



Invitation to MTech Thesis Defense of Sana Akhtar: June 21, 2019 (Friday): 16.00-17.00 IST

In Partial Fulfillment of the Requirements for the Degree of
M.Tech CB

Sana Akhtar (MT17146)

Will defend her thesis

Title: “Computational resources for predicting rare diseases associated mutations in lysosomal enzymes”

IIIT-D Faculty and Students are invited

Date: June 21, 2019 (Friday)

Time: 16.00 – 17.00 IST

Place: CB Meeting Room (3rd Floor, R&D Building)

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| Examiner: | Internal: | Vibhor Kumar |
| | External/Internal: | Vinod Scaria, IGIB |
| | Advisor: | Gajendra P S Raghava |

Abstract

A significant burden of genetic disorders comes from the malfunctioning of the “suicide bags of the cells” called as the lysosomes and are referred to as Lysosomal Storage disorders or LSD’s. LSD’s comprises a major proportion of rare genetic disorders globally. These lysosomes are small storage houses inside the cell, harboring a set of enzymes involved in maintaining the cellular hemostasis. Any malfunctioning in these vital players results in the accumulation of their un-degraded metabolic substrates and eventually manifestation of such monogenic disorders. Detailed and efficient analysis of LSD’s and its associated enzymes is still challenging due to scattered relevant information and the lack of an integrated platform. To fill this lacuna, in this work, we aim to explore the domain of lysosomal disorders from various perspectives. We divide the thesis into three parts: (1) Development of a web repository: RareLSD (2) Benchmarking of existing mutation Prediction tools (3) Development of a disease specific mutation prediction method for monogenic Pompe disorder.