**Tata Institute of Fundamental Research**
Survey No. 36/P, Gopanpally Village, Serilingampally, Ranga Reddy Dist., Hyderabad – 500 046

# Colloquium

## Uncovering the Functional and Clinical Impact of de novo variants in Rare Disease

## Michael Wangler

### Baylor College of Medicine, TX

Rare diseases represent opportunities to understand the genetic mechanisms that underlie human health. Our lab uses genomic sequencing, metabolomics and Drosophila models to understand rare diseases such as peroxisomal disorders. We have focused on a new group of patients with atypical phenotypes with de novo mutations in peroxisomal genes such as DNM1L/drp1. We have also studied a number of de novo mutations for genes like ACTG2 and WDR37 in undiagnosed patients. Across these rare disease genes we have observed that the patterns of Arginine codon usage in human genes is a marker for dominant rare disease genes.

***Monday, Jan 22nd 2024***

***14:30 Hrs (Tea / Coffee 14.15 Hrs)***

***Auditorium, TIFR-H***