

Down Syndrome: A Multidisciplinary Perspective

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Abstract

Trisomy 21, a genetic disorder resulting from a chromosomal abnormality, is one of the most common forms of mental disability in the United States. Individuals with Down syndrome frequently present with a constellation of medical problems including conductive hearing loss and, to a lesser degree, sensorineural hearing loss. As part of a health care team, audiologists must be sensitive to and aware of medical conditions prior to establishing intervention strategies. Medical conditions, by necessity, precede audiologic interventions and, therefore, a close working relationship among team members is critical. Yet, audiologic and communication interventions should be established at the earliest possible time for maximizing an individual's development potential. This article stresses the importance of a multidisciplinary team in the provision of services so that prevention of further disabilities, improved outcomes of medical interventions, and appropriate habilitative and educational planning may ensue.

Key Words: Amplification, audiologic intervention, Down syndrome, hearing impairment

Trisomy 21, a genetic disorder resulting from a chromosomal abnormality, is one of the common forms of mental disability in the United States, with an incidence of approximately 1/700 live births. In 1866, Langdon Down prepared the first adequate description of this syndrome — hence, the name, Down syndrome. The correct diagnosis of Down syndrome can often be made at birth, fostering early identification and immediate habilitative planning.

Otolaryngologic and audiologic problems are common in Down syndrome and constitute potential additional disability. These topics are the focus of this paper and emphasize the importance of early and regular re-evaluation of the audiologic status of every child with Down syn-

drome. Skilled pediatric audiologic and otolaryngologic management in association with appropriate communication intervention can greatly enhance the social and cognitive function of children with Down syndrome.

GENETIC CONSIDERATIONS

The genetic imbalance leading to Down syndrome can be represented by either Full 21 trisomy (nondisjunction), Partial 21 trisomy (translocation), or mosaicism (postfertilization nondisjunction). When nondisjunction occurs during the first meiotic division of reproductive cells, both #21 chromosomes end up in one cell. Instead of both cells having 23 chromosomes, one cell has 24 chromosomes, and the other has 22. The loss of a #21 chromosome makes it highly unlikely for an egg containing 22 chromosomes to survive. However, the egg with 24 chromosomes can survive. Union with a cell containing 23 chromosomes results in a child with 47 chromosomes who has Down syndrome or trisomy 21.

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Down Syndrome A Multidisciplinary Perspective

Harish Pemde, Vikram Datta



Down Syndrome A Multidisciplinary Perspective:

Down Syndrome Qadoshyah Fish, 2008-11 People with Down syndrome can do a lot this is the reality We will share with you in this book stories from people with Down syndrome who have excelled in many areas and live lives like many other people who do not have Down syndrome We will also share with you stories about those with Down syndrome who have had medical problems and issues to deal with yet give joy to those they come in contact with Preface **Down Syndrome, Neurodegeneration and Dementia** Sylvia Eva Perez, Tao Ma, Elliott Jay Mufson, Stephen D. Ginsberg, Aurélie Ledreux, 2022-01-27 **New Perspectives on Down Syndrome** Siegfried M. Pueschel, 1987 **Shafer's Textbook of Oral Pathology - E Book** B Sivapathasundharam, 2016-07-25 Extensively revised and updated contents Advanced information scattered throughout the book in highlighted boxes Removal of outdated data Addition of more than 200 colour pictures Re categorization of cysts of the oral cavity Odontogenic keratocyst and Dentinogenic Ghost cell tumour topic has been updated Nonepithelial Benign and Malignant tumours of the oral cavity are discussed as a separate chapter Topic on stem cells has been revised and updated Neuro Rehabilitation - A Multidisciplinary Approach Mr. Rohit Manglik, 2024-07-30 Presents a multidisciplinary perspective on neurorehabilitation covering physiotherapy occupational therapy speech language therapy and psychological interventions Dementia and Aging Adults with Intellectual Disabilities Matthew P. Janicki, Arthur J. Dalton, 2014-04-23 First published in 1999 Routledge is an imprint of Taylor Francis an informa company HCI for Children with Disabilities Josefina Guerrero-Garcia, Juan Manuel González-Calleros, Jaime Muñoz-Arteaga, César A. Collazos, 2017-05-03 In this book the authors present an HCI principle based approach to develop applications to assist children with disabilities Design knowledge related to developing complex solution for this audience is explained from an interaction design point of view Different methodologies models and cases studies are covered with the aim of helping practitioners to adopt any of the proposed techniques presented in this book HCI methodologies that adopt an agile strategy are presented including novel techniques at different development steps such as board games agile planning agile implementation method engineering As this is a huge research field the authors do not just focus on a specific disability but test their methods in different contexts with excellent results Readers of this book will find both a well organized and structured set of methodologies and also material that has been tested and refined throughout years of research Using detailed case studies the reader is guided towards specific solutions which will also provide insights into how to address related problems **Browse's Introduction to the Investigation and Management of Surgical Disease** Norman L. Browse, John Black, Kevin G. Burnand, Steven A. Corbett, William E. G. Thomas, 2010-10-29 A companion to the award winning Browse s Introduction to the Symptoms and Signs of Surgical Disease this volume provides clear insight into how the pathological features and extent of disease dictate necessary diagnostic investigations and treatment Like its established partner it is a practical and concise textbook that is easy to read and use The content is highly structured with each chapter

divided into three sections Relevant Pathology provides a brief description of the important pathological features of the disease relevant to the clinical diagnosis investigations and treatment Investigations covers significant clinical findings including radiological biochemical haematological immunological pathological and genetic Treatment reviews all aspects of management Other key features include more than 300 high quality clinical photographs and 60 color line drawings to enhance textual explanations as well as learning and review boxes to aid understanding and provide a tool for self assessment The book is essential reading for all medical students undertaking a surgical rotation or preparing for their surgical exams

IAP Colour Atlas of Pediatrics A Parthasarathy,Rohit Agrawal,2012-04 Colour atlas pn paediatric illnesses and management covering both common and rare disorders

The Netter Collection of Medical Illustrations: Integumentary System, Volume 4 - E-Book Bryan E. Anderson,2023-11-24 Offering a concise highly visual approach to the basic science and clinical pathology of the integumentary system this updated volume in The Netter Collection of Medical Illustrations the CIBA Green Books contains unparalleled didactic illustrations reflecting the latest medical knowledge Revised by Dr Bryan E Anderson Integumentary System Volume 4 integrates core concepts of anatomy physiology and other basic sciences with common clinical correlates across health medical and surgical disciplines Classic Netter art updated and new illustrations and modern imaging continue to bring medical concepts to life and make this timeless work an essential resource for students clinicians and educators Provides a highly visual guide to the skin hair and nails from normal anatomy and histology to pathology dermatology and common issues in plastic surgery and wound healing Covers new topics throughout including infantile hemangiomas COVID 19 porphyria cutanea tarda and Muir Torre syndrome Provides a concise overview of complex information by integrating anatomical and physiological concepts with clinical scenarios Compiles Dr Frank H Netter s master medical artistry an aesthetic tribute and source of inspiration for medical professionals for over half a century along with new art in the Netter tradition for each of the major body systems making this volume a powerful and memorable tool for building foundational knowledge and educating patients or staff NEW An eBook version is included with purchase The eBook allows you to access all of the text figures and references with the ability to search make notes and highlights and have content read aloud

Families and Their Children with Down's Syndrome Elizabeth A. Byrne,Cliff Cunningham,Patricia Sloper,1988 The authors present research based on interviews with families in the Manchester Down s Syndrome Cohort given to professionals involved in the welfare of children with Down s Syndrome or in the provision of community care and support for their families

Development of Movement Coordination in Children Geert Savelsbergh,Keith Davids,John van der Kamp,Simon J. Bennett,2013-04-15 Co ordination of movement plays a key role in human development and is an important area in sport and health sciences This book looks in detail at how children develop basic skills such as walking and reaching for objects and more complex skills such as throwing and catching a ball accurately or riding a bicycle Development of Movement Co ordination in Children is informed by five major theoretical perspectives

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IAP Color Atlas of Pediatrics A Parthasarathy,2015-08-31 IAP Color Atlas of Pediatrics is the latest edition of this comprehensive and highly illustrated guide to paediatric care The book is divided into 22 paediatric specialties Within each section topics are organised alphabetically for ease of reference Topics covered include neonatology growth and development nutrition infectious diseases all of the major organ systems of the body from neurology to endocrinology genetics allergy adolescent health and medicine paediatric surgery and orthopaedics This new edition emphasises the early and rapid diagnosis of various paediatric illness and features two new chapters one in the growth and development section and the other in the haematology section concerning blood component therapy Enhanced by 1240 full colour illustrations images and tables this comprehensive atlas is an ideal resource for students of paediatrics and practitioners Key Points Comprehensive and highly illustrated atlas covering all paediatric specialities Range of conditions covered in alpha order 1240 full colour images illustrations and tables Latest edition featuring two brand new chapters First edition 9789350257104 published 2012

Pediatric Neuropsychological Intervention Scott J. Hunter,Jacobus Donders,2007-05-03 This 2007 volume provides an overview of evidence based treatments for common pediatric brain behavior disorders

Developmental Motor Disorders Deborah Dewey,David E. Tupper,2004-09-21 Bringing together leading experts and providing vital insights to guide clinical practice this is the first volume to comprehensively address childhood motor disorders from a neuropsychological perspective The book explores the neural and behavioral bases of movement disorders and summarizes current findings from applied research Existing approaches to assessment and neuroimaging are critically examined and new and innovative methods presented Authors also synthesize the latest knowledge on motor difficulties associated with specific developmental and neurological problems cerebral palsy neuromuscular disease autism brain injury disorders of coordination speech and written language and more Other important topics covered include psychosocial effects of motor skills impairments frequently encountered comorbidities and the status of available intervention approaches

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The New Public Health Theodore H. Tulchinsky,Elena A. Varavikova,Matan J. Cohen,2023-01-21 Selected for Doody s Core Titles 2024 in Public Health The New Public Health has established itself as a solid textbook throughout the world Translated into seven languages this work distinguishes itself from other public health textbooks which are either highly

locally oriented or if international lack the specificity of local issues relevant to students understanding of applied public health in their own setting Fully revised the Fourth Edition of The New Public Health provides a unified approach to public health appropriate for graduate students and advance undergraduate students especially for courses in MPH community health preventive medicine community health education programs community health nursing programs It is also a valuable resource for health professionals requiring an overview of public health Provides a comprehensive overview of the field illustrated with real life specific examples Updated with new case studies and examples from current public health environment in North American and European regions Includes detailed Companion website <https://www.elsevier.com/books-and-journals/book-companion/9780128229576> featuring case studies image bank online chapters and video as well as an Instructors guide

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Pediatric Anesthesia: A

Problem-Based Learning Approach Kirk Lalwani, Ira Todd Cohen, Ellen Y. Choi, Vidya T. Raman, 2018-09-17 Linking existing knowledge to new knowledge by presenting it in the form of a case or a problem is a popular and effective educational approach resulting in better retention of the knowledge and improved ability to apply that knowledge to solve real problems This problem based learning PBL method was introduced into medical education at McMaster University in Ontario Canada in 1969 Since then it has been widely incorporated into secondary undergraduate and graduate education in a variety of disciplines worldwide This new volume for the Anesthesiology Problem Based Learning series reviews pediatric anesthesia utilizing the PBL approach Each chapter deals with conditions and problems in pediatric anesthesia practice presented as a case stem with questions to encourage critical thinking followed by an evidence based discussion and multiple choice questions for self assessment Cases were carefully selected to present a broad systems based tour of commonly encountered clinical cases in pediatric anesthesia The book can be used to review an upcoming clinical case or as a PBL tool The Stem Case and Key Questions and Discussion sections can serve as the basis for interactive learning experiences for study groups or as a broad yet in depth clinical review of the subspecialty for the individual learner Self assessment questions can be used as a measure of knowledge acquisition or simply as a question bank to prepare for examinations

Developmental Behavioral Neuroscience Megan R. Gunnar, Charles A. Nelson, 2013-04-15 This volume provides an introduction to current research on the relation between brain development and the development of cognitive linguistic motor and emotional behavior At least two audiences will benefit from this book psychologists interested in brain development and neuroscientists interested in behavioral development Although each chapter is content oriented the volume as a whole provides a well integrated summary of the latest findings from developmental behavioral neuroscience

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