

## A Rare Case Report: Weill Marchesani Syndrome

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**Abstract:** A 21 year old female came with defective vision for 10 years. She also had short stature (height 126 cm) with short and stubby fingers in both limbs. Anterior segment slit lamp examination revealed shallow anterior chamber, microspherophakia with superonasal subluxation in right eye and superotemporal subluxation in left eye respectively with normal fundus. Keratometry reading and axial length was +44.75D, 22.30mm in right eye and +45.25D, 22.11mm in left eye. Intraocular pressure by Goldmann applanation showed 20 mmhg and 34 mmhg respectively. On UBM with 50HZ probe we found decreased anterior chamber, narrow angle of anterior chamber, increased lenticular sphericity, sparse zonules in both eyes and subluxation of lens in both eyes. She was diagnosed with Weill-Marchesani syndrome (WMS) a connective tissue disorder that usually presents in childhood with short stature and ocular problems. The major features of Weill-Marchesani syndrome are short stature, eye abnormalities, unusually short fingers and toes (brachydactyly), and joint stiffness.

**Keywords:** Weill-Marchesani syndrome, defective vision, microspherophakia

### INTRODUCTION

Weill Marchesani syndrome (WMS) is a rare genetic disorder characterized by short stature, brachydactyly, joint stiffness, and characteristic eye abnormalities including microspherophakia, ectopia of the lens, severe myopia, and glaucoma [1]. The name has been coined after ophthalmologists Georges Weill and Oswald Marchesani who described it in 1932 and 1939, respectively [2].

### CASE REPORT

A 21 year old female came to ophthalmology out patient department with complaints of defective vision in both eyes for past 10 years. She was second child of second degree consanguineous marriage. She was full term normal delivery with normal developmental milestones. She was short stature (height 126 cm) with short and stubby fingers in both limbs. There was no similar history in family. Ocular examination showed visual acuity of 1/60 in both eyes. Refraction was done which showed -20.00DS/-3.00DC 180 axis improving to 6/60 -20.00/-3.00DC 180 improving to 6/60. Anterior segment slit lamp examination revealed shallow anterior chamber, microspherophakia with superonasal subluxation in right eye and superotemporal subluxation in left eye respectively. Keratometry reading and axial length was +44.75D, 22.30mm in right eye and +45.25D, 22.11mm in left eye. Intraocular pressure by Goldmann applanation showed 20 mmhg and 34 mmhg respectively. Pachymetry showed central corneal

thickness 0.52 mm in right eye and 0.53mm in left eye. Fundus examination was within normal limits. On UBM with 50HZ probe we found decreased anterior chamber, narrow angle of anterior chamber, increased lenticular sphericity, sparse zonules in both eyes and subluxation of lens in both eyes. Laboratory investigations revealed absence of homocysteine in the urine, absence of sulphur containing aminoacids and lysine in the urine and normal level of phytic acid in the serum. Patient was advised for lens extraction in both eyes and laser peripheral iridotomy.



Fig. 1: Short fingers



**Fig. 2: Showing short stature**



**Fig. 3: Showing spherophakia with lens subluxation**



**Fig. 4: Showing the lens**



**Fig. 5: Showing subluxation of lens in left eye**

## DISUSSION

Weill-Marchesani syndrome (WMS) is a connective tissue disorder, usually presents in childhood with short stature and ocular problems. The major sign and symptoms include short stature, eye abnormalities, unusually short fingers and toes (brachydactyly) and joint stiffness [3, 4].

WMS can be inherited in either an autosomal recessive or an autosomal dominant pattern caused by mutations in the ADAMTS10 gene that has an autosomal recessive pattern of inheritance. Other cases including those caused by mutations in the FBN1 gene are autosomal dominant pattern of inheritance [3].

Microspherophakia is the most important manifestation of WMS that results in lenticular myopia, ectopia lentis and glaucoma. Loss of vision occurs earlier in WMS than in lens dislocation syndromes. In some cases, lens dislocation and pupillary block appear after blunt trauma resulting in weakening of the zonular fibers [3].

Based on several characteristic clinical signs WMS diagnosis is done. Confirmation of WMS cannot be done by a single test. Exploring family history or examining other family members may help in confirmation of diagnosis [5].

## REFERENCES

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