Rheumatological Aspects of Marfan Syndrome

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Introduction

Locomotor symptoms are frequent and troublesome in the Marfan syndrome [1, 2]. Close involvement with rheumatologists and orthopaedic surgeons may be indicated to manage painful joint hypermobility and its complications in early to midlife, and, with increased longevity, degenerative joint disease in later years. Seventy per cent of children experience symptoms of arthralgia, back pain and ligament laxity and injury [1]. The earliest complaints of the child are often knee pain or ankle pain necessitating rest and analgesia. These symptoms, if not appreciated by parents and teachers, may lead to the belief that the child is not trying to participate in family outings or school sporting activities. It is important to allow the child to participate to the extent of his or her ability, but to permit rest, or to terminate the family outing, if the child cannot keep up. Taking a pushchair on family outings makes it a more pleasant activity for all concerned. Allowing the child to act as a referee or a goal keeper or play the least demanding role on the team at school is helpful. Weak ankles and knees, combined with poor eyesight may make the children appear clumsy in sporting activities, and they soon lose interest. It is important to wear sports shoes with ankle support, and even arch supports and heel cups, to render the gait more controlled. Ankle and knee supports may be worn to stabilise these joints, which are prone to recurrent sprains, and in some cases patellar dislocation. In severe cases, surgical fusion of the ankle, or shortening of the patellar ligaments may be helpful. Patients should be taught to gently reduce recurrent

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dislocations of patellae, fingers and toes. These joints gain stability with passing years. Wrist or finger splints may help to stabilise the loose wrist, thumb or fingers, and render writing neater. If the hand tires, especially during examinations, extra time should be given. Using fat pens or pencils, which may be built up, makes the handwriting more legible. For older children, the use of a laptop computer, or even a tape recorder to record lessons, may avoid the need for handwritten notes.

Preventive Measures

Back and neck pain may be eased by ensuring correct posture. Some patients find learning the Alexander method of correct posture at all times, very helpful. Stretching exercises may be taught by a physiotherapist, and sitting for long periods of time should be avoided. It is essential that the chair and desk are of adequate height for the tall child and adult, so that they do not stoop. It is important that computer screens and keyboards are at the correct height to avoid neck and eye strain.

Medical and Surgical Management

Flat, long, thin feet, often with hammer toes, require very careful shoe fitting with orthotics, and for the largest and most difficult feet, hand-made shoes may be the only solution. Surgery for hammer toes can be helpful, but frequently this deformity recurs after surgery, so it may be best just to purchase shoes with extra depth to allow room for the toes. 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 the age of 50. Protrusio acetabulae is known to occur with increased frequency [3–5].

Because of the overgrowth of ribs, pectus deformities may result, and even without this, 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 medication, review of possible triggering events such as playing tennis, and if all else fails, intra-articular injection with steroids. In general, steroid injections should be used as a last resort, as they suppress collagen production. Certainly repeated injections are discouraged.

Physiotherapists and occupational therapists may be of great assistance, provided that they are informed that they are not dealing with normal joints. Exercises alone will not markedly strengthen genetically weak ligaments or muscles, although they are helpful to maintain optimum strength. Manipulation of the spine and other painful joints should be gentle.

When prescribing medication for acute or chronic joint complaints, in the Marfan patient, the physician should be aware that most patients, although tall, have very little subcutaneous fat. Therefore, a low dose may be appropriate. In addition, gastric irritation with anti-inflammatory medication should be guarded against, with

simultaneous Losec and non-steroidal [anti-inflammatory] medication. Several NSAIDs may have to be tried, starting with the mildest before the most effective one is found.

Should the patient come to surgery, it must be remembered that healing is often delayed, as indicated by a tendency to scar, or to heal with papyraceous scar. Sutures need to be strong, and left in somewhat longer than for the average patient. Antibiotic cover should also be provided to prevent endocarditis, and an experienced anaesthetist should be utilised, as with all surgery, since the combination of high narrow palate, reduced neck extension, and floppy larynx may give rise to difficulty in intubation.

Occupation and Choice of Sports

Fibrillin-1 is an important constituent of bone, cartilage, periosteum, as well as tendons and ligaments and muscle itself. It has a special function in providing insertion of ligaments into bone. This explains the frequency of disability due to pain, dislocation or laxity in the central or peripheral skeleton, as well as skeletal muscle underdevelopment or hypotonia. Muscles tend to be slim and weak, and do respond to regular exercise, but muscle building exercises such as heavy weightlifting should be discouraged, as it is not realistic to expect muscle hypertrophy. It is better to choose sporting activities which are suited to the long, lean frame, such as cycling, basketball or badminton. Occupational choice may also be limited by physical ability, and patients should be encouraged to prolong their academic studies, and enter an occupation which is physically undemanding. Fibrillin deficiency in muscles and ligaments may also play a role in the easy fatigability which affects children and adults alike. Depression tends to occur if the working day takes all the energy a patient has to give. It is better to save some energy for social or leisure pursuits. Similarly, some patients experience an increase in fatigability from the age of 50 onward, necessitating early retirement if they are in a demanding occupation. Informed choice of a less demanding occupation may prolong the working life of the patient.

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