The Spina Bifida: Management and Outcome
The Spina Bifida
Management and Outcome

Forewords by
C. Sainte-Rose
C. di Rocco

Preface by
M. Necmettin Pamir
For 700 little candles having shed light on my path and assisted me to do it right

M.M. Özek

To my mother, to Fabrizia, Francesco and Maria Allegra

G. Cinalli

To my parents, who through their love have allowed me to realise my dreams
To my daughter, the most beautiful of all my dreams

W.J. Maixner
As we stand at the dawn of the 21st century, one may ponder the rationale of writing a book on spina bifida. Once commonplace in European countries prior to the era of ultrasonography, this disease became increasingly rare in developed countries as a result of improvements in antenatal diagnosis, to the point that we believed it to be disappearing. Knowledge of spina bifida and of its treatment, once so richly diffused only 30 years ago, began to fade. Young neurosurgeons who had never seen such a malformation at its initial presentation were hesitant, and did not understand the protean clinical signs of these patients presenting to the emergency department or outpatient clinics. This situation, however, did not last for long. As a consequence of the political and economic events of the final years of the 20th century, the advent of globalisation, and the significant desire for immigration, we realised that spina bifida had not disappeared at all in the rest of the world. Migration was, and is, bringing it back onto our doorstep, to our everyday clinical and surgical practice. It is important therefore, not to lose the knowledge gained by our masters, to try and assemble it in one place in order to understand the disease from its inception in utero through until adulthood and the reproductive age.

The chronology of the book, in reflection of this aim, is well organised, and ranges from history and embryology, to prenatal diagnosis and treatment, perinatal care, initial treatment and management, and middle- and long-term complications, and finally provides insights for the future. The authors, by not merely focusing on the neurosurgical issues but also on the urologic and orthopaedic consequences of this malformation, have allowed for a more global approach to these patients.

Through a greater understanding of the disease, through the improved quality of initial care, and through the dedication of the surgeons and physicians who manage these children from infancy into adulthood, children inflicted with spina bifida, whilst heavily burdened, may nevertheless lead happy and full lives.

Paris, May 2008

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Spina bifida refers to a cohort of pathological conditions that vary in severity from the dramatic and life-threatening myelomeningoceles to relatively mild defects such as dermal sinuses. This book aims to cover the whole spectrum although, as expected, most of its content is devoted to the management of myelomeningocele. With regard to this last type of malformation, the treatment of very few diseases in the history of medicine has been characterized by such an intimate relationship between scientific and technical problems and ethical and social considerations. Myelomeningocele is one of a very few congenital malformations that require the neurosurgeon and parents to face the fundamental dilemma of whether to promote survival, at a very high cost, or to deny treatment in the belief that it is better to prevent the newborn suffering unacceptable and lifelong physical and emotional suffering. It is unusual as it has been associated with such mutually exclusive therapeutic approaches – from treatment refusal to the use of sophisticated surgical and rehabilitative care. The management of the disease has been marked by oscillations between hope and dejection, despair and jubilation. These opposing emotions are also likely to affect the individual surgeon in his/her professional life, with cases of early success turning to late failures and, conversely, cases of satisfactory lives with rich social interaction following initially poor prognoses.

Before the 1960s, in common with other children with hydrocephalus, newborns with myelomeningocele had little hope of survival. Indeed, most of the affected subjects died from the uncontrolled progression of the associated ventricular dilation; death also resulted from sepsis, meningitis, and renal failure. The same secondary complications accounted for the extremely serious disabilities that blighted survivors’ lives, in addition to the congenital neurological, orthopedic, and urologic deficits. It is not surprising, therefore, that at the beginning of the 1970s Lorber commented pessimistically on a series of patients followed in the previous decade. He considered that only 7% of the survivors had acceptable disabilities, while the majority of them “had a quality of life inconsistent with self-respect, learning capacity, happiness, and even marriage”. A generation of pediatricians as well as the public opinion of the time was deeply influenced by the so-called “Lorber’s selection criteria”. Macrocrania, severe paraplegia, severe kyphosis or scoliosis, the presence of concomitant significant congenital anomalies, or the history of birth damage were all regarded as criteria for excluding any active treatment for myelodysplastic newborns. The echo of this negative attitude can also be found in more recent experiences, as demonstrated by the “Baby Jane Doe” case at the beginning of the 1980s, and by the “Baby Rianne” case in 1993. In this context, the protocol approved by the Dutch Association of Paediatrics in 2005, known as the Groningen protocol, on deliberate life termination for newborns with severe forms of spina bifida, is even more significant.
On the other hand, although these years were dominated by this negative attitude in most centres, some neurosurgical departments produced results that were much more optimistic. In a small number of neurosurgical centres, series of surgical unselected patients were managed with more advanced techniques and a multidisciplinary approach. The results unequivocally demonstrated that most of the children operated on could reach normal levels of intellectual ability, and that even individuals with severe myelomeningoceles could, after this treatment, lead meaningful lives.

These patients benefited from the surgical advances made in assuring a more effective closure of the back defect. They also had the advantage of improved control of the impaired cerebrospinal fluid (CSF) dynamics by means of more reliable CSF shunting apparatus, and, recently, endoscopy, early recognition and treatment of complications such as Chiari type II malformation or syringomyelia, and prevention and early treatment of spinal cord retethering. They gained further benefit from timely orthopedic correction for club feet, scoliosis, or kyphosis, as well as adequate prevention of damage to urological function. Furthermore, they also received strong support for reaching independence and social integration from the development of ad hoc rehabilitation and educational programs, which were additionally promoted by the establishment of myelomeningocele clinics; more generally, they also benefited from the combined efforts of multidisciplinary teams assisted by a more sympathetic public opinion.

The spirit that has inspired multidisciplinary myelomeningocele teams over the last two decades can be found in this multiauthored volume devoted to the management of spina bifida. The book conveys all the best information currently available in the field, and integrates the most specialized knowledge, from the basic sciences to the various surgical, medical, and psychosocial skills, in a unique well-organized source of information.

As a result of understanding the role of alpha-feto-protein in preventing spina bifida, and the introduction of routine prenatal ultrasound diagnosis, the number of children born with myelomeningocele is continuously decreasing in many western countries. This phenomenon is likely to result in a decreased interest in the management of spina bifida and there will almost certainly be fewer specialized scientific contributions. Consequently, the experience accrued over recent years faces the risk of not being sufficiently updated. Spina bifida, however, still continues to represent an important problem in several countries where preventive measures have not yet been adopted. In these countries, the need for specialized knowledge will persist for the foreseeable future. The present book then may cover an impending gap by helping to preserve the body of relevant scientific knowledge and expertise acquired over the last three decades in a society that may not have so much need of it in future years. By transmitting this skill and knowledge to countries that need it the book will help these countries to avoid retracing the false steps that in the past prevented many children with spina bifida from reaching the best possible outcomes.

Rome, May 2008

Concezio Di Rocco
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Preface

By M. Necmettin Pamir

This book presents the current understanding, diagnosis and treatment of spina bifida and related pathologies to the neurosurgical literature.

The book has two important qualities: first of all it provides a thorough analysis of the historical experience in the field, and secondly it presents to the reader the most up-to-date conclusions, standards, and trends. Although this disease has been known to mankind since antiquity, the treatment is still not straightforward and experts still disagree on various fields. The discussion of modern diagnostic technologies, treatment modalities, and complications, presented in 39 chapters, will guide the reader to tailor an optimal treatment strategy to their patients. The book covers a wide range of topics in the field, starting from preventive measures and progressing to issues like the social adaptation of the patients. The extremely detailed nature of each chapter will be easily appreciated by the reader, whether this is the neurosurgeon, pediatrician, pediatric neurologist, pathologist, neuroradiologist, or any other professional who is involved in the diagnosis or treatment. Therefore, the reader will find answers to almost all his questions on spina bifida and related pathologies in this book. In this regard, it is a significant contribution to the medical literature.

I take great pleasure in congratulating the editors, Professor Özek, Professor Cinalli and Professor Maixner for the quality of the book and for their superb and unique work. It is an honor for me to introduce this excellent book to neurosurgical literature.

Istanbul, May 2008

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Preface

By the Editors

Spina bifida has been an issue of concern for thousands of years. The treatment for this malady begins at birth, or in some cases even before birth, and continues throughout the patients’ lives. Over years of investigations and studies in our spina bifida outpatient clinics, we have come to realize the importance of understanding and preparing for the long-term difficulties awaiting our patients. As we are aware that the treatment is multidisciplinary, it is no surprise that the spark of an idea for a spina bifida book was generated during meetings between members of the subspecialties in our spina bifida team. With this inspiration, the decision was shaped further and finalized during the 2006 ESPN meeting in Martinique.

With the aim of promoting the academic success of our book, deciding on the authors for the specific chapters and depending on their expertise was of utmost importance. For this reason, we would like to acknowledge the invaluable contribution of all our authors. We express our heartfelt gratitude to Springer-Verlag Italy, and particularly Dr. Donatella Rizza and the whole editorial team for their skillfulness and tolerance. Lastly, we would like to sincerely thank our patients and their families from whose endurance we get all our clinical experience.

May 2008

M. Memet Özek
Giuseppe Cinalli
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