

WEILL-MARCHESANI SYNDROME IN NIGERIA: Report of a Case that Presented with Bilateral Cataract

***NO MAGULIKE** FRCS, Consultant Ophthalmic Surgeon/Senior Lecturer
Department of Ophthalmology, University of Nigeria Teaching Hospital PMB 01129 Enugu

AE AGHAJI FMCOphth, Senior Registrar
Department of Ophthalmology University of Nigeria Teaching Hospital, Enugu

SUMMARY

This study reports a single case of Weill-Marchesani syndrome in an adult Nigerian. This syndrome is typically characterized by progressive joint stiffness, short stature, brachydactyly, microspherophakia and ectopia lentis. The patient had progressive loss of vision from bilateral cataract. To the best of our knowledge, there is no previously reported case of the syndrome in Africa.

Key words: Weill-Marchesani syndrome, Nigerian, cataract

INTRODUCTION

Weill-Marchesani syndrome is a rare connective tissue disorder first described by Weill in 1932 and further delineated by Marchesani.¹ Less than 100 cases have been reported in the literature.² We present here, the case of a fifty-year-old Nigerian male with clinical features of Weill-Marchesani syndrome, who presented with progressive loss of vision due to cataract. To the best of our knowledge, this is the first reported case in Nigeria.

CASE REPORT

A.L., a fifty-year-old casual labourer presented with a four-year history of painless progressive blurring of vision in both eyes, which worsens with distance. There was no diurnal variation. He bumped into objects directly in front of him and at his sides. There was no history of haloes around light or redness of his eyes. There was a positive history of traditional eye medication. Apart from poor vision, he also complained of generalized joint stiffness, which he claimed affected his work.

General examination revealed a short-statured elderly man, 1.58m in height. Other physical findings included brachydactyly, with limitation of abduction of the thumbs, and flexion at the wrists.



Figure 1. Patient's height 1.58m



Figure 2. Digits of the patient showing brachydactyly and limitation of abduction of the thumbs

* Author for correspondence

Ocular examination revealed an unaided visual acuity of counting fingers at 1 metre in each eye. There was no improvement with a pinhole. Other abnormal findings were iridodonesis and a superotemporal subluxated lens in each eye. The lens was opaque in each eye. Both pupils reacted directly and consensually to light (but they were very difficult to dilate with cyclopentolate 1% and phenylephrine 10% or atropine 1% and phenylephrine 10%). The intraocular pressure was 17mmHg in each eye. Funduscopy showed no red reflex in both eyes.

A right intracapsular cataract extraction was performed with a primary scleral-fixated posterior chamber intraocular lens. The intact extracted lens was microspherophakic, measuring about 5.0mm in diameter.

The postoperative condition was satisfactory and his vision in the right eye improved to 6/36 three weeks postoperative, before he was lost to follow-up.

DISCUSSION

Weill-Marchesani syndrome was initially thought to be inherited as an autosomal recessive trait.^{3, 4} More recently, however, both autosomal recessive and autosomal dominant modes of inheritance have been described.^{1, 2} Weill-Marchesani syndrome has also been associated with consanguinity.^{4, 5} In this case, a proper family tree could not be established, as the patient was unable to supply the required information.

Weill-Marchesani syndrome is a rare connective tissue disorder with characteristic systemic and ocular features. The systemic features include short stature, brachydactyly and mental handicap.⁶ Other features include limited joint mobility, especially of the hands and wrists,² and thick inelastic skin.

A few cardiovascular problems have been noted, e.g., pulmonary stenosis and subvalvular aortic stenosis, but these findings have been inconsistent.⁷ There were no cardiovascular or pulmonary abnormalities in this case.

The ocular features include microspherophakia, which may cause the lens to move anteriorly, thus occluding the pupil; inferior lens dislocation; and an angle anomaly secondary to mesodermal dysgenesis.⁶ This patient had bilateral superotemporal subluxated lens with iridodonesis. These signs are more commonly seen in simple ectopia lentis, which is a congenital form without associated systemic problems.⁸

The ophthalmic manifestations are primarily lens related. Progressive² microspherophakia results in lenticular myopia ranging from -3D to -20D. This patient may have been myopic; his vision was worse for distance. Refraction was not possible because of the cataracts.

The formation of the small spherical lens is due to defective development of the zonule.⁹ The lens zonules

may be loose and eventually rupture during the second decade of life, leading to ectopia lentis.²

Following spontaneous dislocation, the lens generally becomes cataractous.¹⁰ In addition, acute pupillary block glaucoma may occur in untreated patients with ectopia lentis as loose zonules allow the lens to displace anteriorly therefore increasing lens-iris apposition.² In this patient, cataract occurred at about the same age as in normal individuals, and intraocular pressure was normal. The extracted cataract showed a small spherical lens.

Ectopia lentis continues to be a therapeutic challenge for ophthalmologists. Cycloplegics or laser iridotomy are the non-surgical methods of relieving angle closure in patients with microspherophakia. Minimal subluxation may cause no visual symptoms, but in more advanced cases, serious optical disturbances arise. Indications for surgery include: diminished visual acuity, lens dislocation into the anterior chamber and lens-induced glaucoma.¹⁰

The diagnostic features in this patient were short stature, joint stiffness, brachydactyly, ectopia lentis and microspherophakia. The mean height of an adult Nigerian is 1.69m SD \pm 0.08.¹¹ This patient measured 1.58m in height. In Weill-Marchesani syndrome, the lens is often dislocated inferiorly. In this case, however, it was dislocated superotemporally, an occurrence which is more often associated with Marfan's syndrome. Recent literature, however, strongly suggests that autosomal dominant Weill-Marchesani syndrome and Marfan's syndrome are allelic conditions at the fibrillin 1 locus, adding to the remarkable heterogeneity of type 1 fibrillinopathies.¹

Weill-Marchesani syndrome, Marfan's syndrome and simple ectopia lentis may have some common ocular features. This may explain why this patient had bilateral superotemporal dislocation of the lens, which is seen in Marfan's syndrome. This, with iridodonesis is also seen in simple ectopia lentis. This patient would have been followed-up to a postoperative refraction to obtain his best corrected visual acuity, unfortunately, he failed to keep further appointments. He may have been satisfied with his vision and gone back to his job. If so, he may have had poor vision prior to the onset of the cataracts.

CONCLUSION

Weill-Marchesani syndrome is a very rare condition with less than 100 cases reported in the literature.²

In the absence of genetic studies, a diagnosis of Weill-Marchesani syndrome can be made in the case of a patient with ectopia lentis who presents with the typical systemic and ocular features of the syndrome, if a high index of suspicion is maintained.

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