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## Cell trafficking and complex lipids: an interconnected network of rapidly expanding disorders

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### Rome, Italy 28-30 November 2024

#### Scientific Organising Committee:

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Information and registration: www.rrd-foundation.org



A course for **general metabolic physicians** with an intermediate to advanced training level, also **clinicians specialized in organellerelated disorders** (i.e. mitochondrial, lysosomal...) **child and adult neurologists** and **geneticists**.

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#### Deadline for registering: 20<sup>th</sup> October

### Course overview and objectives:

Cellular trafficking is essential for maintaining critical biological functions. Mutations in over 370 genes, mostly identified in the last five years, are linked to cellular trafficking disorders. Initially limited to membrane trafficking, recent disease discoveries have unveiled new biological pathways. Complex lipid metabolism is closely related to cell trafficking due to the constant need for lipid supply and remodelling in vesicles and organelle membranes.

Clinically, these disorders often present multisystem symptoms that can resemble mitochondrial, peroxisomal, and lysosomal defects. Metabolic crises, such as rhabdomyolysis, hepatic failure, neurological symptoms, and biochemical abnormalities, can occur. These diseases can affect any organ at any age, though the nervous system is particularly vulnerable.

This course will cover the clinical presentations and mechanisms of these inborn metabolic diseases. Practical workshops will address diagnostic and treatment questions.

- Understanding the biological basis of cell trafficking.
- Understanding the intricate connection between cell trafficking and lipid metabolism.

Fondation d'entreprise

- Recognise cell trafficking disorders as a rapidly emerging category of inborn errors of metabolism with its own biological and clinical characteristics.
- Learn the main clinical manifestations of cellular trafficking diseases.
- Recognise the forms of presentation of metabolic crises in diseases of cellular trafficking.
- Identify the main biomarkers and treatments.

