INTRODUCTION

The scene is a school leavers’ assembly at a 1500-pupil comprehensive school. The assembled pupils have a wide range of prospects: ranging from aspirant lawyers and teachers to those with antisocial behaviour orders. One of them was asked to make a short speech. The 16-year-old begins hesitantly but the 300 gathered listen respectfully. The address is short, clear and poignant, ‘I would like to thank everybody for helping me and being my friend’. There is rapturous applause. The boy leaves the stage, a little self-consciously but with a look of satisfaction. The gathering offers genuine warmth and delight in response to the lad’s words.

The young person in question has Down syndrome and has just passed through mainstream education. One teacher remarked that this pupil had offered more to this school than they had been able to give him. This reflects a sea change in societal attitude to children with Down syndrome over the past 40 years (only after the 1970 Education Act [UK] where children with intellectual disability entitled to an education at all). By the 1980s, ‘Mongol’ was only just leaving common parlance and a prejudicial view often entered medical decision-making processes: suboptimal treatments were reported for heart disease and leukaemia. Fortunately this has changed, reflecting a view that young people with Down syndrome have prospects, can contribute to society, and deserve respect, love and emotional support along with access to appropriate medical care.

Down syndrome is not a medical condition but represents a common recognizable variation of the human form created through a random biological event. Nonetheless, as we will see, people with Down syndrome present with many common medical conditions and some that are more specific to the condition. Doctors have an important part to play in the lives of the people concerned. They help parents readjust to a new set of expectations following the birth of an affected child. They can help with the early identification and treatment of conditions attendant on the syndrome.

We will illustrate through a number of case scenarios that, unfortunately, too often medical symptoms are attributed to the ‘syndrome’. This is termed ‘diagnostic overshadowing’ and results in a delay in the delivery of appropriate medical care. We hope this book will strengthen the knowledge of doctors and other health professionals who have this responsibility. Our book incorporates anecdotes and experiences, some good some bad, of service providers and users which illustrate good and bad practice, and missed opportunities. We hope these vignettes will help the readers reflect on their own attitudes and practice in our shared constant quest for professional development and improvement.

We begin with an overview of our current knowledge of the biology of Down syndrome. It is followed by an important contribution from the UK Down’s Syndrome Association.
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which explores the lives, experiences and at times frustrations of service users and how we might improve our practice. We then take a systematic approach to Down syndrome and its attendant medical problems—the biology, presentation and diagnostic formulations; and how we might intervene in a timely fashion. Presentations can be different in Down syndrome; the important thing is to recognize this so that opportunities to help are not missed.

An underlying theme is how we might initially set the scene for a loving and nurturing environment at home and then develop this into opportunity and encouragement outside the home as the children get older. We believe professionals will best be able to fulfil their part in this task by first examining and reflecting on their own views of disability in general and Down syndrome in particular. An increased understanding of Down syndrome should allow us as professionals to support the person at the centre and their families; and to present information and interventions clearly and positively enabling them to lead a fulfilling life.