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EDITORIAL

Mucopolysaccharidoses (MPS)

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This special edition of the *Journal of Inherited Metabolic Disease* brings together several publications focused on the mucopolysaccharidoses (MPS). The development of enzyme replacement therapy (ERT) for many of these disorders has led to a burgeoning interest in this field, as reflected in this group of articles.

The general theme of phenotypic presentation is highlighted in the report by Hendriksz and colleagues (10.1007/s10545-011-9410-9), who present the natural history and treatment outcomes from a clinical surveillance program for MPS 6 (Maroteaux-Lamy disease). The focus then shifts to organ systems in the MPS disorders, centered primarily cardiovascular manifestations. Three reports firstly by van der Ploeg and co-workers (10.1007/s10545-011-9444-z) summarize the cardiac features of MPS I (Hurler syndrome), MPS II (Hunter syndrome) and MPS VI, and the associated outcomes with ERT. The report by Harmatz and co-workers (10.1007/s10545-012-9481-2) summarizes the long term cardiac outcomes in a cohort of MPS VI patients, while the report of Wynn and colleagues (10.1007/s10545-012-9500-3) highlights cardiac outcomes in a subgroup of MPS I patients who manifest cardiomyopathy. The latter report provides management guidelines and represents an important adjuvant for clinicians dealing with MPS patients. The critical association between the timeline of ERT prior to BMT, and its relationship to ablation therapy geared to optimize outcomes, is highlighted in this paper. Rounding out the discussion of cardiac findings is the report by Braunlin and co-workers (10.1007/s10545-011-9438-x) who overview the endothelial dysfunction of cardiac tissue in MPS patients, which may help to explain the increased risk for cardiovascular complications in these disorders.

The overall theme of systems and organ involvement is continued with reports on the respiratory and central nervous system (CNS) in MPS patients. The respiratory system, poorly understood in most MPS disorders, is closely linked to airway patency in many of these disorders. Berger and colleagues (10.1007/s10545-012-9555-1) examines this topic in conjunction with a supporting article on anaesthesia management by Walker and colleagues (10.1007/s10545-012-9563-1). The latter should become a pivotal "must read" for anaesthetists involved in the management of MPS patients. Moving to the theme of CNS disorders, Cross and co-workers (10.1007/s10545-012-9572-0) evaluate the behavioural phenotypes of several MPS disorders, followed by reports from the Hendriksz (10.1007/s10545-011-9430-5 and 10.1007/s10545-012-9459-0) and Guigliani (10.1007/s10545-012-9559-x) groups that begin to describe the CNS involvement in MPS IVa and MPS VI, which is at variance with the classical concept that these disorders have no impact on the brain. For both disorders, the pathology of the cervical spine has been very topical, and Solanki and colleagues (10.1007/s10545-013-9585-3) address this issue via morphometric evaluation of the spine in MPSV IA patients followed by an overview of management guidelines for spinal manifestations (10.1007/s10545-013-9586-2). The latter paper overviews anatomy, neuroimaging, and the approach to follow-up, and may well become an important future treatment reference for MPS disorders.

Moving from phenotype to genotype, Pollard and co-workers (10.1007/s10545-012-9533-7) present 104 novel mutations in a cohort of 355 MPS patients, representing a significant contribution to the molecular diagnostic armamentarium. Progressing to a discussion of diagnosis, Wood and colleagues (10.1007/s10545-013-9587-1) present an excellent overview on the diagnostic approach to MPS

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IVA (Morquio A syndrome) that further highlights the diagnostic pitfalls and conundrums with this disorder. Wijburg and colleagues continue the theme of diagnosis with two papers evaluating the effects of therapy on urinary glycosaminoglycans and frequently employed biomarkers for disease severity in the MPS disorders (10.1007/s10545-012-9535-5 and 10.1007/s10545-012-9538-2).

Readers who seek a "taste" of basic science and animal models are not forgotten in this issue. Tomatso and colleagues (10.1007/s10545-012-9522-x) report on the micro CT evaluation of the bone dysplasia in different MPS mouse models, while Fildes and co-workers (10.1007/s10545-012-9508-8) characterize the immune system of the murine model of MPS1, with a focus on both T and dendritic cells. Auricchio and co workers (10.1007/s10545-012-9521-y) explore the potential of some MPSVI mutations as potential targets for read through therapies. The effects of disease on the family are not forgotten as well, and Hare and colleagues (10.1007/s10545-012-9558-y) assess the coping strategies of parents of Sanfilippo syndrome in comparison to parental strategies for handling other intellectual disabilities.

The authors and Editorial Board are confident that this special edition will evolve into an invaluable resource for clinicians and laboratory personnel dealing with MPS patients.

Conflict of interest None.