

Management

This involves members of various professions, including physician, orthopaedic surgeon, physiotherapist, occupational therapist and orthotist.

Because of its relatively rapid progression in Marfan syndrome, scoliosis may require early bracing and surgical correction.

Shoes with arch and ankle support, joint supports and even splints may be used to improve joint stability and reduce pain during activity. Flat, long feet, often with hammer toes, require very careful shoe fitting with orthotics.

Arthralgia may respond to rest, joint supports and pain relief. NSAIDs should be varied until one is found which provides adequate pain relief. In general, steroid injections should be used as a last resort as they suppress collagen production. Certainly repeated injections are discouraged.

Patients should be taught to gently reduce recurrent dislocations of patellae, fingers and toes. These often improve with age.

Physiotherapists may be of great assistance, provided that they are informed that they are not dealing with normal joints. Exercise alone will not markedly strengthen genetically weak ligaments or muscles, although regular gentle exercise is helpful to maintain optimal strength. Some patients find learning the Alexander postural technique helpful.

In severe cases arthrodesis may be necessary to accomplish joint stability and freedom from pain. Soft tissue surgery performed to stabilise defective joints often fails with the passage of time, because the primary defect is in the supporting tissues. Healing after surgery is often delayed, therefore sutures need to be strong and left in somewhat longer than for the average patient. Antibiotic cover should be provided to prevent endocarditis and an experienced anaesthetist should be utilized since patients with Marfan syndrome may be difficult to intubate, due to a high palate, limited neck extension and narrow trachea.

Conclusion

Musculo-skeletal problems are common and troublesome in Marfan syndrome. Indeed, recognition by a rheumatologist 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.

Useful References

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Musculoskeletal Problems in Marfan Syndrome

INFORMATION FOR RHEUMATOLOGISTS AND ORTHOPAEDIC SURGEONS

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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 pigeon or funnel deformity, joint hypermobility and dislocations. Dural ectasia occurs in about 75% of patients.

Respiratory

Pneumothorax, asthma, emphysema and bronchiectasis.

Ocular

Subluxation or dislocation of lens, myopia and unstable refraction, detachment of retina, 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 and prenatal diagnosis.

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 surgery.

Rheumatological Aspects

Fibrillin-1, the defective protein in Marfan syndrome, is an important constituent of bone, cartilage, periosteum, tendons, ligaments and muscle. It has a special function in providing insertion of ligaments into bone.

Common problems

- scoliosis / spinal pain
- cervical spondylosis
- arthralgia
- meniscus injury
- ligament injury
- anterior knee pain
- dislocation/subluxation
- pes planus of joints/hammer toes
- myalgia/muscle injury

- osteoarthritis
- Scoliosis is very common in Marfan syndrome, appearing at an earlier age (from 7 years) and often progressing more rapidly than in other conditions. Screening examination using the forward bending test should be carried out once a year between ages 7 and 17.
- Most children's height is above the 97th percentile. If final predicted height is excessive, cyclical hormone therapy to limit the pubertal growth spurt needs to be considered at height 150cm.
- Joints of the extremities show wide variation in mobility, from congenital contractures to marked hypermobility.
- Men with Marfan syndrome are especially prone to spondylolisthesis, and should avoid gaining weight in middle age and heavy lifting at all ages.
- In approximately 5% of all families there is a tendency to early osteoarthritis, and the hip joint may need replacing by age 50. Protrusio acetabuli is known to occur with increased frequency.
- If patients have both rheumatoid arthritis and hypermobile joints, they may have a more serious type of rheumatoid arthritis which leads to early joint destruction.
- 70% of children experience symptoms of arthralgia, back pain and ligament laxity and injury. It is important to allow the child to participate in school and family events to the extent of his/her ability, but to permit rest, and not to prolong activities known to cause distress. Sports which do not traumatise lax joints should be chosen, e.g. swimming and badminton. Weight lifting, long-distance running and rugby are not recommended.
- A tendency to costochondritis, especially in rapidly growing adolescents, may lead to worries about chest pain coming from the heart. This is best treated with reassurance, non-steroidal anti-inflammatory drugs (NSAIDs), and review of possible triggering events.
- Fibrillin deficiency may also play a role in the easy fatigability found in adults and children alike.