Non-necrotising Scleritis and Spherophakia in Marfan Syndrome: A Rare Situation Adhering to Hickam's Dictum

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ABSTRACT

Marfan syndrome is an autosomal dominant, connective tissue disorder that affects various systems of the body including the eyes. We present a case of a 22 year old man with Marfan syndrome presented with a painful red left eye. On examination he was diagnosed to have non-necrotising scleritis and microspherophakia. Non-necrotising scleritis with microspherophakia is a rare and arduous situation. This is an interesting case where Hickam's dictum prevails over its counter argument of Occam's razor principle.

KEY WORDS

Marfan syndrome, Non-necrotising scleritis, Spherophakia

INTRODUCTION

Marfan syndrome is an autosomal dominant, connective tissue disorder primarily affecting the cardiovascular, musculoskeletal and ocular system and is associated with mutation in protein fibrillin 1 (FBN1) on chromosome 15.[1] The main ocular features of Marfan syndrome include myopia, iridodonesis, phacodonesis, ectopia lentis, keratoconus, iris coloboma, cataract, and glaucoma.[2] Microspherophakia can also be seen in Marfan syndrome.[3]

Herein we report an interesting case of Marfan syndrome with bilateral ectopia lentis and non-necrotising scleritis in one eye. Although microspherophakia can be implicated with Marfan syndrome, necrotising scleritis with inflammation certainly cannot. This can be linked with the principle of Hickam's dictum which says 'a patient can have a number of diagnoses and a single diagnosis need not always explain all the pathology'.[4]

CASE REPORT

Twenty-two year old man, a known case of Marfan syndrome (diagnosed on clinical grounds in a tertiary care multidisciplinary hospital), presented with painful red left eye for 5 days. There was no history of trauma, discharge, coloured halos, nausea and vomiting. The best corrected visual acuity was 6/12 with +6 DS in right eye and 6/18 with -4.5/-1.5 DC x 160 degrees in left eye. Slit-

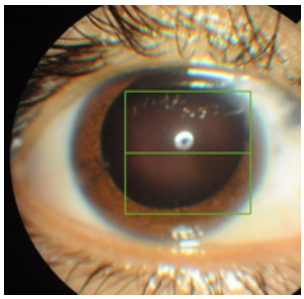

Figure 1. Right eye, aphakia

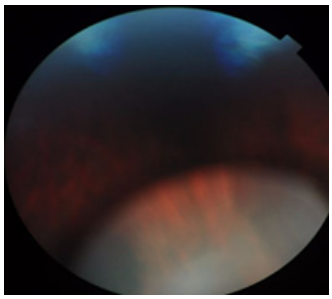

Figure 2. Dislocated lens in vitreous of right eye

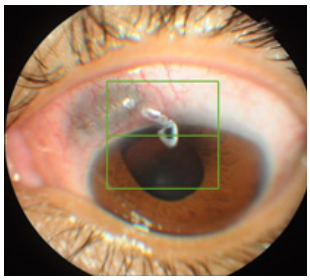

Figure 3. Left eye showing peaked pupil, non- necrotising scleritis with visible uveal tissue in the patient

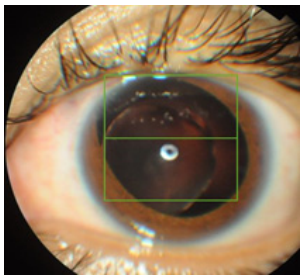

Figure 4. Left eye with microspherophakia

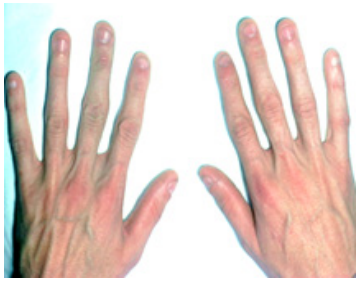

Figure 5. Arachnodactyly in the patient

lamp biomicroscopy revealed clear cornea, deep anterior chamber, round reactive pupils and aphakia in right eye. (Fig. 1) Lens was dislocated and visible in vitreous cavity. (Fig. 2) No reactive vitritis was seen and retina was normal. Left eye revealed conjunctival and ciliary congestion and thinning of sclera superiorly with bluish appearance of uveal tissue (Fig. 3). The appearance and history was suggestive of non-necrotising scleritis. Cornea was clear, anterior chamber was quiet and normal in depth. Pupil appeared to be peaked towards 10'o clock. Iridodonesis and phacodonesis were present and lens appeared spherical in shape. (Fig. 4) The lens edges and zonules were clearly visible on slit-lamp examination after pupillary dilation. Intraocular pressure (IOP) was 12 mmHg in the right eye (OD) and 18 mmHg in the left eye (OS). Central corneal thickness was 526 µm OD and 525 µm OS. Axial length was 26.25 mm OD and 26.22 mm OS. Anterior chamber depth was 2.02 mm OS, Lens thickness after pupillary dilation was 4.9 mm OS. A thorough family history revealed no history of high myopia and poor vision. Systemic examination revealed long slender fingers and an increased ratio of arm span to height. (Fig. 5) Complete blood count was normal with elevated ESR (35 mm/ hr). Rheumatoid factor was negative. Serum homocysteine, serum lysine were normal. Genetic analysis couldn't be performed due to unavailability of the test. We prescribed the patient with topical Prednisolone acetate 1% 4 times a day and atropine eye drop thrice daily in left eye. Tablet Prednisolone 55 mg (1 mg/kg body weight) once daily for 10 days and then to taper 10 mg every week for next 6 weeks. The patient was planned for right eye pars plana vitrectomy, lens retrieval with IOL implantation followed by left eye lens extraction with IOL implantation after resolution of the inflammation. The inflammation did subside after 3 weeks but the patient did not opt for surgery.

DISCUSSION

The above case describes a patient of Marfan syndrome with microspherophakia. Such cases have been described by Muralidhar et al.[3] The most common ocular abnormality in Marfan syndrome is ectopia lentis occurring in 50% to 80% of affected individuals.[5] Presence of ectopia lentis is a major criteria for the diagnosis of Marfan syndrome.[6] In the case presented above, along with microspherophakia and ectopia lentis, non-necrotising scleritis was present. Scleritis is a severe, destructive inflammatory disease affecting the eye wall. In Watson's series, 46% of the patients had an identifiable systemic disease that can be linked to scleritis.[7] Rheumatoid arthritis is by far the most common systemic condition associated with scleritis.[8] The cause of scleritis could not be determined in the case described above. Presence of Marfan syndrome cannot solely explain the presence of such degree of scleritis. Such clinical picture can be explained by Hickams dictum which says 'a patient can have as many diagnoses as he darn well pleases'.[4] Its counter argument Occam's razor principle tells the clinician to look for the most simple and unifying diagnosis which could explain all of a patient's problems.[4]

To conclude, non-necrotising scleritis with microspherophakia is a rare and arduous situation. This is an interesting case where Hickam's dictum prevails over its counter argument of Occam's razor principle.

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