P-033 - ANALYSIS OF GLYCOSAMINOGLYCANS BY TANDEM MASS SPECTROMETRY: REPORT FROM THE MPS BRAZIL NETWORK

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INTRODUCTION: Mucopolysaccharidoses (MPS) are lysosomal storage disorders characterized by progressive accumulation of glycosaminoglycans (GAGs). Several methods, mostly colorimetric, are used for the measurement of urinary GAGs. Thin-layer chromatography and electrophoresis are also used to identify the different species of GAGs. However, the use of these methods is limited by several factors, being false positives and false negatives not rare. Tandem mass spectrometry (MS/MS) is a powerful tool to identify and quantify GAGs, and can be used in a variety of samples, at any age, and at any disease stage.

OBJECTIVE: To evaluate the potential use of MS/MS for analysis of GAGs in a variety of samples.

MATERIALS AND METHODS: Serum, plasma, dried blood spots, urine, cells and other biological samples are incubated with chondroitinase B, heparitinase and keratanase II for disaccharide digestion. Different classes of GAGs are separated by liquid chromatography tandem mass spectrometry. Normal ranges are defined by analysis of age-matched controls.

RESULTS: The use of this new method allowed analyses of samples from patients with clinical suspicion of MPS, and from patients in therapy for treatment monitoring.

CONCLUSIONS: The implementation of the LC/MS/MS method allows the measurement of GAGs in several types of samples, with higher sensitivity and specificity in comparison to the colorimetric methodologies. This novel assay will improve the diagnostic work-up in suspected patients and the follow-up of treatment, and will overcome difficulties in analyses from samples which need to travel long distances and/or cross country borders. Furthermore, the measurement of GAGs in dried blood spots could be explored as an alternative for newborn screening of MPS.