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2nd Department of Ophthalmology, Medical University of Lublin

ANNA MATYSIK, MAREK GERKOWICZ, JACEK CIECHAN

Ocular symptoms in Crouzon's syndrome

Crouzon's syndrome is a craniofacial dysostosis first described by the French neurologist Octave Crouzon in 1912 (1, 3). Its incidence is 1 per 25,000 of live births, its inheritance is autosomally dominant and only rarely recessive (5). Yet, 30–60% of all cases are believed to be caused by new mutations.The essential feature of this disease are developmental disorders of the craniofacial region combined with defects of the visual system which results from abnormal growth of mesenchymal and ectodermal tissue (2).

Normally, final ossification of sutures occurs at the age of 30–40 years while in Crouzon's disease in the last months of fetal life or the first four years of life. Because of craniosynostosis craniostenosis develops. The head shows features of acrobrachycephaly while its width is increased. The middle part of the face is hypoplastic and the nose is contorted, which is called a parrot-beak nose. There is maxillary hypoplasia and mandibular prognasthism. The described disorders are responsible for the so-called frog-like face (3).

Too small skull capacity in relation to the developing brain often causes intracranial hypertension resulting in secondary neurological disorders, such as e.g. defects of hearing and sense of smell. Anomalies also involve the system of vision and include: too big distance between the eyeballs caused by too wide forehead and base of the skull as well as ocular proptosis resulting from shallow orbits (1, 2, 3, 4, 5). The described changes of the skull lead to the positioning of the eyeballs in divergent strabismus and possibility of their spontaneous dislocation through the palpebral fissure to which maxillary hypoplasia also contributes (2, 3, 4). Defect of vision may result from congestive symptoms at the eye fundus as well as from oedema and atrophy of the optic nerves. Optic nerve disorders can also be fostered by deformed, narrow canals of the optic nerves. The other described disorders of the system of vision were: nystagmus, keratoconus, aniridia, blue scleras, glaucoma, and opic nerve atrophy (2, 3, 4). No anomalies involving the bones of extremities or soft tissues have been found in Crouzon's syndrome (2).

The aim of our study was to present ocular symptoms in a 4-year-old boy with Crouzon's syndrome. The child was referred to 2nd Department of Ophthalmology in the first year of life because of exophthalmos. The response of pupils to light and the anterior part of both eyes were normal. No congestive symptoms were found at the eye fundus. Optic nerve discs had distinct borders and were slightly paler. Head ultrasound did not show any focal changes in brain tissue. The ventricular system of the brain with rounded anterior horns of lateral ventricles. Vascular plexi of lateral ventricles had irregular contours. The depth of anterior horns of lateral ventricles was 9 mm. The third and fourth canals were normal. Subscleral spaces had dimensions of 5 mm each and were widened. In subsequent oculist examinations responses of pupils to light were normal and the appearance of the eye fundus did not change.

In March 2004 it was already possible to assess visual acuity. It was V.o.u.=1, exophthalmos was in the right eye 21 mm, in the left eye 20 mm with the distance between eyeballs 100 mm, the angle of divergent strabismus was 16 degrees. The anterior part of both eyes was normal (Fig. 1, 2). The optic disc of nerve II of the right eye had distinct borders, was slightly paler on the temporal side and a bit inclined in the superonasal direction. The disc was distinctly larger than the optic nerve disc of the left eye. In the left eye the disc had distinct borders and was slightly paler on the temporal side. At the fundus of both eyes no congestive symptoms were found (Fig. 3, 4).

Fig. 1–2. Face of the child with Crouzon's syndrome

Fig. 3. The fundus of the right eye

Fig. 4. The fundus of the left eye

Computerized tomography of the head showed craniosynostosis and symptoms of intracranial tightness visible as smoothness of gyruses of cerebral cortex surface and tightening of ventricles (Fig. 5, 6, 7). The child is in good general condition and develops normally. At present there is no need of surgical or pharmacological treatment. He is under permanent pediatric and ophthalmologic care.

In the case history given by the parents no information appeared about similar familial cases.

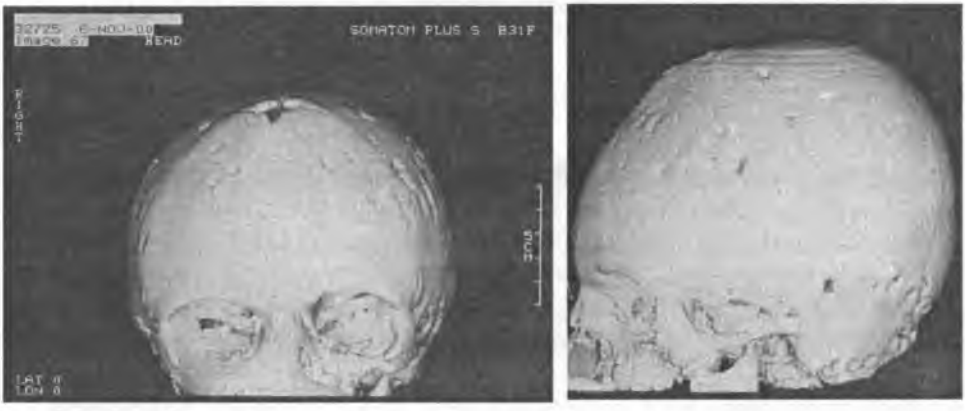

Fig. 5–6. Premature atresia of the fontanelle – CT picture

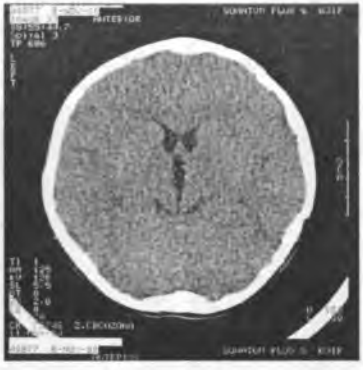

Fig. 7. Smoothness of the gyruses of cerebral cortex surface, tightening of ventricles (symptoms of intracranial tightness) – CT picture

CONCLUSIONS

1. Crouzon's syndrome should be considered one of pathologic conditions causing abnormal growth of the orbits and associated ocular symptoms.

2. Because of possible neurological and ophthalmologic complications the child should be permanently followed up.

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SUMMARY

The aim of the study was to present ocular symptoms in a 4-year-old boy with Crouzon's syndrome. The syndrome belongs to craniofacial dysostoses. Its essential characteristics is craniosynostosis and the formation of tightened skull. Too small skull capacity in relation to developing brain often causes intracranial hypertension resulting in secondary neurological disorders. The following anomalies were found in the described boy: excessive interocular distance, divergent strabismus and exophthalmos caused by shallowing of the orbits. No neurological disorders or optic nerve damage were found The child is under permanent neurological and ophthalmological follow--up.

Objawy oczne w zespole Crouzona

Celem pracy było przedstawienie objawów ocznych u 4-letniego chłopca z zespołem Crouzona. Zespół Crouzona należy do dyzostoz twarzoczaszki. Istotą tej jednostki chorobowej jest przedwczesne kostnienie szwów czaszkowych i powstanie czaszki ścieśnionej. Zbyt mała pojemności czaszki w stosunku do rozwijającego się mózgowia powoduje często wzmożone ciśnienie śródczaszkowe, którego skutkiem są wtórne zaburzenia neurologiczne. U opisywanego chłopca stwierdzono nadmiernie szeroki rozstaw gałek, zez rozbieżny i wytrzeszcz będący wynikiem spłycenia oczodołów. Nie wykryto zaburzeń neurologicznych ani uszkodzenia nerwów wzrokowych. Dziecko jest pod stałą kontrolą okulistyczną i pediatryczną.