Introduction: Gaucher’s disease (GD) is the most common inherited lysosomal storage disorder and presents with a wide clinical spectrum including parkinsonism. Recent clinical, neuropathological and genetic studies have implicated Glucocerebrosidase (GBA) gene mutations in different parkinsonian phenotypes and in Parkinson Disease (PD) susceptibility.

Aim: Describe an atypical parkinsonian syndrome in a patient with GBA gene mutations.

Methods: A 70 year-old man presents with a 4-year history of parkinsonism with a predominance of bilateral akinetic-rigid symptoms and progressive gait disturbance. He had a poor response to conventional anti-parkinsonian therapy, presenting from the beginning dyskinesias and oromandibular dystonia. In the two years prior to the emerging parkinsonian features he starts developing a progressive mood/behaviour disturbance with obsessive-compulsive disorder and akathisia. No other complaints were reported and had never suffered from other disease.

Results: Hoehn-Yahr stage was 3 and UPDRS score was 31. General physical and neurological examination (besides parkinsonian features) was unremarkable. He performed routine blood test. He was also tested for GBA and LRRK2 mutations and revealed two mutations in exon 10 of GBA gene (L444P and A456P).

Conclusion: According to genetic testing a diagnosis of GD type 1 was made. The present case is distinctive in combining atypical parkinsonism with a GBA heterozygous compound mutation found in an older patient without typical GD manifestations. Both mutations have been previously reported. At present, the nature of the association between GD and Parkinson’s Disease remain to be elucidated, nevertheless, GD diagnosis must be considered in the differential diagnosis of atypical parkinsonism.