

Serum Cytokine Profile in a Patient Diagnosed with Dysferlinopathy

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Abstract

© 2017 Svetlana F. Khaiboullina et al. Limb-girdle muscular dystrophy type 2 (LGMD2B) is a mild form of dysferlinopathy, characterized by limb weakness and wasting. It is an autosomal recessive disease, with currently 140 mutations in the LGMD2B gene identified. Lack of functional dysferlin inhibits muscle fiber regeneration in voluntary muscles, the main pathological finding in LGMD2B patients. However, the immune system has been suggested to contribute to muscle cell death and tissue regeneration. Serum levels of 27 cytokines were evaluated in a dysferlinopathy patient. Levels of 8 cytokines differed in patient serum compared to controls. Five cytokines (IL-10, IL-17, CCL2, CXCL10, and G-CSF) were higher while 3 were lower in the patient than in controls (IL-2, IL-8, and CCL11). Together, these data on serum cytokine profile of this dysferlinopathy patient suggest immune response activation, which could explain leukocyte infiltration in the muscle tissue.

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