Case Report

Microspherophakia and Lens Subluxation in Klinefelter Syndrome: A Case Report

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Abstract

Purpose: To report a rare case of microspherophakia and lens subluxation in a young patient with Klinefelter syndrome who underwent cataract surgery, emphasizing the importance of a thorough general health assessment and anamnesis prior to cataract surgery.

Methods: The case concerns a 52-year-old male referred for phacoemulsification under general anesthesia. A review of literature was performed.

Results: Preoperative assessment revealed a corticonuclear cataract in both eyes, with dislocation of the crystalline lens in the left eye in an area of zonular dehiscence. Upon careful examination, both eyes showed a microspherophakic lens with an increased lens thickness and the lens equator being visible over 360° in the left eye. The patient denied any trauma or medical conditions. His medical health record revealed the coexistence of Klinefelter syndrome (47, XXY). The association between Klinefelter syndrome and microspherophakia has only once been reported in the literature. Intraoperatively, a more cautious approach was withheld and a capsular tension ring was used. Postoperative outcome was successful with good visual outcome and no interoperative complications.

Conclusions: This case report highlights the importance of proper preoperative assessment before cataract surgery, especially in unusual cases such as early-onset cataract and/or lens subluxation. In addition, it stresses the importance of a systemic and/or genetic evaluation in patients with microspherophakia and an ophthalmological examination in patients with Klinefelter syndrome.

Keywords: Cataract, Klinefelter syndrome, Lens subluxation, Microspherophakia, Phacoemulsification, Zonular dehiscence

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Introduction

Lens subluxation is associated with multiple ocular and systemic disorders, such as blunt trauma, aniridia, Marfan syndrome, and pseudoexfoliation syndrome. It can cause complications such as pupillary block glaucoma and refractive shifts and can have systemic consequences.[1] To avoid unforeseen perioperative complications (due to underlying conditions) and to provide an appropriate patient management, the preoperative assessment should comprise both a proper medical and ophthalmological anamnesis to define the etiology, as well as a thorough ophthalmological examination.

We present a rare case of lens subluxation with underlying microspherophakia in a patient with Klinefelter syndrome who underwent cataract surgery.

Case Report

A 52-year-old male patient was referred to University Hospitals Leuven for phacoemulsification under general anesthesia because of phobia of needles and medical interventions. He had experienced a gradual visual decline over the past 2 years. The patient did not mention any relevant personal ocular or medical history.

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Upon presentation, the best-corrected distance visual acuity was at Snellen 0.7 in the right eye (sphere −5.25 Diopter [D], cylinder −1.75 D at 8°) and at Snellen 0.3 in the left eye (sphere −7.00 D, cylinder −2.25 D at 171°). Slit-lamp examination revealed a corticonuclear cataract in both eyes. The left lens appeared dislocated in the inferonasal quadrant. Upon close examination, the lenticular equator could be observed over 360° in the left, but not in the right eye [Figure 1]. Both lenses showed a thickened anteroposterior diameter and convex anterior surface. Further ophthalmological examination was unremarkable.

Ocular biometry, using the Zeiss IOLMaster 700, confirmed the increased lenticular thickness (right lens 5.78 mm; left lens 5.77 mm), the decreased anterior chamber depth (right eye 2.47 mm; left eye 2.48 mm), and steep anterior lenticular curvature [Figure 2a and b]. The IOLMaster central corneal topography showed a with-the-rule astigmatism. Further examination of the patient's health record revealed a personal medical history of Klinefelter syndrome (47, XXY).

The patient underwent uneventful phacoemulsification cataract surgery in both eyes under general anesthesia. In both eyes, a very mobile lens complex was noted intraoperatively, with good stability and centration of the capsular bag after implantation of a Bausch and Lomb ACPi-11 capsular tension ring. Because of the zonular instability, a nontoric monofocal implant lens was implanted (right eye: Hoya Vivinex +24.0 D, target refraction −2.5 D; left eye: Hoya Vivinex +22.0 D, target refraction −2.5 D).

The postoperative recovery at 6 weeks was unremarkable with an excellent visual acuity (right eye: distance Snellen 1.0 with sphere −1.5 D, cylinder −1.5 D at 180°, near Snellen 1.0 plano; left eye: distance Snellen 1.0 with sphere −1.25 D, cylinder −1.75 D at 165°, near Snellen 1.0 plano).

Written informed consent for publication was approved by the local ethical committee and signed by the patient.

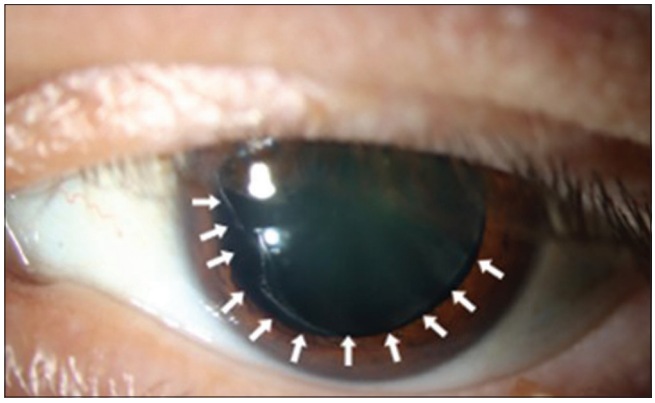

Figure 1: Slit-lamp examination shows a lens subluxation of the microspherophakic lens of the left eye

Discussion

Klinefelter syndrome is a sex chromosome aneuploidy that is characterized by an X chromosome polysomy in men. It is most often the result of one extra X, leading to a 47, XXY karyotype instead of the classic 46, XY in most males.[2,3] It is the most common chromosome disorder in men (1:500–1:1000) and is caused by meiotic nondisjunctions in parental germ cells.[4] First described in 1940, the syndrome has since been associated with multiple comorbidities in various organ systems. The clinical phenotype is very variable due to skewed X chromosome inactivation and varying degrees of hypogonadism. The cardinal symptoms and signs of Klinefelter syndrome are a tall stature, small testes, gynecomastia, infertility, and psychosocial disorders.[2-4] In our patient, several of these features were present, including small testes, infertility, and depression. Various ophthalmological abnormalities have been reported in clinical reports on Klinefelter syndrome, including uveal coloboma, microphthalmia, cataract, diffuse choroidal atrophy, and juvenile glaucoma.[3-7] Since no other apparent cause of early-onset cataract and lens subluxation could be withheld in our case, we performed a literature search in PubMed and Embase to evaluate the possible association of microspherophakia and lens dislocation/lens subluxation in Klinefelter syndrome. To our knowledge, we only found one reported case from 1962 of microspherophakia associated with Klinefelter syndrome.[8] However, it is difficult to suggest a correlation between these ocular manifestations and Klinefelter syndrome since they can also be present in nonsyndromic patients and the link with the presence of one or more additional X chromosomes is not known. Brand *et al*. performed the first systematic and comprehensive evaluation of eye health in 21 patients with Klinefelter syndrome.[4] They found that the presence of a supernumerary X chromosome in Klinefelter patients is associated with a number of morphological and functional ophthalmological features which should raise awareness towards a possibly higher risk of those patients to suffer from ocular disease. However, there is a further need for epidemiological studies on eye health in patients with Klinefelter syndrome.

Microspherophakia is a rare, usually bilateral, developmental disorder of the crystalline lens. It is the result of an embryological failure in which an arrest in development of secondary lens fibers leads to zonular insufficiency. Consequently, there is a lack of tension in the equatorial plane of the lens during growth. This results in a small, spherical lens with a reduced equatorial and increased anteroposterior diameter. It can present either isolated, familial, or as part of a systemic syndrome (e.g., Marfan syndrome, Weill–Marchesani syndrome, Alport syndrome, Lowe syndrome, homocystinuria and mandibulofacial dysostosis).[9,10] Microspherophakia can produce high lenticular myopia and/or defective accommodation. It can also, as in our case, result in lens subluxation which in turn can cause secondary glaucoma through lens block.

Lens extraction followed by in-the-bag intraocular lens (IOL) placement may be indicated to relieve crowding of the angle

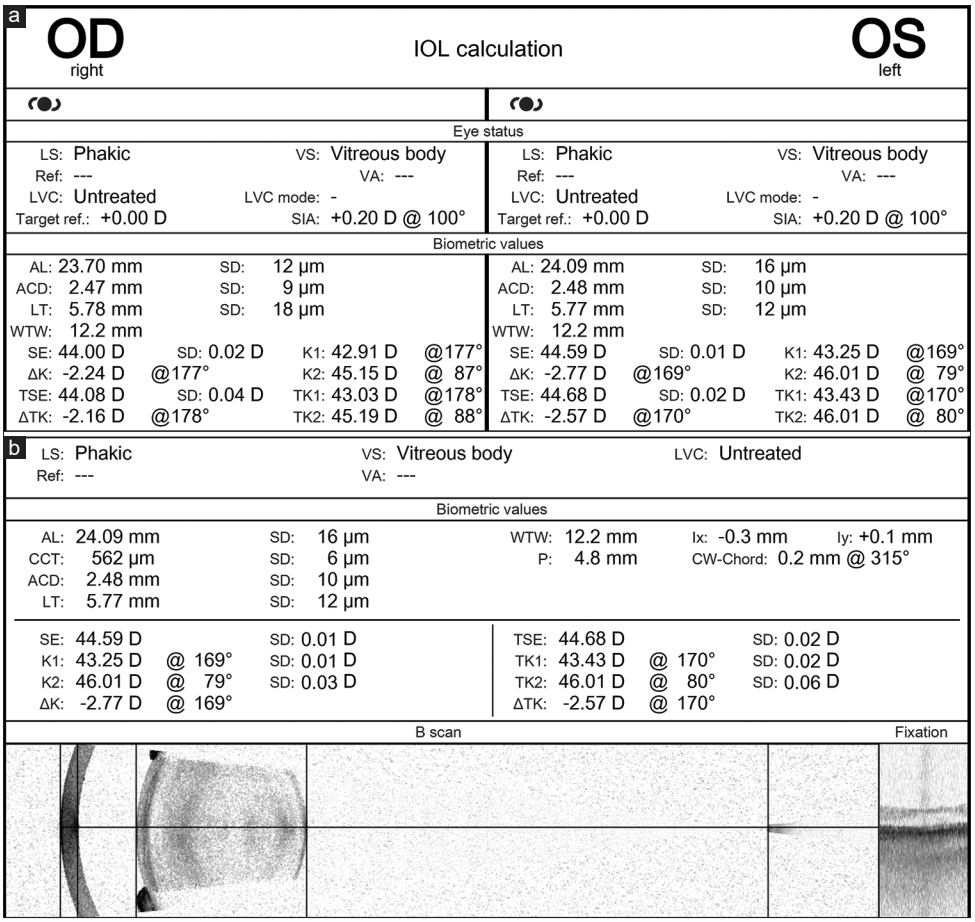

Figure 2: a) Ocular biometry (IOLMaster 700) showing an increased lenticular thickness and decreased anterior chamber depth of both eyes. b) Ocular biometry and B-scan of the left eye showing a steep anterior lenticular curvature

by the spherophakic lens or because of subluxation, cataract, or lenticulocorneal touch.[9,10] However, microspherophakia can present some surgical difficulties/challenges because of the small capsular bag and zonular instability. Therefore, capsular tension rings (with or without capsular tension segment) are often used during surgery in conjunction with the IOL. Other surgical procedures that have been adopted in patients with microspherophakia are scleral-fixated IOL implantation and angle-supported or iris-claw-supported anterior chamber IOL implantation.[9,10]

A thorough preoperative assessment is fundamental for successful surgery. In this case, we present a self-proclaimed healthy man with bilateral cataract. Through careful anamnesis and examination, the diagnosis of Klinefelter-associated microspherophakia could be made, which allowed for appropriate surgery preparation with favorable outcome. In cases of early-onset cataract and/or lens subluxation, a thorough inquiry into systemic disorders such as connective tissue diseases and metabolic disorders is warranted. In addition, this case stresses the importance of a systemic and/or genetic evaluation in patients with microspherophakia and an ophthalmological examination in patients with Klinefelter syndrome.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that his name and initials will not be published and due efforts will be made to conceal his identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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