FACULDADE DE FARMÁCIA
Universidade de Lisboa

SEMINAR

Faculdade de Farmácia da Univ. Lisboa, Auditório
– 16 April 2010, 12:00h –

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METABOLIC FUNCTIONS AND BIOGENESIS OF PEROXISOMES AND ITS RELEVANCE TO HUMAN DISEASES

Peroxisomes are subcellular organelles which are present in every eukaryotic cell except mature erythrocytes and catalyze a number of essential metabolic functions in humans. The importance of peroxisomes is emphasized by the existence of a group of inherited diseases in man in which there is a defect in either the biogenesis of the organelle or one of its metabolic functions. The prototype of the first group of disorders is Zellweger syndrome which is a devastating autosomal recessive disease affecting multiple organs including the brain with usually early death in affected patients. The prototype of the group of single peroxisomal enzyme deficiencies is X-linked adrenoleukodystrophy (X-ALD). The phenotypic spectrum of X-ALD is huge ranging from the rapidly fatal childhood form of the disease (CCALD) to a late onset form called adrenomyeloneuropathy (AMN). In recent years diagnostic methods have become available at the metabolite, enzyme and DNA level for the full spectrum of peroxisomal disorders. Our current stage of knowledge in the field will be presented with special emphasis on the genetic basis of the disorders of peroxisome biogenesis.

Host:
Margarida Silva,
Metabolism and Genetics Group, iMED.UL (Research Institute for Medicines and Pharmaceutical Sciences)

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