Peroxisomal Disorders and Regulation of Genes (Advances in Experimental Medicine and Biology)

Frank Roels, Myriam Baes, Sylvia De Bie

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In most peroxisomal disorders the nervous system is severely affected which explains the clinical and community burden they represent. This is the first book to focus not only on the mutations causing these inherited illnesses, but also on mechanisms that regulate, suppress or enhance expression of genes and their products (enzymes). Indeed since the success and completion of the Human Genome Project all genes (coding DNA sequences) are known. However, of many, their function, and the role of the gene product has not been determined. An example is X-linked adrenoleukodystrophy, the most frequent peroxisomal disorder. Children are born healthy, but in more than 1 out of 3, demyelination of the brain starts unpredictably and they die in a vegetative state. The gene mutated in most families has been known for 10 years; but the true role of the encoded protein, ALDp, is still speculative; and within the same family, very severe and asymptomatic clinical histories co-exist, unexplained by the mutation.

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