

Jeroen Cortier MD², Stijn Vandamme DBS nurse¹, Wim Maenhoudt MD¹, Stephanie Du Four MD PhD¹,
Jeroen Van Lerbeirghe MD¹, Dimitri Vanhauwaert MD¹, Olivier Van Damme MD¹

¹ Department of Neurosurgery, AZ Delta, Roeselare, Belgium
² Department of Neurosurgery, AZ Maria Middelares, Ghent, Belgium

Introduction

Bassen-Kornzweig syndrome or abetalipoproteinemia is a rare autosomal recessive disorder characterized by a malabsorption of dietary fat and fat-soluble vitamins.

The condition is caused by biallelic mutations in the microsomal triglyceride transfer protein gene (MTTP gene) and is inherited in an autosomal recessive manner.

This deficiency can lead to a variety of symptoms, including hematological (acanthocytosis, bleeding tendency), neurological (tremor, spinocerebellar ataxia), neuromuscular (myopathy), ophthalmological symptoms (retinitis pigmentosa).

The thalamic ventral intermediate nucleus (VIM) is a well-established target for deep brain stimulation (DBS) in the treatment of refractory tremor.

Case presentation

We report a 53-year-old male who suffers from abetalipoproteinemia since the age of 17. This 17-year-old patient presented at the time to the neurological outpatient clinic with a disabling intention tremor and ataxia.

A clinical work-up confirmed an ApoB deficiency in the blood serum, acanthocytosis and a vitamin E deficiency.

Despite Vitamin E suppletion, and treatment with propranolol (up to 160mg/day) and primidone (up to 1500mg/day), the patient had a persistent disabling tremor.

A brain MRI (1.5T) showed no abnormalities. Best medical treatment did not suppress the tremor in a sufficient way.

At this time the patient had a score of 95 on the Fahn-Tolosa-Marin scale.

Diagnostic video



SCAN ME

Spiral drawings

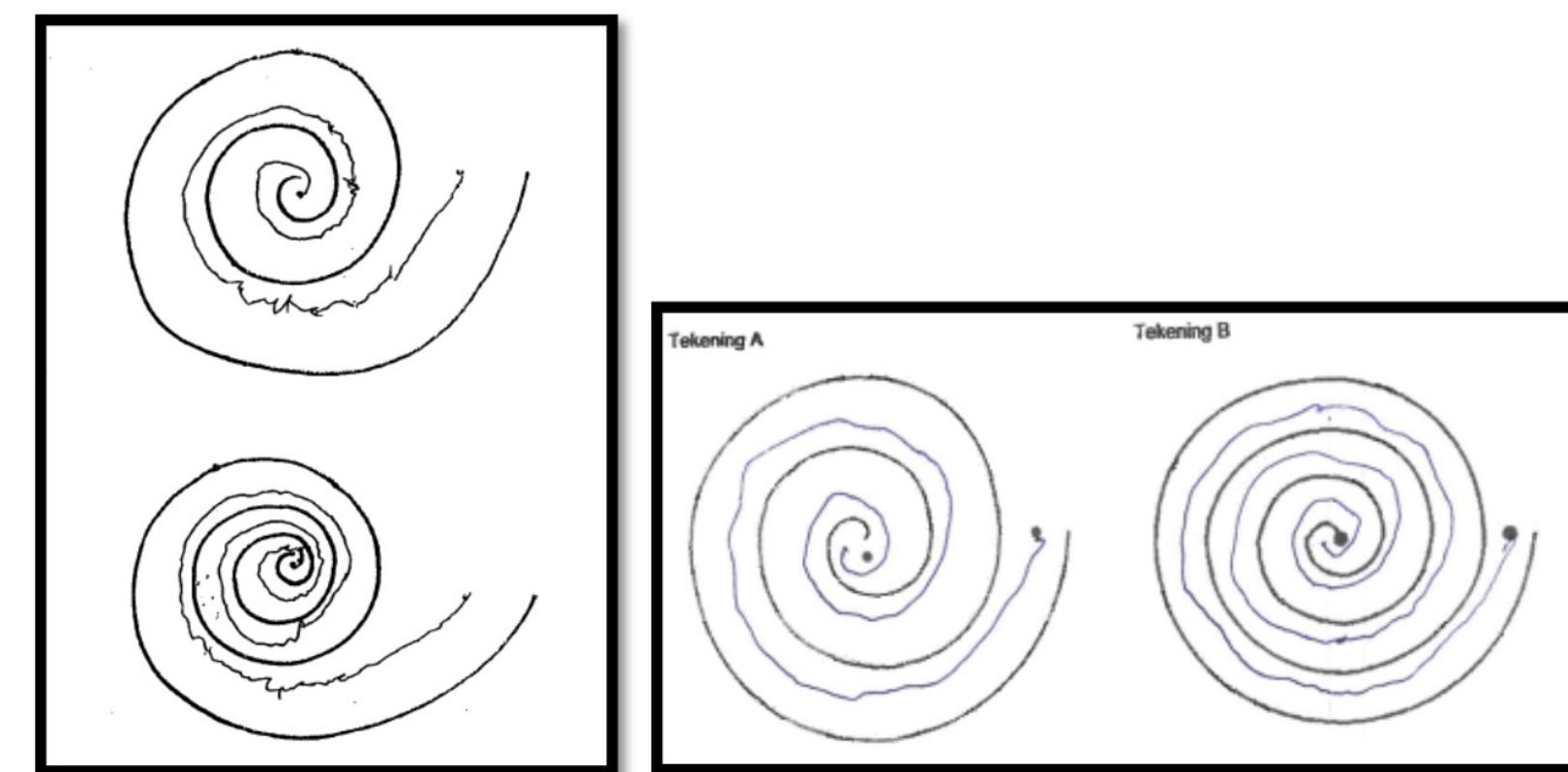


Figure 1 – Spiral drawings pre- (left) and post VIM-DBS (right)

Stimulation parameters

Postoperative settings	
Setting	Left Right
Contacts	Monopolar Bipolar
Current (V)	1- 2- 1- 0+ 3+
Pulse Width (μs)	3.2 3.6
Frequency (Hz)	60 240
	130 130

Figure 2 – Stimulation parameters

Surgery & postoperative care

In 2000, after a full multidisciplinary work-up (neuropsychological-, neurology-, and neurosurgery counseling) deep brain stimulation was proposed.

He underwent bilateral VIM-DBS to treat his disabling refractory intentional tremor at the age of 31.

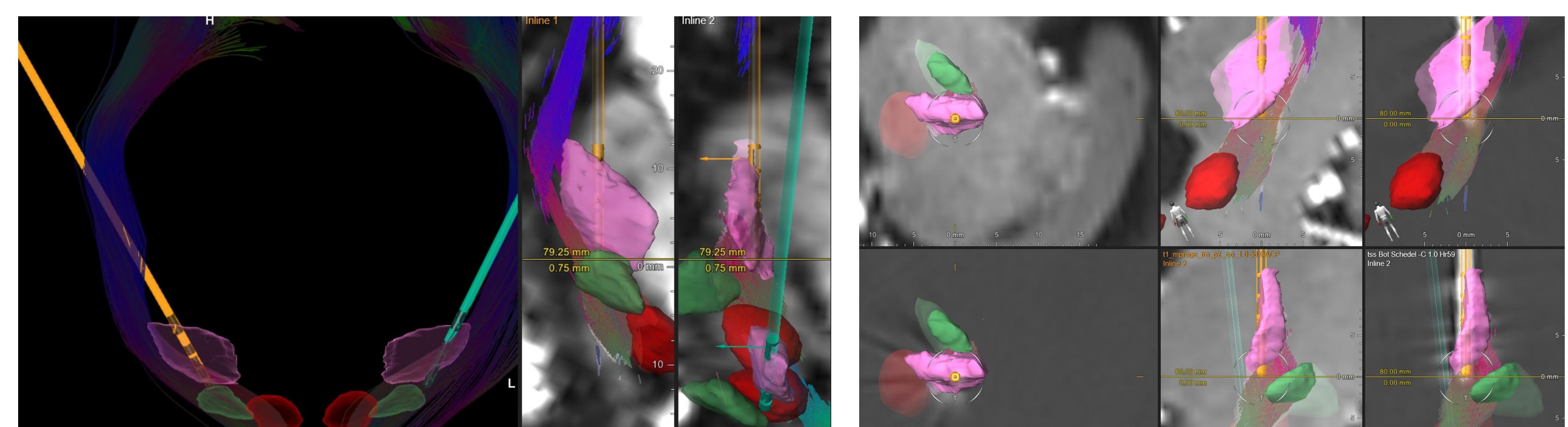


Figure 3 – VIM-targeting and lead positioning using Brainlab software. Blue: Tractography, Pink: VIM nucleus, Green: STN nucleus, Red: Red Nucleus

On the first postoperative day the stimulation was initiated according to the same stimulation parameters in the DBS treatment for essential tremor, i.e. multiple contacts with intermediate high voltage.

He experienced a very good response to his tremor with limited stimulation adaptations postoperatively. For more than two decades follow-up, the treatment significantly improved his ADL functions and therefore also the QoL.

Conclusion

- The VIM target for DBS in the treatment of refractory tremor has been extensively reported in the literature.
- Thalamic VIM-DBS is a safe and effective treatment for a severe, refractory tremor as a neurological symptom caused by abetalipoproteinemia.
- It also highlights the importance of a multidisciplinary follow-up, to adjust and optimize the stimulation/medication balance after VIM-DBS surgery.