

Bacterial Endocarditis Prophylaxis

The prevention of bacterial endocarditis is all-important because of the high incidence of heart valve involvement. High risk patients are those who have had cardiac surgery or a previous attack of bacterial endocarditis. Therefore antibiotic prophylaxis is mandatory for any invasive procedure involving bleeding. Guidelines for antibiotic prophylaxis are periodically updated in the British National Formulary. The possibility of a bleeding tendency, probably due to vascular fragility, has been noted in some affected individuals, and this should be taken into consideration when planning surgery.

General Anaesthesia

Patients with Marfan syndrome are recognised to have a slightly increased morbidity and mortality risk associated with general anaesthesia. Factors contributing to this are cardiovascular abnormalities and arrhythmia, impaired respiratory function, scoliosis, the potential to develop endocarditis, and a tendency to spontaneous pneumothorax especially in adolescence. Rarely, difficulty with intubation has been reported, due to limited neck extension, high palate and narrow trachea.⁶

Pre-operative assessment should include a thorough medical examination with a chest x-ray, electrocardiogram and echocardiogram looking for valvular insufficiency and aortic root dilatation in the sinuses of Valsalva. Any treatment must be carried out in conjunction with the patient's cardiologist.

Conclusion

In summary, the patient's doctor and ENT specialist must be aware of all the problems associated with treating a patient who has Marfan syndrome. Prompt management of ENT infections in childhood, together with surgical removal of tonsils and adenoids, where indicated, and correction of deviated septum or inadequate sinus drainage, will preserve health and hearing.

The classical marfanoid appearance of the face, mouth and ears can be recognised by a physician or

surgeon, and could be the first vital step towards a diagnosis of the underlying condition. If the diagnosis is suspected, the patient should be referred for echocardiography and genetic counselling through the family practitioner.

Useful References

1. Pyeritz R. (1993) The Marfan syndrome. In *Connective Tissue and its Heritable Disorders*. Eds Royce P.M. & Steinmann B. Wiley Liss, London. pp437-468.
2. Child AH & Briggs M, eds. *The Marfan syndrome, a clinical guide*. British Heart Foundation, London. Revised 2002 edition.
3. Crosher R., Holmes A. (1988) Marfan syndrome: Dental problems and management. *Dental Update* 15(3): 120-122.
4. Cistulli P.A. & Sullivan C.E. (1993) Sleep-disordered breathing in Marfan's syndrome. *American Review of Respiratory Disease* 147:645-648.
5. Cistulli P.A. & Sullivan C.E. (2000) Influence of maxillary morphology on nasal airway resistance in Marfan syndrome. *Acta Otolaryngol* 120:401-413.
6. Verghese C. (1984) Anaesthesia in the Marfan syndrome. *Anaesthesia* 33:917-922.

Contacts

Dr. Anne Child MD FRCP
(Medical Director, Marfan Trust)
Cardiac and Vascular Sciences
St. George's, University of London
Cranmer Terrace, London SW17 0RE
Tel: 020 8725 5248
Fax: 020 8725 2653
email: achild@sgul.ac.uk

Marfan Trust
Cardiac and Vascular Sciences
St. George's University of London
Cranmer Terrace, London SW17 0RE
Tel: 020 8725 1189
www.marfantrust.org

ENT Aspects of Marfan Syndrome

INFORMATION FOR ENT SURGEONS

Written by
Anne H. Child MD, FRCP
Mr. C.V. Praveen FRCS
Joanna Rowntree BSc (Hons)
Mr. A.J. Prior FRCS

**Marfan Trust**
Supporting Research into Marfan Syndrome

Registered Charity No: 328070
Sponsored by the Marfan Trust
and the Bluff Field Charitable Trust

**Marfan Trust**
Supporting Research into Marfan Syndrome

What is Marfan Syndrome?

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

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 60% 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 92% of patients, assisting with screening of family members, including prenatal diagnosis.

The Ageing Marfan Population

Modern medical and surgical therapies, combined with refined family screening, have significantly increased the expected life span of people with Marfan syndrome. Health problems in later years are thus becoming of increasing importance.

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 5cm, 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.

ENT Aspects

The clinical findings in the ear, sinuses, nose and throat display the same great variability in expression as found throughout the rest of the body in Marfan syndrome.¹ Important skeletal characteristics which contribute to ENT problems include a long narrow face and skull (dolichocephaly), often associated with mandibular prognathism, high arched palate, crowded teeth,

prominent supra-orbital ridges, deep-set eyes and frontal bossing.³

The ears are often large, simple, low-set and posteriorly rotated. Ear canals can be narrow and angulated upward and forward. Unilateral hearing loss is not uncommon, although the cause is obscure, possibly due to ossicular malformation during development, but most likely due to recurrent or chronic otitis media in childhood.

The nose is long, often beaked and asymmetrical, with narrow cavities and frequently deviated septum, which may lead to complete blockage of one side of the nose. Mouth breathing may result. In addition, sinuses may be narrow and underdeveloped with narrow drainage channels, therefore recurrent sinusitis is a common problem.

Sleep apnoea is a common associated feature, requiring referral for sleep studies.^{4,5}

Frequent unexplained nosebleeds may be a feature in childhood and adolescence, probably due to vascular wall fragility.

Medical Management

Early recognition and prompt medical management of allergic rhinitis or sinusitis may prevent secondary infections. Antibiotics for infection must be used earlier in the sinusitis, otitis or tonsillitis attack, and for longer than in a normal child or adult.

Surgical Management

Grommets for "glue ear" may help resolve the problem of recurrent otitis media. However, early removal of tonsils and/or adenoids is recommended, as surgical experience shows that these structures are surprisingly large and can actually contribute, through obstruction, to chronic otitis and sleep apnoea.

Surgical correction of deviated septum can be very helpful in management of nasal obstruction, mouth breathing and sleep apnoea. Cautery for recurrent nosebleeds may be necessary.