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Neurofibromatosis Type 1 with tumor resection: Case report

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Abstract

Neurofibromatosis type 1 has physical variability or clinical signs associated with the pathology; disease that focal gliomas can occur in the brainstem.

Introduction

Neurofibromatosis type 1, also known as von Recklinghausen disease, is an autosomal dominant familial tumor syndrome. The incidence of NF1 is about 1 in 3,500 individuals. Low-grade gliomas involving the optic pathway and brainstem are the most common central nervous system neoplasms in patients with NF1. Glioblastoma is a common malignant brain tumor in adults but is a rare occurrence in patients with NF1. We report a rare occurrence of glioblastoma in a 60-year-old man with NF1.

Case Report

A 60-year-old Indian diabetic male presented with headache, neck pain, and gait unsteadiness of 3 days duration. On examination, the patient had postural hypotension. Motor examination revealed left hemiparesis. Multiple subcutaneous soft swellings (most of them pedunculated) along with multiple café-au-lait spots were seen all over his body, including the scalp. He also had inguinal and axillary freckling. Ophthalmologic examination was normal. The patient had no bone lesions. There was no family history of neurofibromatosis. Abdominal imaging showed a well-defined cystic lesion in the right adrenal gland with multiple enhancing septations and solid areas with hemorrhage, suggestive of cystic pheochromocytoma. Computed tomography (CT)-brain showed a variegated space occupying lesion (SOL) in the right-frontal-lobe with a large cystic component and an enhancing solid component. Moderate surrounding edema was noticed with mass effect and midline shift to left. Magnetic resonance imaging (MRI)-brain confirmed a predominantly cystic intra-axial right-frontal-SOL with enhancing solid component, perilesional edema, and mass effect.

Mutations of the NF1-gene (a tumor-suppressor-gene) located on chromosome 17q11.2, partly explains the clinical susceptibility for malignancies.The NF1-gene product neurofibromin functions in part as a negative regulator of the p21 Ras proto-oncogene by accelerating the conversion of active guanosine triphosphate bound Ras to its inactive guanosine diphosphate bound form. Active as a result of reduced or absent neurofibromin expression, lead to increased cell growth and facilitates tumor formation.

Molecular analysis of GBMs arising in NF1-patients showed the presence of genetic alterations such as p16INK4A/ARF deletions and p53 mutations. These alterations are believed to cooperate with NF1 in the development of malignant astrocytomas reported that in their mouse model with NF1 and simultaneous loss of p53, most tumors arose in the vicinity of the subventricular zone (SVZ), where the majority of neural precursor cells reside. This indicates:

- Cells in this region of the brain are more susceptible to p53/ NF1-mediated tumor formation
- The micromilieu of the SVZ is more advantageous to the growth of brain tumor precursor cells.

Objectives

To report the results of the evaluation of the peripheral and central auditory system before and after the acoustically controlled auditory training of a patient submitted to tumor resection.

Resumed Report

The patient with 11 years old with neurofibromatosis type 1, at 9 years underwent surgical intervention in the brainstem. School difficulties that accentuated after surgery due to social isolation. The patient presented auditory thresholds within bilaterally normal patterns and adequate middle ear functioning. Auditory brainstem response revealed latency III and V increased and interpeak intervals increased bilaterally. The evaluation of central auditory processing revealed a change in the gnostic processes of auditory analysis and synthesis, in assigning meaning to the auditory information regarding the analysis of the phonemic system of language, analysis and synthesis of the supra-segmental aspects of speech, integration of auditory information with other information sensory and sounds in sequence. Therapeutic intervention was performed through acoustically controlled auditory training, with weekly sessions in an acoustic booth. After the reevaluation, it was verified the reduction of latencies of waves III and V bilaterally. The evaluation of central auditory processing revealed improvement in all auditory skills. These improvements were generalized for academic and social performance.

Conclusion

The child has central auditory processing disorder, which was minimized after acoustically controlled auditory training, showing intervention effectiveness both from a behavioral and electrophysiological assessment point of view.

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