

24

June 2021

Thursday

**WEBINAR**

via Webex\*

45' (talk) + 30' (discussion)

3.00pm - 4.15pm



# Metabolic perturbations and human disease phenotypes

**ABSTRACT**

Many human diseases are caused by mutations that perturb metabolism at the cellular level and result in tissue dysfunction. Some metabolic perturbations result in disease by interrupting the canonical functions of the metabolic network: producing energy, generating precursors for macromolecular synthesis, maintaining redox balance, disposing of waste, etc. Others interfere with processes beyond the conventional metabolic network, interfering with signaling and gene expression networks. Understanding these pathological states of metabolic perturbation may help us develop rational approaches to normalize metabolism and restore health. We study two types of diseases characterized by metabolic dysfunction: inborn errors of metabolism and cancer. I will discuss ongoing work in both these classes of disease that seeks to characterize abnormal metabolic states directly in human subjects, then uses experimental models to explore disease mechanisms and propose potential therapies. I will emphasize methods in metabolomics and stable isotope tracing that allow us observe metabolic phenotypes in intact systems relevant to physiology and disease, highlighting recent work on tumor metabolism in cancer patients and fetal metabolism in mice.



**SPEAKER**

**Prof Ralph DeBerardinis**

Professor, Children's Medical Center Research Institute (CRI),  
University of Texas Southwestern Medical Center

**HOST:**

Department of Oncology (LIH)

**RESPONSIBLE LIH SCIENTIST:**

Johannes Meiser / (johannes.meiser@lih.lu)

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