MUCOPOLYSACCHARIDOSIS (MPS) SCREEN, URINE
(Toluidine blue spot test) **Negative / Positive**

Note: 1. Results should be clinically correlated as individual / biological variations can affect the test

results

2. Genetic counseling available with prior appointment at Department of Genetics,

National Reference Lab, New Delhi

Comments

Mucopolysaccharidosis (MPS) are autosomal recessive disorders caused by deficiency of any of the enzymes involved in the stepwise degradation of dermatan sulphate, heparan sulphate, keratan sulphate or chondroitin sulphate. Affected individuals have a period of normal growth and development followed by progressive disease involving multiple systems. There are 11 known enzyme deficiencies that result in MPS's that include a variety of disorders ranging from MPS I to MPS IX . They vary in severity and produce symptoms like facial coarsening, organomegaly, skeletal changes, cardiac abnormalities and developmental delays. Disease can present itself in early to late infancy or in adulthood.