

Evaluation of reliability for urine mucopolysaccharidosis screening by dimethylmethylene blue and Berry spot tests

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Background: The mucopolysaccharidosis (MPS) are a group of inherited metabolic disorders resulting from the deficiency of the enzyme responsible for intralysosomal catabolism of glycosaminoglycans (GAGs). GAGs are progressively accumulated in multiple tissues and released into the corporal fluids. The first laboratory approximation to MPS diagnosis is the identification of an increased urinary GAG excretion. For this, several semiquantitative and quantitative methods have been developed. The aim of this retrospective statistical study was to evaluate the reliability of MPS urine screening for the semiquantitative Berry spot test (BST) and the quantitative dimethylmethylene blue test (DMB). **Methods:** The 24-h-urine samples (n=246) were tested through BST, DMB, and for GAG excretion pattern by one-dimensional electrophoresis or thin layer chromatography. **Results:** the 204 samples that demonstrated a normal GAG excretion pattern were considered as non-MPS samples. Forty-two samples presente