Diagnosis and Management of Marfan Syndrome

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This book is dedicated to all those with Marfan syndrome – past, present and future – who teach us how to care for them, and inspire our research efforts
To all my colleagues internationally, whose unceasing efforts to improve our understanding of Marfan syndrome in all its ramifications have been willingly shared to speed progress
To my family, especially my husband, Dr. Geoffrey V Child, who have supported me in all my endeavours
To my students, who carry on the battle against disease
And in memory of Professor F Clarke Fraser who first introduced me to the rewarding field of genetic research

Preface

The Marfan Trust has become increasingly active over the past few years. Our support network has grown and we have engaged with many more people through events and fundraising activities. Our website and social media are becoming central in explaining what the charity is doing and how our Marfan patient community and supporters are engaging with us and one another. The Trustees held their strategy meeting early in 2015 to determine the charity's direction and goals over the next few years. The outcome is published on our website. Our three key objectives remain: medical research as a high priority; producing educational literature and raising awareness; and support and guidance provided for our patient community. We are growing as a charity, providing top quality research in our designated laboratory, education, awareness and support in the UK and beyond.

The AIMS (Aortic Irbesartan in Marfan Syndrome) Trial, continuing until 2018, is one of a number of international trials comparing beta-blocker therapy with losartan or irbesartan therapy. This UK trial, co-funded by the Marfan Trust and the British Heart Foundation, is a main priority. With 21 participating Marfan syndrome clinics around the UK, this trial has provided an opportunity for Marfan syndrome patients to have local diagnosis and support. Additional research is undertaken in our laboratory into overlapping syndromes such as ectopia lentis and scoliosis, and translational research, which will ultimately benefit patients through improved clarification of diagnosis and the utilisation of results to develop resources for medical professionals and education. One example of this is our new paediatric guide, formulated from research incorporated into our children's database. We have provided funding support to recruit a number of medical and bioscience students to undertake research work. This has assisted our resident researchers and also introduced a new generation of future medical and scientific professionals to genetics and Marfan syndrome in particular.

Knowledge is everything and the first step to knowledge is awareness. We provide a February Awareness Month over various media platforms. Family Fun Days are held twice yearly and permit families to share problems and solutions. Our updated literature is now available. We are particularly targeting general practitioners in the UK, where there seems to be a lack of awareness and understanding of Marfan syndrome.

This present textbook has been eagerly awaited. It reports new research funded by the Marfan Trust. We are most grateful to the authors for sharing their

knowledge, and we know that increased awareness and improved care of Marfan syndrome patients will result. We cannot thank you all enough.

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Patrons: Sir Magdi Yacoub FRCS, Mr Anthony Latter MA and Lady Maryanna Tavener

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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

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