Hans J. ten Donkelaar
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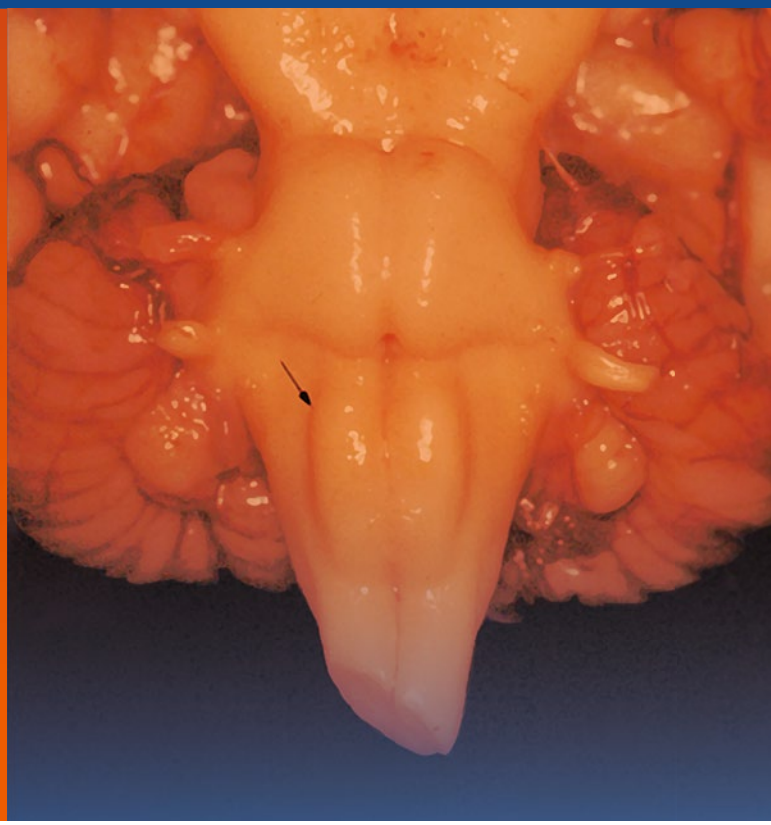

# Clinical Neuroembryology

## Development and Developmental Disorders of the Human Central Nervous System

*Second Edition*

Springer

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Hans J. ten Donkelaar • Martin Lammens
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ISBN 978-3-642-54686-0 ISBN 978-3-642-54687-7 (eBook)
DOI 10.1007/978-3-642-54687-7
Springer Heidelberg New York Dordrecht London

Library of Congress Control Number: 2014940543



Printed on acid-free paper

Springer is part of Springer Science+Business Media (www.springer.com)

# Preface to the Second Edition

Apart from a general updating of the extensive literature on developmental neurobiology, neurogenetics, imaging and developmental neuropathology between 2005 and 2013, more emphasis has been given to: (a) imaging of the embryonic brain (early prenatal diagnosis by ultrasound); (b) imaging of the fetal brain by MRI; (c) DTI studies on the development of major fibre connections such as the pyramidal tract and the corpus callosum; and (d) the impact of newer genetic techniques such as whole exome/genome sequencing. Moreover, new classifications of brain disorders have been implemented such as a new classification of midbrain-hindbrain developmental disorders and entire new families of disorders such as ciliopathies and dystroglycanopathies. Throughout the book, several new Clinical Cases have been added.

Several colleagues kindly contributed as new co-authors their expertise to this second edition, including Eleonora Aronica (Amsterdam), Mireille Bekker (Nijmegen), Kyoko Itoh (Kyoto), Karin Kamphuis-van Ulzen (Nijmegen), Irene Mathijssen (Rotterdam), Ronald Pennings and Hans van Bokhoven (Nijmegen), Patrick van der Voorn (Amsterdam) and Shigehito Yamada (Kyoto). They also contributed new Clinical Cases. For other new Clinical Cases, the help of Remke Dullemond (Rotterdam), Janet Eyre (Newcastle), Floris Groenendaal (Utrecht), Gregor Kasprian (Vienna), Hajime Miyata (Akita), Peter Nikkels (Utrecht), Tetsu Niwa (Yokohama), Andrea Poretti (Zurich), Ritsuko Pooh (Osaka), Goran Simić (Zagreb) and Marjolein Willemsen (Nijmegen) is gratefully acknowledged. New illustrations were also kindly provided by Marco Catani and Michel Thiebaut de Schotten (London), Cyrille Ferrier (Utrecht), Hao Huang (Dallas), Ole Kiehn (Stockholm), Grace Lai (New York), Anna Lavezzi (Milan) and Maria Thom (London). A long weekend with Luis Puelles in Murcia greatly helped the first author to implement new findings on the prosomeric model of the developing brain.

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# Preface to the First Edition

The spectacular progress in developmental neurobiology, the tremendous advances in (neuro) genetics and the high resolution of the modern imaging techniques applicable to developmental disorders of the human brain and spinal cord have created a growing interest in the developmental history of the central nervous system (CNS). This new book provides a comprehensive overview of the development of the human CNS in the context of its many developmental disorders due to genetic, environmental and hypoxic/ischemic causes. The book contains three general, introductory chapters in which an overview of the development of the human brain and spinal cord, a summary of mechanisms of development as obtained in experimental studies in various invertebrates and vertebrates, and an overview of the causes of congenital malformations with some notes on prenatal diagnosis, are presented. The developmental disorders of the human brain and spinal cord are presented in a regional, more or less segmental way, starting with neurulation and the neural tube defects, and ending with developmental disorders of the cerebral cortex. These chapters are abundantly illustrated with clinical case studies with imaging data and, when available, postmortem verification of the developmental disorders involved. The book is intended for advanced medical students, and all those clinicians working with children and adults with developmental disorders of the CNS.

This book would not have been possible without the help of many colleagues in The Netherlands and from abroad. Their help is gratefully acknowledged. Most of the neuropathological material comes from the extensive collections of Drs. Akira Hori and Martin Lammens. Many cases were kindly provided by Drs. Pieter Wesseling (Nijmegen), Gerard van Noort (Enschede), and Kohei Shiota (Kyoto). Photographical assistance was provided by Mrs. Roelie de Boer-van Huizen (Nijmegen), Mrs. Chigako Uwabe (Kyoto) and Richard Rieksen (Enschede). Material for the clinical case studies was provided by many clinical colleagues, including Drs. Ellsworth C. Alvord Jr (Seattle), Harm-Gerd Blaas (Trondheim), Cor Cremers and Hans Cruysberg (Nijmegen), Mark D'hooghe (Bruges), Jennian Geddes (London), Ben Hamel (Nijmegen), Frans Hoevenaars (Nijmegen), Nomdo Jansonius (Groningen), Akiyoshi Kakita (Niigata), Max Kros (Rotterdam), Hajime Miyata (Tottori), Masashi Mizuguchi (Tokyo), Reinier Mullaart,Willy Renier and Jan Rotteveel (Nijmegen), Harvey B. Sarnat (Calgary), Ben Semmekrot (Nijmegen), Waney Squier (Oxford), Sachio Takashima (Fukuoka), Rudy van Coster and Caroline Van den Broecke (Gent), Christl Vermeij-Keers (Rotterdam), Michel Willemsen (Nijmegen), and Mieko Yoshioka (Kobe). Imaging data were kindly provided by Drs. Harm-Gerd Blaas (Trondheim), Berit Verbist (Leiden), John van Vugt and collaborators (Amsterdam), Henk Thijssen and Ton van der Vliet (Nijmegen), and Guido Wilms (Leuven). Several figures were contributed by Drs. Jo Curfs (Nijmegen), Marieke de Heer and Jeannette Hoogeboom (Rotterdam), Raoul Hennekam (Amsterdam), Jan E. Jirasek (Prague), Enrico Marani (Leiden), Loreta Medina (Murcia), Zoltán Molnár (Oxford), Ronan O'Rahilly (Villlars-sur-Glâne), Annemieke Potters (Deventer), Kohei Shiota (Kyoto), Henny van Straaten (Maastricht), Michiel Vaandrager (Rotterdam), Jan Voogd (Oegstgeest), and Shigehito Yamada (Kyoto). Most of the drawings were made by Mrs. Marlu de Leeuw and Mr. Ad Gruter. Financial support was generously provided by the "Stichting Neurologie en Wetenschap" of the Department of Neurology (Head: Prof. Dr. George W.A.M. Padberg) and the Department of Pathology (Head: Prof. Dr. Han van Krieken), both of the Radboud University Nijmegen

Medical Centre, supporting the costs of the drawings. The Japan Society for the Promotion of Science granted the first author a short-term fellowship in May 2004 at the Congenital Anomaly Research Centre (Head: Prof. Dr. Kohei Shiota) of Kyoto University.

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