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## Metabolomics on mouse models of disease for personalized medicine

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etabolomics is a newer omics technology that can Whelp in generating a more comprehensive view of a biological system when combined with the genomic, transcriptomic and proteomic technologies. In this seminar, I will discuss the bioinformatics infrastructure that we have developed at Jackson Laboratory to perform mass spectrometry based metabolomics experiments on mouse based studies. I will highlight some case studies where I have used metabolomics in mouse models of neuromuscular degeneration and patient derived xenograft (PDX) for cancer studies and personalized medicine. Charcot-Marie-Tooth (CMT) disease encompasses a genetically heterogeneous class of heritable motor and sensory neuropathies that result in axonal degeneration in the peripheral nervous system. Charcot-Marie-Tooth type 2D (CMT2D) is caused by dominant mutations in Glycyl tRNA Synthetase (GARS). Samples from the spinal cord of GARS mutant mice and littermate control animals were

compared using mass spectrometry based metabolomics. The changes associated with GARS mutations suggested possible treatment strategies through supplementation. In another case study, Gas Chromatography Mass Spectrometry (GCMS) based analysis was used to derive a metabolic signature of triple negative breast cancer (TNBC) by comparing the metabolites present in three study arms of patient-derived xenograft (PDX) mouse urine. This analysis not only suggested metabolic uniformity of the JAX mice but also showed cancer specific changes in the vehicle (tumor with no treatment) arm, known drug side effects in the treatment arm (tumor engrafted and treated) and the age related changes in the pure (no tumor no treatment) arm.

## Biography

Preeti Bais is a Scientist at Jackson Lab's new research institute- JAX Genomics Medicine (JGM). It is an independent, nonprofit organization focusing on mammalian genetics research to advance human health. She holds a PhD degree in Bioinformatics and Computational Biology. She has worked on projects involving metabolomics analysis of Human Embryonic Stem (hES) and Induced Pluripotent Stem (IPS) cells based assays for drug toxicity screening, autism biomarker detection using blood samples, mouse models of neuromuscular degeneration and cancer drug efficacy testing using orthotropic mouse models triple negative breast cancer.

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