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Foreword

Heme is indispensable for nearly all life forms on Earth. As a prosthetic group of proteins it serves several roles, including electron tranfser, oxygen binding, and even sensing molecules like nitric oxide. Naturally occurring porphyrins are precursors of heme in its biosynthetic pathway and their structures are fascinating and unique.

The study of porphyria, as with other metabolic disorders, deals with gaining a deeper understanding of how defects in the enzymes that generate porphyrins cause disease. Cloning of the cDNA and genes coding for all heme biosynthetic enzymes had great impact on the understanding of the molecular pathology and accurate diagnosis of individual porphyrias, as well as therapy and prevention of life-threatening acute porphyria attacks. The establishment of EPI (European Porphyria Initiative) was an important event in the porphyria field. Creating an available, informative tool for patiens and doctors had already enhanced the well being of our porphyria patients, as well as promoting research collaborations within Europe. On the contrary, very fundamental questions concerning the precipitation of various clinical acute porphyric attacks remain to be understood.

The idea to publish this special issue crystallized during the „Porphyrins and Porphyria 2003“ Meeting, held in Prague. During the preparation of this issue, several important observations were made, including the discovery of the heme intestinal transporter and the solution of several structures of heme biosynthetic enzymes, which aided in understanding their molecular mechanisms of action. The expanding role of heme in controlling numerous basic cellular processes was also reported.

This issue covers diverse topics connected by a unifying theme related to heme in its various roles: biosynthesis, diagnosis of inherited defects in this pathway, recent developments in the field of porphyrin chemistry with relevance to therapy and diagnostics, small excursions into the kingdom of hemoproteins, represented by cytochrome c oxidase, and attempts to place porphyria into the complicated genomic and proteomic network. Support to publish this special issue was provided by grants from the Czech Ministry of Education, Youth and Sports (No. 0021620806), the GIS-Institut Maladies Rares 2006 Research Network on Rare Disorders n°A04155HS, and Orphanet.

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