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An isolated case of microphthalmia

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Objective

Microphthalmia presents a rare congenital (1/7.000 live births) developmental defect of the eye with a structurally disorganized small eyeball with the presence of the optic nerve and chiasma. Although, the precise pathogenesis of this entity remains unknown, various risk factors can contribute to microphthalmia. Most lesions are bilateral and appear to the same degree in both males and females. Anophthalmia, cryptophthalmos, synophthalmia, microhthalmia and cyclopia can be diagnosed prenatally through 2D, 3D ultrasonography at the anomaly scan and through fetal MRI. We report a case of unilateral microphthalmia diagnosed during infancy and moreover we highlight the proper management of such rare cases.

Methods

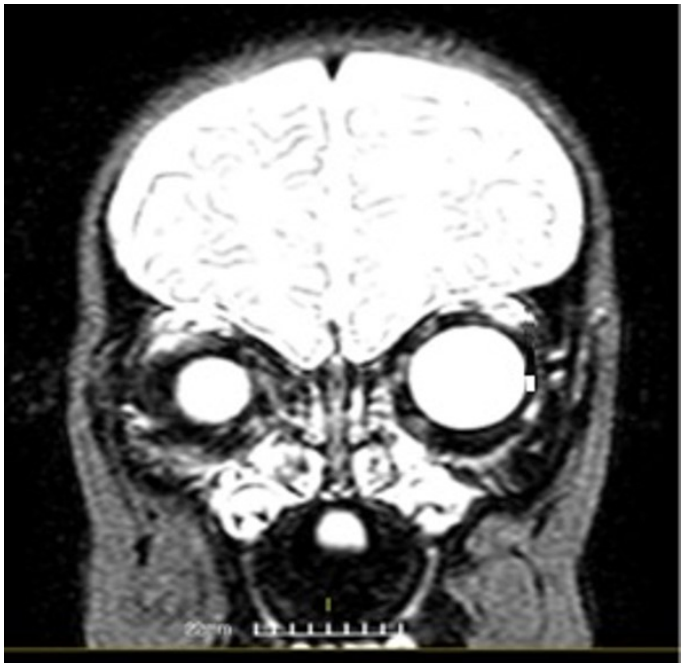
A male fetus was born at 38 weeks and 3 days of the gestational age, by C section (due to previous C section) with a birth weight of 3.150 grs. The pregnant woman has no pathological medical or family history. First trimester screening test: low risk for aneuploidies, anomaly scan: with no obvious structural anomalies.

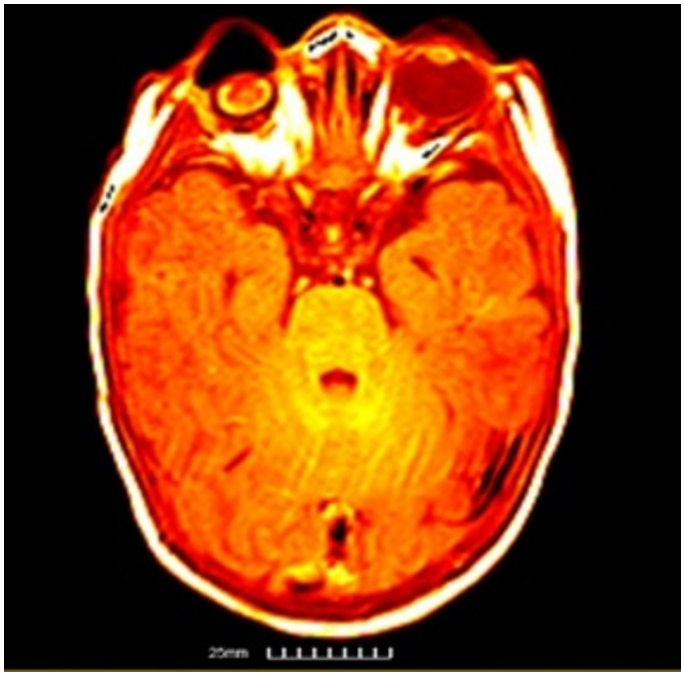
Results

Due to isolated right sided infant microphthalmia, the newborn was referred to the NICU for further management. TORCH screening test results were negative for infection. Infant karyotype, CNS ultrasonography and echocardiography did not indicate noticeable pathology. MRI reported right sided microphthalmia with poorly differentiated anterior segment and lens, and possibly minimal vitreous body lacking the expected signal enhancement in T2 sequence of brain MRI. Asymmetric mild enlargement of the right nasolacrimal duct. Symmetric appearance of both optic nerves. The neonate was scheduled for whole-exome sequencing (WES) examination.

Conclusion

Various environmental factors such as increased maternal age, TORCH infection, hyperthermia, exposure to alcohol, medications and x ray and hereditable factors such as mutations in SOX2 genes can be related to microphthalmia. Most of the cases are sporadic and isolated with a good prognosis. Fraser syndrome (autosomal recessive) presents a syndromic form of microphthalmos characterized by the coexistence of facial clefts, tracheal atresia, renal, heart and finger defects. Another form, Patau syndrome (trisomy 13) is characterized by microphthalmia, CNS, heart, musculoskeletal and cutaneous, genitalia and kidney defects. During pregnancy, a detailed anomaly scan, echocardiography, CNS ultrasonography, fetal karyotyping and MRI should be offered in order to exclude syndromic cases and co-existed pathology. As far as the genetic counseling is concerned, in isolated forms the risk of recurrence is very low compared to hereditary cases. Neonates should be evaluated by multidisciplinary teams including ophthalmologists in order to perform ocular prosthesis aiming to avoid deformities during growth and preserve normal development of bones. In syndromic cases, termination should be offered.

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