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Professor

## Antonella Spinazzola M.D.

Neuroscience and Mitochondrial Medicine
Co-director MSc Brain and Mind Sciences
UCL Queen Square Institute of Neurology,
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# Mitochondrial DNA Disorders: the (long and winding) road to treatment


### *Abstract*

In her seminar, Antonella will guide the audience through the journey of her research in mitochondrial disorders- from genetic discoveries to advances in understanding disease mechanisms, culminating in the identification of therapeutic strategies for two classes of mitochondrial DNA disorders. She will present the recent progress in validating these therapeutic strategies in preclinical models and share the effort underway to translate these findings into clinical trials; thus, offering hope for individual affected by these currently incurable diseases.


### *Speaker*

Research. Antonella Spinazzola is Professor of Mitochondrial Medicine and Neuroscience at University College London where she leads a comprehensive program that has at its core a bidirectional fundamental research/translational approach- from patients to the laboratory and back- to advance patient care.

Antonella's research focus is understanding the role of mitochondria and their genome in human disease. In addition to studying primary mitochondrial disorders, she is investigating the role of mitochondrial dysfunction in common neurometabolic disorders. The goal is twofold: first, to develop therapies tailored for genetic mitochondrial disorders and, second, to leverage the gained insights to develop innovative approaches for common diseases.

### *Biography*

Antonella qualified in Medicine from the Catholic University in Rome, and she trained as neurologist at the "A. Gemelli" University Hospital, in Rome. She then was awarded two consecutive postdoctoral research fellowships to work with Professor Michio Hirano and Professor Salvatore DiMauro at the Houston Merritt Clinical Research Centre, Columbia University (New York City). In the laboratory of Professor Hirano, Antonella and a colleague discovered the first nuclear gene defect causing mitochondrial DNA dysfunction. The breakthrough launched the field of perturbed nucleotide metabolism and mitochondrial disease.

Upon returning to Italy, Antonella held a Senior Clinical and Research fellowship at the National Institute of Neurology "C. Besta" in Milan, where she continued her research on mitochondrial DNA disorders, identifying, among others, the cause of another form of mitochondrial genomic instability disorders.

Her achievements were recognized by the award of a Marie Curie Career Development Fellowship, which allowed her to join the Mitochondrial Biology Unit (MBU), in Cambridge. There she was subsequently recruited by the then Director and Nobel Laureate, Professor John Walker, as a Track Programme Leader, and began to address the fundamental question of how the biogenesis of the mitochondrion is achieved and regulated (2010). Supported by a prestigious MRC Senior Fellowship, Antonella moved her group to the Mill Hill Laboratory in London (2013) and joined UCL Queen Square Institute of Neurology ( 2016) where was promoted Full Professor in 2018.