Weil-Marchesani Syndrome in an Eight Year Old Girl

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Background: Weill–Marchesani syndrome is a rare systemic connective tissue disease. Ocular features include ectopia lentis with inferior subluxation in 50% of cases. Microspherophakia is also common with resultant anterior lens subluxation causing pupil block or complete luxation into the anterior chamber. Systemic features include short stature, brachydactyly, stiff joints and mental handicap.

Case Report: An 8 year old girl, primary one pupil presented to the paediatric eye clinic of Eleta Eye Institute with 1 year history of progressive loss of vision, worse in the left eye associated with progressive enlargement of the left globe. Visual acuity at presentation was hand movement(HM) in the right eye and no light perception(NPL) in the left eye. There was no preceding history of trauma. She was noticed to have short stature like her mother. The right eye had clear cornea and normal anterior chamber depth. There was microspherophakia with antero-inferior subluxation. The intraocular pressure was normal. The left eye was buphthalmic with poor view of the pupil and the lens. She had uneventful right lens extraction at University College Hospital, Ibadan and evisceration of the left eye with an implant. She was commenced on visual rehabilitation with full spectacle correction with distance visual acuity correction of 6/24 and near correction of N18 in her right (only) eye.

Discussion: Weill- Marchesani syndrome(WMS) is a rare disease, which often remains undiagnosed until the patient presents with complications[8]. Management requires timely intervention and close follow up and irreversible blindness may result if not properly managed. Our patient presented with bilateral progressive loss of vision similar to reported cases by Magulike *et al*[4] in Enugu, Nigeria and Tenkir *et al*[5] in Ethiopia. Our patient was a child similar to Pawar *et al*[8] in India, however Magulike *et al*[4] in Enugu, Nigeria and Tenkir *et al*[5] in Ethiopia reported adult cases. This patient presented with microspherophakia,

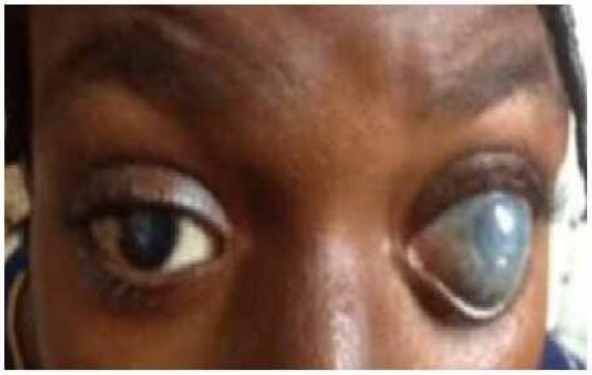

Fig. 1: Clinical photograph of the patient at presentation, showing left buphthalmos and corneal opacification

Fig. 2: Clinical photograph of the patient following right lens extraction and left evisceration.

cataract, lens luxation and short stature similar to Magulike *et al*[4] in Enugu, Nigeria , Tenkir *et al*[5] in Ethopia and Hui *et al*[6]. Secondary glaucoma seen in the left eye of our patient is a common finding in WMS as reported by Tenkir *et al* in Ethopia[5],Hui *et al*[6] and Pawar *et al*[8] in India. Hui et al[6] reported a patient who had trabeculectomy due to the secondary glaucoma as well as cataract extraction with intraocular lens implantation with good outcome. Diagnosis is made based on clinical signs. Family history might be of help in making diagnosis. Treatment is based on presentation while physical therapy and orthopedic treatments are generally prescribed for problems stemming from mobility from this connective tissue disorder[7].

Conclusion: Weill- Marchesani is not unusual in pediatric age group. Its management requires close follow-up and timely intervention to address ocular complications. Irreversible blindness may result if not properly managed.

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