CASE REPORT

Unilateral optic nerve aplasia associated with microphthalmos

Jednostrana aplazija nervusa optikusa udružena sa mikroftalmusom

Gordana Stanković-Babić*, Ana Oros†, Sonja Cekić*, Mlena Vujanović*, Rade R. Babić†

*Clinic for Eye Diseases, Clinical Center Niš, Niš, Serbia; †Faculty of Medicine, University of Niš, Niš, Serbia; ‡Clinic for Eye Diseases, Clinic Center of Vojvodina, Novi Sad, Serbia; §Faculty of Medicine, University of Novi Sad, Novi Sad, Serbia; || Center for Radiology, Clinic Center Niš, Niš, Serbia;

Abstract

Introduction. Optic nerve aplasia is a rare developmental anomaly characterised by the congenital absence of the optic nerve, central retinal vessels and retinal ganglion cells that is seen most often in a unilaterally malformed eye. Case report. We reported a girl with a very rare anomaly of the eye, unilateral aplasia of the optic nerve and microphthalmia. We carried out a complete ophthalmological examination, A- and B-scan ultrasonography, magnetic resonance imaging (MRI) of the orbit and brain, pediatrician, neurological examinations and karyotype determination. The examined child was a third child from the third regular pregnancy, born at term (39 GS, BM 3100 g). Family ocular history was negative. The right corneal diameter was 7.5 mm and left 10 mm. On dilated fundus examination, the right eye showed the absence of optic nerve and central retinal vessels. B-scan echography showed a small right globe (axial length 13.80 mm), normal size left globe (axial length 18.30 mm) and the absence of optic nerve on the right eye. Physical and neurological findings and karyotype was normal. MRI of the orbits and brain marked asymmetry of globe size and unilateral absence of the optic nerve. The patient is under the control of a competent ophthalmologist and prosthetic. Conclusion. Further aesthetic and functional development of a young person is the primary goal in tracking this rare congenital optic nerve anomalies in the malformed eye.

Key words: optic nerve; congenital abnormalities; microphthalmos; diagnostic techniques and procedures; therapeutics.

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Gordana Stanković-Babić*†, Ana Oros‡§, Sonja Cekić*, Milena Vujanović*, Rade R. Babić║

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Apstrakt


Ključne reči: n. opticus; anomalije; mikroftalmus; dijagnostičke tehnike i procedure; lečenje.
acteristics are the absent optic nerve, retinal ganglion cell layer of nerve fibers and blood vessels 10.

Aplasia of the optic nerve can be uni- or bilateral. Bilateral aplasia of the optic nerve can be communicated in an otherwise healthy person 1,2, or accompanied by severe congenital anomalies such as septooptic dysplasia, a fatal outcome in the newborn 4, hydrocephalus, along with other multiple congenital anomalies 5 with congenital hypopituitarism and posterior pituitary ectopia 5. Unilateral aplasia of the optic nerve is often present in the malformed eyes, with no change in the brain tissue 11. Malformations of the eye, other than microphthalmos, are possible even cataracts, malformations of the chamber angle, retinal dysplasia, coloboma of iris and ciliary body, iris hypoplasia, persistent hyperplastic primary vitreus 6, 15.

According to Weiter et al. 15 aplasia of the optic nerve is typically unilateral in otherwise healthy individuals. It shows no gender, neither racial predilection, with no recorded share of hereditary factors and occurs as a result of abnormal ventral invagination 15.

According to many previous statements aplasia of the optic nerve is a part of hypoplasia of optic spectrum. According to the analysis performed by Alqahtani 11, of 42 statements in the literature, 29 are real aplasia of the optic nerve, others are hypoplastic optic nerve and among 4 bilateral aplasia communications only one is true aplasia of the optic nerve 11.

Microphthalmia of the affected eye is often followed by the finding of aplasia of the optic nerve. Microphthalmia sen microphthalmos is also a rare developmental anomaly of the eyeball, which is characterized by the reduced volume of the eye. Microphthalmia may occur isolated or in one third of cases of various syndromes. The average length of the eye at birth (newborn futures) is 17 mm, or about 70% of the size of the adult eye, compared to 23.8 mm which is the average axial length of the adult eye in persons with emmetropia. The diameter of the cornea in the case of microphthalmia is less than 10 mm and the axial axis of the eye is less than 20 mm. Adnexa and eyelids are usually present. In unilateral anatomicies of the eye – orbital no asymmetry is more pronounced with growing up 16.

Case report

We presented a girl with a very rare anomaly of the eye – unilateral aplasia of the optic nerve and microphthalmia. The parents presented their daughter due to the smaller right eye. Ophthalmic examination included: measurement of corneal diameter, motility test, catar test, checking the intraocular pressure of the Goldman tonometer, slit lamp, indirectly ophthalmoscopies (Heine500, Germany) with a magnifying glass +20 D, determination of objective refraction of the left eye (Sol. Atropin 0.1%) to computerized refractometer (Speedy Righton – K), and the control visits and verification of subjective visual acuity for optotypes with pictures, at the distance of 3 m. Ultrasonography of both eyes was carried out in the Departament for Ehosonography, Clinic for Eye Diseases in Niš (Ultrasound A / B Scanner UD – 6000), Tomey, A-scan probe, inflexible, with a frequency of 10 MHz, the top dimension of 5.3 mm ø flat with local anesthia (Sol. Tetracaine) and in the Clinic for Eye Diseases in Novi Sad (AB Sonomed EZ Scan 5500 +, the probe 10 MHz, 30 frames / sec). Additional tests were conducted: endocranial magnetic resonance (MR) by standard protocol (magnet Avanto 1.5 T, Siemens) in the Center for Radiology, Clinical Center Niš, then pediatric, neurological observation and karyotype implemented in the appropriate organizational units in the Clinical Center Niš.

The examined child was a third child from the third regular pregnancy, born at term (39th gestational week, body mass 3100 g). Neonatal period was eventless. Older children were healthy, family ocular history was negative. Ophthalmologic findings: right eye did not fix, more often eso in position. The whole right eye was smaller. The right corneal diameter was 7.5 mm and the left 10 mm. The anterior segment was without any abnormality on both sides. Refraction of the left eye was -1.75 / -1.0 (10°). Intraocular pressure was 18 mmHg bilaterally. Indirectly, ophthalmoscopy showed no optical drive to the right, much of the coverage of the retina which choriocapillaris vascularization in the foramen could correspond to the place of departure of the optic nerve. On the left eye it was the optic nerve head round, clear boundaries, the level of other retinal arteriovenous ratio was normal, the area moved an inch-preserved appearance and reflexes. Ultrasonography, B-scan showed no upright optic taper, the bulb as a whole was smaller (Figure 1), and the left eye – the last segment of echographic neat. A-scan showed axial diameter of the right eye, 13.80 mm and 18.30 mm of the left eye.

After ophthalmological examination we performed additional examinations. Endocranial MR showed not only the eyeball smaller diameter, but also absence of the right optic nerve (Figure 2, a), and no anomalies in the central nervous system (Figure 2, b). Karyotype, pediatric (clinical examination and echosonographic examination of the abdomen) and neurological examination were normal.

We explained to the parents the nature of abnormalities of the right eye in their child. The girl was sent to the
appropriate prosthetic center for further treatment and monitoring.

On subsequent check-ups, in accordance with the objective refraction of the left eye, the girl was given an appropriate adjustment for the left eye -1.0 / -0.50 (10°) (right eye without correction, plan glass), with whom she recorded visual acuity near the maximum for the age (VOS = cc = 0.6–0.8, 3 m, frames).

In the appropriate time period, a temporary ocular prosthesis was changed for the new one. The patient is under control of the competent ophthalmologist and prosthetic (Figures 3, a and b).

Discussion

The eye starts to develop early in about week 4 (day 22) before joining the neural folds in the neural tube. The distal part of optic vesicles (day 32) invaginates and transforms into the optical cup from which later the retina is formed and proximal remains narrow and makes the optical stalk which results in the ocular nerve 17, 18. Embryological disturbance events and interactive process between the different embryonic structures and cell types are the base for the development of congenital anomal 19.

The pathogenesis of optic nerve aplasia remains unclear. Aplasia of the optic nerve was first described 140 years ago by von Graef's, but the cause of it is still unclear. The defective embryonic fissure forms, the failure of mesenchymal angle hyaloid system to infiltrate the embryonic fissure, or primary agenesis of retinal ganglion cells are referred as potential causes.

Animal models showed that aplasia of the optic nerve can occur spontaneously in several animal species, and experimentally can be induced by treating pregnant bunnies with hypoglycemic sulfonamides, creating a deficit of folic acid in pregnant muscle, exposing mice and fetal mice to X-rays, and treating bunnies at the time of fertilization with actinomycin D. Multiple ocular abnormalities are basically the result of the loss of cells: nerve, retinal pigment epithelial cells, corneal endothelium, retinal coloboma, iris hypoplasia, microphthalmia 13.

Aplasia of the optic nerve is characterized by afferent pupillary defect, reduced vision in the absence of light perception, a typical ophthalmologic picture, and fluorescein angiography demonstrates the absence of retinal blood vessels at the side of aplasia. Perimetry is not registered in cell aplasia, ERG and EOG are abnormal. Echography does not show optic taper, and bulb as a whole is smaller. Radiologi-
cal examination shows small optic foramen on the side of aplasia, the orbit is smaller, while documenting the absence of endocranial MRI optic nerve affected side. There are predilections in regard to sex and race of patients with aplasia of the optic nerve. In our case family history was negative as well as genetic examination. Pregnancy has been described as neat, though the possible influence of external factors such as episodes of viral infections in the first trimester, smoking, exposure to acetone could not be excluded. Microphthalmos, enopthalmus and ptosis are the most common associated anomalies with aplasia of the optic nerve, wherein the microphthalmos is found in 20 of 25 releases as a follow-up of abnormalities.

Microphthalmia is located in 3.2%–11.2% of blind children. Predilection with respect to gender or race can be simple or complex, unilateral or bilateral in over 50% of cases associated with systemic abnormalities. Anophthalmia and microphthalmia are estimated to 3–14 to 100.000. Epidemiological data indicate the following risk factors: maternal age (over 40 years of age), multiple pregnancies, children with low birth weight, low gestational age, prenatal infection with rubella, cytomegalovirus, fetal alcohol etc.

Ragge et al. recommend anophthalmia/microphthalmia with a complete ophthalmic examination and electrodiagnostic procedures – flash VEP in anophthalmia/microphthalmia, pattern VEP in detection of a dysfunction of the optic nerve and retinal ERG in case of dysfunction. There is a need to correlate ocular abnormalities with brain development and good radiological perception of such patients. Due to better contrast resolution and the absence of multiphase radiation, MRI with sophisticated radiological imaging is preferable and becoming an imperative in modern ophthalmology. Morphophysiological differences in orbital structures, and the difference in tissue density in anatomical parts of the intra- and extracranal segment, the possibility of perceiving the sellers region, optic chiasm and endocranial structures in the exclusion of the central causes of clinical symptoms and signs, provide an almost perfect MRI visualization of different incipient and advanced pathological states in eye diseases.

In case of microphthalmos, a coordinated team of ophthalmologists, pediatricians, radiologists is needed in neonatal period. A complete pediatric examination and a system to search abnormalities associated with early ophthalmic examination (in case of severe abnormalities of the eye, but during the first 2 weeks of age) are required. Ophthalmologic diagnosis and assessment of vision are crucial for planning further treatment and monitoring of patients. It is necessary for the parents to explain the nature of the anomaly, further observation and monitoring plan for the child, with mandatory genetics consultation.

In pediatrics, renal ultrasonography is required to exclude associated anomalies of the eye and kidney disease, as well as screening for intrauterine infections (rubella, varicella, toxoplasmosis gondii, herpes simplex virus, cytomegalovirus), and testing of other family members in relation to ocular pathology (anophthalmia or microphthalmia, anterior segment malformations, glaucoma, coloboma, retinal dystrophy and optic hypoplasia). Genetic tests are necessary for chromosomal analysis and testing of individual gene is required.

It is noted that the rapid increase in the first three years of life reach 78% of the size of the fourth year, and the size of adults of twenty. In the third month of life, a person reaches 40% of adult size. The rapid growth of persons recorded in the second year to 70% and from 5.5 years 90% of the size of an adult person. For these reasons, it is necessary to monitor and correct developmental anomalies of the eye and/or orbit in the appropriate prosthetic center. To correct the anomaly is to minimize deformity of the face and prevent any consequences of a sociomediically handicapped child, to increase opportunity of education, correct development, socialization and integration of a patient into active working population. Children with this rare anomaly of the optic nerve require to be treated by a team of ophthalmologists, prosthodontists, pediatricians, geneticists, radiologists at an early age.

Conclusion

Further aesthetic and functional development of young people is a primary goal in monitoring aplasia of the optic nerve, rare congenital anomalies in the malformed eye. It is realized through the coordinated team of ophthalmologists, prosthodontists, pediatricians, geneticists at the earliest stage. Pediatric review and search for associated systemic abnormalities, early ophthalmologic examination and assessment of vision are essential for further treatment planning. Parents should be aware of the circumstances, the need and plan for monitoring of their child.

References


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