

# Microphthalmia and Retinoblastoma: the Importance of the Early Diagnosis in the Ocular Losses Prevention

## Microftalmia y Retinoblastoma: la Importancia del Diagnóstico Precoz en la Prevención de Pérdidas Oculares

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**ABSTRACT:** Microphthalmia is regarded as an exaggerated smallness or absence of the eyeball and Retinoblastoma as the most frequent children's eye malignant tumor, which may lead to the eyeball loss if not diagnosed early. The ocular losses are embarrassing to their carriers because they are related to the face, an area in which the sense organs, essential in the human relations, are. This paper aims at showing the importance of the prevention and the reports of microphthalmia and retinoblastoma-rehabilitated children's clinical cases by the use of ocular prostheses.

**KEY WORDS:** microphthalmia, retinoblastoma, oncology, diagnosis, ocular.

### INTRODUCTION

When a baby is born, its parents' largest desire is that it is healthy and physically perfect, but this is not what happens in all the cases. Many times we can see anomalies that could have been avoided if there were an ideal medical follow-up during gestation. Defects, either of congenital origin, trauma or malignancy may cause trouble to their carriers and their family, mainly when the alteration is located in a visible area such as the face (Goiato *et al.*, 2005).

The exaggerated smallness of the eyeball (microphthalmia) can be mentioned as a congenital defect (Wilk-Wilczynska *et al.*, 1966). Among other factors it is caused by intra-uterine infections and rubella, considered the most common etiology (Murray *et al.*, 2002).

The rubella virus is transmitted through breath droplets (Murray *et al.*) and it initially infects the superior airways, is disseminated to the local lymph nodes and, from this point on, the viremia is settled and there is a propagation of the virus to the other tissues of the

organism, including the placenta (when the woman is pregnant) and then to the fetus. The nature of the disturbances varies according to the affected tissue and to the development stage that has been broken (Chantada *et al.*, 2007). The virus may persist in some tissues such as, for instance, the crystalline lens, from three to four years after the baby's birth, but it can also be eliminated one year after birth. Early abortive lysis by phage BF23 in *Escherichia coli* K-12 carrying the colicin Ib factor (Nisioka & Ozeki, 1968).

Microphthalmia is associated with cataract, abnormalities of the optical nerve and glaucoma (Joseph *et al.*, 2006). It was believed that natural infections as well as vaccination would take to a protecting immunity for a lifetime, but recent studies show cases of rubella manifestation in individuals that had already been exposed to it during childhood or that had already received the serum antibody (Sonnenberg *et al.*, 2007). Nowadays, the most effective and commonly used way is vaccination, but this not a 100% safe method.

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Retinoblastoma is the most common and potentially deadly children's malignant eye tumor (Joseph *et al.*). In about 75% of the cases, it affects children which are less than three years old, being also possible to happen on birth (Goddard *et al.*, 1999). It may have hereditary origin or happen sporadically. When it has hereditary origin, it attacks both eyes; in cases of sporadic occurrence, it affects one eye only (Goddard *et al.*). Retinoblastoma diagnosis is very easy to be obtained: by using a picture flash or through the exam of eye core (Ozdemir *et al.*, 2007).

The beginning signs used for the establishment of a differential diagnosis include leucocoria, which is the whitish reflex presented by the eye when the light enters it. When this white stain is visible, more than 50% of the retina is altered. Other signs are: squint, deficient vision, red and aching eyes (lacks reference) (Hafsa *et al.*, 2006; Meier *et al.*, 2006).

This paper aims at: alerting health professionals about the importance of preventing diseases during gestation; alerting health professionals about the importance of an early diagnosis in cases of retinoblastoma; showing the need and the importance of the non-eyeball patients' rehabilitation through ocular prostheses.

## CLINICAL CASES

**Clinical case I.** A six-month old girl presented exaggerated reduction of the left eyeball (Fig. 1). During the exam (anamnesis) it was found out that the mother had not received the vaccine against rubella before gestation and was a carrier of the pathology during pregnancy, which caused the child to be a microphthalmia carrier.

It should be pointed out that a simple action as the mother's vaccination one year or more before gestation could have avoided her daughter's disease. Despite the fact that it is not a 100 percent-safe method, the rate of the disease in individuals that received the serum antibody is small (Hatakeyama *et al.*, 2004). As the prevention was not done by the baby's mother, the only possible and necessary treatment was the making of an ocular prosthesis. This prosthesis has an anatomical, physiologic (occupying the orbit with the deficient eye, the prosthesis provides a symmetrical facial growth) and aesthetics function (Fig. 2).

**Clinical case II.** In this second case, a girl carrying bilateral ocular absence is shown (Figure 3). During anamnesis, it was found that when the patient was born, her vision was perfect, in other words, within normal patterns, but she lost both eyeballs due to a malignant tumor (retinoblastoma), diagnosed late, when the conservative treatment was no longer possible.

If there were a little more careful observation by the parents or pediatrician that assisted the child, those losses would not probably have happened. The diagnosis of retinoblastoma can be obtained easily, through the presence of a "strange" light or white reflex (leucocoria) in the child's eye, which can be obtained by the help of a picture<sup>6</sup>. The interesting point is that this pathology is discovered before the white reflex can be noticed because when that happens, over 50% of the retina is already altered, but a conservative treatment is still possible (Ozdemir *et al.*; Hafsa *et al.*).

The early diagnosis is important, not only because of the maintenance of the vision and the eyeball, but also because in case the pathology to be treated is a tumor, the more quickly it is discovered, the greater the chances of and the smaller the collateral effects caused by the treatment (radiotherapy and chemotherapy) (Sahagal *et al.*, 2006).

As in the case presented the patient didn't have the opportunity to receive the conservative treatment, going through a bilateral nucleation, the possible treatment was the making of bilateral ocular prostheses, that improved aesthetics and promoted asymmetrical facial growth, since the prosthesis occupied the empty spaces (Fig. 4).

## FINAL CONSIDERATIONS

We hope that, with this information, health professionals are more aware of the presented situations and may know how to diagnose and to instruct their patients in a better way, showing them the importance of a prenatal exam and vaccination, as well as being able to show them how simple it is to have an early diagnosis of retinoblastoma using a picture.

The early diagnosis of retinoblastoma is important, not only for the keeping of vision and eyeball, but also, as it is a malignant tumor, for its diagnosis and treatment, in order to have smaller collateral effects.



Fig. 1. Child without ocular prosthesis (microphthalmia).  
Fig. 2. Child with ocular prosthesis.



Fig. 3. Child without binocular prosthesis (retinoblastoma).  
Fig. 4. Child with binocular prosthesis.

When it is not possible to have a conservative treatment, the making of ocular prostheses is very important in order to occupy the orbit, stimulating facial growth. This act may bring aesthetic, anatomical (favoring

facial symmetry) and physiologic benefits, not only in reinserting the children in the social environment, but also in giving them back self-esteem, because they are, in most cases, excluded by society for being handicapped.

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**RESUMEN:** La microftalmia se considera como una exagerada pequeñez o ausencia del globo ocular y el retinoblastoma como el tumor maligno más frecuente del ojo en niños, los cuales pueden conducir a la pérdida del globo ocular si no se diagnostica precozmente. Las pérdidas oculares son desconcertante para sus portadores, ya que están relacionadas con la cara, un ámbito en el que los órganos sensoriales son esenciales en las relaciones humanas. Este trabajo tiene por objeto demostrar la importancia de la prevención y los informes de casos clínicos de niños con microftalmia y retinoblastoma rehabilitados por el uso de prótesis oculares.

**PALABRAS CLAVE:** microftalmia, retinoblastoma, oncología, diagnóstico, ocular.

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