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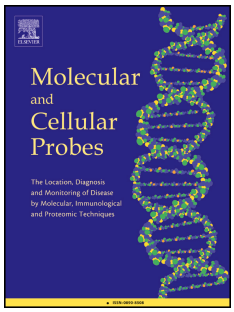

Peroxisomal disorders: Improved laboratory diagnosis, new defects and the complicated route to treatment

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Review

Peroxisomal disorders: improved laboratory diagnosis, new defects and the complicated route to treatment

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Key words: Peroxisomes, Laboratory diagnosis, Fatty acid oxidation, Plasmalogens, Zellweger syndrome, Inborn errors of metabolism, Genetic diseases, Metabolomics, Lipidomics

High lights MCP manuscript

- The peroxisomal disorders comprise a genetically and biochemically heterogeneous group of diseases in man caused by mutations in > 30 different genes.
- The peroxisomal disorders are usually subclassified into two groups including the disorders of peroxisome biogenesis and disorders of peroxisome metabolism.
- Analysis of a set of peroxisomal biomarkers as deduced from studies on Zellweger syndrome provides a good read-out of the metabolic functions of peroxisomes *in vivo* and can be measured in a simple blood sample.
- The clinical, biochemical and genetic spectrum of the various peroxisomal disorders has increased through the years, creating difficulties in the identification of patients, both clinically as well as in the laboratory.
- Whole exome and genome sequencing methods offer new options for patient identification but only when embedded in a larger network of -omics techniques, including Metabolomics.