

Prevention of Thalassaemias and other haemoglobin disorders

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On the cover: Peripheral blood film of a patient with β -homozygous Thalassaemia, kindly provided by Mrs A. Kyrii - head of screening laboratory - Thalassaemia Centre, Nicosia, Cyprus.

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Foreword

Thalassaemia and the haemoglobinopathies are a major health problem, placing an immeasurable emotional, psychological and economic burden on millions of people around the world. In the West, substantial progress has been made towards understanding the pathology of thalassaemia and its treatment. But the fact remains that the treatment of thalassaemia is a costly and painful process. More important still, for many, many patients, particularly in poor countries, knowledge of the disease remains sparse and treatment is an unaffordable luxury.

It is in this context that the role of prevention is best understood.

Over the past 25 years, a number of European countries have seen a dramatic fall in the number of affected births as a result of prevention programmes based on health education, widespread screening and genetic counselling. For many involved in the field of thalassaemia, the experiences of these countries have provided a wealth of expertise from which other countries can - indeed, must - draw.

This book provides a thorough overview of the issues involved in prevention. As such, it represents a major step towards a fundamental goal of the Thalassaemia International Federation (*TIF*) - that is, to establish prevention programmes based on the same high standard of best practice in every country of the world.

Advances in genetic research have made prevention a feasible option everywhere. TIF is determined not only to encourage prevention programmes but also to ensure that lessons already learned are not lost - determined that those countries that can least afford a period of trial and error are provided with the essential knowledge needed to establish an effective prevention programme.

Of course, the issue of prevention is a difficult one, fraught with moral and emotional complexity. But no one is insisting that a

blanket solution be applied everywhere - only that each country take a hard-headed look at the costs of inaction, before assessing what is feasible in a local context, based on well-established international norms.

The fight against disease always requires dedication - both in terms of time and resources. For TIF, the overriding concern is that families should be supported in finding ways to avoid the burden and pain of dealing with a serious genetic disease... not least one that can so easily be avoided.

As Chairman of the Thalassaemia International Federation, I welcome this thorough and wide-ranging book as a vital contribution to the work of TIF, and an invaluable resource for all those working in the field. We are also all looking forward to the next publication which is a companion book to Volume 1, presenting detailed laboratory procedures essential for establishing of prevention programme. On behalf of the Board of TIF, I would also like to express our most sincere appreciation for the hard work and devotion the authors have shown in compiling this publication. This book is the product of many months' effort, reflecting a consensus of ideas born of the authors' considerable expertise in a range of fields.

Panos Englezos
TIF Chairman

Preface

The haemoglobin disorders are the most common clinically serious single gene disorders in the world with an estimated 300,000 affected births each year, of which approximately 60,000-70,000 are β -thalassaemia major. However, these figures are just an estimate - and as there have been few in-depth, reliable epidemiological studies into the incidence of thalassaemia in many parts of the world, they are likely to be a gross underestimate. Although a detailed analysis of molecular variance in the thalassaemias exists in almost every affected country of the world, true gene frequencies for many populations are unfortunately still not available. Indeed, this book indicates how, in addition to preventing new births of affected children, a greater emphasis on prevention is likely to improve knowledge of the epidemiology of thalassaemia through pilot studies assessing the prevalence of the disease in different areas.

The benefits of prevention programmes have been clearly demonstrated by a number of Mediterranean countries over the past 25 years. The purpose of this book is to draw on this experience, and to lay out the issues and approaches involved in preventing new births of children affected with haemoglobin disorders.

The authors present the advantages for establishing prevention programmes, as well outlining the tools required for such programmes to be truly effective - understanding the epidemiology of the diseases concerned and developing appropriate health education programmes that are sensitive to prevailing cultural and religious norms.

As research into genetic disorders continues apace, the screening for and diagnosis of haemoglobin disorders, including prenatal diagnosis, are becoming increasingly significant aspects of prevention - along with genetic counselling and the need for medical specialists to be ever more sensitive to the power and complexity of the information with which parents are being presented. These subjects are addressed in detail, including problems associated with screening for the haemoglobinopathies.

It is hoped that this book together with the forthcoming Volume 2, a companion book to this volume presenting detailed laboratory procedures will be an informative source for public health professionals and policy makers alike, as well as scientists involved in the effort to reduce the suffering caused by single gene disorders. Of course, the issue of prevention is a delicate one. The authors remain sensitive to this fact and have no wish to simplify or minimise the significance of the issues involved. Rather, we hope this book will contribute to constructive debate on the issue while encouraging ongoing pioneering work in the field of prevention.

The Editors

About the publishers

The Thalassaemia International Federation (*TIF*) is a non-governmental not-for-profit organisation in official relations with the World Health Organisation (*WHO*). It is currently represented in 91 countries, with headquarters in Nicosia, Cyprus.

TIF places great emphasis on promoting awareness of thalassaemia amongst the public and health professionals, and on promoting the establishment and improvement of policies aimed at the prevention and medical treatment of thalassaemia. TIF's support for national thalassaemia associations and its regular series of education programmes for scientists and medical specialists involved in thalassaemia have played a key role in fulfilling the Federation's objectives.

As part of its educational programme, TIF produces a range of scientific material on all aspects of the control and clinical management of thalassaemia, translated into a number of languages. More than 40,000 copies of TIF publications are distributed each year to member associations, scientific collaborators in medical centres around the world, medical schools, national health authorities and WHO offices.

In addition to WHO, Hereditary Disease Department, TIF is also in official relations with International Society of Blood Transfusion (ISBT), International Consortium for Blood Safety (ICBS) and WHO, Global Collaboration for Blood Safety (GCBS).

Other TIF publications

- Guidelines for the Clinical Management of Thalassaemia
Available in English, Greek, Spanish, Farsi and Arabic
- What is Thalassaemia?
Available in English, Greek, French and Spanish
- Compliance to Iron Chelation Therapy with Desferrioxamine
Available in English, Greek, Spanish, Italian, Farsi and Arabic
- Blood Kit
Available in English
- TIF Magazine (Quarterly a year)
Available in English
- About Thalassaemia
Available in English

Forthcoming TIF publications

- Prevention of Thalassaemias and Other Haemoglobin Disorders - Vol.2

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TIF can also provide details of national thalassaemia associations and medical centres treating thalassaemia around the world.