Neurocognitive Rehabilitation of Down Syndrome

The Early Years
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Edited by
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To the memory of our friend
Professor Krystina Wisniewski
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Preface

The study of Down syndrome, the most frequent genetic disorder affecting development, has led to a rich body of interdisciplinary research in genetics, neuroscience, psychology, and education. These collaborations have not only promoted a better understanding of the condition itself, but also favored an increasing recognition that many neurodevelopmental disorders have strong genetic components even though their genetic underpinnings still need to be better understood and the characteristics of their neurocognitive developments better specified. Fifty years of intensive research on Down syndrome following the discovery of the genetic basis of the syndrome have lead to a reasonable understanding of many of its major developmental aspects. On this basis, it has become possible to define an interdisciplinary framework for locating, justifying, and assessing early rehabilitative intervention.

The purpose of this book is to represent some of the major ways in which a comprehensive neurocognitive rehabilitation program may be conceptualized and carried out, taking into account the spectrum of specific knowledge available from the genotype, brain development, and the behavioral phenotype. By comprehensive, we mean a neurocognitive approach connected transactionally with the major therapeutic endeavors in neighboring fields such as neurogenetics, experimental environmental enrichment in animal models, molecular and genic therapies (viewed as synergistic with neurocognitive rehabilitation), pharmacology, pediatrics, and cardiology for infants with Down syndrome.

The book is divided in five sections with the aim of helping to orient the reader and classify the wealth of information provided. Each chapter is followed by a summary and a complete list of references.

Section 1 deals with definition, methodology, and assessment issues. Chapter 1 proposes a definition of early rehabilitative intervention, its age limits, objectives, models of action, and target groups. It also examines the practical challenges that early rehabilitation presents in the short and medium term. Chapter 2, summarizes 50 years of practice of rehabilitative intervention for infants and children with Down syndrome and the changes and progress witnessed in this evolution. Chapter 3 reviews current knowledge of the cognitive profile of Down syndrome, discusses recent advances in our understanding of the pathways that may be potential targets for treatment, and details the ideal properties of assessments for these interventions. It also presents the Arizona Cognitive Test Battery, a set of primarily nonverbal neuropsychological assessments, and details additional assessments that could be included in the context of a clinical trial.

Section 2 deals with genetics, brain, and animal models relating to early neurocognitive rehabilitation. Chapter 4 exposes and discusses new experimental perspectives of molecular and genic therapies in Down syndrome. Chapter 5 analyzes the outcomes of a number of recent works on animal models in Down syndrome. The authors discuss the effect of environmental enrichment for alleviating some of the molecular abnormalities found in Ts65Dn mice, suggesting that it might also have therapeutic potential in children with Down syndrome. Chapter 5 examines the question of adequate nutrition and food supplement in the mother and the fetus during fetal (and before for the mother) and postnatal development, showing that it has profound influences on brain and nervous system development.
Section 3 is devoted to pharmacological and medical management and treatment. Chapter 7 analyzes recent advances in pharmacotherapy for children with Down syndrome, dealing particularly with cognitive enhancement. Pharmacological agents targeting GABA and glutamate receptors and dopamine transporters hold promise for advancing toward clinical testing. Chapter 8 centers on early medical caretaking and following up of children with Down syndrome. Chapter 9 discusses the pros and cons of cardiac surgery in infants with Down syndrome in the light of recent progress in surgical techniques and postoperative intensive care. It appears that congenital heart defects in babies with Down syndrome can be repaired with a very low risk of mortality and morbidity.

Section 4 is concerned with an analysis of key aspects of early neurocognitive rehabilitation. Chapter 10 exposes the basic principles of the developmental theoretical approach, which provides a relevant conceptual instrument for assessing development and interpreting the impact of neurocognitive rehabilitation with children with Down syndrome. Chapter 11 is concerned with motor development and rehabilitation. The difficult problem of hypotonia in children with Down syndrome is addressed together with an analysis of technical ways for reducing its negative incidence on neurocognitive development. Chapter 12 focuses on the characteristics of long- and short-term memories in children with intellectual disability and, particularly, children with Down syndrome. Findings suggest that specific intervention procedures can markedly improve their memory development and functioning. Chapter 13 draws on the research literature on typical development to recommend a number of steps and strategies in the very early training of babies and children with Down syndrome and corresponding congenital genetic conditions leading to intellectual disability and language difficulties. The chapter also deals with conventional aspects of language development in children with Down syndrome, such as prelexical and early lexical development, pragmatics, and grammatical patterning. Chapter 14 explores the work relating to speech perception in Down syndrome and argues that in order to be able to design effective early rehabilitation methods, speech perception in this population needs to be more comprehensively investigated. Chapter 15 stresses temperament and character issues in designing effective early rehabilitation programs for infants and children with Down syndrome. Chapter 16 analyzes the roles of parents in participating actively in the training and education of their Down syndrome children, given that the effectiveness of early rehabilitation is highly associated with the impact it has on parents' level of responsiveness to their children. The ways parents interact with young children with Down syndrome account for a major portion of the variability of the cognitive and communication outcomes these children attain during the first years of their lives.

Lastly, Section 5 is on therapeutic perspectives. Chapter 17 analyzes the prospects for genetic therapies in Down syndrome and stresses the necessity to keep providing strong neurocognitive rehabilitation in a future interdisciplinary framework labeled "hybrid genetic–neurobehavioral strategies," meant to improve decisively the biological and psychological functioning of the person with Down syndrome.
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