Brain MRI has a distinct feature with eye-of-the-tiger sign characterized by bilateral hypointensity of the globus pallidus with a central area of hyperintensity on T2-weighted images. Peripheral blood acanthocytosis and low plasma pre beta lipoprotein are supportive evidence. Establishing PANK2 variant on molecular genetic testing is confirmatory.

Management is mainly supportive. Dystonia can be controlled with benzodiazepines, anticholinergics and botulinum toxin. Spasticity can be revealed with baclofen. Stereotactic surgical modalities such as thalamotomy, pallidotomy and deep brain stimulation of the globus pallidus can help to control symptoms.

We present a 5-year-old boy who presented with tiptoe walking who gradually deteriorated neurodevelopmentally. He had declined in academic performance and progressive dystonia, dysarthria and spasticity was there. He had hyperreflexia but parkinsonism, chorea or myoclonus was not evident. The neuro-ophthalmologic examination was normal. Laboratory work up was normal including serum copper, ceruloplasmin levels. Blood smear had no acanthocytes. T2 weighted brain work up was normal including serum copper, ceruloplasmin but parkinsonism, chorea or myoclonus was not evident. The neuro-ophthalmologic examination was normal. Laboratory work up was normal including serum copper, ceruloplasmin levels. Blood smear had no acanthocytes. T2 weighted brain MRI was consistent with eye-of-the-tiger sign. Genomic testing was not performed due to the cost effect.

The child is under multidisciplinary rehabilitation to improve quality of life.

This case report highlights the importance of considering this entity in progressive dystonia with neurodegeneration.