Clinical and Molecular Features of Pompe Patients: Single Center Experience

Arzu Selamioglu¹, Meryem Karaca¹, Mehmet Cihan Balci¹, Hüseyin Kut Kaybeyli¹, Aslı Durmuş¹ and Gulden Gökçay¹
1 İstanbul Faculty of Medicine, Division of Pediatric Nutrition and Metabolism

Introduction:

Pompe disease is a lysosomal disorder caused by deficiency of acid alpha-glucosidase and causes classic infantile, childhood onset or adulthood onset phenotypes.

Objective:

We evaluated the records of 44 patients with Pompe disease.

Results:

Twenty-six (59%) of cases were girls.

Thirty-two (73%) were classical infantile form (IPD). The mean age of symptom onset in IPD patients (n=23) was 2.1 months (0.5-6 months) and the mean age of confirmed diagnosis was 4.8 months (1-12 months). Twenty-four cases died in their first years. Nine patients received alglucosidase alfa.

Five pathogenic variants including c.896T>C, c.2237G>A, c.2078dupA, c.2741delinsCAG and c.2662G>T were identified.

Median age of symptom onset in late onset Pompe disease (LOPD) patients (n=9) was 2.4 years (0.9-9 years) and median age of confirmed diagnosis was 3.2 years (1.2-12 years). Serum creatine kinase level elevation was detected in 9 LOPD patients, and the mean level at diagnosis was 809 (357–1926) U/L. Six patients received alglucosidase alfa of 20 mg/kg eow.

Four pathogenic mutations including c.2558C>A p.A853D, c.670C>T p.Arg224Trp, c.1655T>C and c.1784C>T were identified.

Three (6%) were adult form. At the time of diagnosis, the electromyogram was consistent with myopathy and there was restrictive respiratory failure. The muscle biopsy was suspected Pompe disease. Two patients received the standard dose of alglucosidase alfa of 20 mg/kg eow.

Discussion:

The molecular and clinical features of Pompe disease underlines the importance of early diagnosis and treatment.