Novel paradigms for in inborn errors with muscular and central neuropathology

By Eva Morava and Thomas S. Jacques

In a joint collaboration between the *Journal of Inherited Metabolic Disorders* and *Neuropathology and Applied Neurobiology*, we have drawn together a special, free to access, online collection of 12 articles from both journals, published in the past few years, which could interest the readers of both journals. We have chosen a number of intriguing case demonstrations as well as showcasing neurologic, neuropathologic and metabolic features of rare Mendelian IEMs.

Molecular technologies, *e.g.* whole-exome sequencing (WES), have rapidly accelerated Mendelian disease discovery. In the last decade, several hundred inborn errors of metabolism have been discovered.[1] Most of these disorders show significant central nervous system involvement. Understanding the pathomechanisms of these disorders also revealed new concepts underlying disease progression in metabolic disease.[2-3] These include lessons on the significance of autophagy in the progression of lysosomal disorders such as Danon disease or adult Pompe disease, [4-5] the role of heat shock proteins in the inflammatory aspects in X-linked adrenoleukodystrophy,[6] nitric oxide metabolism in argininosuccinic aciduria,[7] or Bergman glia translocation in vanishing white matter disease.[8]

The discovery of novel disorders has also taught us the importance of energy metabolism for normal synaptic function and that not only proteins but lipids play a major role in the dynamics of synaptic membranes contributing to transport of synaptic vesicles.[9]

In this special online collection we also selected a review on neurologic disease progression in Cerebrotendinous xantomatosis and on inborn errors of Coenzyme A metabolism.[10-12]

We hope you find the collection of articles useful and informative.

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