Familial spinal neurofibromatosis

Familial spinal neurofibromatosis (FSNF) is a rare localized subtype of NF1 which shows neurological symptomatology during adult life.

In FSNF1 multiple, extensive, bilateral spinal root neurofibromas occur leading to signs and symptoms of progressive, segmentally distributed nerve sheath neuropathy in absence of spinal cord symptoms. Very few cases of Non-FSNF1 have been described in literature so far. Most of the published series so far comprised of segmental involvement of the spinal roots by the tumour affecting patients of various age groups [1) 2) 3)].

Pascual-Castroviejo et al. describe a family in which three members in two generations, mother, son and daughter, were affected. The patients, aged 48, 22 and 18 years, had spinal bilateral neurofibromas affecting all spinal roots. Spinal symptoms were not present in any of the patients. However, the son had generalized nerve sheath tumors that caused important signs of peripheral neuropathy. The daughter also had benign tumors that involved the left optic nerve and chiasm and the left cerebellar hemisphere. The spinal neurofibromas underwent an important growth in size between 20 and 22 years of age. A specific mutation G848R, 2542 G > C in NF1 exon 16 was present in all three patients [4)].

1)
Schorry EK, Cawford AH, Egelhoff JC, Lovell AM, Saal HM. Thoracic tumors in children with neurofibromatosis-1. Am J Med Genet. 1997;74:533–537. https://dx.doi.org/10.1002/(SICI)1096-8628(19970919)74:5<533::AID-AJMG16>3.0.CO;2-D. [PubMed]
2)
Leonard JR, Ferner RE, Thomas N, Gutmann DH. Cervical cord compression from plexiform neurofibromas in neurofibromatosis-1. J Neurol Neurosurg Psychiatry. 2007;78:1404–1406. https://dx.doi.org/10.1136/jnnp.2007.121509. [PMC free article] [PubMed]
3)
Tonsgard JH, Kwak SM, Short MP, Dachman AH. CT imaging in adults with neurofibromatosis-1. Frequent asymptomatic plexiform lesions. Neurology. 1998;50:1755–1760.
4)
Pascual-Castroviejo I, Pascual-Pascual SI, Velazquez-Fragua R, Botella P, Viaño J. Familial spinal neurofibromatosis. Neuropediatrics. 2007 Apr;38(2):105-8. PubMed PMID: 17712740.