Megalocornea

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**Definition**

Megalocornea: A term used to describe those eyes that have an enlarged cornea, normal IOP, normal optic disk, and no family history of congenital glaucoma; it often occurs in association with other ocular or systemic abnormalities.

**Etiology**

Megalocornea is a developmental anomaly of unknown etiology. Several mechanisms of development have been postulated including a defect in the growth of the optic cup which leaves a larger space for the development of the cornea.

**Clinical Presentation**

Megalocornea presents as a bilateral enlargement of the cornea, with a horizontal diameter of 12 mm or greater at birth and 13 mm or greater after 2 years of age. There are a few variants of this disorder: autosomal dominant being the least common (and is typically not associated with other ocular abnormalities) and X-linked recessive which is more common (thus, accounting for the increased prevalence in males). The X-linked variant is commonly associated with other ocular findings including iris transillumination defects secondary to pigment dispersion, lens subluxation, arcus, and central crocodile shagreen. Other associations include mental retardation and congenital miosis. The enlargement does not arise from corneal stretching but rather from an overgrowth; and, as such, the endothelial cell count is normal. Corneal clarity and thickness are usually normal as well. This is in stark contrast to the enlarged corneal diameter that can be observed with buphthalmos (which presents as elevated IOP and an enlarged globe). Other findings which differentiate megalocornea from buphthalmos include absence of tears in Descemet’s membrane and lack of optic disk cupping.

In addition to associated ocular anomalies, various systemic abnormalities have been observed including multiple malformation syndromes, connective tissues diseases, dermatologic abnormalities, and genetic diseases. As such, it is prudent to perform a thorough pediatric exam and to consult physicians from other disciplines based on the associated findings.

**Diagnosis**

Various diagnostic modalities can be utilized to differentiate megalocornea from buphthalmos and anterior megalophthalmos:
1. Gonioscopy: lack of a widened ciliary band and lack of excessive mesenchymal tissue in the angle in the presence of an enlarged anterior segment/cornea helps distinguish megalocornea from the other potential diagnoses such as anterior megalophthalmos.

2. A-scan ultrasound biometry: allows for measurements of the axial length of the eye. Increased axial length can be seen in buphthalmos, whereas a normal to short vitreous length can be seen in megalocornea.

3. Specular microscopy: allows for measurement of the corneal endothelial cell count. Normal counts indicate megalocornea, whereas a decrease in the count is consistent with congenital glaucoma.

Pertinent lab testing entails screening for certain genetic markers. The genetic locus for X-linked megalocornea appears to be in the region Xq21–q22.6.

**Differential Diagnosis**

- Buphthalmos
- Anterior Megalophthalmos

**Prophylaxis**

No effective prophylaxis.

**Therapy**

The condition does not usually require treatment except for management of associated complications including high myopic refractive error, juvenile cataract, and glaucoma. Management of the refractive error is usually via spectacle correction. As for cataracts, this can be challenging given the underlying anterior segment dysgenesis. Various case studies have reported on the success of cataract extraction in patients who have megalocornea; however, care must be taken when considering surgical technique as well as intraocular lens size/type. Another frequently associated complication is secondary glaucoma. Per case reports, elevated IOP usually results from associated spherophakia or ectopic lens. Thus, surgical treatment for glaucoma secondary to megalocornea is lens removal; this differs from the conventional treatment for primary congenital glaucoma and buphthalmos which requires angle surgery.

**Prognosis**

Overall prognosis is good; the cornea itself does not continue to enlarge after 2 years of age. There can be associated ocular complications (see sections “Clinical Presentation” and “Therapy”) which are treated accordingly. In the presence of concomitant systemic abnormalities, a more thorough and long-term follow-up needs to be established including scheduled follow-ups with physicians from other specialities.

**Epidemiology**

Males account for ~90 % of cases because of the most common pattern of inheritance (X-linked recessive). According to the literature, 50 % of cases are X-linked recessive and 40 % are autosomal, whereas the remaining 10 % are sporadic. However, the condition itself is rare and the exact incidence is unknown (Fig. 1).
**Cross-References**

- Buphthalmos
- Megalocornea
- Megalophthalmos

**References**


**Megalocornea, Fig. 1** (a) Corneal diameters of 14 mm (b) Gross specimen showing appearance of enlarged cornea and heavily pigmented trabecular meshwork (Sugar, Joel, Wadia, Hormuz P., Vasaiwala, Roshni A. – Ophthalmology, 173–176.e1 © 2014 Copyright © 2014, Elsevier Inc. All rights reserved)