X-linked dystonia parkinsonism

X-linked dystonia parkinsonism (XDP), also known as Lubag Syndrome or X-linked Dystonia of Panay, is a rare x-linked progressive movement disorder with high penetrance found almost exclusively in males from the Panay, Philippines.

It is characterized by dystonic movements first typically occurring in the 3rd and 4th decade of life. The dystonic movements often either coexist or develop into parkinsonism within 10 years of disease onset.

Previous case reports have reported beneficial effects from bilateral pallidal deep brain stimulation (DBS).

Kilbane et al. report the long-term clinical outcomes of 3 patients treated at our center.

All patients presented with medication refractory dystonia and parkinsonism. They were followed prospectively. Clinical evaluations included the Burke-Fahn-Marsden Dystonia Rating Scale (BFMDRS) and the Unified Parkinson's Disease Rating Scale (UPDRS). Adverse events were recorded.

The average length of follow-up was 45.7 months. No serious adverse events occurred. All patients experienced an immediate and sustained improvement in dystonia. Mean percentage improvement in motor subscores of BFMDRS was 63.5% at the last follow-up visit. Parkinsonism was less responsive to neuromodulation, with a mean improvement in UPDRS-III of 39.5%. Standard pallidal stimulation parameters were used. Freezing of gait developed after DBS therapy in 2 patients, stimulation-induced in one and due to disease progression in the other.

Bilateral pallidal DBS resulted in significant and sustained improvement in dystonia and moderate improvement in parkinsonism. Pallidal DBS represents an important treatment option for XPD for the management of motor symptoms [1].

1)
Kilbane C, Witt J, Galifianakis NB, Glass GA, Volz M, Heath S, Starr PA, Ostrem JL. Long-Term Outcomes of Bilateral Pallidal Deep Brain Stimulation for X-Linked Dystonia and Parkinsonism. Stereotact Funct Neurosurg. 2018 Nov 27;96(5). doi: 10.1159/000492823. [Epub ahead of print] PubMed PMID: 30481788.