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Case Report

Sustained Effect of Posteroventral Pallidotomy for X-Linked Dystonia Parkinsonism

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Abstract

Background: X-linked dystonia parkinsonism (XDP, DYT3), a disease endemic to Panay Island in the Philippines, is a progressive adult-onset sex-linked neurodegenerative movement disorder with features of both dystonia and parkinsonism. Medical treatment and botulinum toxin injection (BTX) are of limited benefit. Deep brain stimulation (DBS) of the bilateral globus pallidus internus (GPi) is effective but is expensive and requires specialized equipment, training and after care. Posteroventral pallidotomy (PVP) is an older known ablative procedure which has successfully treated cervical and generalized dystonia, but has not been previously used for XDP. *Case presentation*: Two male adults with genetically-confirmed XDP, presented with progressive and severely disabling dystonias. The mean preoperative Burke-Fahn-Marsden Dystonia Rating Scale (BFMDRS) was 80.5. Both XDP patients underwent a unilateral posteroventral pallidotomy. In the first case, there was a 38.46% decrease in BFMDRS score at 2 weeks sustained to 41.66% decrease (compared to pre-op BFMDRS at 4 years post-surgery). In the second case, there was a 33.73% decrease in BFMDRS at 2 months post-op, and a decrease of 86.74% in BFMDRS score at 3 years post-surgery. *Conclusion*: While not as effective as bilateral pallidal DBS, the benefits of a unilateral PVP are still substantial and sustained while also being less costly and resource expensive.

Keywords

XDP, Dystonia, Parkinsonism, Pallidotomy

1. Introduction

X-linked dystonia parkinsonism (XDP, DYT3) is a progressive adult-onset sex-linked neurodegenerative movement disorder with features of both dystonia and parkinsonism. It endemic to Panay Island in the Philippines and cases described elsewhere have been linked to Filipino ancestry. XDP was first described in 1975 by Lee [1].

Medical therapy and botulinum toxin injection (BTX) are of limited benefit and primarily symptomatic [2, 3]. The only effective treatment at present is bilateral globus pallidus internus (GPi) deep brain stimulation (DBS), a prohibitively expensive surgical procedure [4]. It has become the preferred surgical treatment for virtually all types of dystonia due to its

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superior safety profile compared to pallidotomy, particularly in bilateral cases [5].

Posteroventral pallidotomy (PVP) is an ablative procedure, commonly used in the past in cervical and generalized dystonia. The procedure became less popular after the advent of DBS, but is again recently regarded as a viable alternative to DBS [5-8]. There is no previous record of its use in XDP in the literature.

2. Case Presentation

1. Patient 1

A 44-year-old Filipino male government-worker has an initial presentation of blepharospasm and involuntary beating movements of his right arm. He has maternal origins in Capiz, which is endemic for XDP, had SVA (SINE-VNTR-Alus) repeats on genetic testing, and was diagnosed to have XDP. Over three years, he developed dystonic posturing of the right arm, later generalizing to the neck, shoulder, all extremities, and the trunk. He could walk short distances with difficulty but was primarily wheelchair-borne. He had difficulty in speech and swallowing and required some assistance with feeding. Initial Burke-Fahn-Marsden Dystonia Rating Scale (BFMDRS) was 78. He was treated with Biperiden, Clonazepam, and Zolpidem with limited benefits. BTX injections ameliorated some dystonic symptoms. Due to financial limits, he opted to undergo unilateral PVP instead of DBS.

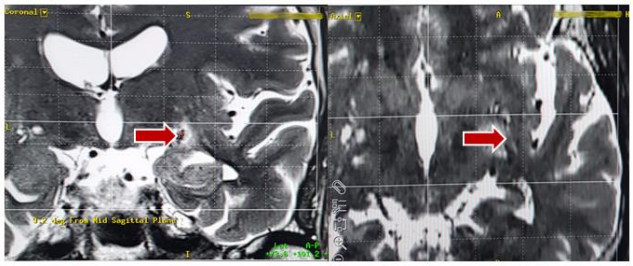
2. Patient 2

A 45-year-old male initially presented with stumbling difficulty in speech followed by neck twisting and hyperextension. He rapidly developed progressive arm dystonia, blepharospasm, and difficulty in swallowing. Truncal dystonia with associated pain resulted in difficulty ambulation with small steps. His origins are from Panay islands, and has a brother who had similar dystonic movements. His gene testing showed SVA repeats. Initial BFMDRS was 55. He was treated with Biperiden and Tramadol. Due to financial constraints and progressive deterioration and cachexia, the family opted to undergo PVP.

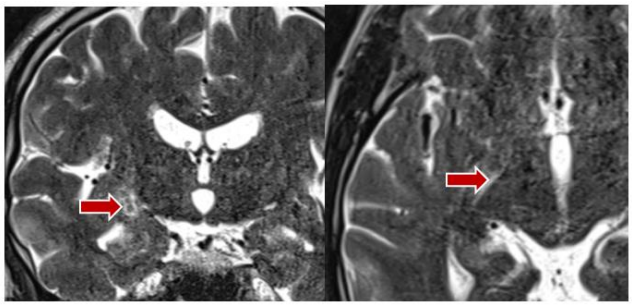
Posteroventral pallidotomy

Both patients underwent PVP using a Leksell stereotactic frame under local anesthesia. Patient 1 had left PVP, while patient 2 had right PVP. A stereotactic computerized tomography (CT) scan (1 mm cuts) was fused with volumetric magnetic resonance imaging (MRI) (2 mm cuts, 1.5 T) and used to identify the coordinates of the GPi. The target was determined on direct inspection of images and using standard offsets from the mid-commissural point. Unilateral frontal incision and burr hole were made and a 2-mm exposed tip lesion electrode was inserted. Impedance testing and macrostimulation were carried out to confirm position and the effect of stimulation.

Motor threshold was determined using 4 Hz, 100 msec macrostimulation with increasing amperage and optic tract scintillations elicited using 133 Hz, 100 msec at around 7.5 mA (2-3 volts). At target, high frequency macrostimulation was performed with intraoperative evaluations for improvement and adverse effects. We performed test lesions for 60 seconds at 46 degrees Celsius and at 60 degrees Celsius. The permanent lesion was made at 76 degrees Celsius for 60 seconds at the selected depth and at -2 mm depth and -4 mm depth. Post-operative MRI verified placement of the lesion in the GPi (see Figure 1).



Patient 1



Patient 2

Figure 1. *Coronal and axial views of post-pallidotomy MRI showing the lesions in the left and right GPi (red arrow) of Patients 1 and 2, respectively.*

3. Results

1. Patient 1

The patient's pre-operative BFMDRS was 78. He had an early improvement to a score of 48 (38.46% improvement from baseline) at 2 weeks post-pallidotomy. Patient then had a sustained improvement ranging from 33.33% to 41.66% over the succeeding months and years (2 months, 4 months, 1 year, 2 years, 4 years) (see Table 1). All the BFMDRS components improved but the greatest improvements were observed in blepharospasm and truncal dystonia (see Table 2 and Figure 2). Although PVP is a unilateral procedure, both left and right extremities improved, expectedly with the right side more than the left. When queried, the patient was appreciative of the improvement, particularly in speech and swallowing. There was also improvement in the activities of daily living (ADL). The patient was capable of limited but independ-

ent walking and transfers. Self-feeding required less assistance and nutrition improved with less choking.

Table 1. BFMDRS scores of Patient 1 pre- and post-pallidotomy and their corresponding % improvement from the baseline.

Timeline of assessment	BFMDRS score	% improvement from baseline
Pre-op	78	---
Post-op	80	-2.56%
2 weeks	48	38.46%
1 month	64	17.94%
2 months	52	33.33%
4 months	53	32.05%
1 year	49	37.17%
2 years	45.5	41.66%
4 years	51	34.61%

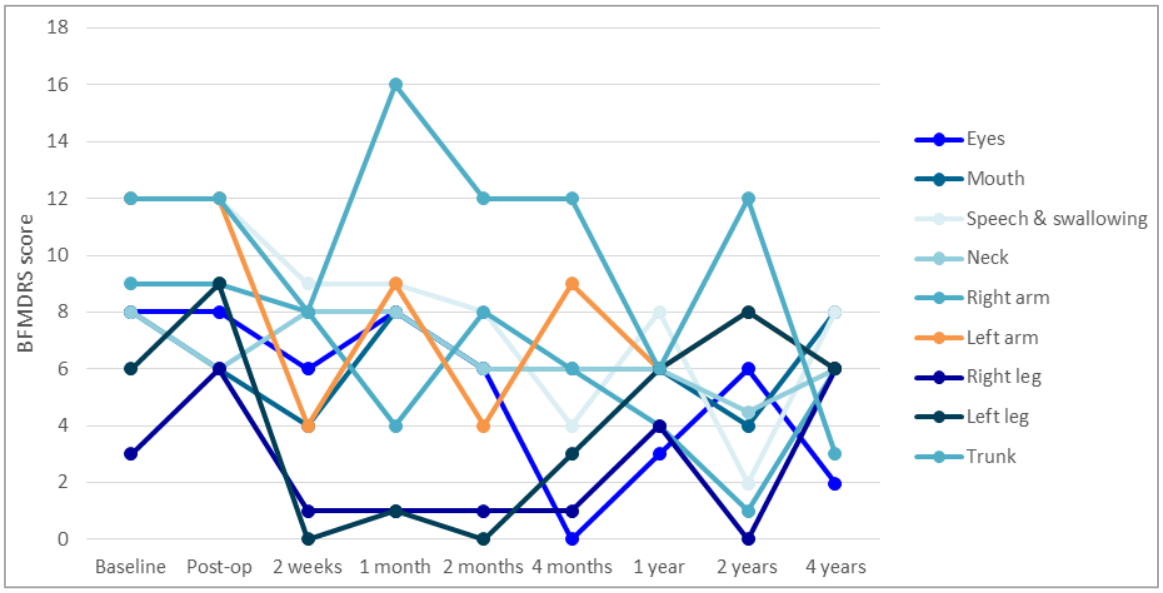


Figure 2. Regional changes in the BFMDRS of Patient 1 after left unilateral PVP.

Table 2. Components of the BFMDRS scores of Patient 1 after left PVP.

Components of BFMDRS	Baseline	Post-op	2 weeks	1 month	2 months	4 months	1 year	2 years	4 years
Eyes	8	8	6	8	6	0	3	6	2
Mouth	8	6	4	8	6	6	6	4	8
Speech & swallowing	12	12	9	9	8	4	8	2	8
Neck	8	6	8	8	6	6	6	4.5	6
Right arm	9	9	8	4	8	6	4	1	6
Left arm	12	12	4	9	4	9	6	8	6
Right leg	3	6	1	1	1	1	4	0	6

Components of BFMDRS	Baseline	Post-op	2 weeks	1 month	2 months	4 months	1 year	2 years	4 years
Left leg	6	9	0	1	0	3	6	8	6
Trunk	12	12	8	16	12	12	6	12	3
TOTAL	78	80	48	64	52	53	49	45.5	51
% Improvement compared to baseline	-	-2.5%	38.46%	17.94%	33.33%	32.05%	37.17%	41.66%	34.61%

2. Patient 2

The patient's pre-operative BFMDRS was 83. He had the earliest recorded improvement to a score of 55 (33.73% improvement from baseline) at 2 months post-pallidotomy. Patient then had a sustained and progressive improvement ranging from 59.03% to 86.74% over the succeeding months and years (3 months, 1 year, 3 years) (see Table 3). In addition, all BFMDRS components improved but the greatest improvements were in the dystonia of right arm and both legs, closely followed by improvements in blepharospasm and oromandibular dystonia (see Table 4 and Figure 3). Just like in Patient 1, both left and right extremities improved despite undergoing a unilateral procedure (right PVP). The patient's truncal pain and corresponding Tramadol dose decreased, as well as his extremity numbness and salivation. His improvements in speech, swallowing and ambulation translated into the improvement in the ADL.

Table 3*. BFMDRS scores of Patient 2 pre- and post-pallidotomy and their corresponding % improvement from the baseline.*

Timeline of assessment	BFMDRS score	% improvement from baseline
Pre-op	83	---
2 months	55	33.73%
3 months	34	59.03%
1 year	25	69.87%
3 years	11	86.74%

Table 4. *Components of the BFMDRS scores of Patient 2 after right unilateral PVP.*

Components of BFMDRS	Pre-op	2 months	3 months	1 year	3 years
Eyes	8	2	2	0	0.5
Mouth	8	6	2	2	0.5
Speech & swallowing	12	6	6	2	1
Neck	8	8	6	6	6
Right arm	8	6	4	1	0
Left arm	12	12	6	4	2
Right leg	6	0	0	0	0
Left leg	9	3	2	1	0
Trunk	12	12	6	9	1
TOTAL	83	55	34	25	11
% Improvement compared to baseline	---	33.73%	59.03%	69.87%	86.74%

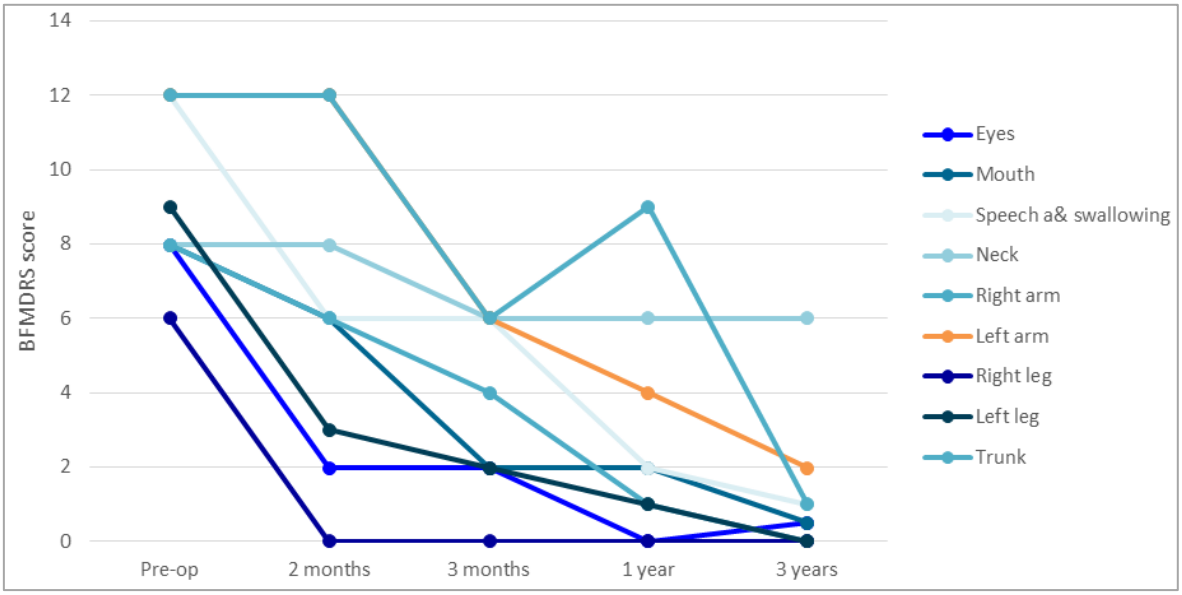


Figure 3. *Regional changes in the BFMDRS of Patient 2 after right unilateral PVP.*

4. Discussion

Medical treatment in advanced XDP is limited and largely ameliorative. Anticholinergics and benzodiazepines are the mainstays of initial treatment but in the advanced severely dystonic stage, zolpidem is the only effective medication [9]. Even this drug typically improves BFMDRS by only a few points and for a few hours [3]. BTX improves specific targeted muscles but benefits are focal and lasts weeks to months [10].

Bilateral pallidal DBS is currently the best treatment for XDP. There is an average of 60-80% improvement in BFMDRS scores [11]. The improvement is sustained [12] showed a positive impact in improving the quality of life among XDP patients [13]. Progression of XDP can be addressed with reprogramming of the pulse generator. The barriers to DBS are high, with a cost of Php 1.8 to 2 million (USD 30,000-35,000) being the greatest. Long term, the risk of hardware and infectious complications are usually quoted as 1-3% [14], but may be as high as 7-10% over multiple years.

With the sustained improvement in BFMDRS of the 2 patients in this case series, unilateral PVP may be considered superior to medical treatment alone, but inferior to bilateral pallidal DBS. Bilateral pallidotomy or additional pallidotomy in the future may further increase this improvement.

The improvement in BFMDRS is sustained in both cases. In patient 1, the latest BFMDRS improvement was 34.61% from baseline at 4 years post-PVP, but the best improvement was observed 2 years post-surgery with 41.66%. In patient 2, the trend of improvement was smoother with lower BFMDRS scores in every follow-up consultation. The best improvement of 86.74% was observed during the latest encounter at 3 years post-surgery. Patient 2's latest improvement is more than twice compared to what patient 1 achieved. There was notable progression in both patients as they became more parkinsonian over time, but the dystonic features remained stable.

In both cases, the need for BTX injection in other areas such as the back and neck were also reduced following pallidotomy. The oromandibular, lingual, and oropharyngeal dystonias which are typically difficult to treat with BTX had also improved. The improvement in dysphagia greatly increased the patient's caloric intake and forestalled the implantation of a tube gastrostomy. Although the overall improvement is insufficient to return the patient to a normal state, it made a difference between complete assistance and mild to moderate assistance in activities of daily living.

Besides costs, PVP is less restrictive in eligibility criteria than DBS. Extreme cachexia, inability to tolerate multiple hours of awake surgery, and psychologic contraindications bar DBS from many patients with advanced XDP. DBS also requires more frequent and regular outpatient follow-up and evaluation in a specialty center, whereas no additional and special follow-ups are required following pallidotomy.

The greatest limitation of pallidotomy in comparison to DBS is its one-shot nature. The improvements in pallidotomy are typically swiftly attained following the procedure but also plateau thereafter. In cases of rapid progression of XDP, there is no additional benefit. In DBS, programming allows maximization and targeting of benefits in the months following surgery. Reprogramming addresses progression of the disease.

5. Conclusion

Unilateral PVP is a viable surgical treatment for XDP. It is not as effective as bilateral pallidal DBS, but is less costly

and has less eligibility restrictions. Bilateral PVP is a possibility for future exploration. As a limitation in this pilot study, the authors were unable to use the XDP-MDSP rating scale, which could have also assessed the parkinsonian features of both patients.

Abbreviations

ADL	Activities of Daily Living
BTX	Botulinum Toxin
BFMDRS	Burke-Fahn-Marsden Dystonia Rating Scale
CT	Computerized Tomography
DBS	Deep Brain Stimulation
GPi	Globus Pallidus Internus
MRI	Magnetic Resonance Imaging
PVP	Posteroventral Pallidotomy
SVA	SINE-VNTR-Alus
XDP	X-linked Dystonia Parkinsonism

Conflicts of Interest

The authors declare no conflicts of interest.

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