JBR–BTR, 2010, 93: 144-145.

MASS IN THE BACK AS MANIFESTATION OF NEUROFIBROMATOSIS TYPE 2

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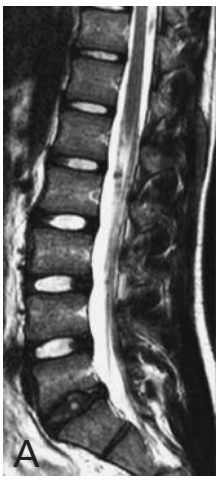
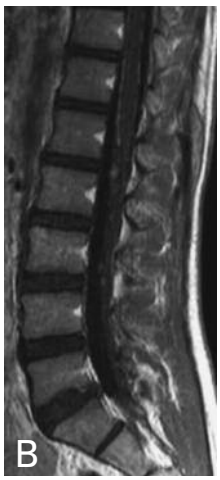
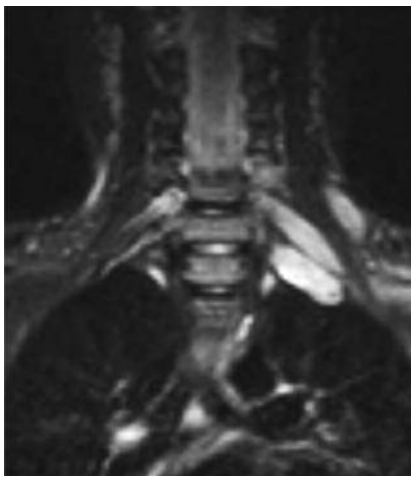
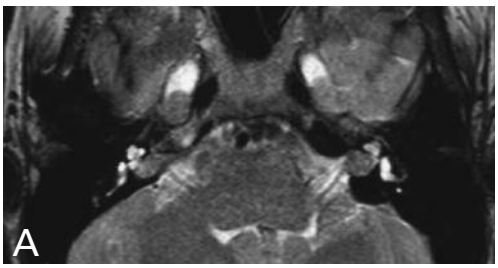
Key-word: Neurofibromatosis

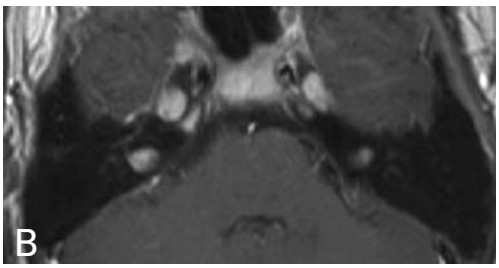
Background: A 13-year-old boy was referred for MR imaging of the thoracolumbar spine to further differentiate a spindle shaped hyporeflective soft tissue mass in the back seen on ultrasonography (not shown) most characteristic for a lipoma.
After a closer look at the medical history we learn that the boy is known since the age of 7 with a problem of paresis with atrophy of the left arm. An earlier MR examination of the thoracic outlet showed bilateral soft tissue masses interpreted as diffuse thickening of the plexus brachialis nerves, thus explaining the patient's complaints, without however a clear etiology being withheld. One year ago a new small mass was excised from the boy's lip. Anatomopathological analysis revealed an interesting diagnosis: submucosal neurofibroma. Now putting these at first apparently independent events in a greater context, we decided to also realize a MR examination of the brain.

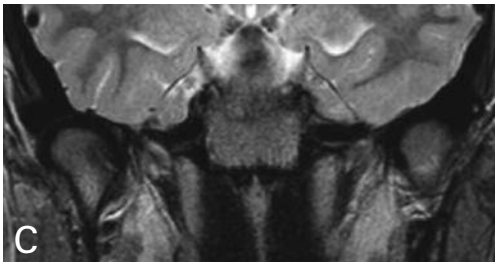












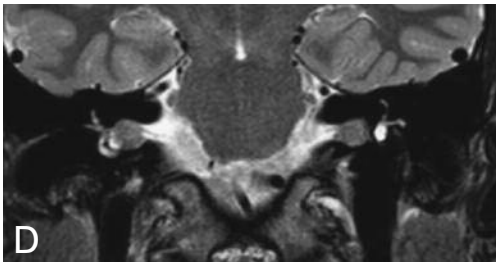


Fig.	1A	1B	2
	3A	3B	
	3C	3D	

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Work-up

MRI of the thoracolumbar spine (Fig. 1) includes a sagittal T2-weighted image (WI) (A) and enhanced T1-WI (B). At the level of the palpable swelling a spindle shaped hypointense, nonenhancing soft tissue mass is recognized. The sagittal T2-WI shows the presence of multiple nodular lesions which are wedged into the roots of the cauda equina (A). There is a clear contrast enhancement after intravenous Gadolinium injection (B).

MRI of the thoracic outlet (Fig. 2) shows hyperintense lesions on T2- and STIR-weighted images at the level of the apex of both lungs.

MRI of the brain (Fig. 3) consists of an axial and coronal T2-WI (A,C,D) and enhanced T1-WI (B) at the level of the internal auditory canal (IAC) and Meckel's cave.

There is a bilateral nodular contrast enhancement in the IAC after intravenous Gadolinium injection. Image of bilateral schwannomas originating from the nervus vestibulocochlearis. At the level of Meckel's cavum 3 enhancing nodules are seen as well as bilateral nervus trigeminus schwannomas.

Radiological diagnosis

Based on the pathognomonic findings on MRI of the brain and the clinical characteristics the patient was diagnosed with *neurofibromatosis type 2* (NF2).

Discussion

Neurofibromatosis type 2 (NF2) is in this case differentiated from neurofibromatosis type 1 (NF1) by the pathognomonic finding of bilateral acoustic neurofibromas on the MRI of the brain. The presence of cutaneous neurofibromas and peripheral nerve sheath tumors predominantly point in the direction of NF1, but the absence of café au lait patches and the visualization of neurofibromas/schwannomas on the roots of the cauda equina make NF2 a very likely diagnosis.

Neurofibromatosis (NF) is an autosomal dominant disorder that affects bone, nervous system, soft tissue, and skin. At least 8 different clinical phenotypes of neurofibromatosis have been identified. The most common types are NF1 (von Recklinghausen disease) and NF2. NF1 has a birth incidence of one in 2500. NF2, is a lot rarer with a birth incidence of about one in 33.000-40.000. In this case, familial anamnesis for the same clinical entity was negative so the diagnosis of a de novo mutation is plausible.

In literature 50% of the NF2 cases appear to be new germ line somatic mutations.

The National Institute of Health (NIH) Consensus Development Conference formulated following diagnostic criteria for NF2: (1) bilateral acoustic nerve (nVIII) masses seen with appropriate imaging techniques (e.g. CT or MRI) or (2) a first-degree relative with NF2 and either unilateral acoustic nerve mass, or two of the following disorders: neurofibroma, meningeoma, glioma, schwannoma, or juvenile posterior subcapsular lenticular opacity.

NF2 mainly presents in young adults and predominantly features symptoms associated with vestibular schwannomas as hearing loss and vertigo. Complaints due to cranial neuropathies, cataract, cutaneous lesions and peripheral muscle weakness can also exist. On the other hand, in children, the disease presentation

is somewhat different. In a large UK database Evans et al observed that in the pediatric onset cases of NF2 significantly less children had hearing loss and vertigo complaints and that relatively more children presented with cutaneous lesions and other neurological deficit.

To define the clinical spectrum of the disease, gadolinium-enhanced MRI of brain and spine with special attention for the cranial nerves is required. In addition, neurological, dermatological and ocular examinations are advised as well as a systematic follow-up in time. Finally, genetic staging of patient and family should be considered. NF2 is a severely debilitating disease, which is worse with childhood onset, and multidisciplinary management at specialty treatment centres is required.

Bibliography

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