Introduction

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Anne H. Child

Rare or Common?

Marfan syndrome is generally thought to be rare, but clinicians who have seen how the gene expression can shade into normality in affected members of a family, suspect that its overall frequency could be underestimated, as a recent collaborative publication states [1]. The fact that the syndrome presents in many different ways may also hinder its recognition. Marfan syndrome patients may first be referred to any one of a number of specialists. Each doctor is likely to be familiar with a particular aspect of the syndrome, but less familiar with its other features. The aim of this guide is to pool the knowledge available from different experienced specialists, and to make it available to the many clinicians likely to encounter a patient with this condition.

Co-ordinated Care

Who should care for the Marfan syndrome patient? I believe the answer is that we should all do so. Whoever first suspects the diagnosis should pursue the question until it is either proved or disproved, and should consider what other specialist opinions might be required. All patients should be referred for genetic counselling, both to help with diagnosis, and to identify other affected family members, as well as provide guidance about the risk to future children. Each patient is best cared for by a team, led by one co-ordinating physician who is familiar with all aspects of the condition. This is likely to be a geneticist or cardiologist. The rise of aortopathy

A.H. Child, MD, FRCP
Cardiovascular and Cell Sciences Research Institute,
St George's, University of London, London, UK
e-mail: achild@sgul.ac.uk

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diagnostic and follow-up clinics is to be applauded. As a result, patients can be referred locally rather than having to travel long distances for management.

International Collaboration and Patient Support

National and international patient support group networks have been set up, and active research is being undertaken into the cause, early diagnosis, treatment, and prevention of Marfan syndrome. A summary of diagnosis and management is provided in Chap. 2, Table 2.2. Subsequent chapters show that the management of this condition can be much improved by wider knowledge of its principal features, most of which can now be effectively treated, resulting in more normal lifespan [2]. The discovery that the underlying connective tissue defect in Marfan syndrome results from a primary deficiency of fibrillin-1 has led to attempts at more fundamental methods of prevention and treatment, such as the international trials of losartan and irbesartan aimed at preserving the aortic media [3].

First Gene for Familial TAAD

International collaborative groups aimed at discovering new genes and new treatments are proving effective [3]. Marfan syndrome has led the way in this field of Familial Thoracic Aortic Aneurysm and Dissection (FTAAD), and clinicians experienced in managing this condition will be called upon to manage many similar genetically-determined aneurysms in future [4].

References

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