DOI: 10.7860/JCDR/2015/11932.5906

Ghent Criteria an Aid to Diagnose Latent Systemic Diseases in Marfan Syndrome

GOMATHI RAMLINGAM[1], UMA MAHESWARI T. NATARAJASUNDARAM[2]

Keywords: Autosomal dominant, Connective tissue disorders, Steinberg sign,Walker sign

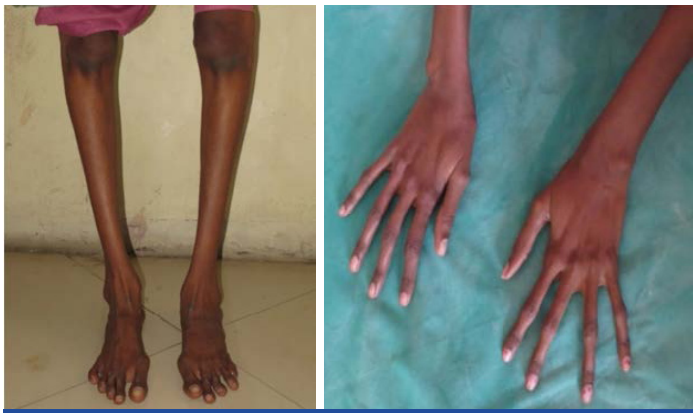
A 23-year-old tall female reported with a chief complaint of intermittent vague pain in the left lower posterior teeth region for the past two years. Her past medical history revealed that the patient has cardiac problem and visual impairment since birth and had not undergone any treatment for the same. Family history revealed that her parents and siblings are apparently healthy with no similar complaints.

General examination revealed height measurement of 174.5 cms (upper segment-72.5 cms and lower segment -102 cm, long and slender extremities [Table/Fig-1] with spider- like fingers and toes (arachnodactyly) [Table/Fig-2], positive Walker's sign [Table/Fig-3], Steinberg's sign [Table/Fig-4] and pectus carinatum [Table/Fig-5]. Facial profile examination revealed dolicocephalic face, sunken eyes, incompetent lips, malar hypoplasia [Table/Fig-6]. Review of systems revealed impaired vision (myopia)

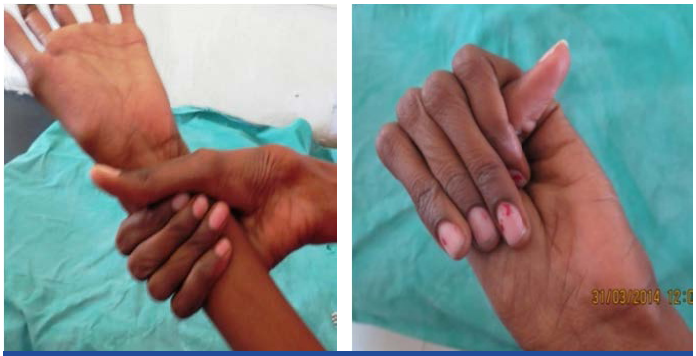
On intraoral examination patient had narrow upper and lower arch with crowding of teeth, high arched palate and multiple dental caries and root stumps [Table/Fig-7]. Correlating the history such as congenital cardiac anomaly, visual impairment and characteristic features such as dolicocephalic face, high arched palate, positive Walker's and Steinberg's sign the case was provisionally diagnosed as Marfan syndrome with a differential diagnosis of Ectopia Lentis syndrome, Ehlers-Danlos syndrome, Loeys-Dietz syndrome, Beals syndrome.

Orthopantomogram (OPG) revealed root stumps 16,37,46 and coronal radiolucency in 36 involving pulp associated with diffuse periapical radiolucency suggestive of periapical abscess, coronal radiolucency in 26,27,47 involving pulp suggestive of dental caries, impacted 48, prominent antegonial notch bilaterally, malformed right condyle and flattening of left condyle [Table/Fig-8].

On referral to cardiologist patient was subjected to Echocardiogram which revealed mild mitral valve prolapse with mitral regurgitation and mild tricuspid valve prolapse with tricuspid regurgitation, aortic dilatation with Z score=2. Ophthalmologist report revealed bilateral ectopia lentis and myopia (dioptre -4).

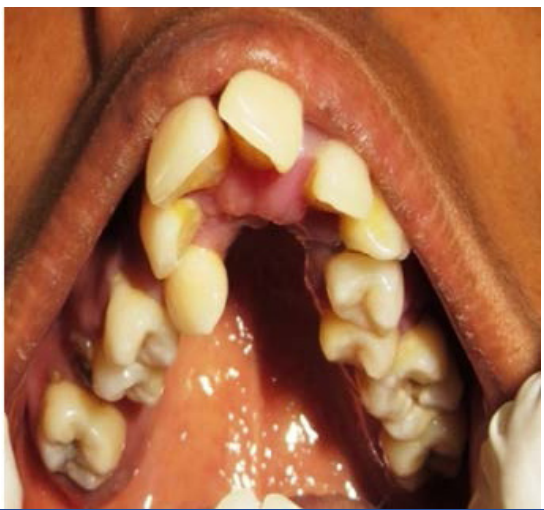


[Table/Fig-1]: Thin and slender lower limb **[Table/Fig-2]:** Arachnodactyly

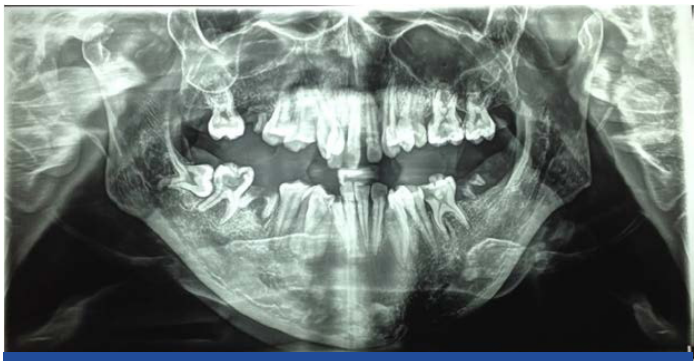


[Table/Fig-3]: Walker's sign (the distal phalange of the first and fifth fingers of the hand overlap when wrapped around the opposite wrist) **[Table/Fig-4]:** Steinberg's sign (the thumb projects beyond the ulnar border while completely opposed within the clenched hand)

[Table/Fig-5]: Pectus carinatum **[Table/Fig-6]:** Facial profile showing dolicocephalic face, incompetent lips, malar hypoplasia



[Table/Fig-7]: Showing crowding of teeth in upper arch and high arched palate



[Table/Fig-8]: OPG reveals alteration of condylar morphology in right and left TMJ, impaction, root stumps, dental caries

Based on the revised Ghent criteria [1] in the absence of family history with aortic dilatation (Z=2), ectopia lentis and systemic score of 9 (wrist and thumb sign-3, pectus carinatum-2, scoliosis-1, facial features-1, myopia>3 diopter-1, mitral valve prolapse-1) the case was confirmed as Marfan syndrome. Patient was adviced extraction of root stumps 16,37 and 46 under antibiotic prophylaxis (Amoxicillin 2g one hour prior to extraction) and endodontic management of 26,27,36,47. On regular review visits for endodontic treatment, apprehensive and less sociable nature of the patient was evident.

Many case reports of Marfan syndrome reported in the literature though was diagnosed based on Ghent criteria, the systemic score, aortic Z score, dioptre value has not been reported for all the cases [2-4]. In the present case the diagnosis of the common and other hidden systemic manifestations was arrived based on revised Ghent criteria and the aortic Z score of 2, dioptre value of -4 and systemic score of 9 were calculated. Unique features such as bilateral condylar malformation, prominent antegonial notch which were not reported in the other cases was observed in the present case.

REFERENCES

[1] Bart L Loeys, Harry C Dietz, Alan C Braveman, et al. The revised Ghent nosology for the Marfan syndrome. *J Med Genet*. 2010;47:476-85.

[2] AK Randhawa, C Mishra, SB Gogineni, S Shetty. Marfan syndrome: Report of two cases with review of literature. *Nigerian Journal of Clinical Practice*. 2012;15(3):364-68.

[3] Maysah Faisal Al-Mulla. Marfan syndrome: A Case Study. *International Journal of Scientific Study*. 2013;1(1):38-47.

[4] Bauss O, Sadat-Khonsari R, Fenske C, Engelke W, Schwestka-Polly R. Temperomandibular joint dysfunction in Marfan syndrome. *Oral Surg Oral Med Oral Path Oral Radiol Endod.* 2004;97(5):592-98.

PARTICULARS OF CONTRIBUTORS:
1. Post Graduate, Department of Oral Medicine and Radiology, Saveetha Dental College and Hospitals, Chennai, Tamilnadu, India.
2. Professor, Department of Oral Medicine and Radiology, Saveetha Dental College and Hospitals, Chennai, Tamilnadu, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Gomathi Ramlingam,
No 20, Adhibagawan Nagar, 2nd Cross Street, Sakthi nagar, Porur, Tamil Nadu-600116, India.
E-mail: gomu23@yahoo.com

FINANCIAL OR OTHER COMPETING INTERESTS: None.

Date of Submission: **Nov 01, 2014**
Date of Peer Review: **Feb 10, 2015**
Date of Acceptance: **Feb 20, 2015**
Date of Publishing: **May 01, 2015**