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Type 2 Neurofibromatosis in a Patient Originating in a Twin Pregnancy - Case Report

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I. INTRODUCTION

Type 2 NF is a dominant autosomal hereditary disease triggered by a mutation which inactivates the tumor-suppressor gene on the 22p12 chromosome. Gene mutations may also occur randomly, in infrequent instances of the disease.

The disease's occurrence is 30-40 in 100.000, evenly split among genders and half of the patients have first degree relatives who also carry this disease.

The role of hormones and growth factors in the disease's pathogenesis is currently unknown.

The disease is characterized by the development of benign tumors, mostly in skin, eye, bone and nervous tissue. About 20% of the tumors can undergo a malign sarcomatous transformation. The neurofibromas in skin tissue grow during late childhood and teenage years. About a third of the patients exhibit apparent cutaneous indicators, while roughly two thirds of the patients require examinations to identify them. These skin neuromas are pressure-sensitive and painful to the touch. Neurological effects typically start around the age of thirty, by exhibiting symptoms triggered by the development of a uni- or bilateral neuroma on the acoustic-vestibular (VIII) nerve: progressive hear loss, vertigo, gait and standing trouble,. Other typical symptoms may include optic nerve gliomas, with blindness, optical atrophy and exophthalmia. The emergence of a neuroma may also affect the trigeminal nerve. In rare situations, the debut can be a spinal neuroma. The neurological symptomatology may evolve through the onset of multiple cranial or spinal meningioma which triggers epilepsy, radicular or medullar compression, mental retardation, cranial and peripheral nerve affections or other effects. The number and size of tumors increases with time. Seventy percent of meningiomas are small, located fronto- basally.

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Silvio's aqueduct stenosis may also occur, along with secondary internal hydrocephaly.

There are also abortive forms of the disease, which require the investigation of heredo-colateral history.

The Manchester Diagnosis Criteria for Type 2 NF:

1. Bilateral vestibular schwannoma, or first degree relative with Type 2 NF with unilateral vestibular schwannoma.
2. Two of the following: meningioma, schwannoma, glioma, neuromas, posterior subcapsular cataract.
3. One vestibular schwannoma and two of the following: meningioma, schwannoma, glioma, neuromas, posterior subcapsular cataract.
4. Two or more meningioma and one unilateral vestibular schwannoma or two of the following: meningioma, schwannoma, glioma, neuromas, posterior subcapsular cataract.

Acoustic-vestibular bilateral schwannoma is the most representative for the Type 2 NF.

Existing at birth, the disease may sometimes become apparent only at puberty, when it starts evolving very quickly.

The clinical diagnosis is confirmed via neuroimaging: MRI, CT-scan.

The neurosurgical treatment aims the excision of surgically-reachable tumors. The typical medication includes antiepileptic drugs, and radiation therapy is also an option.

It is important to obtain genetic advice, although males suffering from Type 2 NF usually have low fertility. This case study will showcase a 26 year old female patient diagnosed with type 2 NF, while her twin sister is healthy and does not present lesions under neuroimaging examination.

D.A., female, 26 years old, originating from twin birth, was diagnosed at birth with infant encephalopathy, and later diagnosed with slight mental retardation.

Her family history indicated that the father exhibited left ear deafness and several angiofibroma-type elements at chin level, although he has rejected any neuroimaging investigations. In October 2014, after an acute vestibular syndrome, he had a MRI evaluation and he was diagnosed with an infiltrativ glioma of the middle brain. Her father's brother also exhibited many skin neuromas, not investigated through neuroimaging techniques.

Starting from puberty, the patient exhibited neurofibroma-type cutaneous lesions located on the upper back, whose number and size have increased in time.

In 2006, the patient exhibited increasingly severe gait trouble which became paraplegic spasms,

neuro-clinical examinations suggesting potential medullar compression. Dorsal vertebo-medular MRI revealed a tumorous formation in the T9 medullar region, which was surgically removed following a psamomatose meningioma:



Figure 1 : Dorsal psamomatos meningioma-MRI

In 2008, the tumor re-emerged at the same level, which was again addressed through a new surgical procedure.

In 2009, the patient was diagnosed with a cranial-spinal meningioma which was partially excised.

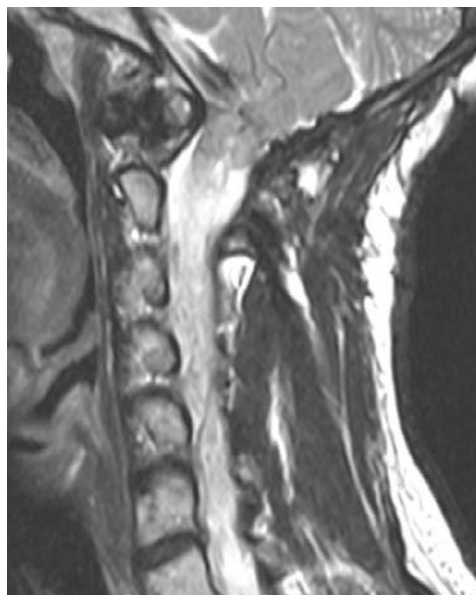


Figure 2 : Junctional meningioma

In March 2012, the patient exhibited loss of hearing with her right ear , diminished visual accuracy for her left eye and oculomotor nerve paresis. Her

cerebral MRI found a plated meningioma in the fronto-basal right region, multiple bilateral supratentorial meningioma smaller than 10 mm in diameter, a right

cerebellar meningioma and a ponto-cerebellar right angle expansion process, affecting the right internal

auditory conduit, probably a neuroma located on the acoustic-vestibular nerve.



Figure 3: Cerebellar tumor, right ponto-cerebellar angle tumor

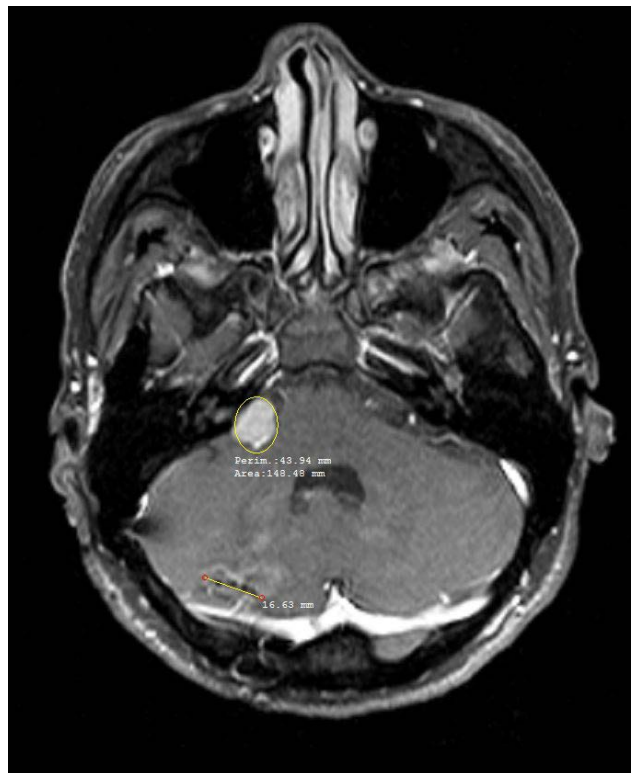


Figure 4: Right pontocerebellar angle tumor



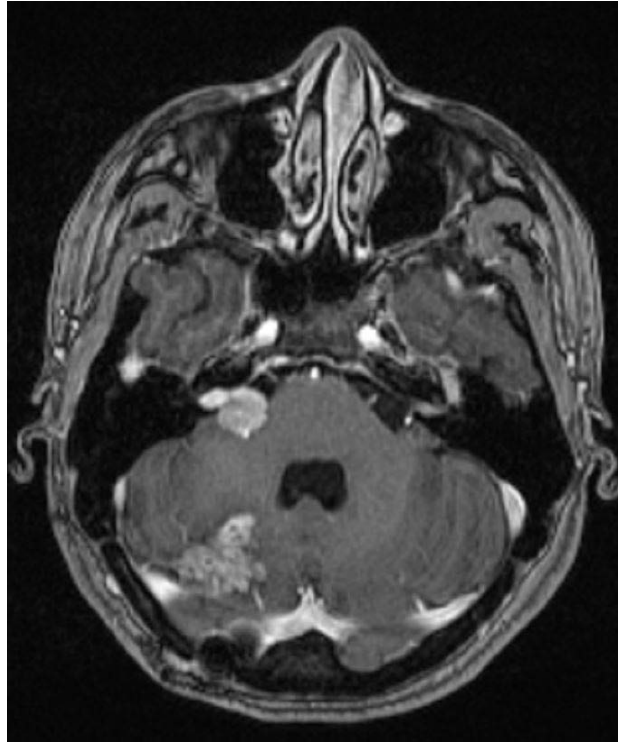


Figure 5 : Cerebellar tumor and right ponto-cerebellar angle tumor

In June 2012 a 50 Gy dose of radiation was used to address the cerebellar tumor process.

In October 2012, a brain and spinal neuroimaging reassessment revealed the relapse of the cranio-spinal meningioma which encased the vertebral artery at the right parasagittal bulbous-spinal junction up to the C1-C2 level, the growth of the cerebellar right angle expansion process, the emergence of an intensely gadolinium-absorbing in the Meckel cavum, probably a left trigeminal neuroma, growth of the front-basal meningioma. The cerebellar meningioma remained the same, multiple small bilateral supratentorial meningioma, with the emergence of six new lesions.

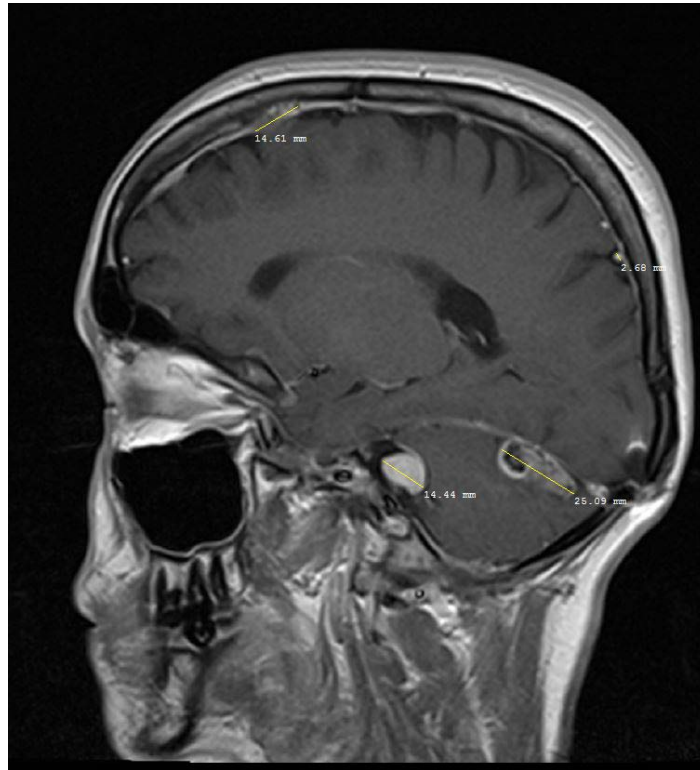


Figure 6 : Multiple small bilateral supratentorial meningioma

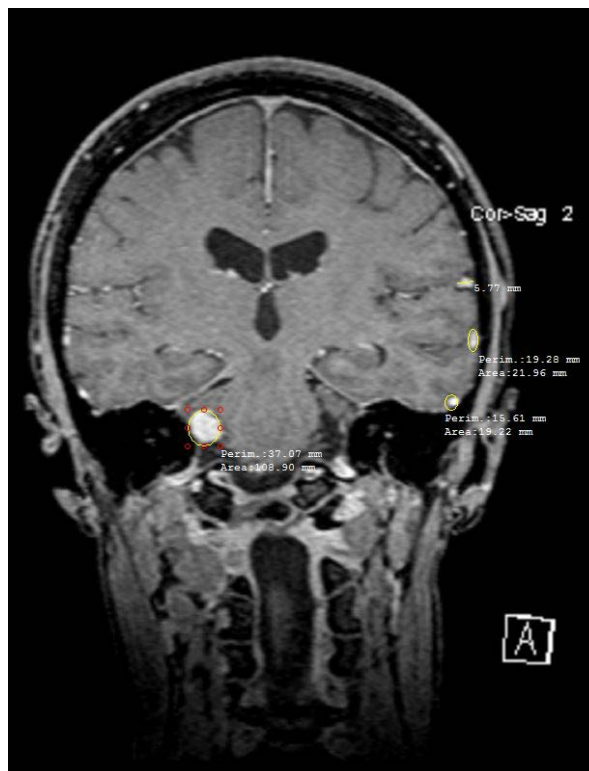


Figure 7 : Fronto-basal meningioma and multiple supratentorial meningioma





Figure 8 : Optic nerve glioma

Gamma Knife surgery was considered at that point, however the patient is currently treated solely on a symptomatic base only, and is periodically subjected to medical checks. She has no seizures, but there are important gait disturbances.

- Raymond D. Adams, Maurice Victor, Allan H. Ropper- Principles of Neurology, Sixth Edition, The McGraw- Hill Companies, USA, 1011-1014.

II. DISCUSSION

In conclusion, following the Manchester criteria the patient could be diagnosed with Type 2 NF, probably inherited from her father. The illness started at birth, as the patient was early diagnosed with infant encephalopathy). However, the illness manifested itself clinically during puberty, first being revealed as spinal meningioma, later the front-basal meningioma emerged and eventually the characteristic acoustic-vestibular nerve neuroma and the rest of the supratentorial meningioma showed up.

The difference between this particular case and other related ones is that the patient is one of two twins. While her twin sisters skull-spine neuroimaging examination confirmed a perfect health, the patient suffered from infant encephalopathy, mental retardation, and later exhibited symptoms corresponding to Type 2 NF, with multiple neurological symptoms determined by the intra-cranial and intra-spinal expansion processes. In this case, Type 2 NF, a dominant autosomal hereditary disease, has only affected one of the 2 twins (whether they are monozygotic or not is not clear).

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