P-046 - MUCOPOLYSACCHARIDOSIS: IS LESS DIAGNOSED THAN BELIEVED?

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**INTRODUCTION:** Mucopolysaccharidosis is a progressive, chronic and rare lysosomal storage disease due to accumulation of glycosaminoglicans (GAGS) caused by deficiency of any of the catabolic enzymes. It compromises several organs and systems such as CNS, bone, liver, cornea, hearing with variable severity. Because of this and low prevalence of the disease, early diagnose turns difficult. **OBJECTIVE:** Assess the existence of patients with undiagnosed MPS disease at Genetic Unit in Complejo Médico Policia Fedral Argentina Hospital Churruc-Visca (CABA - Argentina). **MATERIALS AND METHODS:** A retrospective analysis of 491 clinical histories of patients attended in 2016 were made. 52 of them were selected through stablished clinical criteria. Selected patients were analyzed through dried blood spots (DBS) for enzymatic activity for different MPS. All of the patients included in the study signed informed consent. **RESULTS:** From 52 patients selected only 42 were contacted of whom 22 attended clinical consultation for clinical reevaluation. Only 16 were selected for enzymatic activity test in DBD. One positive test was detected for beta-D-glucuronidasa deficiency related to MPS VII diagnose in a 12 year old patient. **CONCLUSION:** Through this analysis we can conclude there´s MPS subdiagnosis in the Churruca-Visca hospital population. As a consequence of 491 clinical histories revaluation a 12 year old patient was diagnosed for MPS VII. These results make us ask ourselves how many non diagnosed MPS patients exists and about the real world wide prevalence of MPS.