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# Preface

The following papers were invited during a three-day workshop at the Swedish Rett center in June 2003. This beautiful situation among the lakes and mountains of northern Sweden provided an ideal environment for detailed consideration and discussions of the early deviations in Rett disorder. Due to the time lag between the meeting and publication, some papers contain the experience gained over the intervening period since the meeting. However, all were influenced by those presentations and discussion, and since the theme is a powerful one it was agreed to publish them together in this volume, following independent peer review.

Onset is clearly of the greatest importance to a correct understanding of any disorder and in the Rett disorder this has presented difficulties because the attractive appearance and quietly suboptimal behaviour of the child are such that the first signs are readily overlooked until the sudden episode of developmental regression which may not occur until the second year or later if at all. Objective evidence of the earliest effects of the disease on the brain has therefore been hard to display although experienced parents and developmental health professionals have consistently held that these children do show signs from birth. This workshop was planned in order to examine the existing evidence about the period of early childhood, the implications for the later course of the disease and for present, and potential intervention.

Some of the original papers provide information which relies on parents' and physicians' reports. Studies by Leonard et al, Larsson et al and Kerr & Prescott have taken this approach. A further valuable resource has been a collection of more than 80 very early family videos donated for this research. This video material was recorded by parents as part of their family archive, in most cases without initial awareness of the presence of a developmental disorder. It is therefore truly objective and although not standardized can be readily compared with video of healthy children taken under the same conditions. Studies by Einspieler et al and Burford have used this material, applying methods well tried in other conditions. The paper by Julu and Witt Engerström, continuing earlier work on the brainstem defects in Rett, presents an analysis of the several types of autonomic abnormality which become obvious at regression and are believed to account for some sudden, unexpected deaths.

Early anatomical and neurochemical changes in the human brain can only be deduced from the studies of autopsied brains of mid-childhood to adult life. Papers from Armstrong and Kaufmann et al, present this evidence and consider the light thrown by it on earlier events. Papers by Segawa and Nomura focus on early neurological and neurophysiological features and discuss the pathophysiology. Trevarthen reviews the psychological literature comparing Rett and autism, curiously and significantly different and yet parallel conditions in their pervasive and developmental character.

Finally, short case reports present pilot studies by Wigram & Lawrence, Lotan & Shapiro, Budden et al and Elefant & Wigram describing interventions found to be of value in the early stages and throughout the disorder.

One result of this workshop has been to encourage further research on the earliest changes in Rett disorder and to plan a further international workshop on early intervention, physical and pharmacological. It has also stimulated lively discussion on valid objective measures with which to evaluate the efficacy of any treatment regime.

Together these papers represent the first collection of essays dealing specifically with the early signs of Rett disorder and the neuroscience which underlies that. In doing so they finally lay to rest the early misconception that the brain shows no signs of the disease at birth and highlight the importance of pursuing the investigation of this very early period which determines the character of much that follows.

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*GUEST EDITORS:*
Yoshiko Nomura*
Alison Kerr
Ingegerd Witt Engerström


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* Corresponding author. Tel.: +81 3 3294 0371; fax: +81 3 3294 0290
E-mail address: nomura-y@segawa-clinic.jp