The eyes hide surprises: unilateral microphthalmia with severe bilateral colobomatous involvement

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Obstetricia y Ginecología, Complejo Hospitalario Universitario Insular Materno Infantil de Canarias, Las Palmas de Gran Canaria, Las Palmas, Spain.

Case report

A 28-year-old mother with personal history brachydactyly. Her husband had mild microphthalmia, operated for strabismus; the mother, grandmother’s husband presented microphthalmia without blindness.

Ultrasound

- **20 + 5 weeks.** Bilateral microftalmia (Figure 1).
- **23 weeks.** Echogenic structure behind the left sense. Right eye normal.
- **28 weeks.** Normal neurosonography.
- **33 weeks.** Retinal non-attachment. Normal optic chiasma, optic nerve <p5 mesure in the left side.

**Follow up:** bilateral microphthalmia, more evident in the left side, with normalization of the right globe mesuremente (Figure 2).
- **41 weeks.** Delivery.

Ophthalmologic examination: the right eye microphthalmia with microcornea with corioretinal coloboma, complete chorioretinal colobomatous atrophy. On the left eye: a persistence of fetal vitreous with complete chorioretinal atrophy and persistence of hyaloid artery (Figure 3).

RMN: left microphthalmia with primary vitreus persistent (Figure 4).

Genetic study

- Mother translocation: 46, XX, t (13; 18)(q32; q23).
- Patient: Duplication-deletion Syndrome involving chromosomes 13 and 18.

The patient is currently 1 year and two months and presents blindess, psychomotor delay, deafness.

**Conclusion**

Normal ultrasound is not a guarantee that vision will be intact in an eye that appears normal. Some abnormal echographic findings such microphthalmia it can make us suspect that a severe ocular pathology could hide in the eye.

Figure 1

Figure 2

Figure 3

Figure 4