Williams Syndrome and Zonulopathy

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Abstract

A 61-year-old male with only a past medical history of Williams syndrome presented with bilateral visually significant cataracts. Although pre-operative evaluation revealed no signs of zonular compromise, marked instability of the zonules was noted during cataract extraction with intraocular lens placement in both eyes, requiring the use of a capsular tension ring and three piece lens. Williams syndrome may be associated with zonular weakness. When treating these patients, surgeons should be prepared to deal with zonulopathy with the use of capsular support aids.

INTRODUCTION

Weakness of the zonular apparatus has well-known associations with systemic conditions including Marfan’s syndrome, homocysteinuria, and pseudo-exfoliation. Here we present a possible association between Williams Syndrome and zonulopathy, which to our knowledge has not been previously reported.

Individuals with Williams syndrome have a characteristic "elf-like" facial appearance and often exhibit a cheerful and outgoing personality. Other common manifestations include heart defects, hypotonia, failure to thrive, and developmental delay [1-3]. Reported ocular manifestations of Williams syndrome include strabismus, amblyopia, hyperopia, stellate iris pattern, and tortuosity of retinal vessels [2,4-5]. It is estimated that Williams Syndrome occurs in approximately 1 in 10,000 live births [6].

CASE PRESENTATION

A 61-year-old male presented to the Gavin Herbert Eye Institute with worsening vision which was reported by caretakers to be affecting daily activities. Pertinent medical history includes a prior clinical diagnosis of Williams Syndrome, not confirmed with molecular testing. Review of systems and family history were non-contributory. Best correct visual acuity was count fingers at four feet in the right eye and 20/100 in the left eye. Slit lamp exam revealed bilateral cataracts. The right eye cataract was 3+ nuclear sclerotic with 3+ central anterior cortical changes. The left eye cataract was 3+ nuclear sclerotic. Of note, the patient was a high myope with an axial length of 26.77mm in the right eye and 27.09mm in the left eye. Pre-operative evaluation by the retina service revealed only peripheral pigmentary changes. No other abnormal findings were noted. Specifically, the patient did not exhibit any signs of zonular weakness including subluxation, phacodenesis, or pseudoexfoliation material on the anterior lens capsule. In addition, there was no trauma noted on history. After discussion of risks, benefits, and alternatives, it was determined that cataract extraction with intraocular lens placement would be indicated in both eyes.

During surgery, initiation of the capsulorhexis revealed striiae in the anterior capsule. As the capsulorhexis was continued, marked mobility of the lens was observed in all quadrants. At this time, use of a capsular support system (capsular hooks) was considered, but was ultimately determined to be unnecessary. The capsulorhexis was completed, nuclear disassembly performed using a horizontal chop method, and the cortex was removed without complication. A capsular tension ring and a three-piece lens were then inserted into the capsular bag. Cataract extraction and intraocular lens placement in the fellow eye revealed similar findings and was handled in the same manner.

DISCUSSION

Pre-operative evaluation of our patient did not reveal any signs of zonulopathy and subsequent review of the literature did not uncover any reports of zonulopathy in patients with Williams syndrome. Of course, our report here merely suggests an association, as the causes of zonulopathy are varied and can escape even the most thorough pre-operative evaluation, especially in a patient where medical history could not be accurately obtained from the patient himself.

The pathology of Williams syndrome is rooted in the deletion of 27 genes from chromosome seven, one of which is the gene for elastin. The deletion of this gene has been implicated as the cause of the cardiac defects and other connective tissue abnormalities observed in these patients. Histological examination of zonular fibers has revealed that although elastin is not present in the
zonules themselves, it is present at the zonular connections proximally with the ciliary process and distally to the lens capsule [7]. This is a possible explanation for zonulopathy in our patient. We suggest here that Williams syndrome is akin to diseases such as Marfan's syndrome, which manifest in a wide range of connective tissues, including the zonules.

A number of techniques exist for addressing zonular instability. In our case, a capsular tension ring and a three-piece lens were used. Evidence has shown that expansion of the capsular bag allows the zonules to recoil and have more strength. Perhaps more importantly, the presence of a capsular tension ring and three-piece lens allow for more options if subsequent dislocation or subluxation of the intraocular lens were to occur in the future.

CONCLUSION

Williams syndrome may be associated with zonular weakness, indicating the use of capsular support systems that aid in actual cataract extraction as well as give options if future intraocular lens repositioning is needed.

LITERATURE SEARCH

The authors conducted a MEDLINE search using the PubMed database (National Library of Medicine) through June 18, 2016. The search was limited to articles in English or articles with English abstracts. Various combinations of keywords were used, including Williams, syndrome, zonulopathy, zonular, apparatus, instability, zonules, anterior segment, and capsular tension rings.

REFERENCES