Nursing considerations for people with Marfan syndrome

Author: Robyn Backhouse, BSc (Hons), DipHE, RGN, is senior staff nurse, cardiac intensive care unit, Leeds General Infirmary.


People with Marfan syndrome present with a range of health problems that require specialised nursing, medical and surgical care. They face many challenges throughout their lives – some of which may be life-threatening – and require continuing support and education from a specialist team. Nurses play a pivotal role in the care of people with Marfan syndrome and their families, encompassing not only medical and surgical interventions, but aspects of lifestyle such as diet, exercise and hobbies, as well as family planning and psychological well-being.

Marfan syndrome was first described in 1896 at a meeting of the Medical Society of Paris when Bernard Marfan presented the case of a five-year-old girl with disproportionately long limbs. In later years, several other people studied the same girl and added to his findings, noting her malaigned dorsal spine, thoracic asymmetry, arachnoidactyly (long digits), cardiovascular abnormalities (including aortic dissection) and dislocation of the ocular lens (Marfan Association UK, 2004). They called the condition hyperchondroplasia, as they wrongly believed it to be the opposite of achondroplasia – the disorder of bone formation that causes dwarfism.

Today, much more is known about Marfan syndrome. It has an incidence of about one in 5,000 people of all races and both sexes, and affects many organ systems, with severity differing greatly between individuals (Marfan Association UK, 2003).

What is Marfan syndrome?

Marfan syndrome is caused by a deficiency of microfibrillar fibres, which form the framework for connective tissue (Dietz, 2003). Connective tissue provides structural support and shape to organs, muscles, blood vessels and the entire body (Canadian Marfan Association, 2004). Abnormality in these fibres causes them to elongate progressively, stretching and weakening, leading to a range of health problems (Box 1) with severity varying between individuals.

Connective tissue is also important for other functions, including antenatal development and postnatal growth, cushioning of joints and enabling the passage of light through the eye. All organs that contain connective tissue can be affected, especially the skeletal system, ocular system, cardiovascular system, pulmonary system and nervous system (National Marfan Foundation, 2003). People affected are often unusually tall with long, thin extremities and chest deformity (Child, 1998). They usually develop stretch marks at an early age, especially over the shoulders, hips and lower back.

Marfan syndrome is an autosomal dominant genetic condition and so each child of a Marfan sufferer has a 50 per cent chance of inheriting the syndrome (Canadian Marfan Association, 2004; Marfan Association UK, 2003). In 75 per cent of cases it has been inherited from an affected parent, while the remaining 25 per cent result from spontaneous gene mutation of either the egg or sperm with no previous family history of the condition. The severity of symptoms varies but, on average, one in ten of those affected have serious health problems (Marfan Association UK, 2003).

Presentation and diagnosis

Diagnosis is often difficult due to the variation of clinical manifestations. It generally involves careful physical examination focusing on the eyes, skeletal and cardiovascular system. Dilatation of the aortic root is a reliable sign, present in over 90 per cent of people with Marfan syndrome and detectable very early in life by echocardiography (Pyeritz, 1998). Since 75 per cent of affected people have an affected parent, family history is an important consideration. But the condition can also occur as a result of a new, spontaneous genetic mutation.

Recent studies have identified the chromosomal and gene component of connective tissue (fibrillin) responsible for Marfan syndrome, and this has led to successful blood and skin testing and even antenatal diagnosis for some families (Marfan Association UK, 2003). This could have an important impact on prospective parents with regard to family planning, counselling and decisions regarding the pregnancy itself, and specialist nurses often work closely with families at such times.

Skeletal problems include curvature of the spine (scoliosis/kyphosis), an abnormally shaped chest, tall stature and loose joints, which often cause joint pain and dislocation. Treatment for skeletal deformities includes bracing, physiotherapy and surgery (Watson, 1998). Severe skeletal deformities may themselves impair cardiac or lung function and require careful observation (Pyeritz and Conant, 1998). Other manifestations may include spontaneous pneumothorax, early emphysema, congenital lung abnormalities, right ventricular enlargement (due to pulmonary hypertension caused primarily...
by lung disorders), dislocation of the ocular lens and retinal detachment (Marfan Association UK, 2003; Bellamy, 1998; Calver and Jones, 1998).

The primary cause of death in people with Marfan syndrome is aortic disease, so this is one of the most important areas of investigation and treatment (Moir, 2003). Gross dilatation of the ascending aorta and incompetence of the aortic valves occurs and patients often present with aortic reflux, as an aortic dilatation itself is usually asymptomatic. The remainder present as emergencies with either a ruptured aorta or a dissecting aneurysm. Dissection of the coronary artery may also occur and this, too, requires prompt surgical repair (Leatham, 1998). Another complication is mitral valve insufficiency, which may be associated with cardiac dysrhythmias and can lead to heart failure (Leatham, 1998). Echocardiography is widely used to monitor aortic and cardiac size and function (Child, 1998; Leech, 1998).

Prognosis and treatment

Prognosis is based on the aortic root diameter, with a greatly increased mortality rate if the diameter is more than 4cm (Thelan et al, 1998). Where widening of the aortic root is evident, beta blockers may reduce the risk of development of an aortic aneurysm, as they help to control heart rate and decrease blood pressure on the weakened aortic wall. This slows the dilatation of the aortic root and limits the spread of aortic dissection (Marfan Association UK, 2003; Ramstead, 1998). Elective surgical repair appears to be highly successful once an aneurysm has developed, but delay in cardiac assessment may allow undetected aortic dissection or aneurysms to rupture. These situations are surgical emergencies with high mortality rates (Child, 1998).

Elective cardiac surgery should be discussed in depth with the affected person and family to ensure they are able to make an informed choice based on sound and current knowledge of the condition and treatment alternatives. Most people who have had elective aortic graft surgery over the past 15 years have not needed additional surgery. However, those who have undergone emergency surgery following dissection have frequently required further surgery (Professional Advisory Board, 2003). While this seems a good argument for elective surgery, which may lengthen the person’s life significantly, there is always a risk that it will be unsuccessful and either shorten life or even cause death. A great deal of support and unbiased information is necessary to enable patients and families to make the right decision based on their individual beliefs, values and circumstances, and a nurse with specialist knowledge may be able to assist them.

Management of the condition

Because Marfan syndrome affects numerous body systems, nursing care depends largely on the presenting condition. If the patient has cardiac complications, corrective surgery appears to be the only treatment currently offered other than medication. Following cardiac or aortic surgery, the patient will usually be transferred to intensive care, where nursing care will focus on haemodynamic monitoring and organ support. Nursing care and management of any patient following aortic root replacement, with its associated coronary artery re-implantation and cardiopulmonary bypass, is extensive and requires specialist nursing knowledge. The aim should be to provide holistic care, particularly when the person’s condition is chronic, as they will need appropriate education and a lot of support, both initially and throughout their life.

Priorities of care for a patient with an aortic aneurysm are similar to any other with this condition. It includes urgent resuscitation when needed, pain relief, high-flow, high-concentration oxygen and maintenance of a systolic blood pressure no higher than 120mmHg (Adam and Osbourne, 1997). One of the main aims of nursing care of people with Marfan syndrome is to provide education and support for both them and their loved ones. Some lifestyle changes will probably be necessary to minimise the health problems their condition causes, and these may need to take place over a long period of time (Pyeritz and Conant, 1998). Both affected people and their families will need guidance and up-to-date information about these changes. Nurses can provide emotional support, which will be vital during any hospital admissions, and assist with education on subjects such as family planning and genetic counselling, fitness and exercise, diet and nutrition, and any special precautions or follow-up treatment and examinations required.

It is important not to cause undue anxiety when examining and diagnosing people with Marfan syndrome.
When people receive their diagnosis, it is often already apparent to them that they have health problems or abnormalities and it may be a relief to give a name to their concerns. However, they may also feel confused, stunned, frightened, hopeless or frustrated by the sudden perceived loss of health. They may even deny the existence of the condition, the need for appropriate treatment or that they should avoid vigorous activities or occupations and make other lifestyle changes.

Nurses can help them to accept their condition by providing support, understanding and up-to-date information – particularly since there are often rapid improvements in the treatment available (Pyeritz and Conant, 1998). In addition to their symptoms, they may experience economic, social, emotional or psychological problems, for which they need the support of knowledgeable and supportive professionals who can provide emotional support and practical help and advice.

Adolescents

Teenagers and young adults can look different from their peers due to their skeletal deformities and often excessive height. They are frequently extremely self-conscious about their appearance, and their need to be accepted within their peer group may cause depression or psychological or behavioural disturbances. They may have negative feelings about treatment and check-ups, which they can see as unwelcome intrusions into their lives that make them feel different from their friends at a time when they need acceptance as part of the group. Consequently, adolescents with Marfan syndrome may fail to take medication or follow lifestyle advice as a form of rebellion against their condition. In such situations they will need additional support and education to help them through this difficult period (Pyeritz and Conant, 1998).

Young people with Marfan syndrome are often angry about their situation, especially if they have had to sacrifice a favourite sport that has been important to them. They may project this anger towards health care staff, their parents or themselves. It may help them to talk to someone of a similar age who also has Marfan syndrome, as this may make them feel less isolated and different. Nurses can arrange such peer support, as well as providing the latest information on treatments and prognosis, which may help them to adjust and become more accepting of their condition and its impact on their lives.

Parents

Parents of affected children may need a lot of support, as they may feel guilty about passing on the condition, even if they are not themselves affected. Feelings of anger and sadness may follow. They may become overprotective, and this may prevent their children from becoming self-sufficient and independent in later years.

Although few young children need significant restrictions on their activities, parents may be advised to channel their children’s interests into appropriate non-contact sports and activities at an early age – this can be easier than telling a teenager they must give up their favourite activity (Pyeritz and Conant, 1998). It is essential for their psychological health and emotional development that children with Marfan syndrome lead as normal a life as their condition allows (Pyeritz and Conant, 1998), and nurses can help parents to adjust to their fears and allow their children to become as independent as possible. They can also put parents in touch with various organisations that can provide further support to help them overcome feelings of guilt, combat the loneliness of dealing with the situation and discuss experiences and coping strategies with other families in similar situations.

Lifestyle

It is important for people with Marfan syndrome to remain as active as possible, since exercise helps to improve muscle tone and cardiovascular function. However, exercise must be appropriate to the individual’s condition. They should be able to take part in non-competitive sporting activities, but must be allowed to stop whenever tired. They should avoid strenuous activities such as contact sports like football, basketball and boxing, isometric exercises and exercise at maximal capacity to reduce the risk of eye or skeletal injury and aortic damage due to increased cardiac performance (Marfan Association UK, 2003; Child, 1998; Pyeritz and Conant, 1998). Fatigue may be a problem, especially when long periods of concentration are required, so adequate periods of rest are advised to prevent over-exertion or exhaustion (Marfan Association UK, 2003).

People with Marfan syndrome are recommended to have a balanced, nutritious, healthy diet, with restrictions on salt, cholesterol and animal fats. Vitamin, mineral and dietary supplements and protein derivatives have shown

---

**Box 2. Recommended Investigations**

**ECG and Echocardiogram**

To observe for dysrhythmias, mitral valve prolapse and aortic root dilatation.

**Chest X-Rays**

To observe for signs of pneumothorax – especially in tall pubertal males.

**CT or MRI Scans of the Aorta**

On diagnosis of Marfan and periodically in patients with ectasia or dissection.

**Skeletal X-Rays**

Particularly of the chest and back.

**Slit-Lamp**

To detect lens dislocation.
no real benefit, so are not specially recommended. Smoking destroys elastin, which is already deficient in Marfan syndrome. It also increases risks in surgery and postoperative recovery, so it should be avoided completely (Marfan Association UK, 2003; Pyeritz and Conant, 1998).

**Family planning**

Whether or not to have children is a personal decision that should be made solely by prospective parents with Marfan syndrome, who should be helped to understand and acknowledge the potential risks. They should be referred to a genetic counsellor, who can provide guidance on starting a family – both the potential problems associated with pregnancy and the risks to future children (Child, 1998). Genetic counsellors can also assist in identifying other affected family members. Genetic counselling should involve not only the affected person but also their partner and other close family members, as it will have a huge impact on all their lives.

If the potential father is badly affected with Marfan syndrome, artificial insemination using donor sperm may be considered as a safer option (Child, 1998), though this may not be acceptable to many couples for personal reasons. Risk relates not only to the possibility of passing the condition on to the children, but also to the physical and haemodynamic stress placed on the potential mother’s own body. If the aortic root is only minimally dilated, the risk of dissection appears low, so prospective mothers should ideally undergo an echocardiograph before deciding whether or not to conceive, then at six to eight-weekly intervals during pregnancy.

If women conceive, they should receive care under monitoring and medical support throughout their pregnancy. If the aortic root shows evidence of progressive enlargement during the first half of the pregnancy, termination should be considered; if it occurs later, early delivery is advised. In all cases, the baby should be delivered by the least haemodynamically stressful method. Beta blockade is not advised during the early stages of pregnancy due to potential risks to the foetus, but is not contraindicated in late pregnancy if the mother’s blood pressure rises (Pyeritz, 1998).

Women with severe heart valve problems should not attempt to have a baby as it places them at serious risk (Pyeritz and Conant, 1998). The risk of aortic dissection is low in women who have had no previous aortic dilatation, though care should still be taken. Even if there is only slight aortic enlargement, the risk of dissection during pregnancy is higher and increases as the aorta enlarges (Professional Advisory Board, 2003). Women and their partners may need a great deal of support at this time, especially if it is clear that pregnancy is contraindicated or that termination should be considered.

**Other issues**

Patients who have undergone aortic or cardiac valve surgery need to consider a number of precautions, which nurses can discuss with them. Good dental hygiene, including flossing, is important to reduce the risk of bloodstream infections via the oral mucosa – although it is important not to make the gums bleed. Prophylactic antibiotics are recommended prior to dental treatment and other invasive procedures to guard against the development of endocarditis (Professional Advisory Board, 2003; Child, 1998). Many patients will require lifelong anticoagulation therapy following valve surgery and this is associated with numerous complications and risks of which patients and families must be fully aware. Multidisciplinary evaluations of the cardiovascular system are advised every two years until the age of 11, and annually thereafter. A number of regular investigations are recommended (Child, 1998; Marfan Association, 2003) (Box 2).

Nurses with specialist knowledge can explain to people with Marfan syndrome and their families why these regular examinations are necessary and what they involve, as a greater understanding of the procedures and potential benefits will promote acceptance.

**Conclusion**

Although Marfan syndrome is a serious condition with major complications that can occur at any age, the prognosis is vastly improved with modern treatments (Pyeritz, 1998). As complications develop with age, the multidisciplinary team and patient together can make plans for future requirements and treatments, facilitating a more relaxed approach to the condition. Effective treatments have been developed for most complications associated with the syndrome, and have resulted in a decrease in morbidity and mortality. Recent advances in medical and surgical treatment have seen the average lifespan for people with Marfan syndrome rise from 45 years in 1972 to 70 years in 1997 (Professional Advisory Board, 2003). Such positive facts should be stressed to patients, who may become unduly depressed if they gather outdated information suggesting a much worse prognosis (Pyeritz, 1998).

The nurse’s role in caring for people with Marfan syndrome is varied and depends largely upon each individual’s symptoms and particular health problems. Apart from specialised care afforded to each affected organ, the nurse’s main role is to provide support and education – both for patients and their families and loved ones. Feelings of isolation, resentment for being ‘abnormal’ and depression can be common among this group of patients, so nurses should focus on positive aspects of people’s lives by encouraging them to join groups appropriate to their physical capabilities, follow a healthy diet and lifestyle, and lead as full and active a social and personal life as possible.

Family planning, support and regular health reviews are essential to prospective parents, who require appropriate guidance and advice from their nurse. Advances in treatments have greatly improved quality of life and extended lifespan for those with Marfan syndrome, proving that they do have a future to look forward to. ■