Omics approaches to mitochondrial diseases

May 4 2021

Florian Schober from the research group Inborn Errors of Endocrinology and Metabolism, the Department of Molecular Medicine and Surgery, will defend his thesis "Systems biology of mitochondrial dysfunction" on May 7, 2021.

What's the main focus of your thesis?

Mitochondria are the powerhouse and central metabolic platform of our cells. When they don't function accordingly, many different organs can be affected at any age, but how mitochondrial malfunction causes human disease is not well understood. In the past two decades, new powerful methods termed omics have been developed that can measure thousands of molecules in cells within a short time. I explore these techniques and combine them with genetic models of human disease, for instance fruit flies and mice, to capture the systemic biological consequences of mitochondrial malfunction.

Which are the most important results?

We found that tiny modifications of proteins in mitochondria, methylation and phosphorylation, can modify the function of the whole organelle. We saw that the abundance of the amino acid methionine that is present in most food that we eat controls the efficiency of energy production in mitochondria. Using omics techniques, we uncover a novel layer of complexity in the cell that extends far beyond genes, RNA and proteins.

How can this new knowledge contribute to the improvement of people's health?

We could show in a fly model that a group of young patients with mitochondrial disease can profit from adjusted diets, and this finding might even be of importance to better target cancer cells. It is important to carefully test our results that stem from laboratory models until they are useful for medical care. But this clearly shows that basic research is a very important step towards clinical applications.

What are your future ambitions?

I became excited about the idea that large amounts of data and powerful bioinformatic tools can tell us how real-life biology works. From here, I will move on to the laboratory of Prof. Matthias Mann at the Max Planck Institute of Biochemistry in Munich. I will apply the knowledge that I acquired during my doctoral studies at Karolinska Institutet to understand how single cells in human tissue interact with each other in order to answer one important question: Why does disease only affect some specific cells, and not others? If we manage to find an answer, we will be one step closer to target the most daunting and complicated cases of human disease.

More information: Systems biology of mitochondrial dysfunction. news.ki.se/dissertation-florian-schober

Provided by Karolinska Institutet