

**Thursday June 28, 2018 – 10h00**

Conference room AI 1153 (\*)- EPFL - Lausanne

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## **“The Role of Oxygen in Disease and Metabolism: Developing Hypoxia as a Treatment for Mitochondrial Dysfunction”**

**Host:** Prof. Johan Auwerx

### **Abstract:**

Mitochondrial disease affects 1 in 4800 live births, often leading to death within the first few years of life. Currently, there are no proven therapies that work broadly for this class of disorders. We perform a genome-wide screen and find that the cellular hypoxia response is protective against mitochondrial dysfunction. Moreover, chronic hypoxia extends the life of a mitochondrial mouse model of Complex 1 deficiency by five-fold. Starting hypoxia therapy at a late-stage of disease can even reverse the neurodegenerative phenotype and MRI-detectable lesions in this animal model. At the other extreme, mild hyperoxia greatly exacerbates disease and leads to death within several days.

These findings have now led to a phase 1 clinical trial in healthy volunteers, with the ultimate goal of human translation. We believe we have identified a new mode of treatment that will be broadly applicable to different forms of mitochondrial dysfunction, ranging from rare inborn errors of metabolism to more common, age-associated pathologies. We believe that “turning the oxygen dial” to low or high oxygen will serve as a novel therapeutic for a range of conditions in the coming years.

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