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MARFAN SYNDROME – A CASE REPORT

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Abstract: Marfan syndrome is a spectrum of disorders caused by a heritable genetic defect of connective tissue that has an autosomal dominant trait. In this syndrome, defect is isolated to FBN1 gene. The main features of Marfan syndrome include disproportionate skeletal growth with dolichostenomelia, ectopia lentis, fusiform and dissecting aneurysms of the aorta. Craniofacially features include dolichocephaly, prominent supraorbital ridges, long face, downslanting palpebral fissures, hypoplastic malar eminence, high palatal vault, mandibular prognathism. This report describes a case of 26 year old male patient with typical orofacial findings in Marfan syndrome with extensive review of literature.

Index Terms - Marfan syndrome, genetic defect, dolichostenomelia, mandibular prognathism

I. INTRODUCTION

Marfan syndrome(MFS) is also known as Marfan – achard syndrome. It is caused by a heritable genetic defect of connective tissue, the defect is isolated to FBN1 gene on chromosome 15, bands q15–q23[(1)]. MFS is a rare pleiotropic disease, accounts for 1 in 5000 live births[(2)]. Diagnosis of MFS is made on clinical basis. The Ocular, cardiovascular, and musculoskeletal abnormalities are considered the "classic triad" of MFS[(3)]. It also commonly affects lungs. Affected individuals often are tall and slender, have arachnodactyly, scoliosis, and either a pectus excavatum, pectus carinatum, or ectopia lentis in eyes[(4)]. Common orofacial features include dolichocephaly, enophthalmos, downward slanting palpebral fissures, malar hypoplasia, maxillary retrognathia, mandibular retrognathia, skeletal class II malocclusion, and hypermobility of the temporomandibular joint. Dentally, long narrow maloccluded teeth, large positive overjet, posterior crossbites, periodontal disease are also characteristic of MFS[(3)]. This paper describes a case of 26 year old male patient diagnosed as MFS based on craniofacial features with review of literature.

II. Case report:

A 26 year old male patient presented with the chief complaint of difficulty in closing the mouth since childhood. His past medical history reveals, history of tuberculosis 2 years back and taken medication for 6 months. His personal history reveals history of delayed milestones of the child. Patient also had a habit of mouth breathing since childhood. Family history reveals history of consanguineous marriage of his parents. Patient had one elder sister, she died at the age of 1 year and 1 younger sister, she died within 1 week of birth due to systemic complications.

On General examination, Thin built, short stature and poorly nourished (Height - 159 cm tall and weighed - 35 kgs), Difficulty in speech and in pronouncing some words, learning and listening difficulties present.

On Extraoral examination, Gross facial asymmetry noted, long face, Dolicocephaly, prominent supraorbital ridges, hypoplastic malar eminences, broad nose, protruding ears, Pectus excavatum, arachnodactaly, pes planus, hyperextensibility of joints.(Figure 1-3)

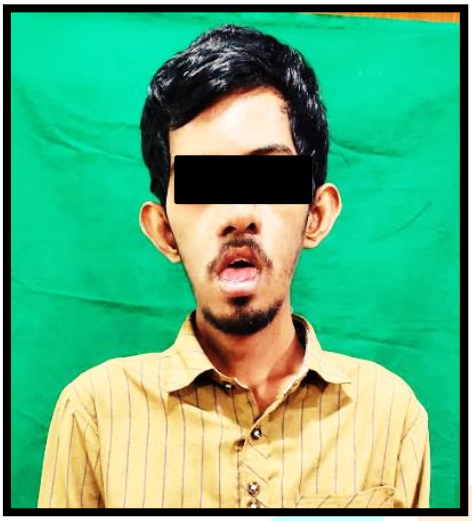

Figure 1: Profile photo

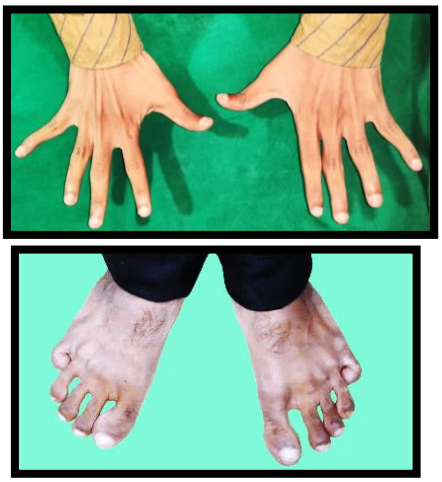

Figure 2: Arachnodactaly, Figure 3: Pes planus

On Intraoral examination, DC: 37, Root stumps: 47, Missing: 48, Labially placed: 23, V maxillary arch, high arched palate, U shaped mandibular arch, Mandibular prognathism, anterior open bite and macroglossia with protrusion of tongue. (Figure 4)

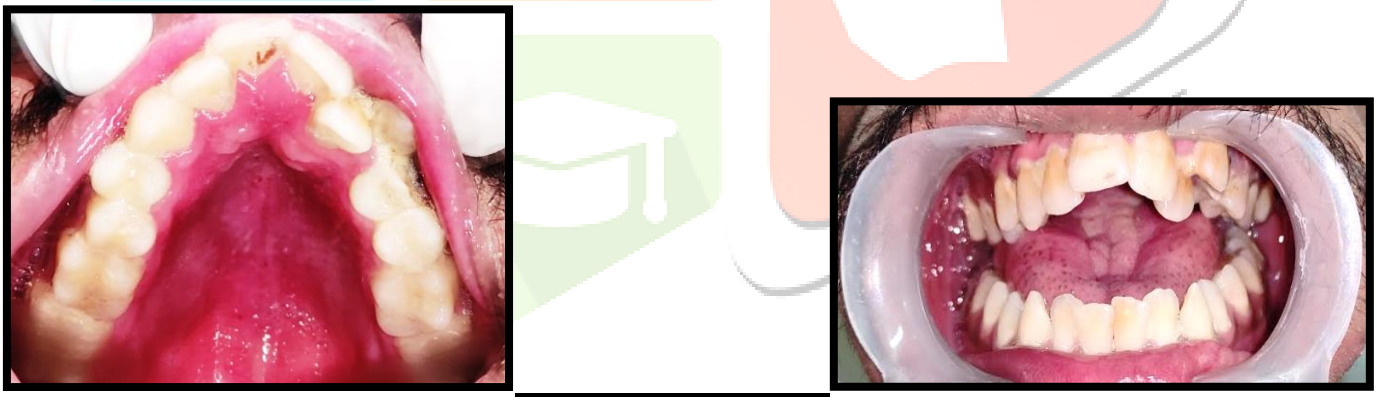

Figure 4: Intra oral pictures

By correlating the patient's history and clinical findings, we arrived at a provisional diagnosis of Marfan syndrome.

III. Investigations:

OPG reveals, Zone 1 reveals number of teeth present are 32. In 47, 48, complete loss of coronal structure with remnants of radicular portion. Suggestive of Root stumps in relation to 47,48. Zone 4 reveals thinning of coronoid process on right and left sides and deepening of sigmoid notch on left side. (Figure 5)

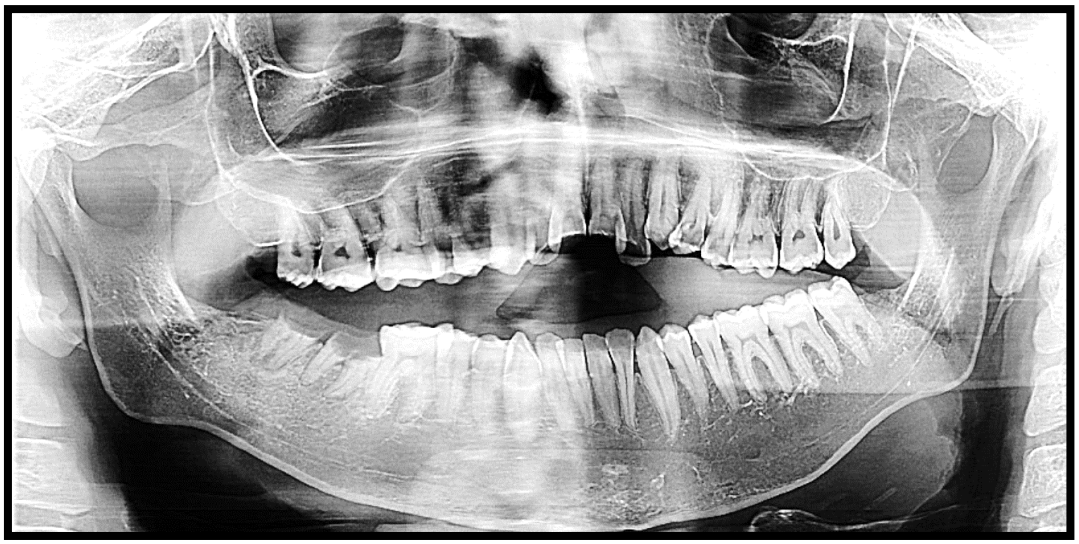

Figure 5: OPG

Patient was subjected to Lateral cephalogram, which revealed hypoplastic maxilla, anterior open bite, class III malocclusion, mild mandibular prognathism. (Figure 6)

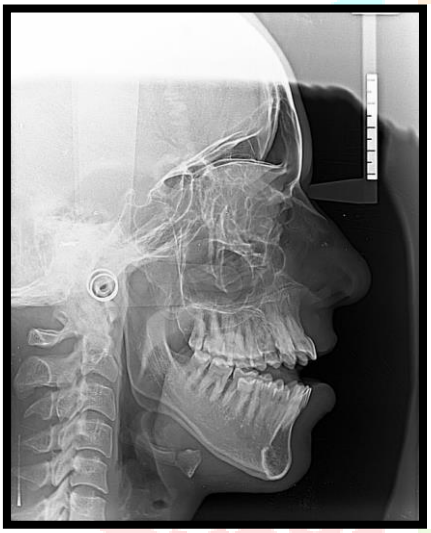

Figure 6 : Lateral cephalogram

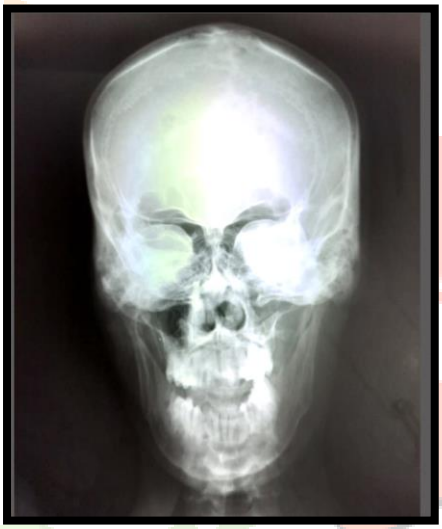

Figure 7: PA Skull

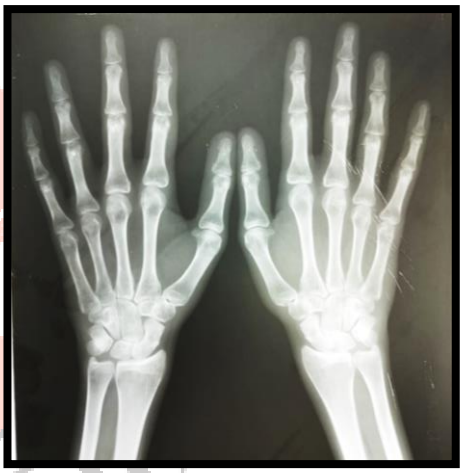

Figure 8: Hand wrist radiograph

Posteroanterior view of skull was taken, which revealed increased skull height, hypoplasia of maxillary sinus, absence of malar prominence, deviation of nose to right side, thinning of condyle on right side, hypoplastic maxilla, mandible deviated to right side. (Figure 7). Patient was subjected to Hand wrist radiograph (Figure 8). It shows increased metacarpal index (value – 9.2), which is also a characteristics in marfan syndrome.

By correlating patient's history, clinical and radiographic findings, we arrived at a final diagnosis of Marfan syndrome. Patient was advised for orthodontic correction of malocclusion and he is kept under regular follow-up.

IV. DISCUSSION:

In 1991, fibrillin-1 gene mutation on chromosome 15 was identified as a cause of Marfan syndrome, but molecular testing was not as diagnostically useful as was originally hoped[5]. Recently, mutations in the transforming growth factor b-receptor 2 (TGFBR2) gene on chromosome 3 and in the TGFBR1 gene on chromosome 9 were seen in some families with apparent Marfan syndrome. These "Marfan syndrome type 2" families seem less likely to have ectopia lentis[6]. Most Marfan syndrome patients are usually diagnosed incidentally when they present for a routine examination. It affect different parts of the body, including the heart, blood vessels, lungs, eyes, bones[7].

Cardiovascular disease is the cause of 90% of deaths among patients with this condition[8]. Cardiovascular abnormalities include aortic root dilatation, aortic regurgitation, aortic dissection, and aortic aneurysm[9]. But these abnormalities were not seen in this case.

On the skeletal level, disproportionate growth of the long bones is the most prominent feature of this syndrome. The other important characteristics are tall stature with the lower segment of the body greater than the upper segment and long, slender limbs, or dolichostenomelia; deformities of the chest like pectus excavatum or pectus carinatum and scoliosis. Other less common manifestations include hypermobility of joints, flat foot (pes planus), reduced extension of elbows (<170°)[6,10]. In the present case, most of this features are seen except scoliosis.

Ectopia lentis (subluxation of lens) is the most common ocular feature of Marfan syndrome which is present in approximately 60% to 80% of patients[11]. But these findings are not seen in this case.

Orofacial features of Marfan syndrome include dolichocephaly, malar hypoplasia, long and narrow face, frontal bossing, prominent supraorbital ridges, maxillary and mandibular retrognathia, skeletal malocclusion, hypermobility of the temporomandibular joint[12]. Westlig et al. found that 50% of the patients with Marfan syndrome had high and deep palates[13]. All these features were positive in this present case except mandibular retrognathia instead patient had mild prognathic mandible, this is also noted in some cases of marfan syndrome.

In the literature, there are plenty of articles with systemic manifestations of Marfan syndrome have been published. however, less number of articles are available based on the orofacial findings of this syndrome.

Early diagnosis of this syndrome can be done by Chorionic villus sampling (10-12 wks), Amniocentesis(16-18wks), ultrasonographic analysis of limb lengths at third trimester and other later diagnosis include Lumbosacral dural sac dimensions by MRI. But dural ectasia is a good marker for Marfan syndrome[14].

Management of this syndrome involves multidisciplinary team including cardiologist, geneticist, orthopedist, ophthalmologist, cardiothoracic surgeon, obstetrician and dentist. As a preventive measure, β blocker can be used to reduce the risk of aortic dissection and prophylactic early aortic root surgery can be planned. For ocular complications, such as Lens subluxation, the first line of management is correcting the refractive error with eyeglasses. If the lens is bisecting the pupil, eyeglasses and removal of the crystalline lens with artificial intraocular lens replacement is advised[15]. Like our case for dental management, orthodontic correction of malocclusion is most commonly needed in these patients. This patients also require oral monitoring to improve or maintain gingival health[10]. Antibiotic prophylaxis should be used for oral procedures, such as orthodontic banding, tooth extraction, and periodontal treatment[16,17]. Patient lifespan is now almost similar to persons without MFS, although cardiovascular impairment is still the commonest cause of mortality, mainly because of sudden death in an undiagnosed patient.

V. CONCLUSION:

Marfan syndrome is one of the most common syndrome associated with single gene defects. It is characterized by diverse clinical manifestations. Genetic testing is nonspecific, and the diagnosis is based on clinical criteria. Early diagnosis of Marfan syndrome is essential to ensure adequate therapy and increase the affected individual's life expectancy. Therefore, dentists also play an vital role in diagnosing and providing adequate care for such individuals.

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