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Early Diagnosis of Mucopolysaccharidoses in Pediatrics

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Main category: Case series

Disease category: Complex molecule and organelle metabolism

Introduction: Mucopolysaccharidoses (MPSs) are a group of Lysosomal Storage Disorders with multisystem involvement, presenting different degrees of severity and evolution. At early disease stages and late onset forms, diagnosis can be postponed for years or even missed. The FIND PROJECT was designed to claim awareness to the red flags of MPSs at pediatric age and to provide a useful tool for physicians to diagnose these pathologies, since most of them are amenable to enzyme replacement therapy.

Methods: MPSs clinical suspicious were addressed by performing seven distinct enzymatic assays in dried blood spots, in order to understand whether any of those specific enzymes was deficient. For positive cases, the identification of glycosaminoglycans and the molecular study is carried out.

Results/Case report: In the first eight years of the project, we have identified 12 patients (five MPS I; one MPS II; two MPS IIIB, one MPS IVA, two GM1 and one MPS VI) out of the 385 samples studied. In the majority of the patients identified, the age of diagnosis was less than 3 years of age, which is much lower when compared to the mean age of diagnosis of 6 years old, reported by Pinto et al, 2004.

Conclusion: These results, shows that this project was successful also in its educational component, by raising the concern and awareness for these multisystemic pathologies that are linked to high morbidity.