insufficiency and aortic root dilation. Any treatment must be carried out in conjunction with the patient's cardiologist. Antibiotic prophylaxis is recommended for surgery.

Working and Retirement

Easy fatiguability may become an increasing problem. Solutions may include living close to work, working part-time or from home. Letters from specialists can support early retirement on grounds of ill-health, if pension schemes permit. Disability pension may increase income.

Travelling

A letter describing cardiac status, including aortic root diameter should be carried while on holiday abroad, and travel plans should include knowledge of the nearest centre of medical and surgical excellence. Travel insurance may be difficult to obtain.

General Advice

Treatment is available for all aspects of Marfan syndrome and is improving yearly. Patients should therefore not worry unduly about affected children or grandchildren, as research advances will include less invasive surgical techniques and specific therapy to strengthen the aortic wall.

Coping with the problems of old age can provide a positive role model for younger affected family members. Just because a person has Marfan syndrome, does not mean that a person will escape the ordinary diseases of old age. These must be tackled with just as much care as for anybody else: we must not blame everything on Marfan syndrome!

Patients with Marfan syndrome reaching their 50s and 60s are a pioneer generation helping us to write a fascinating new chapter in the natural history of Marfan syndrome.

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Growing Older with Marfan Syndrome

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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. Mutations can be found in the fibrillin-1 gene in 92% of patients, allowing confirmation of the diagnosis and 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. Lifelong annual surveillance of the entire aorta is advised because of the risk of progressive dilatation 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. Surgical repair or replacement removes the greatest risk, however further dissection may occur in the remaining aorta.[3]

The diagnosis of Marfan syndrome may be made as a result of increasing aortic and/or mitral regurgitation in midlife.

Arrhythmia occurs in 40% of patients with Marfan syndrome at any age[4]. Most frequently this involves premature atrial or ventricular beats, but more serious rhythm disturbances may require medical or surgical therapy.

The risk of left ventricular failure is slightly increased due to fibrillin-1 deficiency in the heart muscle[5].

Antibiotic prophylaxis is recommended for dental extraction and surgery, to reduce the risk of infective endocarditis.

Other Problems

Problems in the normal older population may present for treatment at an earlier age in Marfan syndrome patients. These include:

- Varicose veins and haemorrhoids
- Hernias
- Slipped disc
- Prolapse of uterus, bladder or rectum
- Urinary problems (stress incontinence)
- Delayed healing

Irritable Bowel Syndrome

Occurs in approximately 20% of people with Marfan syndrome, probably due to the thin, lax, redundant bowel wall. IBS often appears for the first time in middle age, when normal ageing causes weak connective tissue to become even thinner.

Respiratory Disease

Problems are much more common due to lack of fibrillin-1 in lung tissue, and patients should not smoke.

Bone and Joint Problems[6]

Joint pains may become an increasing problem in later life, and can be treated with the usual non-steroidal anti-inflammatory medication. Joint and arch supports may also help to relieve symptoms. A small proportion of patients develop early onset osteoarthritis. Maintaining fitness is very important and suitable activities include swimming, walking, badminton and golf.

Anaesthesia

Patients with Marfan syndrome are recognised to have a slightly increased morbidity and mortality risk associated with general anaesthesia[7].
Pre-operative assessment should include a thorough medical examination with a chest x-ray, electrocardiogram and echocardiogram looking for valvular