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## Case reports on Low Vision Assessment & Management of Marfan Syndrome (MFS) Patients, in a Tertiary Hospital in Kano, Nigeria.

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### Abstract

Marfan syndrome (MFS) is an inherited disorder that affects the heart, joints, skeleton, skin, and eyes. People living with MFS are described as tall, long, slender built in appearance, with arachnodactyly, chest wall deformities, and scoliosis. A long, narrow face with deep-set eyes, down-slanting palpebral fissures, flat cheekbones, and a small chin are the facial features often found in people with MFS. The onset varies from infancy to all ages with most cases being diagnosed in the first two decades of life. In children, ocular findings are microspherophakia, congenital/infantile glaucoma, high refractive error at a young age, uveitis, retinal detachments, and enophthalmos secondary to the absence of retrobulbar fat. The basic management procedures include topical dilating agents for chronic dilation of the eye to increase pupillary size for aphakic correction, thorough and careful refraction, the use of contact lenses and/or glasses, removal of the dislocated lens, cataract surgery and treatment of amblyopia. Low vision assessments were carried out on the patients. The approach to the management of the two patients, whose cases are being reported, was different for each case and differed from conventional low vision management. Spectacle corrections of OD -20.00DS (1.76LogMar 6/36 near: 0.80M@23cm) and OD +7.5.0DS (6/9 1M@12cm) were issued for Cases 1 & 2 respectively. 8X Telescope (6/9) and magnifiers (4X Handheld N5 and spectacle +16 DS 0.50m/ N5) were recommended for cases 1 and 2 respectively. Low vision and rehabilitation were found to maximize visual functioning and are reliable options for MFS.

**Keywords:** Low vision, magnifier, Marfan syndrome, hereditary.

### Introduction

Marfan syndrome is a dominantly inherited disorder of the connective tissue caused by mutations in the gene encoding fibrillin-1

(FBN-1).<sup>1</sup> In the diagnosis of MFS, family history is a relevant positive indicator to consider, while genetic confirmation is valuable beyond estimation. Seventy-five percent of the patients inherit this condition from one of the

1. Lee SY, Ang CL. Results of retinal detachment surgery in Marfan syndrome in Asians. *Retina*. 2003; (2) 24–9.

affected parents, while 25% are thought to occur as a result of sporadic mutations.<sup>2</sup> The major problems caused by MFS have to do with the heart and blood vessels. These patients are often first found by eye doctors, because of short-sightedness or dislocated lenses which can cause major problems in the eye.<sup>2</sup>

In addition to the eye changes, patients with MFS show abnormalities in two areas: The cardiovascular and skeletal systems.<sup>2</sup> Excessive height (caused by excessive length of the distal limbs), anterior chest deformities, loose-jointedness, and scoliosis, are the changes in the skeletal system. The increased arm span in relation to body height, and an elongated lower segment (pubis to sole) compared with the upper segment (pubis to vertex) are unusual skeletal proportions.<sup>2</sup> Marfanoid Habitus is a term used to describe the slender, long, tall,<sup>3</sup> build associated with a wide range of skeletal findings. A long, narrow face with deep-set eyes, flat cheekbones, a small chin, and down-slanting palpebral fissures are the facial features seen among MFS patients.<sup>3</sup>

The zonule of the eye consists of radial fibers that connect the ciliary body to the crystalline lens. Zonules are primarily made up of FBN-1 and therefore mutations to progressive ectopia

lentis, which is seen in up to 65% of patients living with Marfan syndrome can occur.<sup>4,5,6</sup> Ectopia lentis and aortic root aneurysm (z-score > 2) or dissection are considered the two cardinal features of Marfan syndrome as agreed upon in the 2010 Ghent nosology.<sup>7</sup>

Marfan syndrome onset varies from infancy to all ages with most cases being diagnosed within twenty years of the patient's life. The progression may be in a subtle way but can also be acute when there is a late onset of dislocation. Approximately 6% of patients with MFS develop lens dislocation during their lifetime.<sup>2</sup> Open-angle glaucoma, retinal detachments without lens dislocation, pre-senile cataracts, rapid and total lens dislocation in the sixth or seventh decades in the absence of prior diagnosis of lens dislocation are the late onset of ocular complications.<sup>2</sup>

MFS diagnostic criteria consist of major and minor manifestations in different organ systems (Ghent Criteria).<sup>7</sup> The main clinical manifestations are aortic root aneurysm/dissection and ectopia lentis which may be uni- or bi-lateral and vary in severity from absent to total.<sup>2</sup> The third major criterion is genetic testing which is also crucial. The diagnosis of MFS is made when any two of the following three features are present:

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6. Loeys BL, Dietz HC, Braverman AC, Callewaert BL, De Backer J, Devereux RD, et al. The revised Ghent nosology for the Marfan syndrome. *Journal Medical Genetics*. 2010; 47: 476–485.
7. Chandra A, Ekwalla V, Child A, David Charteris D. Prevalence of ectopia lentis and retinal detachment in Marfan syndrome. *Acta Ophthalmologica*. 2014; 92: e82–e83.

Aortic root enlargement (Z-score  $\geq 2.0$ ), ectopia lentis, and a disease-associated form in the FBN1 gene, in the absence of relevant family history.<sup>8</sup> It is good to note that the dislocation, though predominantly superior, can occur in all directions, as well as posteriorly.<sup>2</sup> Other ophthalmologic features suggestive of Marfan syndrome include enophthalmos, miotic pupils, difficulty in completely dilating the pupils, enlarged corneal diameters, flattened corneal curvatures,<sup>9</sup> hypoplastic iris with peripheral iris transillumination, astigmatism and increased axial lengths.<sup>9</sup>

Patients with MFS are at risk of the following ocular problems which include high refractive errors, glaucoma,<sup>10</sup> presenile cataracts, chronic retinal detachments, potential long-term ocular complications, retinal detachments,<sup>11</sup> mild to severe vitreous degeneration,<sup>12</sup> uni- or bilateral amblyopia,<sup>11</sup> intercalary staphyloma, strabismus,<sup>13</sup> total lens dislocation, inadvertent bleb-wound herniation, chronic intraocular inflammation, secondary glaucoma, buphthalmos, and phthisis.

There are specific ocular findings in children (birth or within the first two years of life), which include enophthalmos secondary to the absence

of retrobulbar fat,<sup>13</sup> congenital or infantile glaucoma, microspherophakia which is typically observed in the Weill Marchesani Syndrome, retinal detachments, uveitis, and high refractive error at a young age.

The population incidence is 2–3 per 10,000.<sup>5</sup> Since FBN1 was linked with MFS in 1991,<sup>13</sup> more than 800 mutations in this gene have been identified.<sup>5</sup> The prevalence of MFS is unclear.

The goals of ocular management are the achievement of excellent and equal vision in both eyes, straight eyes, prevention or treatment of retinal detachments, controlled glaucoma, and informed patients and families.<sup>14</sup> Basic management includes careful and thorough refraction and the use of contact lenses and/or glasses, topical dilating agents for chronic dilation of the pupil to increase the pupillary size for aphakic correction, treatment of amblyopia, removal of the dislocated lens and cataract surgery.<sup>13</sup> Genetic counseling services can help families in understanding their diagnosis, make better informed decisions about future family planning and screening other family members. The eye is just one part of this complex genetic disease.

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2. Akram H, Aragon-Martin JA, Chandra A. Marfan syndrome and the eye clinic: from diagnosis to management. *Therapeutic Advances in Rare Disease*. 2021; 2. doi:10.1177/26330040211055738
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## Case Presentation:

### CASE 1

An 11-year-old boy primary 5 school pupil with a history of Marfan syndrome was brought by his mother to the Optometry Clinic of the Department of Ophthalmology, Aminu Kano Teaching Hospital, Kano, reporting a progressive decrease in the boy's vision. He had been seen already by Ophthalmologists in the Ophthalmology Clinic. The mother was told that his crystalline lenses were subluxated in both eyes. He did not report any problem before now with his vision until recent months when he could not copy anything from the blackboard. Medical history indicated Marfan syndrome with no heart, lung or any organ abnormality associated. He was not on any medications and follow-ups as the parents could not afford it. There was no known family members with Marfan syndrome.

### Clinical Examination Results

- External Exam: Long slender face.
- Visual Acuity: Right eye (OD) 3/60 Left eye (OS) 3/60
- Pupils: No anisocoria and no relative afferent pupillary defect detected.
- Motility: Ocular motility full and free Both eyes (OU).
- Anterior segment: Inferiorly subluxed lenses OU.
- Anterior Chamber: Angle was deep OU and there was no lens apposition to the cornea in either eye.
- Dilated funduscopy: Posterior segment was normal OU with no peripheral retinal degeneration as documented by the Ophthalmologist.

**Objective refraction (Retinoscopy):** OD -20:00DS 6/36 OS -30.00DS 6/36

- Subjective refraction: OD -18:00DS 6/36 OS -20:00DS 6/60 OU 6/24

Considering the clinical findings, the patient was referred to the Low Vision Clinic.

### Low Vision Assessment

- Visual acuity using Log Mar Chart  
Distance: OD 1.76LogMar (Equivalent of 6/300) OS 1.99LogMar (Equivalent of 6/480)  
Near: OD 0.80M@23cm; OS 2.00M@20cm
- Contrast sensitivity OD and OS: 0/25@1m; 0/25@0.75m; 22/25@0.50m

- **Low vision calculations:**

$$\begin{aligned} \text{Distance Magnification:} \quad \text{Equivalent Ratio (ER)} &= \text{Actual VA/Desired VA} \\ &= 300/36 \quad = 8.3X = 8X \end{aligned}$$

- For Low vision therapy, the better-seeing eye was used.
- Patient's OD was trained using the 8X handheld telescope and was able to read 6/9.
- Near: Patient's vision was good at near, he was able to read N8 OD and N10 OS. He reads better with the material closer to him. Hence, no aid was prescribed for him at near.

### Treatment Plan

- Spectacle correction for regular wear was issued (OD -18:00DS and OS -20:00DS)
- 8X handheld telescope was recommended to enable him to read 6/9 at distance with the OS.
- The patient was requested to come back after 6months for follow up.
- He was counseled on his condition.

### CASE 2

A sixteen-year-old boy, who has been visiting Aminu Kano Teaching hospital (where he was referred from) since he was five years old and was diagnosed with MFS. The report had it that he had done several refractions in the past which revealed subnormal vision that hindered adequate visual participation in the classroom. He had been counseled on his eye/visual condition and how assisted devices and lifestyle modification can help him function as independently as possible.

### Clinical Examination Results

- External Exam: Long slender face, very tall and slender body build with long arms, could not stand or work without the aid of crutches.
- Visual Acuity (VA):
 

|         |              |    |      |
|---------|--------------|----|------|
| OD 6/60 | With Pinhole | OD | 6/18 |
| OS 6/60 | With Pinhole | OS | 6/36 |
- Pupils: No anisocoria and no relative afferent pupillary defect detected.
- Motility: Ocular motility full and free OU.
- Anterior segment: Inferiorly subluxed lenses OU.
- Anterior Chamber: Angle was deep OU and there was no lens apposition to the cornea in either eye.

- Dilated funduscopy: Posterior segment was normal OU with no peripheral retinal degeneration.

**Objective refraction (Retinoscopy):** OD + 11.25DS 6/18 OS +11.75DS 6/36

- Subjective refraction: OD = +7.5DS 6/9; OS +7.5DS 6/24; Aided Near VA: OU: N 36

Considering the clinical findings, the patient was referred to the Low Vision therapy clinic for near correction.

### Low Vision Assessment at Near

- Visual acuity using Log Mar Chart:

Near: OD 1M@12cm OS 2.00M@10cm

- Contrast sensitivity OD and OS: 7/25@1m; 5/25@0.75m; 9/25@0.50m;

The same result was obtained for each eye and the contrast was reduced.

- **Low vision calculations:**

Distance: (ER) = Actual VA/Desired VA

For low-vision therapy, the better-seeing eye is used for the calculation.

Equivalent Ratio (ER) = Actual VA /Desired VA = 1M/0.5M = 2X (Near)

Equivalent Viewing Distance (EVD) = Eye to image distance / Enlargement ratio

$$EVD = 12/2 = 6\text{cm}$$

Equivalent Viewing Power (EVP) = 1/ EVD = 1m/6cm = 100cm/6cm

$$=16.66\text{D} = 16.5\text{DS}$$

In all, they came for their follow-up and training for three different occasions according to the low vision clinic's protocol and the VA remained the same.

### Treatment Plan:

- Spectacle correction for regular wear was issued for his distance correction (OD = +7.5DS and OS +7.5DS)
  1. 4X handheld illuminated magnifier for near work
  2. 16.00 DS Spectacle Magnifier for reading at 6cm.
  3. The patient was asked to return after 6 months for follow-up.
  4. He was counseled on his condition.

He was supposed to use 2X Magnifier (Handheld) to read 0.63 or N5 from the calculation but he read N24 which was not sufficient for near vision. Hence, a 4X illuminated handheld magnifier was given to him which enabled him to read N5.

## Discussion

Low vision history taking is a little different and more comprehensive than the normal routine case history. It provides information on the patient's goals as well as the need for mobility, medical rehabilitation and training, and psycho-social counseling. This is an important part of the low vision assessment and provides an opportunity for the patient and clinician to get to know each other. With the myriad of low-vision devices available, having a goal-oriented low-vision history ensures that the practitioner can demonstrate the devices that are most appropriate for the patient's stated needs and desires. The patients under review were students, hence performing classroom activities was their main goal.

When examining someone with low vision, log MAR charts are used as they give better measures of acuity for both far and near distances. Its advantages include a geometric progression of letter sizes, an equal number of letters per line, a scoring system, portability, and allowing testing distances to be varied.<sup>14</sup> In both cases, the subjective refraction results were recommended for both patients as it improved their distance vision (from 3/60 to 6/36 and 6/60 to 6/9 respectively). Low vision assessment was later carried out on them: Case 1 (far and near distances) and Case 2 only at near distance.

Contrast sensitivity is the ability to detect objects at low contrast.<sup>15</sup> It is an important test carried out to consider if light and other non-optical devices will be prescribed for the patients for visual enhancement. The Lea Contrast Sensitivity chart was used to carry out the test. Contrast sensitivity of both patients was reduced due to some conditions which affect people living with MFS, these include high refractive error, ectopia lentis, and aortic root aneurysm.<sup>16</sup> Similarly, the visual fields were also poor from the ophthalmologist's reports which may also be due to the same reasons above.

The magnification power and focal distances of optical low vision devices were calculated using the enlargement ratio, which is equivalent to the magnification using the formula:

$$\text{Equivalent Ratio (ER)} = \frac{\text{Actual or Reference VA at far or near/Desire or Goal VA at far or near}}{\text{as the case may be.}}$$

In case 1: an 8X handheld telescope was recommended as it improved the vision from 3/60 to 6/9, while he brought his reading materials/books closer to the eyes to enable him read. 4X handheld illuminated magnifier for near work and 16.00 DS Spectacle Magnifier for reading at 6cm were also recommended for the patient in Case 2. This agrees with the studies reported in India, Ghana, and Nigeria.<sup>17,18,19</sup> The

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two patients attended their follow-up/training for three different appointments according to the low vision clinic's protocol. This was to make sure they were able to learn how to focus, fine-tune, and properly handle the devices to maximize vision.

Genetic counseling services were included in the protocol to help the families understand their diagnosis and the future impact of consanguine marriage which is also part of their culture. This will help to reduce the incidence of MFS since it is hereditary.

Low-vision devices are prescribed to help patients

maximize their vision and live independently. This varied from patient to patient such that no two cases were the same due to the function and need of the patient. Case 1 needed a Telescope for his distance vision while he was counseled to bring his reading material closer to the eye. Unlike case 2, that had no need for distance low vision devices but rather near devices. There was no indication by the ophthalmologist for cataract surgery for ectopia lentis for the two cases which made low vision assessment and management a significant procedure for the management of MFS.

### Conclusion and Recommendations

Marfan syndrome requires multidisciplinary care; therefore, Optometrists need to co-manage such patients with other health practitioners. Low vision and rehabilitation maximize visual functioning and thus the need to effectively implement assessment of functional vision in the management of MFS. In line with that, genetic counseling services should be included as this will help families understand their diagnosis and make better-informed decisions about future family planning as well as screening other family members.

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