List of Tables:

1. Diagnosis of Lysosomal Storage Disorders in Australia

2. Comparison of Incidence/prevalence of lysosomal storage diseases in different country

3. Relative frequency of LSD in Portugal

4. Lipidoses: comparison of data in different populations

5. Mucopolysaccharidoses: comparison of data in different populations

6. Glycoproteinoses, mucolipidoses, glycogenosis type II and neuronal ceroid lipofuscinoses: comparison of data in different populations

7. Number of patients and relative frequency of different categories of lysosomal storage disorders.

8. Related Enzyme Activities in Children with Lysosomal Storage Disorders

9. Different Types of LSDs Diagnosed in north Indian study

10. Specific classifications and features of MPS

11. Enzyme activity of β- Glucosidase obtained by Leukocyte lysate in controls and Gaucher Patients

12. Observed value of enzyme sphingomyelinase in controls and Niemann Pick Patients

13. Enzyme Chitotriosidase level in Controls and Gaucher Patients
14. Enzyme Chitotriosidase levels after one and two years of 1st visit in Gaucher Patients

15. Expression of Biomarker Chitotriosidase enzyme in controls and Niemann Pick Patients

16. Biomarker CCL18/PARC levels in controls and Gaucher Patients

17. Biomarker CCL18/PARC levels on 2nd and 3rd visit in Gaucher Patients

18. Expression of Biomarker CCL18/PARC in controls and Niemann Pick Patients

19. Enzyme α-L-iduronidase levels in controls & MPS Type I (Hurler) Patients

20. Enzyme Iduronate 2 sulfatase levels in controls & MPS Type II (Hunter) Patients

21. Biomarker Heparin Cofactor II Thrombin levels in controls and MPS Type (Hurler) Patients

22. Biomarker Heparin Cofactor II Thrombin levels in controls and MPS Type II (Hunter) Patients

23. Biomarker Heparin Cofactor II Thrombin levels after one and two year of MPS Type I (Hurler) Patients

24. Biomarker Heparin Cofactor II Thrombin levels at 2nd & 3rd visit of MPS Type II (Hunter) Patients

25. Average value and Standard Deviation of GAG in 60 Normal Urine Samples

26. Comparison of GAG content with Heparin Cofactor II Thrombin in MPS type I & II patients at 1st visit and their subsequent visits after one year and two years.
List of Figures:

1. The structure and function of lysosomes.

2. Enlarged vacuoles in a lysosomal storage disease. The electron microscopy picture of a leukocyte cell from a patient affected with alpha-mannosidosis.

3. A possible roadmap of the pathology of lysosomal storage disorders.

4. The biochemical and cellular basis of lysosomal storage disorders.

5. Putative generation mechanism and structure of the human CCL18 gene.

6. Inhibition of Thrombin by Heparin Cofactor II.

7. Relative rate of lysosomal storage disorders in the Czech Republic.

8. Schematic summarizing the concept of the lysosome.

9. Gaucher cell (bone marrow film from a Gaucher patient).

10. Metabolic pathways of glycosphingolipids (GSLs).

11. The GBA gene location on the long (q) arm of Chromosome 1.


13. Expression and role of CCL18 under physiological and immunopathological condition.

15. Autosomal recessive inheritance pattern.

16. Activity of Acid Sphingomyelinase.

17. The Sphingolipid metabolic pathway.

18. The SMPD1 gene location on the short arm of chromosome 11.

19. Cholesterol movement within lysosomes.

20. Cholesterol traffic and Niemann-Pick disease.

21. The NPC1 gene location on the long arm of chromosome 18.

22. The NPC2 gene location on the long arm of chromosome 14

23. Structure of the GAG linkage to protein in proteoglycans.

24. Diagnostic algorithm for mucopolysaccharidoses type II (MPS II).


26. Model for activation of Heparin Cofactor II.

27. Activation of HCII and antithrombin by vascular glycosaminoglycans.
28. A - Normally, the lysosomal enzymes are synthesized in the endoplasmic reticulum (ER) and transported to the Golgi apparatus, where they receive the mannose-6- phosphate marker that is essential for receptor-mediated sorting into the lysosomes.

B- The mutant enzyme is misfolded and retained in the ER, enzyme activity is lacking in the lysosome.

C- N-butyldeoxynojirimycin increases the stability of the mutant enzyme that now is able to enter the Golgi apparatus and after binding the Mannose-6- phosphate receptor-becomes active within lysosome.


30. Box plot demonstration of enzyme β- Glucosidase in leukocyte lysate in controls and Gaucher’s Patients.


32. Box plot demonstration of enzyme Chitotriosidase in controls and Gaucher Patients at 1st visit (Baseline) and at different time intervals in GD patients


34. Box plot demonstration of BiomarkerCCL18 level in controls and Gaucher Patients at 1st visit (Baseline) and at different time intervals in GD patients
35. Box plot demonstration of enzyme $\alpha$-L-iduronidase levels in controls & MPS Type I (Hurler) Patients.

36. Box plot demonstration of enzyme Iduronate 2 sulfatase levels in controls & MPS Type II (Hunter) Patients.

37. Box plot demonstration of Biomarker Heparin Cofactor II Thrombin levels in controls and MPS Type I Patients at 1st visit (Baseline) and at different time intervals in MPS I patients

38. Box plot demonstration of Biomarker Heparin Cofactor II Thrombin levels in controls and MPS Type II Patients at 1st visit (Baseline) and at different time intervals in MPS II Patients

39. A typical straight line assay calibration curve of Heparan Sulfate.

40. Age wise distribution of urinary GAG in normal subjects.