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Cytokines in Glycogen Storage Disease type 1 patients: a controlled cross-sectional study

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Introduction: Glycogen Storage Disease (GSD) type 1 is a genetic disorder of metabolism due the deficiency of glucose-6-phosphatase catalytic activity or glucose-6-phosphate exchanger SLC37A4 activity. The disease mainly affect the liver, but harms the whole body if untreated. The patients are predispose to recurrent infections, adenomas, inflammatory bowel disease and osteoporosis, among others. Aim: To determine the cytokine profile in a sample of GSD type 1 patients in comparison to healthy controls. Methods: Observational, cross-sectional, controlled study, with convenience sampling. Eighteen GSD type 1 patients (GSD type Ia= 13; type Ib= 5; female= 11; median age 14 years, IQR=12) and 18 healthy controls, sex-age-matched were included. Patients recruited from the outpatient clinics of the Medical Genetics Service at HCPA, Brazil. The inclusion criteria for patients were: a) having a biochemical and/or genetic diagnostic for GSD type 1; and b) being aged 3 years or older. The healthy controls (HC) were recruited by invitation among the population of RS. The quantification assay was realized through EMD Millipore's MILLIPEX® MAP Human Cytokine kit. All samples were measured in duplicates for 5 cytokines (G-CSF, INFy, GRO, MDC/CCL22 and IL17A). Measurements with divergence ≥ 30% between duplicates would be excluded from data analysis, as well as data of their respective pair (control/patient). The results were compared using non-parametric test for independent samples, U-Mann-Whitney. Statistical analyses (p≤0.05) were performed with IBM SPSS Statistics for Windows software, version 22 (IBM corp., NY). Results: All patients were on cornstarch therapy. Patients with GSD type 1b were also receiving G-CSF. Patients and HC did not differ regarding sex or age. None of the samples presented divergence≥ 30% for duplicates. Patient and HC were statistically different only for MDC/CCL22 (Median for patients= 427.61 pg/ml; for controls= 674.04 pg/ml; p=0.003). MDC/CCL22 levels did not differ between GSD type 1a and 1b patients. Comparison between GSD type Ia and Ib patients showed G-CSF is higher in the later ones (median for Ia=29.26 pg/ml; for 1b= 178.89 pg/ml; p= 0.001). Conclusion: Our findings do not suggest the presence of an inflammatory status in GSD type 1 patients. However, the low levels of MDC/CCL22 may suggest they are prone to infections and should be better evaluated. G-CSF was higher in GSD 1b patients probably because they were receiving G-CSF. Keywords: Glycogen Storage Disease, cytokines, cross-sectional study