Case report

A CASE REPORT OF MARFAN'S SYNDROME AND A REVIEW OF THE LITERATURE

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ABSTRACT

BACKGROUND: Marfan's syndrome is an autosomal dominant, multisystemic disorder of the connective tissue with variable inheritance patterns. Its cardinal features affect mostly the cardiovascular, ocular and skeletal systems. It is panethnic with no sex predilection and may be diagnosed prenatally, at birth, childhood or well into adulthood.

OBJECTIVE: This article aimed at enlightening dental professionals on the presentation and early recognition of patients with Marfan's syndrome.

CASE REPORT: An 11-year old female previously undiagnosed of any syndrome, presented to the dental clinic with a resolving dental abscess on tooth 26 and dental caries of 16, 36 and 46. She showed features of skeletal disproportion, arachnodactyly (long slender fingers with prominent finger joints), flat feet and pectus carinatum (pidgeon chest). Facial examination revealed a convex profile and elongated face, there was no dental crowding. Her dental abscess was managed by extraction with antibiotic cover while other carious lesions were restored. She was then referred to an ophthalmologist and a cardiologist for further examinations and investigations, which revealed she had features of Marfan syndrome.

CONCLUSION: This case report demonstrates the early recognition of signs and symptoms of this syndrome and the referral of the patient to other professionals for management to prevent severe life threatening complications associated with the syndrome.

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INTRODUCTION

Marfan's syndrome is an autosomal dominant, multisystemic disorder of the connective tissue. It is associated with mutations in fibrillin and occasionally the TGFBR1 or 2.[1,2] It is a rare condition, which has a worldwide incidence of 7-17/100,000 births,[3] and a prevalence of approximately 1 in 5000 births [4] with 26.0% of cases having spontaneous mutations i.e no family history of the disease[5]. It is panethnic with no sex predilection and may be diagnosed prenatally, at birth or well into adulthood.

The syndrome affects mainly the skeletal, ocular and cardiovascular systems. Some characteristic features include: a) skeletal; a tall and thin body, with long limbs, skeletal disproportion, arachnodactyly, hyperflexibie joints, pectus deformities (carinatum or excavatum), and sometimes scoliosis: b) ocular; lens dislocation and myopia: c) cardiovascular; progressive aortic dilatation associated with aortic valve incompetence, mitral valve prolapse and incompetence. Other features include high arched palate with dental crowding and malocclusion, skin striae distensae, recurrent hernia or recurrent pneumothorax.

Fibrillin-1 gene mutation on chromosome 15q21.1 was identified as the cause of the syndrome [6] although molecular testing is not as diagnostically useful as hoped. The Ghent nosology is used in the clinical diagnosis of the syndrome but may be unreliable in children, thus evaluations are suggested to be repeated periodically at ages 5, 10, 15years till 18. [1] Differential diagnosis for tall young persons with Marfan-like skeletal features includes homocystinuria, Beals syndrome, Marshall-Stickler syndrome, Ehlers-Danlos syndrome and MASS phenotype.

CASE REPORT

An 11-year-old female patient reported to the Paediatric Dental Clinic, University of Benin Teaching Hospital, Benin, Nigeria with a complaint of toothache of two weeks duration and facial swelling of one day duration on her upper right quadrant. Medical history revealed that patient had diminished vision for which an ophthalmologist was consulted. The family history of the patient was noncontributory.

On general examination, the patient had tall stature with long, slender limbs, thin body habitus with increased arm span-to-height ratio (166cm: 161cm) [Fig 1], long, slender spidery fingers (arachnodactyly) with prominent finger joints (Fig 2), flat feet and pectus carinatum. Facial examination revealed a convex profile, elongated face, slight facial asymmetry as a result of a resolving dentoalveolar abscess.

Intraorally, high arched palate there was mild spacing in the upper and lower anterior segments (Fig 3). She had permanent incisors, laterals, canines, premolars and first permanent molars present. There was a dental abscess on tooth 26, while 16, 36 and 46 were carious. Dental treatment rendered to the patient involved the extraction of abscessed 26 under local anaesthesia with antibiotic cover (Patient was placed on antibiotics and analgesics orally before the procedure), composite restorations were done on the carious 16, 36 and 46.

The patient was then referred to the ophthalmologist for ocular examination, which revealed transparent and intact lens (no lens dislocation). Although the patient had normal macula she was observed to be myopic with a visual acuity of 6/18; 6/24 in the right and left eyes respectively. Pinhole visual acuity was 6/6 in both eyes with intraocular pressures of 20 and 22mmHg. She showed early signs of glaucoma with pathologically cupped discs with a cup to disc ratio of 0.65 in the posterior segment. There were features of dilator pupillae hypoplasia in both eyes with irregular dilation of the right eye and poor dilatation in the left eye. Refraction revealed a refractive error that was corrected with -1.25 dioptres sphere in both eyes bringing her corrected visual acuity to 6/5. Duochrome test: saw letters on red background clearer than green. Cardiovascular examination revealed murmurs on auscultation but parent refused echocardiography to be done. Parental

counseling is still ongoing. The clinical findings following skeletal and ophthalmological assessment were suggestive of Marfan syndrome.

Figure 2: Showing long slender fingers (arachnodactyly) in patient

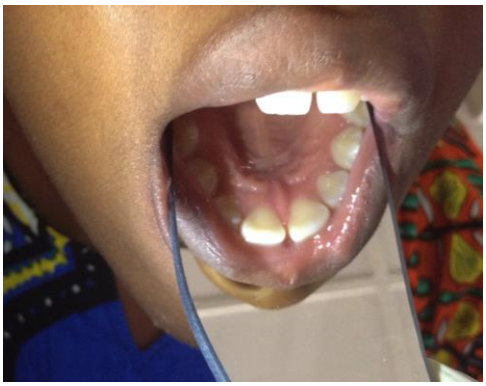

Figure 3: High arched palate in patient with Marfan syndrome.

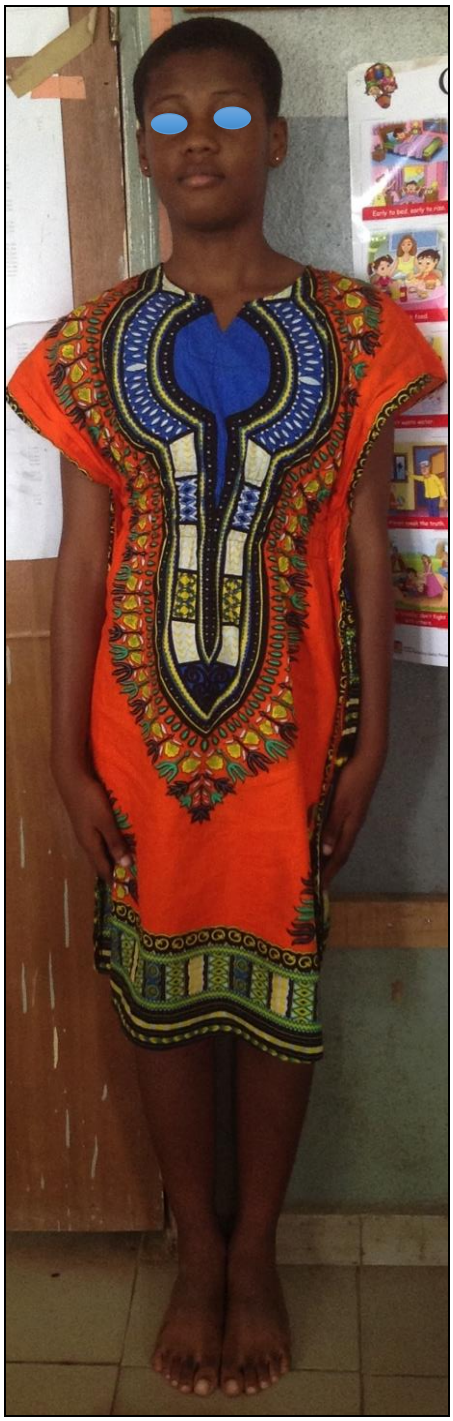

Figure 1: Showing skeletal disproportion and pectus carinatum

DISCUSSION

The period of childhood growth and development is a very challenging one as it involves intense physical and intellectual demands, self-assessment, and judgment by peers[7]. These processes may be affected by a diagnosis of Marfan syndrome. In managing a patient with Marfan syndrome the major goal is to prevent life threatening complications through early diagnosis and treatment, which may in turn improve prognosis and prolong life.[8]

Cardiovascular complications are the major cause of morbidity and mortality with aortic aneurysm which could result in aortic dissection, rupture or both being the most life threatening.[9] Patients with Marfan syndrome who have cardiac or valvular involvement or who have had heart surgery may be at increased risk for bacterial endocarditis thus to prevent it antibiotics may be needed prior to dental or surgical procedures. Regular monitoring of the heart and aorta using imaging techniques should be done to evaluate the size, determine dilation and its progression.[9] Beta blockers have been suggested as drug therapeutic treatment for these patients early in the course of the disease before advanced dilation of the aorta occurs to achieve good results.[10] To prevent cardiovascular complications exercise restriction is a preventive measure and patients should avoid exhausting exercises and activity.[11]

Ocular assessments should be carried out regularly. Identifying ocular pathologies early such as glaucoma, lens dislocation and refractive errors and managing them will help preserve and maximize visual function. Individuals with the Marfan syndrome are at increased risk for glaucoma, cataract formation, and retinal detachment, even in the absence of ectopia lentis.[7] Myopia is a common feature in these patients with the impairment ranging from mild to severe.[8]

Skeletal abnormalities should alert the dentist or physician to the typical physical appearance of those with Marfan syndrome; these include the characteristically tall and thin physique with disproportionately long slender arms, legs and toes, the arm span is greater than the height. Hyperflexibility of joints such as the wrist, carpals and temporomandibular joints. The sternum may protrude (pectus carinatum) or indent (pectus excavatum) with or without abnormal spinal curvatures (kyphosis, scoliosis, thoracic lordosis or kyphoscoliosis). Early evaluation and routine checkup by an orthopedic surgeon is indicated in these cases to prevent severe skeletal deformities.

Oral features include a typically long and narrow face, high arched palate and malocclusion presenting with crowding of the teeth. Severe periodontitis has also been reported [12] these should be managed early by the paedodontist and orthodontist to avoid teasing and development of low self-esteem. In children with Marfan's syndrome, a preventive management may include using battery-operated brushes, adapted manual toothbrushes, fluoride products, and antibacterial mouth washes and toothpastes should be performed.[13]

Sensitivity to cosmetic issues related to Marfan syndrome should be discussed with resultant solutions as regards surgeries. A role model or age-matched peer with the Marfan syndrome to discuss frustrations and opportunities with an affected child [12] may go a long way to helping the child cope with the disease and reduce emotional impact.

In conclusion, this case report demonstrates the early recognition of signs and symptoms of this syndrome and the referral of the patient to other professionals for management to prevent severe life threatening complications associated with the syndrome. As in this case, the dentist may be the first port of call for this patient and a high index of suspicion is crucial to its diagnosis. The dental findings may aid in early diagnosis of this syndrome and it may aid in prompt multidisciplinary treatment reported.

Conflict of Interest: None declared

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