Summary and Conclusion
1. In the present study, the study group comprised of 175 individuals, which consisted of 76 clinically suspected MPS patients, 40 (25 children +15 adults) controls and 59 family members constituting 29 families.

2. Quantitative and qualitative analysis of urinary GAGs helped in the classification of the 76 clinically suspected MPS patients to their MPS types.

3. Analysis of urinary N-acetyl glucosamine sulfates helped in the differential diagnosis of MPS III patients. Out of 38 MPS III patients 16 were classified as MPS III A / D and 22 as MPS III B / C.

4. Estimation of enzyme activities in normal children and adults indicated that the normal ranges of heparan sulfamidase, N-acetyl glucosaminidase, GALNS and acid phosphatase were comparable to the literature range. However, the range of β-galactosidase was found to be lower in our population.

5. These lysosomal enzyme assays confirmed 9 MPS IIIA, 11 MPS IIIB, 12 MPS III C/D and 14 MPS IVA patients out of 70 analysed. The remaining 24 were confirmed to be nonMPS patients who may be suffering from other lysosomal / genetic disorders.
6. Urinary glucosamine sulfate analysis helped in subtyping MPS III patients further. Thus 38 MPS III patients were distributed as 9 III A, 11 III B, 8 III C, 4 III D, 3 IIIA/D and 3 III B/C.

7. The reanalysis of urinary GAGs from enzymatically confirmed patients revealed that a combined qualitative and quantitative analysis is needed to avoid false positive and false negative results.

8. Activity of HSS and GALNS in obligate heterozygotes (parents) overlapped with that of the control range, whereas activity of NAG in obligate heterozygotes did not overlap with that of control range, confirming its usefulness in carrier detection.

9. Analysis of clinical features among MPS III patients showed that the subtypes of MPS III do not vary from each other with their clinical features. The overlapping clinical features observed among the 76 patients indicated that diagnosis of MPS based on clinical features would be highly erroneous.

10. The higher percentage of consanguinity among the MPS families suggested that this may be a contributing factor for increasing the percentage of this rare single recessive gene disorders in this population.

11. No conclusion could be drawn from the community data. It is only a documentation of results.
12. Mutational analysis of SGSH gene showed exons II and VIII to be mutation negative in all 9 MPS IIIA patients analysed under our experimental conditions.

13. Exon VI of one MPS IIIA patient showed the presence of a polymorphism, ins ct (676-37) in intron 5 and a silent mutation at codon 227, ccc-cct (P227P).

14. The total data from the laboratory for the past 10 years shows MPS III to be the most prevalent type followed by MPS IV. MPS I and II were of intermediate prevalence and MPS VI was found to be the least prevalent. We have not come across any MPS VII patients.

15. Among the MPS III subtypes 34% were MPS III B, 28% were MPS III A followed by III C and D respectively. Analysis of more number of patients can only reveal the most common MPS III subtype in our population.