Case report

Anophthalmia and microphthalmia: A clinical case with a prenatal diagnosis

Fátima Silva a,*, Marta Osório b, Conceição Brito b, Ana Sousa b, Cristina Godinho b, Francisco Valente b, Anabela Gomes c

a Department of Obstetrician/Gynecology, Centro Hospitalar Vila Nova de Gaia/Espinho, Vila Nova de Gaia, Portugal
b Department of Obstetrician, Prenatal Diagnosis Unit, Centro Hospitalar Vila Nova de Gaia/Espinho, Vila Nova de Gaia, Portugal
c Neonatology, Centro Hospitalar Vila Nova de Gaia/Espinho, Vila Nova de Gaia, Portugal

ARTICLE INFO

Article history:
Received 24 May 2013
Accepted 22 July 2013
Available online 24 October 2013

Keywords:
Anophthalmia
Microphthalmia
Ultrasound

ABSTRACT

Introduction: Anophthalmia and microphthalmia are serious eye malformations that describe, respectively, the absence of an eye and the presence of a small eye within the orbit. These conditions are the most frequent congenital eye malformations in the newborn, and the precise etiology remains unknown, but they may occur in isolation or as part of a syndrome, with chromosomal, monogenic and environmental causes already identified. The pre- and post-natal diagnosis can be made using imaging techniques and genetic analysis. The authors present a clinical case diagnosed prenatally.

Case report: A 35 year-old primiparous woman, with normal first trimester examinations and karyotype 46,XX, in whom the ultrasound at 18/23 weeks revealed an asymmetry of ocular globes, with a small diameter of the left orbit. This study was complemented with foetal magnetic resonance that confirmed a severe microphthalmia in the left eye, with difficulty in observing the integrity of the optic nerve. The interruption of the pregnancy was discussed, but the parents did not want this. The remaining routine examinations were normal. The pregnancy was terminated at 37 weeks, palpebral closure with absence of ocular globe of the left eye and opacification of cornea and microphthalmia in the right eye was confirmed.

Conclusion: This case shows that a difficult diagnosis of a rare pathology can be made in utero by ultrasound. However, some are impossible to diagnosis, and this must be explained to parents.

© 2013 Asociación Española de Diagnóstico Prenatal. Published by Elsevier España, S.L. All rights reserved.

Anophtalmia y microftalmia: caso clínico con el diagnóstico prenatal

RESUMEN

Introducción: Anophtalmia y microftalmia son malformaciones graves en los ojos que describen, respectivamente, la ausencia de un ojo y de la presencia de un ojo pequeño dentro de la órbita. Estas condiciones son la malformación ocular congénita más frecuente en el recién nacido, y la etiología exacta se desconoce, pero pueden ocurrir en forma aislada...
Introduction

Anophthalmia and microphthalmia are eye malformations that are responsible for visual impairment, or reduced vision, in newborn and children. Anophthalmia refers to complete absence of the globe in the presence of ocular adnexa (eyelids, conjunctiva and lachrymal apparatus), and microphthalmia is defined as a globe with a total axial length that is at least two standard deviations below the mean for age. Microphthalmia can be classified according to the anatomic appearance and the severity of the reduction of the globe: severe microphthalmia refers to a globe with a corneal diameter less than 4 mm and a total axial length less than 10 mm at birth.1

These ocular malformations can be unilateral or bilateral, and can be isolated or occur with other malformations as part of a syndrome. The precise aetiology is heterogeneous and remains unknown, but there are environmental agents that may be involved, genetic factors, and probably vascular disruption. The known environmental causes include rubella, alcohol, thalidomide, retinoic acid and hydantoin. The heritable causes include the chromosome abnormalities (aneuploidy, deletions, rearrangements), and single gene disorders (syndromic or non syndromic).

The prenatal diagnosis, that is now available, permits the early diagnosis of these diseases and the awareness to other malformations or signs that may preview the prognosis, which must be carefully explained to parents. The sonography has a vital role in in utero diagnosis, complemented by magnetic resonance.2,3

Case report

We present a case of a primipar a woman, 35 years old, with no relevant medical history. This pregnancy was planned, followed in private Obstetrician up to the 20th week.

The first trimester exams were normal, including the 12th week ultrasound. Because of the age and maternal anxiety, the expectant performed amniocentesis at 15th week, for cytogenetic study, that revealed a normal cariotype: 46,XX.
The intervention.

Discussion/Conclusion

Anophthalmia and microphthalmia are heterogeneous diseases and it is extremely difficult to predict the morbidity in the future of these children.

This case is an example of a rare pathology which diagnosis can be made in prenatal period. This intra uterine diagnosis is not easy or frequent, however, the advances in technology of ultrasonography machines, like the use of 3D images, help and permit this anticipation.

In the case reported only the diagnosis of ocular malformation was made in uterus, because we verified that the newborn had anophthalmia in the left eye and microphthalmia in the right eye, not the left microphthalmia suspected by the prenatal ultrasounds and by the foetal magnetic resonance, which had supported our diagnostic hypothesis of isolated unilateral microphthalmia. Although, we consider that the prenatal finding of ocular malformation allowed us to alert the parents for this pathologies and prepare them for the treatments that can be offer in the neonatal and paediatric life.

The aetiology of these congenital ocular malformations has a very important role in the prognosis; however we know many causes that are not totally understood, mostly the genetics, which remains as the most likely cause for the appearance of this malformation in the presented case report.

The management of anophthalmia/microphthalmia is mostly supportive, but many surgical interventions have been proposed in the last years to obtain a good cosmetic outcome with perocular symmetry. Early intervention and therapy to optimise psychomotor development, educational endeavours, life skills and mobility are essential. The treatment may include evaluation by an oculoplastic surgeon in order to analyse prosthetic intervention and surgery.

Actually this child remains waiting for surgical intervention, in order to improve her quality of life.
Ethical disclosures

Protection of human and animal subjects. The authors declare that the procedures followed were in accordance with the regulations of the responsible Clinical Research Ethics Committee and in accordance with those of the World Medical Association and the Helsinki Declaration.

Confidentiality of Data. The authors declare that they have followed the protocols of their work centre on the publication of patient data and that all the patients included in the study have received sufficient information and have given their informed consent in writing to participate in that study.

Right to privacy and informed consent. The authors declare that no patient data appears in this article.

Conflict of interests

The authors declare that there are no conflicts of interest.

References