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Title: Chitotriosidase activity and CHIT1 gene polymorphism in sarcoidosis

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Body: Introduction Among several biomarkers of sarcoidosis activity chitotriosidase (CTO) has been found to be useful. Some subjects have a 24-base pair duplication in the CTO gene (CHIT1) that results in the production of inactive enzyme which might interfere with the validity of the marker. Aims The study was conducted 1) to confirm previous observations of increased CTO activity in patients with sarcoidosis and 2) to evaluate influence of CHIT1 polymorphism on CTO activity. Methods The study comprises 47 patients with newly diagnosed active sarcoidosis (27 females, 20 males, average age 42,3 years). CTO activity was determined in serum and BAL using a standard fluorimetric method. CHIT1 genotyping was done by polymerase chain reaction (PCR). Results A normal CHIT1 genotype (NN) was present in 61,7% of the subjects, 34.0% were heterozygotes for defected allele (ND) and 4,3% were homozygotes (DD). The mean serum CTO value was 736 nmol/mL/h (± 583) and was increased in 93,6% of patients. The mean BAL CTO value was 9,01 nmol/mL/h (± 12.8). There was a correlation between serum and BAL CTO activities (r 0,710, $p < 0.001$). There was no significant difference between NN and ND subjects in serum CTO activity (855 ± 657 nmol/mL/h, 612 ± 348 nmol/mL/h, p 0,331), but a significant difference in BAL CTO activity (12,24 ± 15,02 nmol/mL/h, 4,31 ± 5,07 nmol/mL/h, p=0,001). There was no CTO activity in mutation homozygotes. Conclusions The results confirm previous observations that the CTO activity is increased in patients with sarcoidosis. Unexpectedly, there was no significant difference in serum CTO activity between CHIT1 normal homozygotes and heterozygotes which warrants further study.