

## CASE REPORT

# Marfan syndrome in a Ghanaian male: The diagnostic challenges

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## Key Clinical Message

Marfan syndrome (MFS) is an autosomal dominantly inherited condition that has varying phenotypic expressions. This case report describes one such African patient, from Ghana, who had typical clinical and imaging traits of MFS but was first diagnosed incidentally at the age of 23 years. In this report, we explore the challenges of early diagnosis in this population.

## KEYWORDS

Ghanaian, Marfan syndrome

## 1 | INTRODUCTION

Marfan syndrome (MFS) is an inherited connective tissue disease that occurs following an autosomal dominant gene mutation in the fibrillin-1 gene (FBN1).<sup>1</sup> The protein produced by this mutated gene is an essential component of most connective tissue and being structurally abnormal, results in a wide range of specific ophthalmological, skeletal, and cardiovascular abnormalities that characterize MFS.<sup>1</sup> The disease was discovered when Antoine-Bernard Marfan diagnosed a 5-year-old named Gabrielle who presented with skeletal signs.<sup>2</sup> Current studies estimate

the prevalence of MFS at 6.5/100,000.<sup>3</sup> Experts, in 1986, at Berlin created the first clinical criteria for diagnosing MFS known as the Berlin Nosology.<sup>2</sup> A new criterion was detailed in 1996 (Ghent I criteria) on account of high false positive results. In 2010 the Ghent 1 criterion was modified to include specifically FBN1 mutation, aortic root dilatation, and ectopic lentis as the mainstay of MFS diagnosis (Ghent II). The formulation of this nosology was essential for the avoidance of inconclusive diagnosis and differentiation from conditions presenting with similar manifestations.<sup>1,2</sup> Clinical manifestations of this disorder include cardiovascular, ophthalmic, musculoskeletal,

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craniofacial, and cutaneous abnormalities.<sup>4</sup> Among the cardiovascular manifestations, aortic dilatation, and mitral regurgitation from mitral valve prolapse (MVP) occur frequently.<sup>5,6</sup>

In this case report we describe the incidental finding of a young African male with classic Marfan's syndrome but initially diagnosed at the age of 23 years. We further explore the barriers to early diagnosis in our part of the world.

## 2 | CASE REPORT

The patient presented to the cardiologist as a referral from the general practitioner, with a 3-month history of palpitations. The palpitations were not associated with dizziness or loss of consciousness but associated with dull left-sided chest pain which was non-radiating and not associated with exertion, meals, breathing, or movement. He had progressively worsening fatigue on moderate exertion of a 2-month duration but had no associated cough, dyspnea, or pedal swelling. Prior to this, he had no significant medical complaints except for frequent visits to see the optician over the past 5 years resulting in the need to use spectacles.

He admitted to blurred vision for 5 years which had worsened over the period requiring the use of glasses for short-sightedness but had no noticeable eye discoloration, redness, dryness, or tearing.

Since his adolescence, he had been noticeably more flexible than his peers, allowing him more fluidity as a drummer. He had no other joint pain, rash, or dislocation.

There were no abdominal or neurological symptoms of note.

The patient had a strikingly tall physique and was the tallest member of his family. There was no history of heart disease among the immediate family; however family members were bespectacled and had no distinguishing physical features.

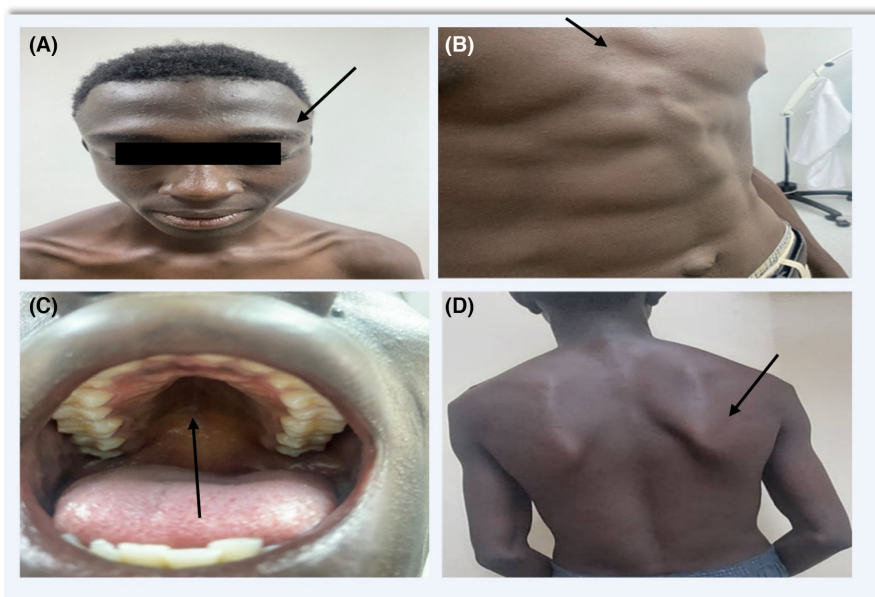
He denied the use of alcohol, tobacco, or illicit drugs and was currently studying computer science at the university.

### 2.1 | Physical exam

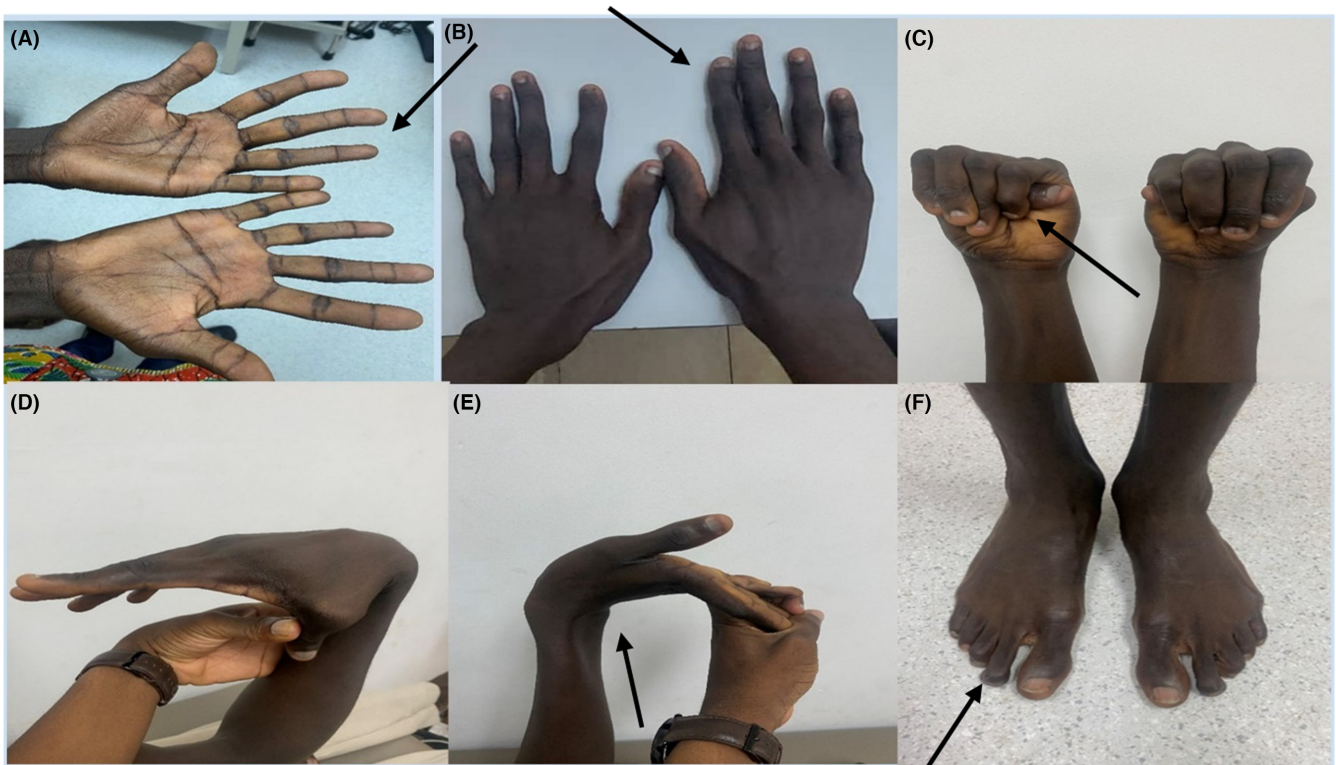
The facial features revealed a narrow, anteroposteriorly elongated cranium (dolichocephaly) and leptoprosopic (elongated) face with malar hypoplasia (underdevelopment of the cheekbones), mandibular retrognathia (small lower jaw), (Figure 1A) and macrostomia (Figure 1C). There was proptosis of the right eye, prominent eye ridges and no ophthalmoplegia. Beighton's score was 8/10.

Intraoral examination revealed mild overcrowding of his dentition with good oral hygiene but a high-arched palate (Figure 2C).

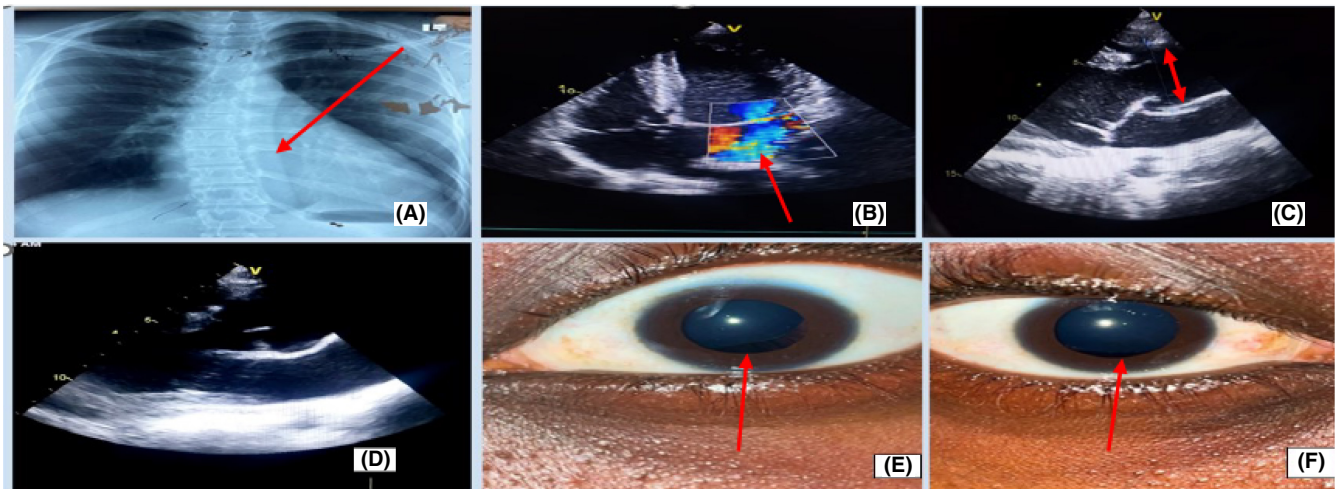
He had disproportionately long arms and legs (dolichostenomelia) with an arm span of 201 cm, a height of 182 cm and an index of 0.905 (positive ape index). His fingers were long and spidery (arachnodactyly) (Figure 2A,B), and Walker wrist sign and Steinberg thumb sign (Figure 2C) were positive. The patient demonstrated increased joint laxity (Figure 2D,E). He was flat-footed with elongated toes (Figure 2F). There was mild swelling of the proximal and distal interphalangeal joints with no tenderness noted. Also noted was mild kyphoscoliosis of the thoracic spine, with a stooped posture. There was a prominent right-sided scapula (Figure 1D) with pectus excavatum (Figure 1B).



**FIGURE 1** Sample images to demonstrate patients' physical appearance. Dolichocephaly, leptoprosopic type malar hypoplasia, mandibular retrognathia, and downward slanting external palpebral fissures (A). Pectus excavatum (B). Macrostomia, high-arched palate, and mildly overcrowded dentition (C). Prominent right-sided scapula and scoliosis (D).



**FIGURE 2** Sample images to demonstrate patient's physical appearance. Long and spidery fingers (A and B). Positive Steinberg thumb sign (C). Joint laxity (D and E). Flat foot and elongated toes (F).



**FIGURE 3** Sample pictures from imaging and eye assessment done. Scoliosis and Cardiomegaly (A). Mitral regurgitation (B). Dilated aortic root and prolapse of anterior valve leaflet (C and D). Ectopia lentis for right and left eye (E and F), respectively.

The patient had a regular pulse at the time of the exam with an undisplaced apex beat and normal heart sounds but had a Grade 2 mid-systolic murmur with no radiation. He had a normal respiratory, abdominal, and neurological exam.

Chest x-ray showed scoliosis and cardiomegaly (cardiothoracic ratio of 0.76) with normal lung fields (Figure 3A) and the electrocardiogram showed sinus rhythm with high voltages and normal ST/T waves.

The Echocardiogram report revealed moderate anterior MVP (Video S1) in the A2 region with moderate eccentric mitral valve regurgitation (Figure 3B). There was mildly dilated left ventricular and atrial chamber size and a severely dilated aortic root (5.9 cm) and 1.98 when indexed to his body surface area (BSA) (Figure 3C,D). A plain CT scan of the chest confirmed severe aortic root dilatation.

The patient was referred to the ophthalmologist and the findings were as follows:

**Visual acuity:** This was found to be myopic in both eyes and worse in the left eye. The unaided vision was 6/18 and 6/60 in the right and left eyes, respectively.

**Cornea:** Found to be thinner than the average. Central corneal thickness was 457 and 476 in the right and left eyes respectively.

**Iris:** There were no transillumination defects noticed in either eye.

**Lens:** The patient had ectopia lentis in both eyes. The lens in the right eye was subluxated superiorly and superonasally in the left eye (Figure 3E,F).

The patient was started on oral Carvedilol 6.25 mg daily by the cardiologist and is currently on 3 monthly reviews with both the cardiologist and the ophthalmologist. The palpitations have since subsided and the easy fatigue improved.

### 3 | DISCUSSION

We described the case of a young African male diagnosed with classical MFS syndrome using the Ghent 11 nosology. The diagnosis of MFS using this criterion involves the observation and imaging of distinct ophthalmologic, musculoskeletal, and cardiovascular abnormalities.

MFS is an autosomal dominant disorder with a mutation in the FBN 1 gene resulting in a broad spectrum of phenotypic expressions. Typically, the condition presents with skeletal, ophthalmologic, and cardiovascular abnormalities. Due to the varying extent of phenotypic expression and rate of progression, the time of diagnosis varies and requires a high index of suspicion especially in the younger age groups.

In a study by Faivre et al., the median age at diagnosis was 6.5 years in a study that looked at 320 patients less than 18 years of age. In this study, 14% had moderate MFS, 14% severe MFS, and 35% had probable MFS with skeletal abnormalities, showing up as positive thumb and wrist signs (83%) and high arched palate (70%).<sup>7</sup>

The diagnosis in childhood may be missed as most diagnostic features are age-dependent hence the Ghent 11 Criterion may have limited application in this population.

The Ghent 11 nosology for diagnosis among adults includes the detection of any of the following:

1. Aortic root dilatation and ectopia lentis
2. Aortic root dilatation and FBN1 mutation

3. Aortic root dilatation and systemic abnormalities
4. Ectopia lentis and FBN1 defect in a known<sup>2</sup>

In low and middle-income countries (LMIC) the diagnosis of MFS will most likely be clinical using the Ghent 11 criterion. This is due to the general unavailability/inaccessibility of genetic testing to the majority of patients. There may therefore be a diagnostic challenge in patients who have features that are not very diagnostic.<sup>1</sup> This is demonstrated in the limited availability of published data in the age range among people of African descent.

MFS is sporadic in 25%–30% of patients having no family history. These patients tend to present with severe manifestations of the disease and have worse outcomes.<sup>8</sup> In this case, neither the two siblings nor the mother had any typical features of MFS.

### 4 | OCULAR MANIFESTATIONS

The FBN 1 gene produces microfibrils that support the lens and keep it in position, the microfibrils are also found in the iris, Schlemm canal and cornea. The defect in these microfibrils is associated with ectopia lentis (lens subluxation), the development of miosis, wider corneal diameter, and pupillary hypoplasia.<sup>9–11</sup>

Myopia is the commonest ocular manifestation of the disease, being associated with elongation of the globe with 60% of these having concomitant lens dislocation.<sup>12,13</sup> The incidence of ectopia lentis in MFS varies from 30% to 72% in different studies it may however be first seen in the second decade of life.<sup>14–16</sup>

Less common ocular manifestations are early cataracts, retinal detachment, and glaucoma. Ocular abnormalities such as myopia, astigmatism, anisometropia, ectopia lentis, and retinal pathologies may result in amblyopia. The earlier the onset and the longer it goes untreated, the harder amblyopia is to correct.<sup>17</sup> Therefore, patients with MFS should be screened annually and examined for refractive errors and other ocular pathologies. Eyeglasses are the first step to correct blurred vision caused by subluxated lenses.<sup>18</sup> Removal of a dislocated lens and the possible insertion of an artificial one may be considered but not usually required.<sup>14</sup>

The indications for surgery in lens dislocation are inability to achieve good corrected visual acuity, risk of amblyopia in children, posterior dislocation of the lens into the vitreous cavity, anterior dislocation of the lens with or without secondary glaucoma, impending to complete lens dislocation, lens-induced glaucoma or uveitis, and cataract.<sup>19</sup>

Glaucoma may develop at a later age and may be a result of phacolytic glaucoma from the dislocation of a matured lens.<sup>20,21</sup>

## 5 | SKELETAL ABNORMALITIES

The system with the largest variety of clinical manifestations is the skeletal system though it scored less in the Ghent nosology compared to cardiovascular or ophthalmologic manifestations. This is likely due to their non-specific nature and the possibility of varied differential diagnoses to explain their presence.

Skeletal abnormalities include craniofacial, ribcage, limb, and vertebral defects with differing levels of impact on the patient. In one study, typical foot deformities such as hindfoot deformities were not limiting compared to controls although the cases had longer and narrower feet.<sup>22,23</sup> In the same study, Lindsey et al. note however the social limitation of patients with MFS finding well-fitted shoes on account of the associated claw and hammer toes.

Two major chest wall abnormalities have been documented in MFS. Pectus carinatum also called the pigeon chest, is thought to be more specific for MFS<sup>8</sup> and scores higher in the Ghent nosology than the pectus excavatum (funnel chest). The impact of chest wall abnormalities on cardiovascular signs and symptoms in patients with MFS included increased chest pain attributable to mediastinal compression. The significance of this can be evaluated by imaging studies.<sup>24,25</sup> For, these patients, however, cosmesis tends to be the more common reason for surgical intervention.<sup>26</sup>

Scoliosis is the lateral deviation of the vertebra with axial rotation and vertebral body compression.<sup>27</sup> Management remains a challenge with outcomes of bracing, (the mainstay and initial treatment option) showing varied outcomes and impact on rates of progression.<sup>28–30</sup> Factors that affect the rate of progression rates adversely include earlier age of onset, intolerance of braces, and increases in the rigidity of the curve.<sup>31,32</sup> Surgical interventions are reserved for severe scoliosis.

The usefulness of craniofacial abnormalities in making a diagnosis is subjective, being dependent on the experience of the examiner. These features include enophthalmos, down slanting of the external palpebral fissures compared to the internal, malar hypoplasia, and dolichocephaly (long skull).<sup>33</sup> Other orthodontic abnormalities, not incorporated into the Ghent nosology are a high-arched palate and dental overcrowding.

It is important to note that despite the reduction in the bone mineral density reduction noted in the wrist, hip, and femur of patients with MFS, its impact on the occurrence of fractures is not fully understood.

## 6 | CARDIOVASCULAR MANIFESTATIONS

MVP and mitral valve regurgitation are common CV complications in patients with MFS. Estimated at 40%–68% in adults compared to about 30%–38% in children with MFS and 1%–2% in the general population.<sup>34</sup> Aortic regurgitation occurs following the dilatation of the aortic root and also abnormal valve cusp structure leading to aortic valve prolapse. Moderate to severe AR has been documented in a study to be an independent predictor of CV events including dilatation, dissection, and the need to have surgical intervention.<sup>35</sup> Eventually, the patient with MFS dies from aortic complications such as dissection or heart failure following volume overload from long-standing aortic and mitral regurgitation.

In the management of MFS, it has been documented that the impact of prophylactic pharmacotherapy with B Blockers on the risk of complication is not insignificant.<sup>36</sup> Randomized controlled trials (RCTs) that compared the impact of ARBs and BB found no statistically significant difference in the rates of clinical outcomes such as aortic regurgitation or the need for surgery. Some studies have found an added benefit of concomitant use of BB and ARB therapies.<sup>37,38</sup>

## 7 | SCREENING FOR MARFAN SYNDROME IN GHANA

The reporting of diagnosed cases of MFS on the continent has been low. The factors that fuel this pattern are likely multifaceted and may include low detection rates, varying severities in disease expression making the recognition difficult and the largely inaccessible means of genetic testing should the need for that in diagnosis be required.

There is no single laboratory test that completely makes the diagnosis of MFS, and other conditions also have the mutation in the FBN 1 gene. The diagnosis therefore requires a thorough physical examination, a family history of the condition and investigations that assess the possible involved organ systems.<sup>32</sup>

In Ghana, the detection of the clinical features may be the most reliable means of screening for MFS. A high index of suspicion by a well-trained clinician will help initiate further testing to make the diagnosis. This is an inexpensive means of initial detection of MFS but will probably be most effective in patients who have florid phenotypic features. By default, the use of this means alone excludes detection in neonates and children in whom these may be obscure.

Assessment of the extent of organ system involvement in MFS is another crucial part of diagnosis and another potentially significant limitation in Ghana. Cardiovascular

involvement requires the use of echocardiography and possibly CT scanning to not only detect but to continually monitor patients and diagnose life-threatening complications such as aortic dissection, should they occur. Across Ghana, these services are limited to a few large cities and so in most instances geographically, and very often financially inaccessible to patients.

Trained cardiologists, ophthalmologists, orthopedic surgeons, and various other specialities who form part of the multi-disciplinary team needed to care for the patient with MFS, are inadequate in number and found in only a few specialized centres.

The need for genetically detected FBNI gene, by the Ghent nosology, is limited to special situations and therefore not mandatory in all cases of MFS. Even though genetic testing is less available than diagnostic tests in Ghana, it should not be a limitation in the detection of MFS especially in patients with a typical phenotypic appearance and in whom some further evaluation can lead to a diagnosis.

## 8 | CONCLUSION

This case report shows how typical MFS presents and how the diagnosis can be made with a high clinical index of suspicion and some level of diagnostic testing. This is the first write-up on a Ghanaian patient with MFS and highlights the fact that diligent assessment may uncover several other patients in our society, with possibly, many of them not requiring genetic testing to make the diagnosis. Following this we have evaluated the limitations in the Ghanaian society that may fuel the low rates of detection. In this report we emphasize on the meticulous use of the physical exam primarily to detect possible MFS, and then supportive tests for the extent of organ involvement and further monitoring in low-resource settings.

### AUTHOR CONTRIBUTIONS

**Aba Folson:** Conceptualization; investigation; supervision; writing – original draft; writing – review and editing. **Kwabena Oteng Agyapong:** Writing – original draft; writing – review and editing. **Dzifa Dey:** Writing – review and editing. **Philip Eghan:** Conceptualization; writing – original draft. **Billey Quay:** Writing – review and editing.

### ACKNOWLEDGMENTS

The authors would like to thank the patient for his cooperation during the assessment and writing of this report.

### FUNDING INFORMATION

There was no funding for this work.

### CONFLICT OF INTEREST STATEMENT

The authors have no conflict of interest.

### DATA AVAILABILITY STATEMENT

The data that support the findings of this study are openly available on <https://doi.org/10.22541/au.169865516.64105131/v1>.

### CONSENT

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

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## SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

**How to cite this article:** Folson AA, Agyapong KO, Dey D, Eghan P, Quaye B. Marfan syndrome in a Ghanaian male: The diagnostic challenges. *Clin Case Rep.* 2024;12:e8494. doi:[10.1002/ccr3.8494](https://doi.org/10.1002/ccr3.8494)