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(CASE REPORT)

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Congenital Megalocornea, Microspherophakia, and Lens Subluxation Leading to Secondary Glaucoma in an Infant: A Case of Suspected LTBP2 Mutation

Ghizlane Daghouj, Sara Ennaki *, Asmaa ElKettani, Zyad Laftimi, Loubna El Maaloum and Bouchra Allali

Pediatric ophthalmology department; 20 August Hospital-Hassan II University, Casablanca.

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Abstract

We report the case of a 6-month-old infant with congenital megalocornea, microspherophakia, and bilateral lens subluxation, leading to secondary glaucoma. Genetic context included consanguinity, suggesting an LTBP2-related condition. The patient underwent bilateral lensectomy with anterior vitrectomy, combined with trabeculectomy and peripheral iridectomy in the right eye. Postoperative intraocular pressure normalized in both eyes. This rare entity should be distinguished from primary congenital glaucoma, as the surgical approach differs. Regular intraocular pressure monitoring is essential in children with megalocornea and lens anomalies to ensure early detection and treatment of secondary glaucoma.

Keywords: Microspherophakia; Megalocornea; Ectopia Lentis; Secondary Glaucoma; LTBP2 Mutation; Case Report

1. Introduction

This is a rare eye disorder caused by recessive mutations in the LTBP2 gene. It is characterized by congenital megalocornea, often associated with spherophakia and/or lens ectopia. These lens anomalies can lead to pupillary block and secondary glaucoma[1].

It is important to distinguish this condition from buphthalmos due to primary congenital or infantile glaucoma. The treatment differs: lens removal is indicated in this condition, while angle surgery is the mainstay in primary congenital glaucoma[2].

Other features may include deep anterior chambers, axial myopia, iridodonesis, plateau iris, miotic and oval pupils, and signs of ocular irritation such as conjunctival injection and corneal edema[3].

2. Case description

We report the case of a 6-month-old female infant referred to our ophthalmology department by her pediatrician for leukocoria, with a suspected diagnosis of congenital cataract. The child had no significant medical history. The parents were first cousins, but there were no dysmorphic features or abnormal physical findings.

On ophthalmologic examination, pursuit, threat, and dazzle reflexes were present. Cycloplegic refraction under 0.3% atropine showed -4.00 (-5.00 to 175°) in the right eye (OD), with inconsistent and unreliable measurements in the left eye (OS).

* Corresponding author: Sara Ennaki

Anterior segment examination of both eyes revealed bilateral megalocornea (14mm OD and 13,5mm OS), microspherophakia, and ectopia lentis. A posterior polar cataract and phacodonesis were also noted (Figure 1). Intraocular pressure (IOP), measured under sedation using a Tono-Pen, was 19 mmHg in the right eye and 20 mmHg in the left eye.

Gonioscopy revealed broad peripheral anterior synechiae involving more than 180° of the angles in both eyes, with iris processes visible over the trabecular meshwork (Figure 2-3).

Fundus examination was normal in both eyes. Ocular ultrasonography revealed no additional abnormalities, with axial lengths of 18 mm in the right eye and 17.9 mm in the left eye.

Surgical intervention was performed in both eyes. The right eye (OD) underwent combined cataract and glaucoma surgery, including lensectomy, anterior vitrectomy, trabeculectomy, and peripheral iridectomy. The left eye (OS) was managed with cataract surgery alone, consisting of lensectomy and anterior vitrectomy. Postoperative follow-up showed favorable outcomes with normalization of intraocular pressure (<6 mmHg) and stabilization of corneal diameter in both eyes.

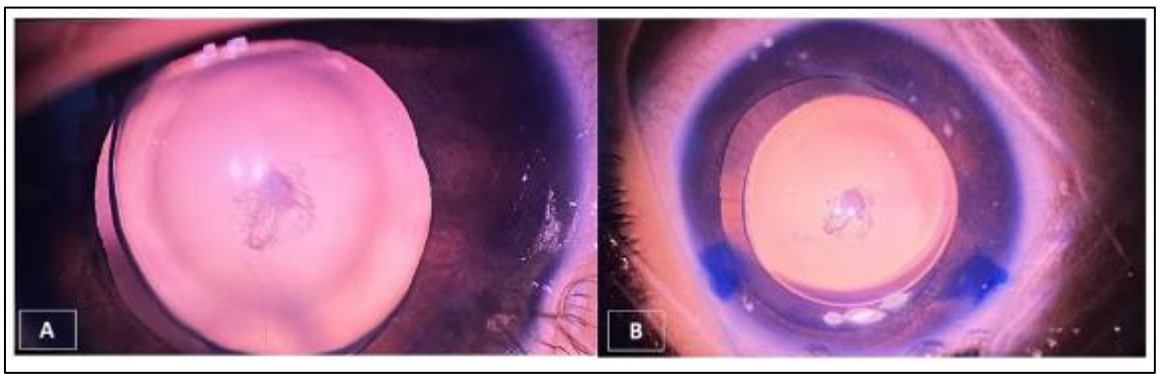

Figure 1 Anterior segment image of both eyes OD(A) and OS(B) showing microspherophakia and cataract. Zonular fibers are clearly visible around the equator of the crystalline lens

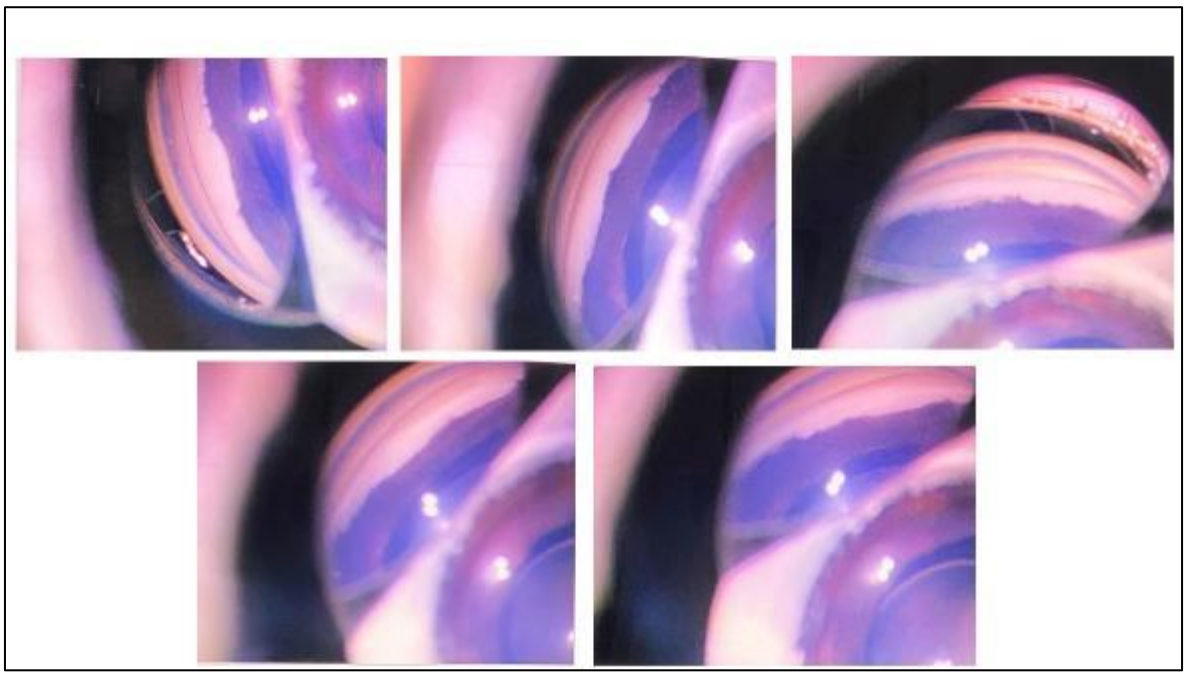

Figure 2 Gonioscopy OD showing goniosynechiae involving more than 180 degrees of the angle, with iris spicules

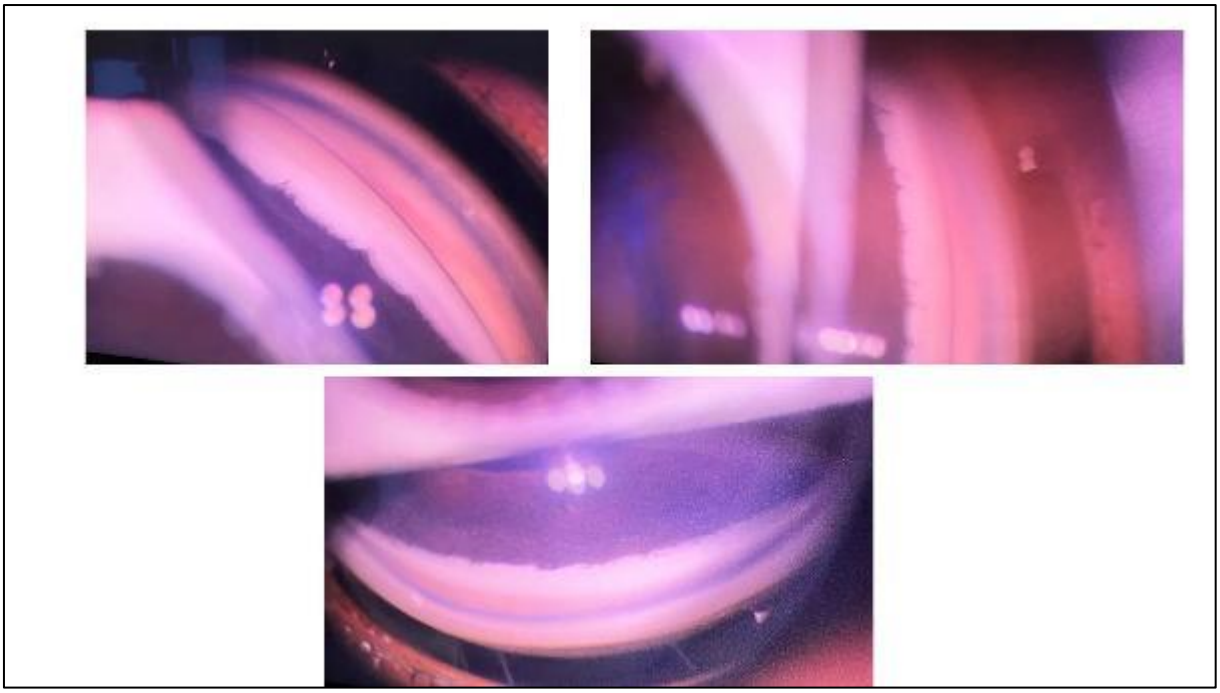

Figure 3 Gonioscopy OS showing goniosynechiae involving more than 180 degrees of the angle, with iris spicules

3. Discussion

Congenital megalocornea with childhood-onset secondary glaucoma due to spherophakia and/or ectopia lentis is a rare, non-syndromic condition caused by autosomal recessive mutations in the LTBP2 gene[4]. This gene encodes a member of the latent transforming growth factor-beta-binding protein family, which is expressed in the anterior segment of the eye, particularly in the ciliary body[1,3].

This clinical entity must be distinguished from buphthalmos associated with primary congenital or infantile glaucoma, as the initial surgical management differs significantly. In the case of LTBP2-related ocular disease, lens removal is the cornerstone of treatment, whereas angle surgery (such as trabeculotomy or goniotomy) is typically indicated in primary congenital glaucoma[2].

Recurrent episodes of pupillary block are common in these patients and can lead to the early onset of secondary glaucoma. Complete anterior dislocation of the crystalline lens may also occur during early childhood[2]. These clinical features may lead to misdiagnosis as primary congenital glaucoma, especially in infants presenting with corneal enlargement, elevated intraocular pressure, or cloudy corneas.

Monitoring intraocular pressure is crucial in any child presenting with megalocornea, microspherophakia, and/or ectopia lentis, even in the absence of elevated IOP at first presentation. Early recognition of the underlying lens pathology allows timely intervention and may prevent irreversible optic nerve damage.

In a study by Desir et al., three children from a consanguineous Moroccan family and one child from a consanguineous Macedonian family were described with congenital megalocornea and secondary glaucoma associated with spherophakia and/or ectopia lentis due to LTBP2 mutations[5]. Interestingly, the oldest child in the Moroccan family had a tall stature and narrow face suggestive of Marfan syndrome, although no other criteria were met and FBN1 gene testing was negative[5].

Additional case series have emphasized the need for careful differentiation between LTBP2-related glaucoma and other developmental glaucomas, particularly in settings of consanguinity and early-onset symptoms[2,6].

Intraocular pressures should be followed-up in young children with an ocular phenotype consisting of megalocornea, spherophakia and/or lens dislocation[5].

4. Conclusion

LTBP2 mutations are a recurrent cause of primary congenital megalocornea with zonular weakness, often leading to spherophakia, lens dislocation, and secondary glaucoma in childhood.

This condition must be clearly distinguished from buphthalmos due to primary congenital or infantile glaucoma, as the surgical approach differs: lens extraction is the preferred initial treatment in LTBP2-related cases, while angle surgery is indicated for primary congenital glaucoma.

We emphasize the importance of regular intraocular pressure monitoring in children presenting with the ocular phenotype of megalocornea, spherophakia, and/or lens subluxation, in order to detect and manage secondary glaucoma early.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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