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Preface

Special issue: Molecular basis of NCL

The neuronal ceroid lipofuscinoses (NCL), also known as Batten disease, are a group of rare and devastating neurodegenerative disorders that can strike at any age, from before birth up to late in life. They are inherited, usually in an autosomal recessive manner, and at present are incurable, with no treatment even to halt disease progression.

This special issue was initiated by the NCL Foundation of Germany and arises from the last international meeting on the NCLs—the 10th international congress, NCL-2005—which was held in Helsinki, Finland, and attended by experts from around the world, and comprises a series of reviews. One review (Haltia) deliberately looks to the past, so that relevant knowledge and experience in an increasingly molecular age are not forgotten, but most look forward as progress is made in understanding the molecular basis of the NCLs. The majority of reviews are deliberately co-authored to reflect the cooperative international expertise that exists, a particular feature perhaps of research into a rare disease that is still such a challenge to understand. The aim of these reviews is to provide articles that summarise the current status of the NCL field.

The molecular basis for the NCLs is not fully understood. This collection of diseases was first recognised as a separate group in the 1960s, and is now known to cover disease caused by mutations in seven human genes, with more expected to exist. Mutations in many of these genes may be severe to mild, resulting in a range of ages of onset and rates of disease progression, and even disease of a very different clinical course (Siintola et al.). The diagnosis of NCL is on the verge of a small revolution as more genes are identified, and the variety of disease phenotypes is expanding, making this a challenging area for clinicians faced with diagnosis of such a rare disease, albeit with some common forms (Williams et al.). Mutations in other genes cause NCL or NCL-like disease in animals. One recent focus of research has been to identify animal models for each NCL disease, both small (Cooper et al.) and large (Tammen et al. and Houweling et al.) and to study these to understand the disease itself (Jalanko et al.). Future work will increasingly use these to develop and test out new therapeutic ideas (Hobert and Dawson). Another recent focus has been the development of simple model systems (Pearce et al.) as well as studying mammalian cells (Kyttala et al.) to better understand the function and the cell biology of each NCL gene. Keeping up with progress in clinical, research, professional advances and support for families in the NCL field is a challenge, but fortunately Internet access to clinical and scientific publications and the many web sites containing useful information has made this easier for all concerned.

The future looks brighter for the NCLs, more than a hundred years after they were first described, than it did several decades ago. Whilst this issue cannot include all aspects and details of the NCLs, it is hoped that these articles will provide good summaries for those new to the field, as well as bringing together important features for those more familiar, and perhaps making succinct predictions about future hopes. A cure for NCL does not yet exist, but work leading towards this is in place and being done. Progress will continue to be reported at biennial international meetings, the next one being held in Rochester, New York, USA, in July 2007.

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