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Pseudoexfoliation syndrome in a patient with subluxation of the lens secondary to homocystinuria

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ABSTRACT

This article aims to comprehensively describe a particular case of bilateral intraocular subluxated lens with pseudoexfoliation syndrome in a Moroccan woman with homocystinuria. We report a case of a 56-year-old patient who has a cataractous subluxated lens along with whitish deposits. The association of PEX syndrome with hyperhomocysteinemia has rarely been found. It worsens the lenses' zonular stability and causes multiple complications.

KEY WORDS: homocystinuria, pseudoexfoliation, subluxated lens, cataract, historical case, ophthalmology, eye surgery

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INTRODUCTION

Various conditions can leads to of the lens, such as Marfan syndrome or rarely homocystinuria. The latter is an autosomal recessively inherited defect in the transsulfuration pathway (type-I) or methylation pathway (Types II or III).

This article aims to comprehensively describe a particular case of a bilateral intraocular subluxated lens with pseudoexfoliation syndrome in a Moroccan woman with homocystinuria.

CASE DESCRIPTION

We report a case of a 56-year-old patient with a history of mental retardation due to hyperhomcysteinemia. She is monitored in internal medicine with poor therapeutic compliance. Her story dates back to the age of five when she had an acute drop in visual acuity following minimal blunt trauma to her OS, then, subsequently, two years later, she had a contralateral drop in visual acuity. The patient's family consulted an ophthalmologist who recommended surgery. However, due to unfavorable socio-economic conditions, she could not have it. Many years after the incident, she consults for a chronic bilateral decline in her visual acuity.

The ocular examination finds visual acuity quantified by counting the fingers at 3 meters in both eyes. The IOP was 15 mmHg. On her left eye, the examination of her anterior segment finds clear conjunctiva and cornea, a normal depth of her anterior chamber, and, after pupil dilation, a cataractous lens subluxated infero-temporally, as well as stretched zonules on the opposite side all covered with PEX deposits. There where whitish deposits in the pupillary circumference and on the surface of the lens related to a PEX syndrome *(figure1).* The iridocorneal angle is open with the distinction of a discontinuous pigmented line at the level of the Schwalbe ring. The right eye is similar to the OS with a lens subluxation and pseudo-exfoliations deposits **(***figure 2).* Her fundus examination was normal.

Subsequently, the therapeutic decision was to perform a cataract surgery and the placement of an iris fixation implant. There were no postoperative incidents

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FIGURE 1. Photo of the left eye showing a subluxated lens with PEX deposits and stretched zonules

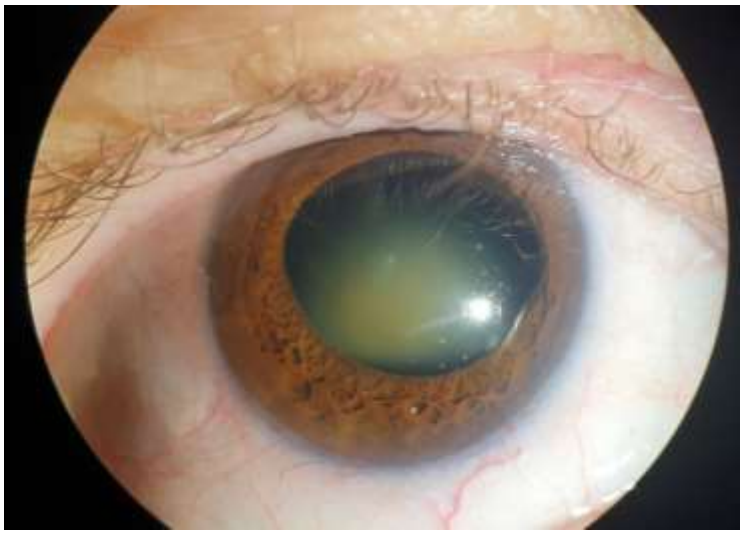

FIGURE 2. Photo of the right eye with a subluxated lens

DISCUSSION

The displacement of our patient's natural crystalline lens from the patellar fossa is initially due to homocystinuria discovered after a mild trauma. However, it worsened by the presence of a pseudoexfoliation syndrome.

Homocystinuria is an autosomal recessively inherited defect in the transsulfuration pathway (type-I) or methylation pathway (types II and III). Its incidence varies between 1 in 50,000 and 1 in 200,000 [1].

In this disease, the self-interaction properties are altered due to homocysteinylation of fibrillin-1 because of a reduction of disulfide-bonded C-terminal fibrillin-1 multimers [2], which results to zonular weakness and progressive ectopia lentis.The manifestations occur afterbirth and are nonspecific. The diagnosis is usually made when the subluxation of the lens happens. It can be spontaneous or it can be caused by a mild blunt trauma. Approximately 40% of 5-year-old patients with untreated homocystinuria have crystalline lens subluxation, and almost all patients have it by the age of 25 years. In 50% of the cases, the lens is dislocated down and nasally. Posterior dislocation occurs in 20% of the cases and the anterior one in 10% leading to an elevated IOP and corneal decompensation [1]. Consequently, there will be a severe lens-induced myopia and iridodonesis. Skeletal abnormalities resembling to those of Marfan syndrome are associated. Mental retardation is common and the children have blue eyes and a malar flush. There is a high risk of thromboembolic accidents. The homocysteine in serum and urine is elevated. The treatment includes a low methionine and high cysteine diet and a supplementation of pyridoxine [3].

Hiller R. et al have reported in their studies that both Homocystinuria and Pseudoexfoliation (PEX) syndrome have a genetic polymorphism (Lysil oxidase like 1 gene). With the latter homozygous presence, there is a risk of homocysteine metabolism disorder and aggregation of elastic components of PEX [4].

However, unlike homocystinuria, Pseudoexfoliation (PEX) syndrome is an age-related systemic syndrome that targets mainly ocular tissues [4]. Its prevalence increases with age and depends on geography and ethnicity. It is commonly found in patients between 69 and 75 years [4]. There are numerous risk factors associated with the origin of PEX deposits: age, genetic predisposition, ultraviolet radiation, oxidative stress, chronic inflammation caused by herpes viruses, hepatitis C virus and Helicobacter Pylori bacteria [5,6].

It is due to the deposition of white material from the lens, mainly on the lens capsule, ciliary body, zonules, corneal endothelium, iris and pupillary margin. PEX deposits are made of a protein core, a mass of conjugated complex sugars (Proteoglycan), glycoasminoglycans, non-collagen ingredients basement membrane and elastic microfibrile [7].

In a Moroccan study of patients operated for cataracts, PEX syndrome was detected in 9.8% of patients while the mean age was 69.3 years old [8].

The association of PEX syndrome with hyperhomocysteinemia has been found. It is due to a disturbed folates status in the body leading to aggregation of elastic components of PEX. However, hyperhomocysteinemia exists in patients with POAG and normotensive glaucoma [4]. Initially the syndrome is manifested unilaterally before becoming bilateral. There are no or few symptoms. [4]

The diagnosis PEX syndrome is made during the clinical examination. Whitish deposits can be seen in the pupillary margin. After a pharmacological mydriasis, we can recognize "3 ring" sign on the surface of the lens: A central disc, then, a peripheral ring covered with PEX material and an annular zone between them. There can be some places of iris transparency due to the friction of the iris and the deposits during pupillary light reactions, which leads to pigment dispersion. Thus, we can observe pigmentations in the anterior chamber and the angle at the level of the Schwalbe's line or in front of it (Sampaolesi line) as well as a corneal edema. Lens capsule is thin and zonules are weakended leading to dislocations after a mild trauma. Cataract is frequently associated and there is a high rate of conversion to a glaucoma which responds poorly to the treatment [4]. Care should be taken during lens extraction, which has a higher chance of complications (lens dislocation, acute elevated IOP, iris bleeding and inflammation…) [9].

Blunt trauma is the most common cause of unilateral lens dislocation [10]. Blunt force in anteroposterior direction leads to equatorial expansion, which disrupts the zonular fibers and dislocates the lens [11]. In diseases that cause zonular fragility (such as Marfan syndrome, homocystinuria or pex syndrome), a mild blunt trauma can cause subluxation of the lens [12,13].

CONCLUSION

This association between PEX syndrome and homocystinuria is a rare one. It causes lens dislocation and compromises the stability of the capsular bag after lens extraction especially when there is an associated cataract [4].

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