



Genetics, Bioinformatics, & Systems Biology Colloquium

presents

Roberto Bonelli, PhD
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LIVE
IN-PERSON
with Pizza

*Live stream via Zoom available

 **Thursday**
Mar. 14, 2024  **12PM**  **Leichtag Auditorium**  **Zoom**

Integrating Genomics, Metabolomics and Retinal Imaging to Decode a Rare Retinal Disease

Macular Telangiectasia Type 2 (MacTel) is a rare retinal disease, affecting approximately 0.1% of the population. Despite ongoing clinical trials, effective therapeutic interventions for MacTel remain unavailable. Historically, the etiological mechanisms of MacTel were elusive, but recent advancements have begun to elucidate this area. This presentation will offer a comprehensive overview of various studies encompassing genomics (including SNPs and WES), metabolomics (both targeted and untargeted), and imaging data. These studies have explored the genetic underpinnings of MacTel, evaluated its potential metabolic aspects, and employed methods such as Mendelian randomization, in-vivo, and in-vitro research to identify probable causative factors contributing to the disease's onset and progression. The presentation will also highlight MacTel unique position as a condition that straddles the line between common complex disorders and those of Mendelian nature.

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