P-044 - MORPHOLOGICAL FINDINGS IN BLOOD SMEARS EXAMINATION OF PATIENTS WITH MUCOPOLYSACCHARIDOSIS TYPE VII (MPSVII)

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INTRODUCTION: Mucopolysaccharidosis VII (MPS VII) is an ultra-rare inborn error of metabolism caused by the deficiency of lysosomal enzyme Beta-glucuronidase (GUS). Phenotype may vary from severe form characterized with non-immune hydrops fetalis, skeletal dysplasia and mental retardation, to attenuated forms with skeletal abnormalities mainly. Biochemical diagnosis is made by the measurement of GUS activity in dried blood spots or leukocytes, and finding the causal mutations in GUSB gene. Cytomorphological alterations in leukocytes of patients with different lysosomal disorders, including mucopolysaccharidosis, have been reported. OBJECTIVE: The aim of this study was to evaluate leukocyte cytomorphology in blood smears from patients with diagnosis of MPS VII

MATERIALS AND METHODS: Peripheral blood from 5 MPS VII patients (ages: 2, 4, 12, 13 and 22-year-old), were obtained by venipuncture and blood smears were stained with May Grunwald - Giemsa. Leukocyte cytomorphology was evaluated at 100X by optical microscopy. RESULTS: Blood smears from 5 MPS VII patients were analyzed. All of them presented cytomorphological alterations in leukocytes from peripheral blood. Alder-Reilly inclusions were observed in neutrophils and eosinophils mainly, and occasionally were found in lymphocytes and monocytes. Also, neutrophils showed thicker azurophilic granulations similar to the toxic granulations seen in patients with severe infections. Lymphocytes and monocytes observed presented cytoplasmic vacuoles and some of them showed granular inclusions. CONCLUSIONS: Cytomorphological alterations were observed in peripheral blood smears from MPS VII patients. These alterations were found in different types of leukocytes, in a greater or lesser degree of complexity, regardless age of the patients. Blood smears examination could contribute with information to support diagnosis of MPS VII, as well as it could be an alert to suspect this disorder.