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Poster Abstract: Diagnostic

Dried Blood Spot for Screening for Late-Onset Pompe Disease: A Spanish Cohort

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BACKGROUND

Pompe disease is frequently underdiagnosed and diagnosed with a big delay, usually measured in years. An early diagnosis is important because enzyme replacement therapy seems to be more effective in less affected patients.

PATIENTS AND METHODS

To screen patients suffering from late-onset Pompe disease (LOPD), we performed a dried blood spot (DBS) in 248 patients: 146 patients with unclassified limb–girdle myopathy and 202 with asymptomatic hyperCKemia.

RESULTS

Twenty patients had abnormal DBS results, in whom genetic studies were performed. LOPD was confirmed in 16 cases: 11 with proximal myopathy and 5 with asymptomatic hyperCKemia. The c.-32-13 T>G was the most common mutation found. The mean diagnostic delay was 14 years.

CONCLUSIONS

DBS is a reliable tool for screening for Pompe disease. We recommend its use in the diagnostic work-up of patients with unclassified proximal weakness and/or hyperCKemia of unknown origin.

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