SEIZURE FREEDOM WITH VNS IN NEUROFIBROMATOSIS TYPE 1: A CASE REPORT


Background

Neurofibromatosis type 1 (NF1) is an autosomal dominantly inherited disorder characterized by multiple café-au-lait macules, Lisch nodules, axillary or inguinal freckling, neurofibromas, distinctive bone abnormalities, and optic pathway gliomas. Seizures occur in 4-7% of individuals with NF1, mostly due to associated brain tumors or cortical malformations. Epilepsy associated with NF1 is usually well controlled with anti-epileptic drugs. However, in some pharmacoresistant patients vagus nerve stimulation (VNS) presents a complementary treatment option.

Case presentation

We present a 24-year-old male patient with NF1 who had focal autonomic seizures with impaired awareness, as well as focal motor seizures with retained awareness, in addition to moderate intellectual disability. MRI abnormalities included infiltrative changes of medulla oblongata, pons and cerebellum, as well as signal intensity changes with mild compression in the apex of the temporal lobes, insular cortex, putamen and medial part of the frontal lobe, all more prominent on the right, along with right mesial temporal sclerosis. Also, increased signal intensity with signs of left hippocampal enlargement was found. Since the patient had been diagnosed with a pharmacoresistant form of multifocal epilepsy, after an extensive preoperative evaluation VNS implantation was performed. In the 3.5-year follow up period, complete seizure freedom was achieved.

Conclusion

This case illustrates that NF1 may be associated with refractory epilepsy and suggests that, in such cases, vagus nerve stimulation therapy should be considered as a treatment option for better seizure control. Further carefully designed studies are needed.

Keywords

Neurofibromatosis, Pharmacoresistant epilepsy, Vagus nerve stimulation, Seizure freedom

Declaration of patient consent

The patient has given his consent for his images and other clinical data to be reported in this case presentation.