**P-055 - SELECTIVE RETROSPECTIVE SCREENING DETECTS FIVE NEW CASES OF MUCOPOLYSACCHARIDOSIS TYPE VII**


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**INTRODUCTION:** Mucopolysaccharidosis type VII (MPS VII; Sly syndrome) is a highly heterogeneous, ultra-rare lysosomal storage disorder, caused by the deficiency of β-Glucuronidase (βGUS). Patients’ phenotypes vary from severe forms with non-immune hydrops fetalis (NIHF), skeletal dysplasia and mental retardation to milder forms with fewer manifestations. Epidemiological data of this disorder is really scarce and underdiagnosis of this disorder is suspected. Enzyme replacement therapy for MPS VII is available since 2017. **OBJECTIVE:** To present results from a retrospective screening of MPS VII in patients with clinical features compatible with MPS spectrum, in whom other MPS had been ruled out previously. **MATERIALS AND METHODS:** For this purpose, we analyzed 584 dried blood spots samples from Argentinean patients with clinical suspicion of MPS, collected between 2017 and 2018, where MPS I, II, IVA, IVB and VI were ruled out previously. βGUS enzyme activity was measured by fluorometric method with deproteinization. β-Galactosidase activity was also measured as a control enzyme, to assess quality of DBS samples. GUSB gene was analyzed by Next Generation Sequencing. **RESULTS:** We detected 4 patients with βGUS deficiency in DBS, from 584 samples analyzed. Ages of positives cases were 4, 11, 12 and 12 years old. The fifth case was a 2 years old girl, sister of one of the MPS VII patients diagnosed in this study, found from familiar screening. All five patients has a clinical course similar to MPS I, including the same degree of clinical heterogeneity. None of the patients had NIHF. **CONCLUSIONS:** We detected 4 patients with βGUS deficiency in cases where others MPS but no MPS VII were suspected. Also, we could detect an additional case by familiar screening. These results clearly remark the importance of ruling out MPS VII in patients with phenotype and clinical features compatible with MPS spectrum, since the different types of MPS may have similar manifestations and share some signs and symptoms.