

Blood film examination for vacuolated and PAS-positive lymphocytes as diagnostic screening test for patients with late onset Pompe disease (LOPD)

Alba Migliorato¹, Daniela Parisi², Olimpia Musumeci², Federica Montagnese², Marco Marino², Anna Ciranni², Carmelo Rodolico², Antonio Toscano²

¹ Department of Biomedical Sciences and of Morphological and Functional Imaging, University of Messina, Messina, Italy - ² Department of Neurosciences, University of Messina, Messina, Italy

Pompe disease is an inherited metabolic multisystemic disorder resulting in glycogen storage in different tissues, caused by a deficiency of the lysosomal enzyme acid alfa-glucosidase. Glycogen storage is often a morphological marker in muscle biopsy of Pompe patients but it could be also present in other tissues. Abnormal cytoplasmic vacuolation of lymphocytes, detectable on routine blood film examination, has been recently proposed as possible screening in these patients.

We examined blood smear of 16 LOPD patients, aged 14-71 years. The cohort phenotype encompasses 3 patients with presymptomatic hyperckemia, 2 with myalgia and fatigability, and 11 with proximal muscle weakness. Among those, 6 patients are on ERT treatment. We collected also peripheral blood films from 20 healthy controls and from 12 patients affected by other muscle glycogenoses. Blood sample was followed by preparation of four blood films: two of them were stained by May-Grunwald/Giemsa (MGG) and the other two by PAS. To investigate the diagnostic value of the test, we quantified the percentage of vacuolated lymphocytes and the percentage of PAS-positive lymphocytes.

The mean values of PAS-positive lymphocytes in LOPD patients (25.6%) were significantly higher than those of healthy controls (4.6%) and of patients with other muscle glycogenoses (4.8%). In this group of patients, we have shown that PAS-positive cytoplasmic vacuolation of lymphocytes in peripheral blood films could be considered as a reliable screening tool to support an early diagnosis of Pompe disease.

Keywords

Diagnosis of Pompe disease; vacuoles; PAS-positive lymphocytes.