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Bilateral Globus Pallidus Internus (GPi) Deep Brain Stimulation (DBS) for a 4 year-old girl with GNAO1 Mutation Related Status Dystonia: Case Report and Literature Review.

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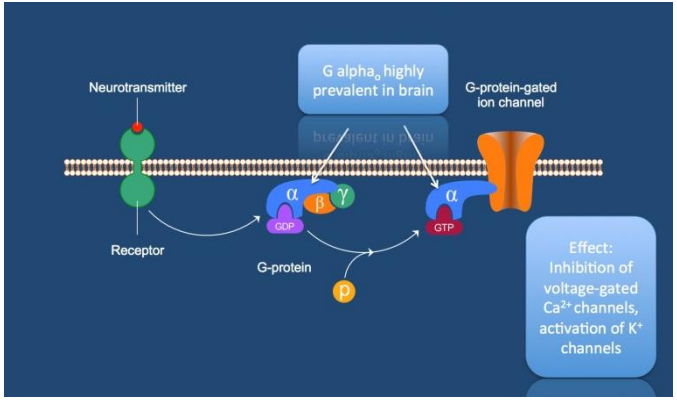
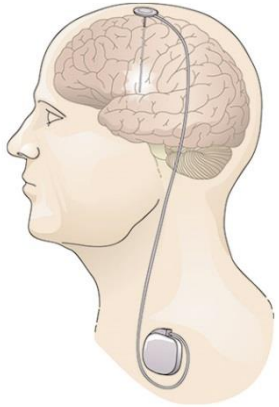
Introduction

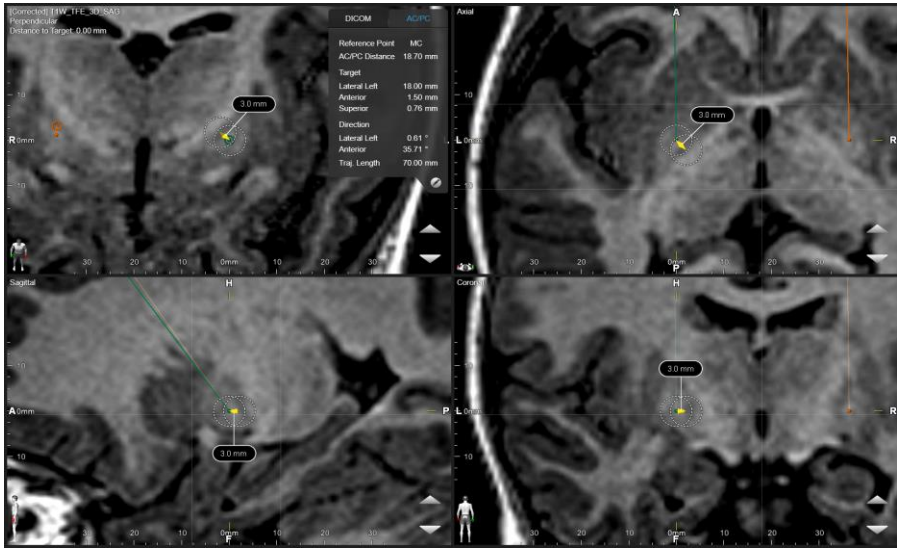
- GNAO1 codes for the Ga0 subunit of a G protein coupled receptor, which is prevalent in the central nervous system
- Patients with GNAO1 mutation present with neurodevelopmental delay and hyperkinetic movement disorder which can be complicated by life-threatening exacerbations (status dyskinesia) refractory to medical treatment.
- DBS has been emerging as an effective treatment to abolish dyskinetic crises
- 15 cases have been reported in the literature as of 2019



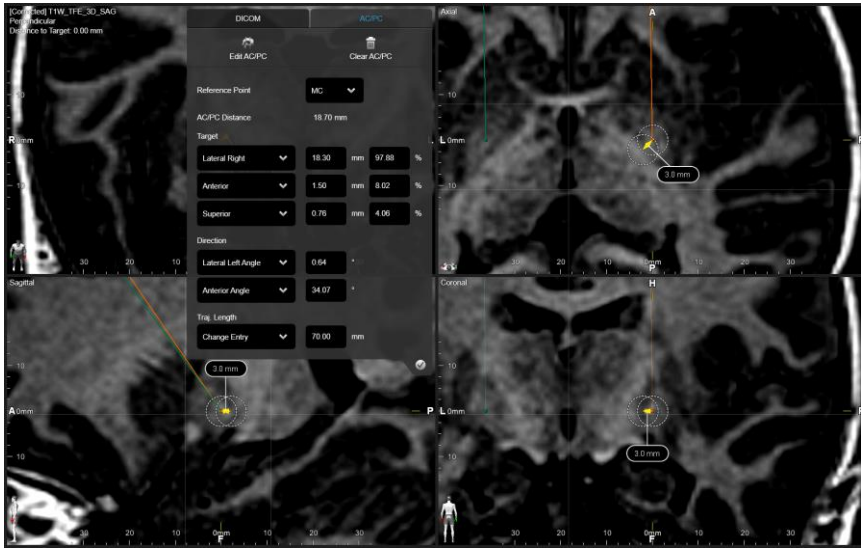


Case Report

- Guo CY, 4-year-old girl with de novo GNAO1 mutation
- Developed generalized dyskinesia affecting the neck and all limbs at age 3-4 months. Also noted to be global developmental delayed by age 7 months. Limited improvement with medications including Levodopa, Artane and Clonidine.
- Suffered from a severe exacerbation of dystonia triggered by a viral infection in August 2019
- Refractory to medical treatment and complicated by rhabdomyolysis requiring paediatric intensive unit care.
- In view of the poorly controlled dyskinesia, she was referred to us in October 2019 for consideration of DBS.
- Pre-operative MRI DBS protocol performed under sedation on January 15, 2020 – DBS plan finalized, kept under sedation
- Bilateral GPi DBS performed January 16, 2020 (frame-based stereotactic surgery, frontal burr hole, MER, macro-stimulation, directional lead, Vercise Gevia IPG)



Left GPi targeting



Right GPi targeting

Leksell Vantage Frame

Vercise Gevia IPG, rechargeable

Case Report

- Extubated and DBS switched-on on postop day 1
- Significant improvement of hyperkinetic movement
- Discharged on postop day 8
- Medications stepped down
- Nasogastric tube removed 1 month postop
- Further improvement in hyperkinetic movement noted with subsequent programming of DBS

	PICU stay (Aug 2019)	Prior to DBS (Jan 2020)	10 months Post-op (Oct 2020)
Abnormal Involuntary Movement Scale (AIMS)	4/4	3/4	2/4
Fahn-Marsden Dystonia Rating Scale	108/120	106.5/120	64.5/120

Score comparison

Case	Contact	Amplitude	Pulse Width	Rate
+	1-	3.9mA	60us	130Hz
+	10-11-12-	3.0mA	60us	130Hz

DBS parameters, 9th November 2020

Discussion

- Issues encountered on long term follow up
  - Abrupt interruption of stimulation can result in recurrence
  - Improvement in hyperkinetic and choreatic component, but increased muscle tone and dystonic posturing
- Challenges of DBS in young children
  - Higher complication rate in children vs adults – infection/hardware related complications due to excessive movement in early postop period → lead fixation and wound management
  - Lead migration during growth – projected increase in distance between entry point and target of 5 to 10 mm was found from age 4 to 18 years → potential loss of the DBS effect with recurrence

Conclusion

- 4 year old girl with GNAO1 mutation associated severe hyperkinetic movement disorder underwent bilateral GPi DBS with good outcome up to 10 months follow up