PO07

Differential Diagnosis of α-Mannosidosis in MPSs

P. Gaspar [1]; D. Gonçalves [1]; R. Neiva [1]; L. Silva [1]; L. Vilarinho [1]

[1] Unidade de Rastreio Neonatal, Metabolismo e Genética, Instituto Nacional de Saúde Doutor Ricardo Jorge, Porto, Portugal;

Presenting Author - paulo.gaspar@insa.min-saude.pt
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Introduction: Mucopolysaccharidosis (MPSs) and oligosaccharidosis, two subgroups of lysosomal storage disorders (LSDs), face diagnostic challenges due to their wide spectrum of clinical presentations and overlapping symptoms. One of the oligosaccharidose is α-mannosidosis, an extremely rare and often undiagnosed disorder. It is characterized by a deficiency in the enzymatic activity of α-mannosidase, which is responsible for cleaving mannose from N-linked oligosaccharides. This study aimed to investigate the activity of α-mannosidase in dried blood spot (DBS) samples that had undergone screening for MPSs.

Methods: Three enzymatic assays were performed in the DBS received (α-mannosidase, β-mannosidase, β-galactosidase). Results were obtained through a calibration curve of 4-methylumbelliferone and expressed in nmol\h\spot. Molecular characterization of the MAN2B1 was performed in the suspicious samples.

Results/Case report: Among the 400 samples analysed, we identified eight cases with reduced α-mannosidase. The molecular findings confirm the biochemical data. The main features observed in those patients were skeletal abnormalities, coarse facies and cognitive impairment.

Conclusion: Since there is an overlap of symptoms in these two subgroups of disorders, differential diagnosis of oligosaccharidosis in children having suggestive symptoms of MPSs is crucial. An early treatment may significantly delay or prevent the onset of the major clinical signs, substantially modifying the natural history of the disease namely in the attenuated forms. Although rare, α-mannosidase should be considered as differential diagnosis for MPSs.