Mosaic Neurofibromatosis Type 2 with Subdural Hematoma and Hydrocephalus: What Therapeutic Strategy?

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Abstract:

Neurofibromatosis type 2 (NF2) is a rare autosomal dominant genetic disease with a birth incidence of 1 in 25,000. Nearly ¾ of the NF2 gene mutations are sporadic, affecting the Merlin protein on chromosome 22q. Hence, the disease is a tumor suppressor gene on chromosome 22q, encoding Merlin protein.

We report the case of a 63-year-old man who was diagnosed with spastic tetraparesis over six months. The patient had anterior cervical meningioma, calcified right anterolateral meningioma of the foramen magnum, posterior meningioma, schwannoma of the posterior root of C7, and multiple schwannomas of the cauda equina. The diagnosis is based on the Manchester criteria (1992), which include bilateral vestibular schwannomas, first-degree relatives with NF2 and unilateral vs., first-degree relatives with NF2 OR unilateral VS AND two of the following: meningioma, cataract, glioma (intramedullary ependymoma), schwannoma, or multiple meningioma (2 or more). The patient underwent total surgical excision of the cervical meningioma, and regular MRI monitoring allowed for assessment of the lesions' evolution.

***Keywords**: neurofibromatosis type 2, vestibular schwannomas, meningioma*

Introduction :

Neurofibromatosis type 2 (NF2) is a rare autosomal dominant genetic disease with an incidence at birth of around 1 in 25,000. However, nearly ¾ of the mutations in the NF2 gene responsible for the disease are sporadic. . NF2 is a tumor suppressor gene located on chromosome 22q encoding Merlin protein

To make a diagnosis of NF2, a patient must meet one of the following criteria: 1. Bilateral vestibular schwannomas (VS) 2. A first-degree relative with NF2 AND unilateral VS. 3. First- degree relative with NF2 OR unilateral VS AND two of the following: meningioma, cataract, glioma (intramedullary ependymoma), schwannoma. 4. Multiple meningioma (2 or more) AND two of: unilateral VS, cataract, glioma (intramedullary ependymoma) schwannoma(1,3).

The severity score is genetic. Patients with a severe genetic score are affected at a younger age with a large number of tumors requiring a statistically higher number of interventions than patients with average or mild scores (severe score: complete truncation of the NF2 gene between lesions 2, 13, not severe: mosaic or splice-site mutation exon 8 to 15 or large deletions(2).

The prognosis is functional, particularly hearing, and in severe forms the prognosis may be serious. The improvement of surgical and radiotherapy techniques and the appearance of new drugs are revolutionizing the management of this disease.

Observation :

We report the case of a 63-year-old man, received for spastic tetraparesis of progressive onset over 06 months with headaches for 01 month, neck pain as well as bilateral. The patient had no particular pathological history, the clinical examination was unremarkable, the neurological examination revealed a vestibular syndrome: tinnitus, hypoacusis. The labyrinthine Romberg sign, the deviation of the index fingers, the lateropulsions when walking, occur on the injured side, without facial paralysis, a sublesional syndrome: spastic tetraparesis with sharp reflexes

with sensory disturbances: tingling and hypoesthesia of the 04 limbs.

Brain Magnetic Resonance Imaging : showed the tumoral iso signal in intermediate T1 in heterogeneous T2, the posterior of 24 by 22 mm, a thickening of the choroid plexus with moderate hydrocephalus.

The presence of a right hemispherical subdural collection of right vestibular of 10 mm, tissue filling of the middle cell, it is associated with a right lateral peribulbar formation the patient was operated on as a first "emergency" with a trephine hole for the subdural hematoma, then we continued the exploration by spinal MRI.

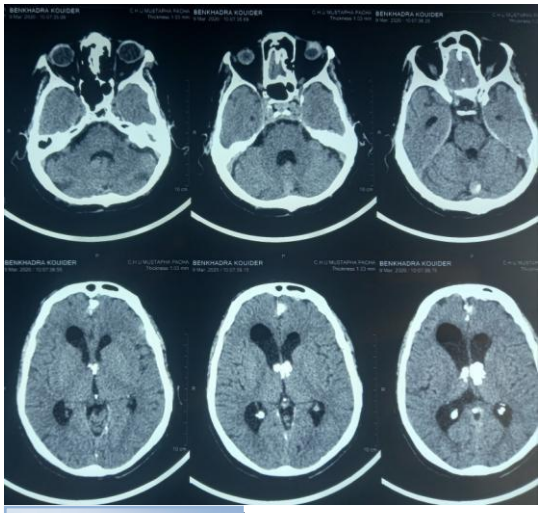

Figure 01

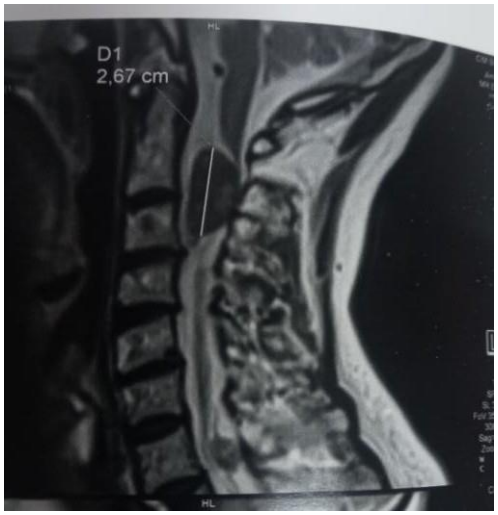

Figure 02

Figure 01: Control brain CT after the 1st intervention

Figure 02 : MRI of the bulbo-medullary junction in T2 sagittal cut

Objective spinal cord MRI: Anterolateral cervical meningioma facing C2 C3 of 27 by 15 mm repressing and compressing the spinal cord **(Figure 02)** , calcified right anterolateral meningioma of the foramen magnum measuring 10 mm, 02 minimal posterior meningioma facing C4 and D5, schwannoma of the posterior root of C7, with multiple schwannoma of the cauda equine **(Figures 03 and 04).**

For cervical location: The patient benefited from a total surgical excision of the cervical meningioma after a C2 C3 laminectomy, dural opening with the demonstration of a yellowish white meningioma which represses the spinal cord on the left, then we begin coagulation of the capsule tumor with protection of the marrow with cotton, dissection of the meningioma in the different surfaces with opening of the serrated ligament then at the end of the dissection

of the meningioma with the spinal cord while always respecting the cleavage plane, tumor excision after coagulation of the base of insertion, the post-operative consequences were simple and one month later the examination showed an improvement on the functional level **(Figures 05 and 06).**

For vestibular localization: the patient is thus referred to a radio surgery department for focused mono-fractionated irradiation

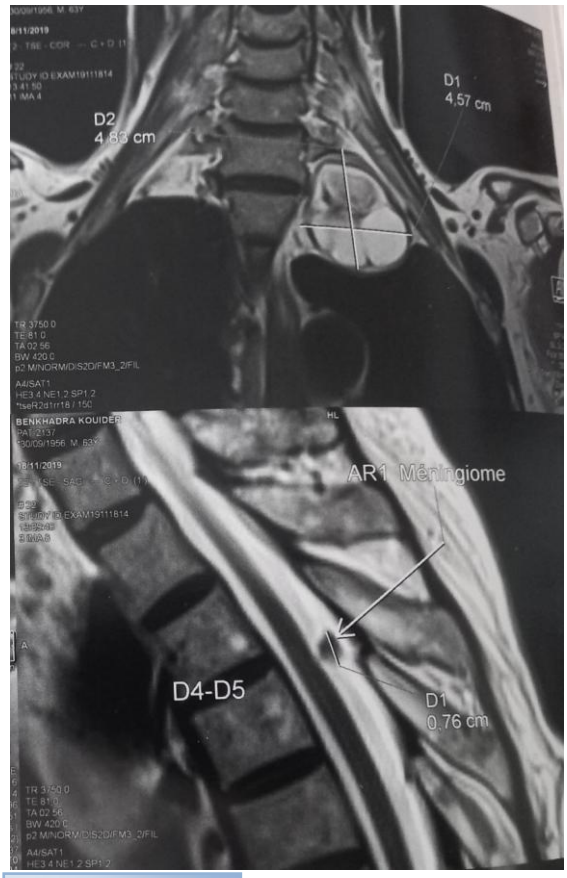

Figure 03

For other locations: Regular MRI monitoring makes it possible to assess the evolution of the lesions

- For extramedullary spinal tumors, surgery is the indicated treatment if symptoms of spinal cord compression appear
- For peripheral schwannomas, in the event of isolated swelling or moderate functional discomfort, simple clinical monitoring is recommended. The risk of malignant transformation of peripheral schwannomas is very low. In the event of significant functional discomfort linked to pain or sensory symptoms, surgical intervention is proposed.

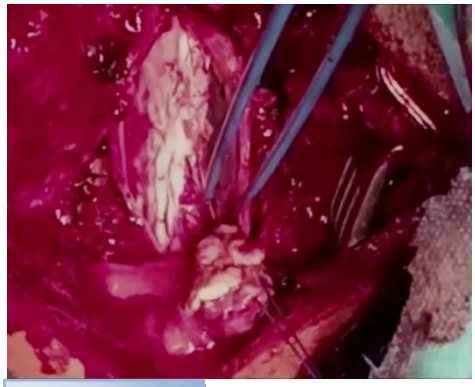

Figure 05

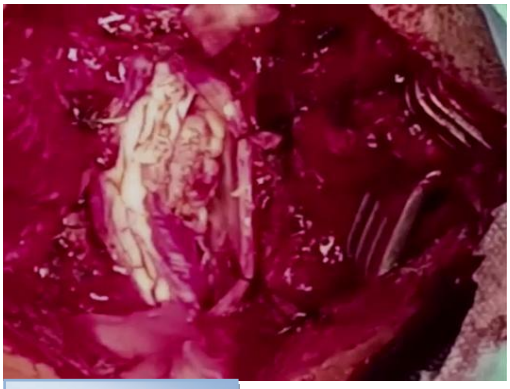

Figure 06

Figure 05: feeder vessel coagulation

Figure 06 : appearance of the marrow repressed by the meningioma

Discussion :

To date, there is no consensual and validated strategy to oppose the SV of NF2, the evolutionary and extension profile of which is extremely variable and the prevalence low(5). When there is no acute revelation of NF2 or deficiency, initial monitoring is most often decided to avoid any functional deficit not justified by the severity of the tumor progression. It also allows us to appreciate the natural history of each tumor and its clinical impact. The wishes of the patient and his family (children), fully informed of the prognosis of his disease according to the profile of extension and evolution observed and the experience of the certified medical-surgical team are essential to offer patients the solution.
The presence of psychologists in the multidisciplinary team managing NF2 is important, even essential, because it makes it possible to offer global listening to the patient and to support them throughout the stages that will mark his illness, as soon as the diagnosis is announced.

Genetic counseling is provided by a physician trained in genetic counseling or a genetic counselor. Genetic counseling can be repeated as many times as necessary. It is aimed at all patients who request it. The genetic counseling interview is particularly important and necessary:

Following the announcement of the diagnosis, whether it concerns children (genetic counseling for parents) or adults of childbearing age or who already have children(2).

As for follow-up, patients must have at least one annual consultation with a doctor coordinating care with, in principle, a brain MRI, and at least pure and vocal audiometry, if the patient is not deaf. Spinal MRI, if there were lesions initially, must be repeated every 3 years when there are no symptoms(7).

Conclusion :

The management of NF2 patients requires an experienced oto-neurosurgical team, within the framework of "NF2 clinics". Prolonged annual clinical and paraclinical monitoring is desirable. A family screening protocol is proposed. Classically, only symptomatic tumors are treated. Some suggest an early active approach to vestibular schwannomas in order to preserve hearing. When treatment is indicated, surgery is the standard treatment for tumors. The auditory brainstem implant has a special place in the hearing rehabilitation of NF2 patients.

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