Imbalance in a Child with Gorham-Stout Disease: Would It Be Ataxia?

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A 6-year-old boy was transferred to the emergency department complaining of an imbalanced gait with a few days of evolution. He had been previously observed in another hospital. The symptoms were described as acute ataxia and an investigation excluded posterior fossa lesions. Concerning his medical history, this boy was diagnosed with Gorham-Stout disease at 3 years old and with focal epilepsy, and he was treated with oxcarbazepine and zolendronic acid. He had also been under treatment with sirolimus, but the drug was suspended about one year before the present episode.

Upon admission, the neurological examination did not confirm the existence of any ataxia. In fact, a right palpebral ptosis was identified as well as an anisocoria (right > left). The exam of ocular movements revealed a limitation on the adduction, supraversion, and infraversion of the right eye (Fig. 1) and the child was referred to a diplopia consistent with paresis of the right oculomotor nerve. A brain imaging study was performed, revealing inflammatory changes in the sphenoid bone and abnormal cavernous sinus on the right side (Fig. 2). Alternating occlusion of the eyes was recommended, and the gait pattern immediately improved.

Third cranial nerve palsy is uncommon in children and it is mostly congenital.[1] Clinical presentation varies depending upon the location and type of lesion.[2] Of particular interest is our patient diagnosis of Gorham-Stout disease affecting the bones at the base of the skull and the trajectory of the nerve. Gorham-Stout disease is characterized by extensive and progressive osteolysis and angiomatous proliferation with a benign origin, and mostly affects bones in the maxillofacial region and upper extremity.[3,4]

This clinical case shows a rare neurological involvement in a rare disease,[1,2] in which the physical examination was crucial for a correct diagnosis and treatment. It reinforces how important it is to correctly characterize clinical deficits, thereby making an adequate topographic diagnosis.

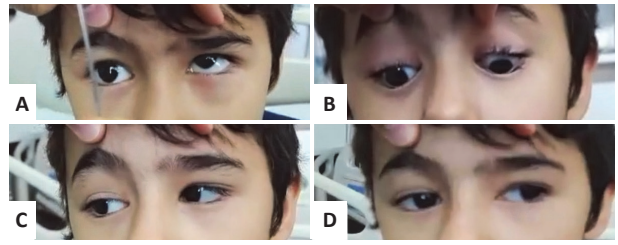

Figure 1. Examination of eye movements, revealing a paresis of the muscles innervated by the right oculomotor nerve: A. Paresis of the supraversion of the right eye (*superior rectus* muscle); B. Paresis of the infraversion of the right eye (*inferior rectus* muscle). C. Absence of any limitation in the abduction of the gaze, on the right (*lateral rectus* muscle, innervated by the abducens nerve). D. Limitation of right eye adduction (*medial rectus* muscle). These movements generated complaints of vertical (A, B) and horizontal (D) diplopia. No sensory changes have been identified in the right ophthalmic nerve territory and the child did not report a headache or facial pain.

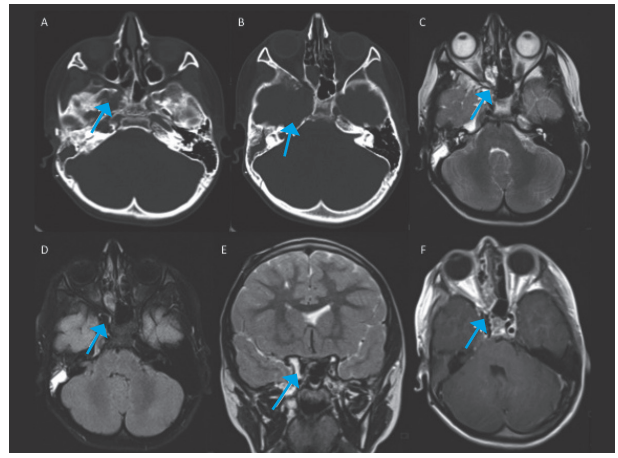

Figure 2. Computed tomography (A, B) and magnetic resonance imaging (C-F) scans. Destruction of bone trabeculae in the right mastoid, in a probable relationship with a local inflammatory/osteolytic process, destruction of the right greater sphenoid wing (A, arrow) and right petrous apex (B, arrow). Axial T2 (C) and fluid attenuated inversion recovery (FLAIR) (D) revealing the extension of the osteoclastic process to the right cavernous sinus, whose anatomy is altered (coronal T2) (E), having ceased to be a venous structure (axial T1 after gadolinium administration) (F). The arrows (C-F) point to the right cavernous sinus, the differences observed in comparison with the contralateral structure being particularly evident. There is no engorgement of the structures that drain the right cavernous sinus, namely the ophthalmic veins and the spheno-parietal sinus is only slightly enlarged. Being a slow process, compensation mechanisms have naturally been developed, which can pass through the left cavernous sinus, since they communicate with each other. Although not visible in the image, the right internal carotid artery is patent and has a normal path.

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WHAT THIS REPORT ADDS

- Gorham-Stout disease can produce osteolytic lesions in the skull and facial bones, leading to focal neurological signs.
- The correct clinical characterization of the neurological deficits is of paramount importance to make an adequate topographic diagnosis.
- Imaging techniques are important to confirm or refute the topographic diagnosis, also allowing you to assess the degree of disease activity.

Conflicts of Interest

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Confidentiality of data

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