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ROLE OF GENERAL PAEDIATRICIANS AND LABORATORY PITFALLS IN DIAGNOSING MUCOPOLYSACCHARIDOSIS

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Mucopolysaccharidosis (MPS) are a group of lysosomal storage disorders.They are characterised by progressive accumulation of glycosaminoglycans (GAG) in
various organs and tissues depending on the type of disease (MPS I - MPS IX).
MPS I is one form of MPS and is characterized by a deficiency of the enzyme alpha-l-iduronidase. The presenting symptoms of this disease are non specific and vary greatly on a patient to patient basis. Early recognition of the symptoms and diagnosis is critical to prevent irreversible damage from occurring, now that new therapeutic options like enzyme replacement therapy has become available. 72 % from the MPS I patients seeked first the opinion of a general paediatrician who plays a key role in referring suspected patients for diagnosis.
As there is still no optimal test for primary screening for lysosomal storage diseases, laboratory diagnostics is performed as part of selective screening for hereditary metabolic disorders.
Early and accurate diagnosis of MPS is usually based on initial test on glycosaminoglycan excretion in urine. A variety of techniques are used by different laboratories, but experience with affected cases is often quite limited, leading to difficulties in interpretation and delays in diagnosis.
Using examples from our experience, we may demonstrate that ignorance of pre-analytic conditions and incomplete analytic implementation of laboratory
diagnostics may lead to false results
Collaboration between clinicians and medical biochemistry specialists is therefore necessary for correct interpretation of obtained findings.
While identifying MPS patients may be challenging, a simple blood test can be performed for confirmation.