Psychosocial Aspects of Marfan Syndrome

A pamphlet for Child Psychiatrists and Child Psychologists

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Marfan Trust
Supporting Research into Marfan Syndrome

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What is Marfan Syndrome?

An inherited disorder of connective tissue that affects many organ systems including the skeleton, lungs, eyes, heart and blood vessels.

- Caused by a mutation in the gene for fibrillin-1 on chromosome 15.
- Can affect both males and females of any race or ethnic group.
- Around 10,000 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 occurs in about 75% of patients.

Cardiovascular:
Dilatation of ascending (and sometimes descending) aorta, incompetence of aortic and mitral valves, aneurysm and rupture of aorta.

Respiratory:
Pneumothorax, bronchiectasis, emphysema and asthma.

Ocular:
Dislocation of lens, myopia and unstable refraction, detachment of retina, 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 2 out of 3 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.

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.

Psychosocial Aspects

Children and adolescents with Marfan syndrome look and feel different and restrictions are often imposed on them because of their poor eyesight, lax and painful joints and cardiac problems. School absence may be frequent because of hospital appointments and corrective surgery for skeletal, ocular or cardiac problems. In addition, other affected members of their family may have been acutely or chronically ill, required heart, spinal or ocular surgery, or even died suddenly and unexpectedly, possibly at an early age.

Despite normal intellectual and gross motor development, children often fail to perform to the best of their ability because of physical limitations: Short-sightedness and clumsiness (due to lax joints and a long thin body build) are the main problems[1]. Nevertheless, most children are able to attend a mainstream school, albeit often with special help (from statementing) in the classroom[2].In a survey of 101 children (60 males and 41 females) aged 4-16 years with Marfan syndrome, compared to an age and sex-matched control group of 117 children, the following results emerged[2]:

- Children and young people with Marfan syndrome have significantly more behavioural problems. Parental report (Child Behaviour Checklist) shows that children with Marfan syndrome tend to internalise their worries, resulting in withdrawal, physical complaints, anxiety and depression. Conversely, they are not inclined to externalise their problems nor be aggressive or antisocial.

- Children with Marfan syndrome may have low self-esteem with respect to social and physical competence, and general self-worth. They feel they are not liked by others and are not sure of themselves (Harter Self-esteem Questionnaire). Answers on The Child Depression Inventory convey a similar theme; the children indicate that they do not like themselves and they wish they had more friends.

- During the pubertal growth spurt, which typically starts and finishes earlier than in the normal population, features of Marfan syndrome may appear or worsen. Affected children realise more acutely that they are different and have difficulty with their body image. Not surprisingly they may be teased or bullied at school, the commonest forms being name-calling, theft of belongings, and being left alone in the playground. They also tend to have fewer friends than their peers (Olweus Bullying Questionnaire).

- Other factors contributing to psychological problems are a prolonged period of dependence on parents (e.g. due to difficulties obtaining employment or visual problems that prevent driving) and easy fatiguability which limits occupational choices.

Suggestions

- Teachers and parents alike should be made aware of the behavioural and emotional problems identified in this pamphlet. Simply knowing that these features may be expected in Marfan syndrome may improve both the home and school situations.

- Early identification and prompt intervention may reduce the suffering caused; professional psychological counselling may help to improve both self-image and social interaction. Teachers and the rest of the family may need to be involved.

- Children should be taught about Marfan syndrome from an early age and should learn to become comfortable in educating their classmates. Parents and doctors alike should be sensitive to cosmetic issues and should anticipate and/or initiate discussions regarding potential solutions such as contact lenses or breastbone (pectus) repair[3]. Time off school should be minimised by asking for hospital appointments and operations to be arranged whenever possible during holidays.

- A positive self-image of the child may be fostered by praising the child's talents. Amongst peers, the child's image may be improved by demonstration of artistic or musical ability or computer skills, for example, to compensate for possible lack of physical ability.

- A sense of humour is vital for coping with teasing, which appears to be limited to school years.

- The opportunity to discuss problems with others of the same age can be very beneficial; contacts may be made via Dr Anne Child.

Useful References

1. Hofman K. J., Bernhard B.A., Pyeritz R.E., Marfan syndrome: neuropsychological aspects. American Journal of Medical Genetics 31:331-8, 1998.
2. Dawkins J., Rowntree J., Spender Q. & Child A. Unpublished data.
3. National Marfan Foundation (USA) Website: www.marfan.org
4. Marfan syndrome: a booklet for teachers. Available from Dr A. Child

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