

Visual Outcome in Weill Marchesani Syndrome – A Rare Case Report

Archana Anand Pawar¹, L Usharani², Sonam Gelek Palzang³,
Vipin Kumar Singh⁴

^{1,2,3,4}Department of ophthalmology, Regional Institute of Medical sciences, Imphal, India.

Abstract : Weill Marchesani syndrome is a rare systemic connective tissue disease characterized by short stature, brachydactyly, ectopia lentis and spherophakia. The disease can result in visual impairment often due its associated complications such as lenticular myopia, lens dislocation and pupillary block glaucoma. These patients typically present in childhood and often permanent vision loss is already set in at the time of initial presentation. Many a times the condition often remains unrecognised until complications occur. Here is a rare case report of Weill Marchesani syndrome in a 14yr old female who was being treated as a case of high myopia for about 5 years and the underlying syndrome was detected only after glaucoma had set in.

Keywords: brachydactyly, ectopia lentis, glaucoma, Weill Marchesani

I. Introduction

A number of congenital and hereditary conditions have been described in the literature having associated ocular manifestations. Some of these conditions are amenable to treatment such as congenital cataract, congenital glaucoma, ectopia lentis and others. While the others are more challenging and still need more research. Weill-Marchesani syndrome is a rare systemic connective tissue disease characterized by short stature, brachydactyly, ectopia lentis and spherophakia. The diseases can often lead to blindness resulting from high myopia, glaucoma and lens dislocation, which is potentially treatable if appropriately managed in right time.

Both autosomal recessive as well as autosomal dominant mode of inheritance are known to occur. Most of the cases described in literature have presented with various degrees of loss of vision. This a case report of a patient presenting with features of Weill Marchesani syndrome and the purpose of reporting this case is to review this rare conditions and to lay emphasis on its early detection to prevent permanent vision impairment

II. Case Report

Case History

A 14 year old female, earlier diagnosed as a case of high myopia for the past 5 years presented with history of eye ache for a duration 1 week

On examination

BCVA(best corrected visual acuity) was 6/12 part in both eyes with a myopic correction of -16 diopter spherical lens in right eye and -14.75 diopter spherical lens correction in left eye. External ocular adnexa were normal. Positive findings on slit lamp examination included bilateral shallow anterior chamber (van Herick Grade I) and a sluggishly reacting, non-dilating pupil with irregular pupillary margin was seen in both eyes. An anteriorly subluxated microspherophakic lens causing pupillary block was noticed bilaterally. Posterior segment revealed presenile vitreous degeneration and glaucomatous disc changes more marked in the right eye was noticed. (Fig 1 a&b)

Investigations

IOP (intraocular pressure): (R) eye: 28 mm Hg and (L) eye: 22 mm in Hg

Gonioscopy: narrow angle in all quadrants in both eyes

HFA (Humphrey Field Analyzer): glaucomatous field defects (right>left) (Fig 2 a&b)

Axial length: (R) eye: 22.65mm and (L) eye: 21.82mm (asymmetric axial length)

Ultrasound B Scan: bilateral dense vitreous floaters

On general physical examination patient was also noticed to have short stature (Fig 3), Brachycephaly (Fig 4), short stubby fingers and toes (Fig 5) with brachycephaly and brachydactyly on Xrays. Systemic examination was within normal limits. Blood investigations including complete blood counts, blood sugar, Liver function test, kidney function test were within normal limits. Screening of the family members was done. Patient

was the youngest of the three siblings. No evidence regarding presence of the disease in any of the family member was seen.

Diagnosis

With above mentioned clinical findings a clear cut diagnosis of Weill Marchesani syndrome with subluxated microspherophakic lens leading to pupillary block glaucoma was made.

Management

Patient was prescribed full correction of myopia and IOP was controlled using a combination of timolol maleate and brimonidine eyedrop. Patient was advised surgery for extraction of the ectopic lens and correction of the pupillary block glaucoma, however the patient did not give consent for surgery and subsequently lost follow up.

III. Discussion

Weill Marchesani syndrome is a very rare disease with a prevalence of 1:100,000 populations and hence, it often remains undiagnosed until the patient presents with complications. It is characterized by abnormalities of the lens, unusually short stature, brachydactyly, and joint stiffness. The ocular problems most commonly reported include microspherophakia (small spherical lens), lenticular myopia, ectopia lentis (abnormal position of the lens), and glaucoma secondary to pupillary block from anteriorly subluxated lens or dislocated lens in the anterior chamber, which can lead to blindness. The similar positive findings in this case led to our diagnosis in addition to presenile vitreous degeneration and asymmetric axial length.

Mandal SK et al[1] and el Kettani A et al[2] have reported the occurrence of glaucomatous optic atrophy in their case reports of Weill Marchesani syndrome as also seen in our case. They also reported association of cardio vascular abnormalities with this disease entity, however no such finding was detected in our case. M.L. Kulkarni et al[3] and Cem Evereklioglu et al[4] have reported the occurrence of this disease in 6 patients each, belonging to the same family. Both studies focused in determining the inheritance pattern of the disease and concluded it to be autosomal dominant in nature. Recently, Verloes et al[5] described Weill Marchesani like syndrome in 3 generations of one family and proposed a new name, GEMSS syndrome (glaucoma, ectopia, microspherophakia, stiff joints, short stature) for dominantly inherited Weill Marchesani like syndrome to distinguish it from the classical recessive Weill Marchesani syndrome.

In the absence of any complication patient can be managed with myopic correction and regular follow up. However early detection and removal of an ectopic lens decreases the possibility of pupillary block and glaucoma. Medical treatment of glaucoma is mostly unsatisfactory and may warrant surgical intervention in the form of peripheral iridectomy and trabeculectomy in advanced chronic angle closure glaucoma

IV. Conclusion

Most of the cases of Weill Marchesani Syndrome remain undiagnosed, often being treated as myopic eyes until complications such as secondary glaucoma and lens dislocation occur. As permanent visual loss of various degrees is already set in at their initial visit, awareness of this condition and importance of diagnosing at an earlier stage cannot be underestimated. Proper patient counseling and screening of family member is an essential part of management of this condition.

References

- [1]. Mandal SK, Mondal SS, Mani S, Chatterjee S, Chatterjee K, Bhattacharya R, et al, Case report of two sisters suffering from Weill-Marchesani syndrome with pulmonary stenosis, *Indian J Med Sci*, 64(3), 2010, 140-3.
- [2]. el Kettani A1, Hamdani M, Rais L, el Belhadji M, Rachid R, Laouissi N et al, Weill Marchesani syndrome. Report of a case, *J Fr Ophtalmol*,24(9), 2001, 944-8.
- [3]. M.L. Kulkarni, V. Venkataramana, C. Sureshkumar, Satishchandra, Weill-Marchesani Syndrome, *INDIAN PEDIATRICS*,28, 1995, 923-26.
- [4]. Cem Evereklioglu, Ibrahim F Hepsen, Hamdi Er, Weill-Marchesani syndrome in three generations, *Eye*, 13, 1999, 773-77.
- [5]. Verloes A, Hermia JP, Galand A, Koulischer L, Dodinval P, Glaucoma lens ectopia-microspherophakia-stiffness shortness (GEMSS) syndrome: A dominant disease with manifestations of Weill Marchesani syndromes, *Am J Med Genet*, 44, 1992, 48-51.
- [6]. Puri LR1, Sharma H, Aryal S, Weill- Marchesani syndrome: a rare case report, *Nepal J Ophthalmol*, 4(2), 2012, 336-8.
- [7]. Jensen AD, Cross HE, Paton D. Ocular complications in the Weill-Marchesani syndrome. *Am J Ophthalmol*,77, 1974, 261-269.



Fig 1 a & b: Subluxated Microspherophakic lens causing pupillary block in right eye (a) and left eye (b)

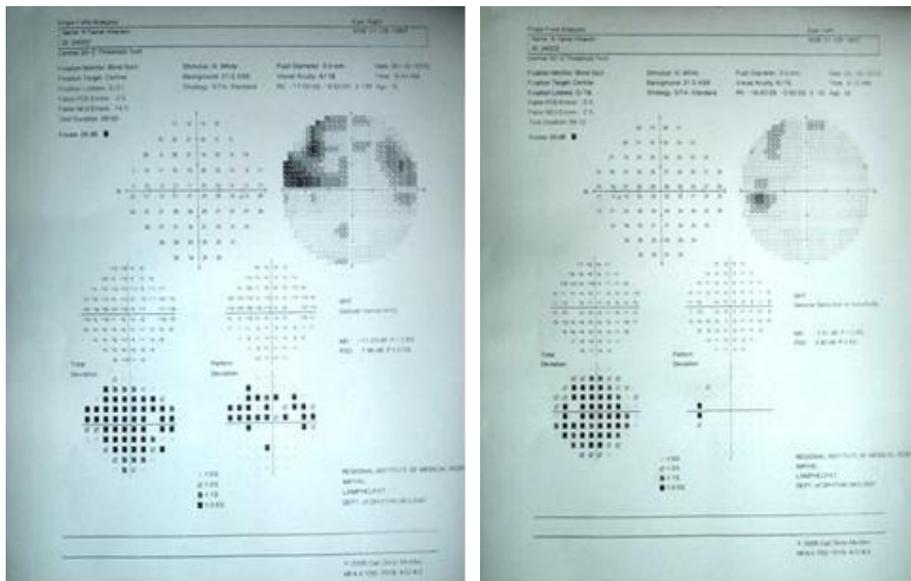


Fig 2(a & b): HFA perimetry report



Fig 3: Short stature

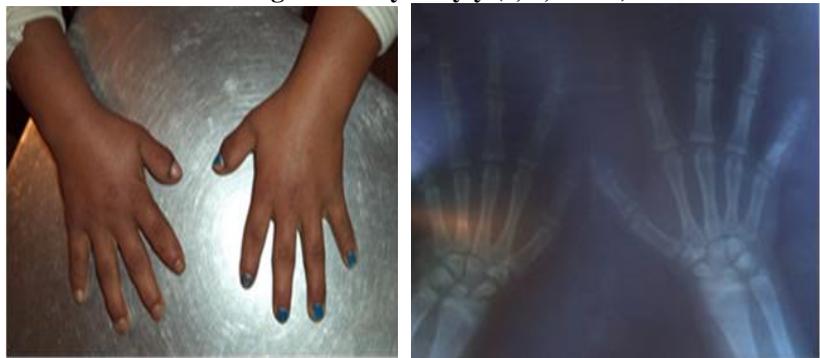
Fig 4: Brachycephaly (a & b)



(a)

(b)

Fig 5: Brachydactyly (a, b, c & d)



(a)

(b)



(c)



(d)