Pompe Disease: Single Center Experience

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Pompe Disease, type II glycogen storage disease, is caused by mutations in the GAA gene leads to deficiency of acid alpha-glucosidase, a lysosomal enzyme that breaks down glycogen. Severe myopathy, muscle weakness and myocardial hypertrophy occurs in the absence of enzyme with lysosomal accumulation of glycogen. (1,2) Enzyme replacement therapy with recombinant human acid alpha-glucosidase was shown to improve cardiomyopathy, cardiac function and gain of gross motor milestones. (3) Herein we report a single center experience, Gazi University Medical Faculty Inherited Metabolic Disease Department, in Pompe Disease.

Thirteen patients had diagnosis of Pompe Disease since 2009. The age on diagnosis was 1 to 42 months (mean:7,3 months). There was 8 girls and 5 boys and consanguineous marriage was seen in eleven family. Except for the patient who was diagnosed at 42 months of age, they all had symptoms like hypotonia and cardiac hypertrophy before the first year. Therefore they all considered as classic infantile onset Pompe disease. Serum creatine kinase activity was elevated at the time of diagnosis as 319 to 922 U/L. Cardiac hypertrophy and/or cardiomyopathy was detected in all patients but 7 of them were needed medical support. In their physical examination mild to severe hypotonia was seen for all but only 5 of them had delay motor milestones. CRIM status could be measured in 10 patients, in which 7 patients had CRIM positive status. Among three patients with CRIM negative status, two received prophylactic immune tolerance therapy and one as treatment for high anti-drug antibodies titers. Except one patient who died before starting treatment, 12 patients had enzyme replacement therapy. Four of them had severe infusion reactions, and needed desensitization protocol which provide a successful management. Our own experience has been found to be compatible with the literature and constitutes an important example for Pompe Disease.(4)