Dental Guide
Marfan Syndrome

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An inherited disorder of connective tissue that affects many organ systems including the skeleton, eyes, heart and blood vessels.\(^{1,2}\)

- Caused by a mutation in the gene for fibrillin-1 on chromosome 15.
- Can affect both men and women of any ethnic group.
- Around 18,000 people in the United Kingdom have Marfan syndrome.

**Salient features**

**Skeletal:** may have a tall, thin physique, with long limbs and fingers, scoliosis, narrow chest with breastbone (pectus) deformity, joint hypermobility and dislocations. Dilation of the lumbar dural sac seen on MRI scan occurs in about 65% of patients.

**Cardiovascular:** dilatation of ascending (and sometimes descending) aorta, incompetence of aortic and mitral valves, aneurysm and rupture of aorta.

**Respiratory:** Pneumothorax, asthma, emphysema and bronchiectasis.

**Ocular:** dislocation of lens, myopia and unstable refraction, retinal lattice degeneration, retinal detachment, squint (strabismus), glaucoma.

**Dental:** high arched palate, crowding of teeth.

**Genetic:** males and females are affected equally frequently. Each child of an affected parent has a 50% chance of inheriting Marfan syndrome. In 25% of cases neither parent is affected; however, apparently unaffected parents should be screened carefully as the severity and pattern of disease are variable, even within one family.
Diagnosis

Diagnosis is made after careful physical examination and echocardiography, demonstrating classical features in two out of three major systems (eyes, heart, skeleton), preferably with a family history. Diagnosis can be confirmed within a family by genetic linkage studies. Mutations can be found in the fibrillin-1 gene in 97% of patients, assisting with screening of family members.

Cardiac problems

The most serious problems occur in the heart and blood vessels. The aorta is usually wider than expected and is more fragile. The dilatation tends to be progressive, leading to aortic regurgitation and dissection. Surgical repair is recommended when the aortic root reaches 4.8cm, or earlier in cases with a family history of early dissection. Beta-blocker therapy can delay dilatation. Mitral valve prolapse is often also present. Antibiotic prophylaxis is recommended for dental extraction and all surgery, to prevent endocarditis.

Dental Aspects

The most common oral sign is a high, arched palate. This can be a very striking feature, the difference from normal being demonstrated in [Figures 6 and 7 next page]. Collapse of the upper dental arch, and in some cases severe malocclusion and open bite, tend to be associated with a high palate.

Other important characteristics include a long, narrow face and skull (dolichocephaly), often with mandibular prognathism, prominent supraorbital ridges, deep-set eyes and frontal bossing. Weakened capsular ligaments and hyperextensibility of muscle can contribute to dysfunction and habitual dislocations or subluxations of the temporomandibular joint.

Developmental abnormalities may also be evident, the most common being the presence of supernumerary teeth. Rare cases of congenital absence, incomplete development, crown dysplasia, enamel hypoplasia, dentinogenesis imperfecta and multiple odontogenic cysts have also been reported. There is a possibility that the fibrillin defect could contribute to slight relapse after orthodontic treatment, and greater periodontal problems.
Dental Management

Good oral hygiene is essential, along with regular routine dental check-ups. Patients may also need orthodontic and possibly oral surgical care. Careful planning from an early age can lead to successful aesthetic results and help prevent more serious dental problems occurring. The prevention of bacterial endocarditis is all-important because of the high incidence of heart valve involvement and abnormalities of the great vessels in Marfan syndrome. Antibiotic prophylaxis for any invasive dental procedure such as scaling or tooth extraction should be considered and these guidelines are periodically updated in the British National Formulary and Dental Practitioners Formulary. However, these measures for antibiotic cover are no longer routine for all patients and advice should be sought from the medical team regarding this if there is any doubt prior to dental surgical intervention.
General Anaesthesia

Patients with Marfan syndrome are recognised to have a slightly increased morbidity and mortality risk associated with general anaesthesia, due to high narrow palate, reduced neck mobility and narrow trachea causing difficulty with intubation, and increased risk of arrhythmia during anaesthetic. The contributing factors to this are cardiovascular abnormalities, impaired respiratory function, scoliosis, the potential to develop endocarditis and a tendency to spontaneous pneumothorax. Rarely, difficulty with intubation has been reported due to limited neck extension, a high palate and a narrow trachea. Pre-operative assessment should include a through medical examination with a chest x-ray, electrocardiogram and an echocardiogram. Any treatment must be carried out in conjunction with the patient’s cardiologist.

Conclusion

In summary, the dentist must be aware of all the problems associated with treating a patient with Marfan syndrome. Prevention of tooth decay, regular check-ups and careful forward planning all reduce the need for more elaborate and potentially dangerous procedures. The classical marfanoid appearance of the face and mouth can be recognised by a dentist, and could be the first vital step towards diagnosis of the underlying condition. If the diagnosis is suspected, the patient should be referred for echocardiography and genetic counselling through the family practitioner.
Raising Awareness:

Many people affected by Marfan syndrome, mostly children, remain undiagnosed due to symptoms not being recognised by doctors and family members. We are campaigning to raise awareness of Marfan syndrome and its signs so more people can be diagnosed and get their treatment programme commenced as quickly as possible.

Useful References


