Novel causes, mechanisms and therapeutic strategies in mitochondrial disease

Propositions belonging to the thesis

Novel causes, mechanisms and therapeutic strategies in mitochondrial disease

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1. Whole exome sequencing (WES) analysis is the preferred, first, strategy to identify the genetic basis in mitochondrial disease, and is superior to panel-based approaches *(this thesis)*

2. As WES is able to identify all genetic causes in multigenic disease, it is essential to evaluate all genetic variants in the context of the disease symptoms *(this thesis)*

3. Testing therapeutic interventions for mitochondrial gene defects should be performed on patient material, within his or her unique genetic context *(this thesis)*

4. Zebrafish is a suitable model to simulate the mtDNA bottleneck and the transmission of mtDNA in human situation *(this thesis)*

5. Complex I deficient patients should be functionally tested for responsiveness prior to prescription of a high-fat diet, as palmitic acid improves mitochondrial respiration only in specific complex I defects *(valorisation)*

6. In 2020 more than 95% of the disease causing mitochondrial gene defects will be identified

7. Exome data should not be limited to identifying the disease-causing gene defect, but should also be evaluated for clinically actionable information and personalized medicine

8. Whole genome sequencing (WGS) data provides also valuable information for healthy individuals

9. Ethical questions need to be dealt with before the patient can take advantage of all information present in the personal genome

10. If you lack technique you lose the freedom to create *(Paco de Lucia)*

11. You are never more than a half-step away from a right note *(Victor L. Wooten)*