



University of Cyprus
Department of Biological
Sciences

Postgraduate Seminars

Seminar Series 2018 - 2019

Assist. Prof. Cybel Mehawej

Faculty of Medicine

&

Director of Functional Genetics and Immunogenetics Unit,

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“Approaches for Novel Gene Discovery in Genetic Disorders”

Wednesday, 20 March 2019, at 17:00

Building CTF 01, Room 110, Panepistimioupoli Campus

This seminar is open to the public

The past few decades have witnessed a tremendous development in the field of genetics. The implementation of next generation sequencing (NGS) technologies revolutionized the field of molecular biology and made the genetic information accessible at a large scale. However, connecting a rare genetic variation to a complex phenotype remains challenging. Indeed, identifying the cause of a genetic disease requires a multidisciplinary approach, starting with the establishment of a clear phenotype with a detailed family history and ending, in the majority of cases, with functional assays that are crucial for the validation of the pathogenicity of a mutation. Different approaches for gene hunting are herein discussed and examples from challenging cases are elaborated.