Summary, Concluding Remarks and Futurology
The results presented in this thesis can be summarised as follows:

1) A total of 95 clinically diagnosed MPS children from 5 different hospitals in Madras city which cater to the South Indian population, were referred to our laboratory.

2) Mainly Morquio and Maroteaux-Lamy patients were chosen for the present study. This constituted 46 out of the 95 patients referred to us.

3) Based on this, a study group comprising of 120 individuals, which constituted 46 clinically diagnosed MPS children, 39 controls (28 children and 11 adults) and 35 family members constituting 12 families, was chosen.

4) Normalisation of urinary GAGs to urinary creatinine helped in overcoming the problem of 24 hrs urine collection.

5) The alcian blue complex formation method used for the isolation of urinary GAGs helped in the isolation of total GAGs without any preferential size loss.

6) The acid alcian blue method used for the quantification of urinary GAGs was superior since it detected KS also.
7) Quantitation of urinary GAGs in these patients indicated that Morquio patients excrete 3 fold excess and other MPS types excrete a 6 fold excess of urinary GAGs compared to the normals.

8) Qualitative analysis of urinary GAGs by discontinuous cellulose acetate membrane electrophoresis, helped in differential diagnosis and grouping of the patients to different MPS types.

9) Sequential thin layer chromatographic analysis substantiated the differential diagnosis and specifically aided in the grouping of Morquio patients.

10) The differential diagnosis highly reduced the number of enzyme assays to be performed on these patients for the definitive diagnosis of their types and subtypes.

11) The dextran sedimentation method used for the isolation of leukocytes aided in obtaining a 70% yield and provided a 99% erythrocyte free leukocyte preparation.

12) The leukocyte lysates had protein concentrations ranging from 120-400 μg protein/ml blood and no major changes were observed in the protein ranges between normal and MPS patients.

13) Estimation of lysosomal enzyme activities in normal children and adults indicated that the normal ranges of GALNS, aroylsulfatase A and acid phosphatase did not show any significant difference between
our population and others. On the other hand, β-galactosidase and arylsulfatase B were significantly lower in our population.

14) Estimation of α-L-iduronidase, GALNS, β-galactosidase, arylsulfatases A & B, β-glucuronidase and acid phosphatase in the leukocyte lysates helped in the identification and confirmation of 16 Morquio A patients, 4 Morquio B patients and 3 Maroteaux-Lamy patients.

15) These analyses helped in the identification of non-MPS children who have overlapping clinical features with MPS children.

16) With respect to clinical features, no major differences were observed between our population and others studied world over.

17) Estimation of GALNS activity in 20 obligate heterozygotes indicated that this procedure cannot be used as a fool-proof method for the detection of Morquio A carriers. Though, it can indicate the probable carriers in most cases.

18) Analysis of consanguinity and MPS indicated a high degree of consanguinity among the parents of MPS children suggesting a correlation between consanguinity and MPS.
CONCLUDING REMARKS AND FUTUROLOGY

Mucopolysaccharidoses is a rare genetic disorder and epidemiological data are highly scarce. This scenario is very true in our country, where no systematic study has been conducted on this life threatening disorder. Hence, our laboratory has been highly interested in studying the prevalence and statistical distribution of this group of lysosomal storage disorders in our population. The present study and other data available from the laboratory indicate Morquio syndrome, particularly Morquio A syndrome to be a highly prevalent and Maroteaux-Lamy syndrome to be a low prevalent types in our population.

Morquio syndrome A being the most prevalent type of MPS in our population, a study on carrier detection for this syndrome was undertaken. A sincere attempt was made at correlating the GALNS levels with the carrier status of Morquio syndrome A. This study suggests that GALNS levels can only indicate the probable carrier but cannot be used as a fool-proof method for carrier detection.

Carrier detection being an important aspect in the abolition of the disease, in future attempts should be made at the mutational analysis of these patients to identify the mutations in our population which lead to GALNS deficiency. Since at present there is no cure for this disease, carrier detection and genetic counselling to prevent unwanted births are a must to alleviate the human suffering.