocular muscles in such cases is unknown. It may be that an abnormal mutual stimulus between the orbital bones and the extraocular muscles arrests the growth of the ocular muscles.

There are several surgical methods for the prevention of ocular and visual complications. They include complicated procedures, such as orbital decompression by the removal of orbital walls,^{13 18-22} midfacial advancement,^{18 19 23} and horizontal maxillary osteotomy with advancement and bone graft implantation.¹⁸⁻²³ Each of these surgical procedures has both advantages and disadvantages.^{4 12 24}

We used a simple surgical technique—pulling forward the Tenon sheath above and below the eyeball and suturing it to the globe anterior to the equator. By this procedure we improved the cosmetic appearance of the patient, prevented luxation of the eyes, and reduced the exophthalmos.

It is of primary importance to evaluate the presence of all extraocular muscles in Crouzon's disease before beginning complicated neurosurgical and plastic reconstructions. In cases of muscle deficiency in

craniofacial dysostosis we suggest this surgical treatment, which will improve the position of the eyes without the harm of other and more complicated procedures.

References

- 1 Walsh FB, Hoyt WF. *Clinical Neuro-Ophthalmology*. 3rd ed. Baltimore: Williams and Wilkins, 1969: 1: 694–5, 700–4.
- 2 Stewart R. Craniofacial malformations: clinical and genetical considerations. *Pediatr Clin North Am* 1978; **25**: 485–515.
- 3 Gorlin RJ, Sedano H. Craniofacial dysostosis—Crouzon's syndrome. *Minn Med* 1972; **17**: 269–70.
- 4 Sanders B, Calcaterra TC, Beumer J. Orbital decompression and infraorbital augmentation for correction of exophthalmus secondary to Crouzon's disease. *J Oral Surg* 1978; **36**: 233–7.
- 5 Crouzon MS. Dysostosis craniofaciale héréditaire. *Presse Med* 1912; **73**: 737–9.
- 6 Newell FW. *Ophthalmology Principles and Concepts*. 4th ed. St Louis: Mosby, 1978: 248.
- 7 Hussain MK, Waspy IA. Crouzon's disease: A report of a typical case. *Bull Ophthalmol Soc Egypt* 1971; **64/68**: 253–63.
- 8 McCarthy JG, Coccaro PJ, Epstein F, Converse JM. Early skeletal release in the infant with craniofacial dysostosis. *Plast Reconstr Surg* 1978; **62**: 335–46.
- 9 McCarthy JB. New concepts in the surgical treatment of the craniofacial synostosis syndromes in the infant. *Clin Plast Surg* 1979; **6**: 201–26.
- 10 Whitaker LA, Schut L, Kerr LP. Early surgery for isolated craniofacial dysostosis (improvement and possible prevention of increasing deformity). *Plast Reconstr Surg* 1977; **60**: 575–81.
- 11 Diamond GR, Katowitz LA, Whitaker LA, Quinn GE, Schaffer DB. Variation in extraocular muscle number and structure in craniofacial dysostosis. *Am J Ophthalmol* 1980; **90**: 416–8.
- 12 Mathews D. Craniofacial surgery. Indications, assessment and complications. *Br J Plast Surg* 1979; **32**: 96–105.
- 13 Miller M, Falk E. Strabismus associated with craniofacial anomalies. *Am Orthopt J* 1975; **25**: 27–37.
- 14 Edgerton MT, Jane JA, Berry FA, Marshall KA. New surgical concepts resulting from cranio-orbital-facial surgery. *Ann Surg* 1975; **182**: 228–39.
- 15 Kushner J, Alexander Jr CH. Crouzon's disease (craniofacial dysostosis): modern diagnosis and treatment. *J Neurosurg* 1972; **37**: 434–41.
- 16 Poswillo D. Pathogenesis of craniofacial syndromes exhibiting colobomata. *Trans Ophthalmol Soc UK* 1976; **96**: 69–72.
- 17 Weinstock FJ, Hardesty HH. Absence of superior recti in craniofacial dysostosis. *Arch Ophthalmol* 1965; **74**: 152–3.
- 18 Murray JE, Swanson LT, Strand RD, Hricke GM. Evaluation of craniofacial surgery in the treatment of facial deformities. *Ann Surg* 1975; **182**: 240–65.
- 19 Monasterio F, Fuente del Campo A, Corrillo A. Advancement of the orbits and the midface in one piece combined with frontal repositioning for the correction of Crouzon's deformities. *Plast Reconstr Surg* 1978; **61**: 507–16.
- 20 Sanders B, Calcaterra T, Beumer J. Orbital decompression and infraorbital augmentation for correction of exophthalmos secondary to Crouzon's disease. *J Oral Surg* 1978; **36**: 233–7.
- 21 Derome P, Rougerie J, Tessier P. Craniofacial dysmorphism—Les dysmorphies cranio-faciales. *Gaz Med Fr* 1973; **80**: 5243–50.
- 22 Converse JM, Wood SD. Craniofacial surgery for ocular hypertelorism and craniofacial stenosis. *Trans Am Acad Ophthalmol Otolaryngol* 1973; **77**: 352–67.
- 23 Wexler MR, Peled I, Rouso M, et al. Surgical treatment of craniofacial dysostosis and hypertelorism. *Harefuah* 1980; **97**: 193–8.
- 24 Whitaker LA, Schut L, Randall P. Craniofacial surgery present and future. *Ann Surg* 1976; **184**: 558–64.