ATLANTO AXIAL INSTABILITY DUE TO NEUROFIBROMATOSIS : CASE REPORT

L. M. VERAS, J. CASTELLANOS, G. RAMÍREZ, A. VALER, J. CASAMITJANA, F. GONZÁLEZ.

Neurofibromatosis is an autosomal dominant genetic disease, characterized by café au lait spots, neurofibromas and several bony anomalies. Deformities of the spine are the most frequent alterations. Involvement of the cervical spine has been studied less frequently.
The case of a 16-year-old male patient affected by neurofibromatosis, with cervical pain without neurological symptoms is presented. Xrays, CT-scan and MRI demonstrated the presence of cervical kyphosis, occipitoaxial instability and atlantoaxial instability with subluxation. Posterior occipito-C2 fusion was performed with prior placement of a halo-vest. The outcome at four years was good with solid occipito axial fusion, moderate loss of cervical spine flexion and moderate-to-severe limitation of cervical spine rotation. The incidence and variety of alterations of the cervical spine in patients affected with neurofibromatosis is discussed, as well as the results obtained by the treatment.

Key words : neurofibromatosis ; cervical spine instability ; occipito cervical arthrodesis.
Mots-clés : neurofibromatose ; instabilité du rachis cervical ; arthrodèse occipito-cervicale.

CASE REPORT

A 16-year-old male patient with neurofibromatosis diagnosed in infancy was referred to our hospital complaining of cervical pain of several months' duration without any neurological symptoms. Physical examination revealed multiple café

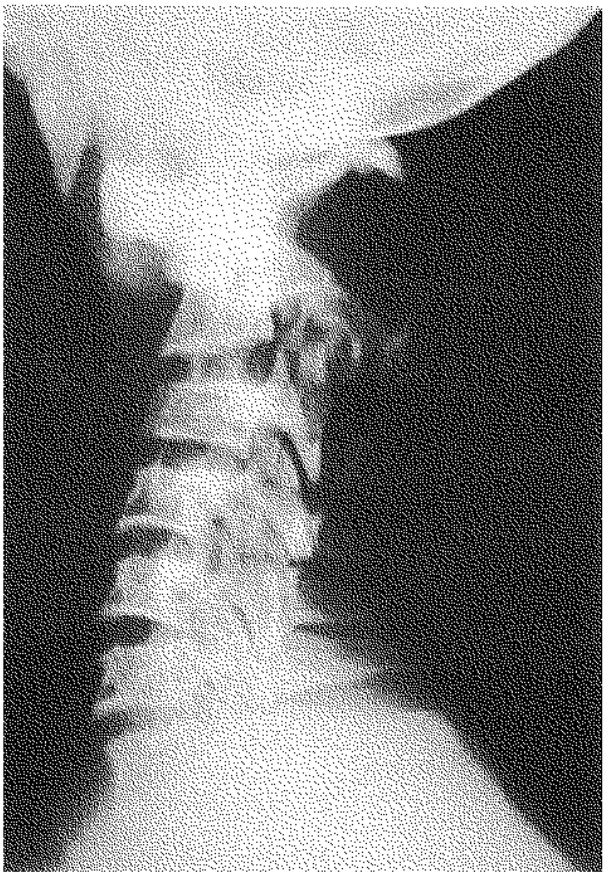

Fig. 1. — Lateral xrays of patient one week before fusion, showing cervical kyphosis and atlanto axial subluxation.

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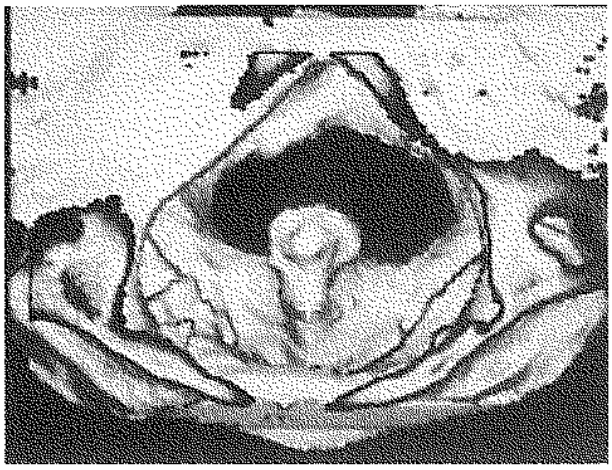

Fig. 2. — Three-dimensional CT-scan reconstruction that shows the increased atlanto axial distance (10 mm), facet subluxation and morphologic alteration of the arch of the atlas.

au lait spots and subcutaneous neurofibromas. Xrays showed cervical kyphosis and an increased distance between the anterior arch of the atlas and the odontoid process (fig. 1). The lateral dynamic xrays showed atlantooccipital and atlantoaxial instability, with mild alteration of the relationship of the atlas to the axis. CT-scan confirmed an increased distance (10 mm) between the anterior arch of the atlas and the dens (fig. 2). Furthermore it showed a facet joint subluxation of C1-C2. MRI demonstrated the atlantoaxial subluxation with a decreased spinal canal diameter at this level and slight narrowing of the cord (fig. 3).

Surgical treatment consisting in occipito-C2 arthrodesis was proposed, and a halo-vest was placed a few days prior to operation. The patient lay prone on the operating table with the head supported in the halo-vest. Posterior fusion was performed using a corticocancellous autograft obtained from the posterior iliac crest and stabilized with a titanium wire loop from the occiput to the posterior arch of C2. At 11 weeks CT-scan showed fusion of the arthrodesis, and the halo-vest was removed. The patient wore a cervical Philadelphia collar for 5 months. One year after surgery, CT-scan showed complete fusion of the graft with large bony bridges to the occipital bone as well as to the posterior arch of C2. At four year follow-up, the patient was painfree with moderate limitation of

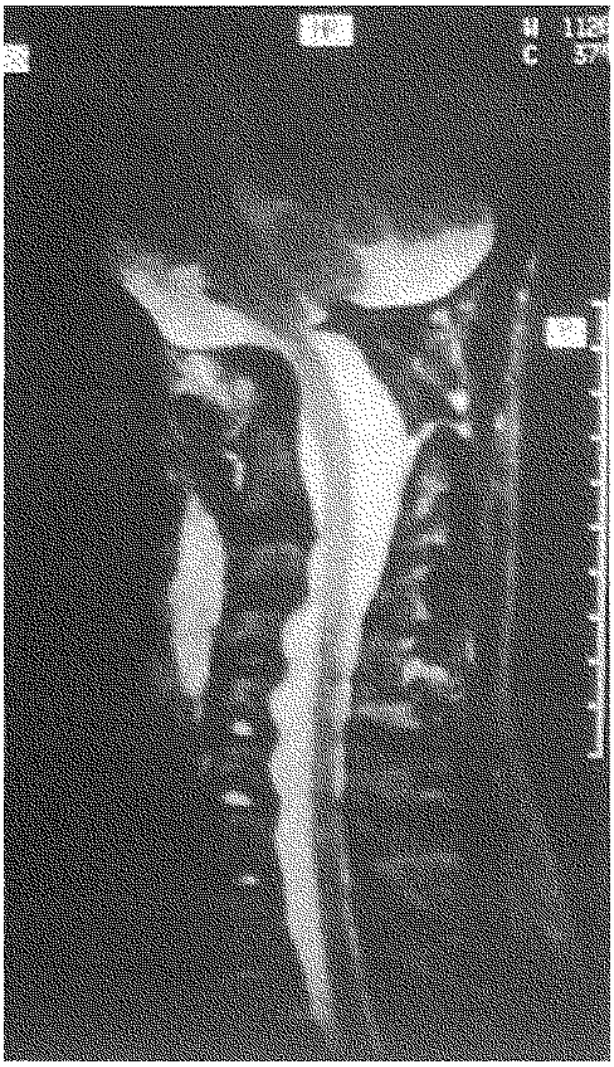

Fig. 3. — Cervical MRI. Note the dural ectasia and the narrowing of the cord without alteration of the MRI signal.

cervical spine flexion and moderate-to-severe restriction of cervical spine rotation. Xrays showed a solid occipitocervical fusion (fig. 4).

DISCUSSION

Frederich von Recklinghausen described neurofibromatosis for the first time in 1882. It is an autosomal dominant inherited disease that affects the connective tissue derived from the three embryonic layers; hence it is multisystemic (6). There are two varieties: type I, the most frequent, affects 1 in

5000 individuals, and it is the result of an abnormality in the long arm of the 17th chromosome. The criteria for diagnosis are two or more of the items shown in table I. Type II is not associated with skeletal alterations.

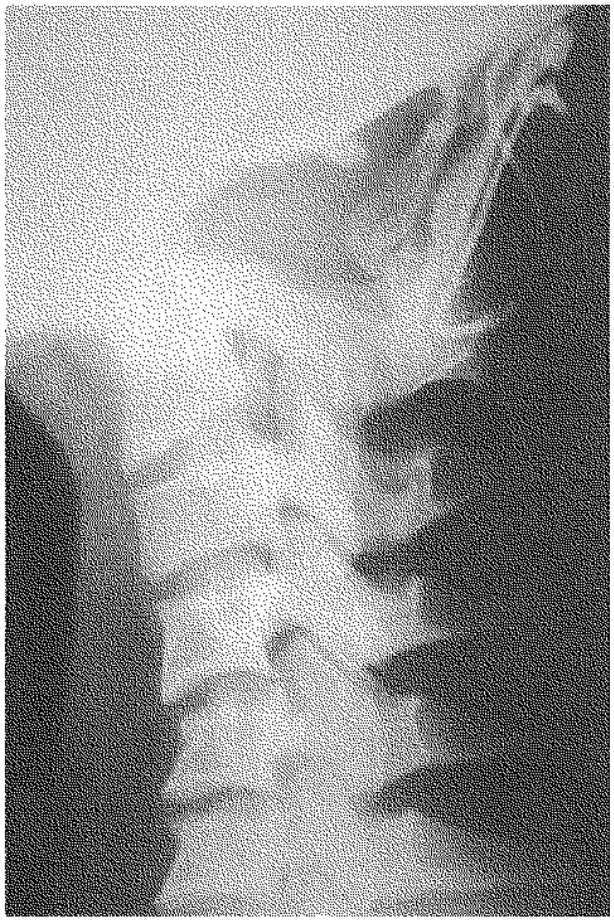

Fig. 4. — Lateral xrays 4 years after surgery showing a solid posterior arthrodesis.

Spinal deformities are the most frequent skeletal alterations in neurofibromatosis. Their incidence is difficult to determine because of the high number of undiagnosed patients with minimal symptoms, varying according to the series between 10 and 60% (6).

Cervical alterations have been described less frequently in the literature (1, 3, 9). Klose reported the first case of cervical alteration by neurofibromatosis in 1926 (9). In a review of 34 patients who had scoliosis or kyphoscoliosis secondary to neurofibromatosis, Yong-Hing et al. (9) found 15 (44%) patients affected with cervical lesions, many of them asymptomatic. Neck pain is the most common presenting complaint in neurofibromatosis of the cervical spine, but neurologic alterations, including complete or incomplete cord lesions and/or nerve root deficit may occur (1, 6, 9). However most of the patients are asymptomatic (3, 9). This is probably because the relationship of the medullary cord to the spinal canal at this level is less critical.

Table I. — Criteria for diagnosis of type I neurofibromatosis. National Institutes of Health Consensus Development Conference.

1	Six or more café au lait spots with diameter >5mm. in children and >15 mm. in adolescents and adults.
2	At least 2 fibromas or any type of plexiform neurofibroma.
3	Freckling in the axillary or inguinal regions.
4	Two or more iris harmartomas (Lisch nodules).
5	A distinct osseous lesion such as cortical thinning with or without pseudarthrosis.
6	A first-degree relative with neurofibromatosis type 1.

Atlantoaxial instability has been associated with a number of conditions including Down's syndrome, congenital scoliosis, osteogenesis imperfecta, Marfan's syndrome, Morquio's syndrome, Larsen's syndrome, short or webbed neck, Sprengel's deformity, congenital heart defects and neurofibromatosis (8). Atlanto axial stability depends on the transverse and alar ligaments and less so on the apical ligaments. An atlantoaxial distance measured on dynamic lateral xrays greater than 3.5 mm is considered abnormal in an adult, and subluxation of more than 10 to 12 mm implies destruction of the entire atlantoaxial ligamentous complex. Anterior atlanto axial subluxation may result in compression of the cervical cord between the posterior arch of the atlas and the dens. During flexion of the neck the spinal cord is particularly vulnerable to compression as the atlas slides forward in relation to the axis.

Atlantoaxial dislocation in neurofibromatosis has been previously reported in three patients by Isu *et al.* in 1983 (5). In 1992, Craig (1) reported

another case who was treated successfully by skull traction and posterior atlantoaxial arthrodesis without complications. Yong-Hing *et al.* (9) reported a case of rotatory atlantoaxial subluxation in 1979.

Treatment of atlantoaxial instability is dependent on the etiology and extent of the instability as well as the patient's activity level (2). Fielding et al. (2) have stated that even when there is minimal instability, trivial trauma superimposed on an already weakened and compromised structure can be catastrophic. They also suggest that indications for spinal fusion are instability with persistent cervical symptoms, evidence of cord or brain involvement or an atlantoaxial distance greater than 5 mm. Reduction of the deformity is associated with a risk for cord damage. We attempted to reduce the subluxation in our case with traction, but did not obtain an anatomical reduction. Recently, a case of a traction-induced rupture of an extracranial vertebral artery aneurysm associated with neurofibromatosis has been reported as a rare and not previously reported complication (4).

The management of these patients is determined to prevent progression of deformities and neurological worsening. It is difficult because of the paucity of symptoms and nonspecific nature in the early stage of the disease. Although one would expect that occipitocervical arthrodesis leads to severe cervical spine limitation of motion, we believe that the loss of atlantooccipital motion is compensated by the increased mobility at the cervical segments caudal to the mass of fusion. Also the lack of atlantoaxial rotation in our patient was very well tolerated. Many techniques of occipitocervical fusion have been described including the use of various devices for internal fixation and anterior approaches, but all of them may be associated with serious complications. In our patient, we obtained fusion without complications; this was also the case for the 13 patients reported by Wertheim and Bohlman (7) using this operative technique. We think that instrumented fusions are risky owing to the inherent malformations associated with neurofibromatosis. Furthermore it is advisable to perform xrays of the cervical spine in all patients affected with neurofibromatosis who undergo general anesthesia for any reason or cranial traction for treatment of curvature of the spine (1, 3, 9). MRI assessment is recommended to evaluate unrecognized intraspinal lesions.

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SAMENVATTING

L. M. Veras, J. Castellanos, G. Ramírez, A. Valer, J. Casamitjana, F. González. *Atlanto-axiale instabiliteit tengevolge van neurofibromatose. Een geval.*

Neurofibromatose is een autosomaal dominant erfelijke ziekte, gekenmerkt door café-au-lait-vlekken, neurofibromata en allerhande botafwijkingen. Misvormingen van de wervelkolom werd niet vaak bestudeerd. De auteurs bespreken het geval van een 16 jarige jongen met cervicale pijn zonder neurologische afwijkingen. Röntgenfoto's, CT-scan en kernspintomogrammen wezen op cervicale kyphosevorming, occipito-axiale instabiliteit, en atlanto-axiale instabiliteit met sublu-

xatie. Een halo-vest werd aangebracht, waarna een dorsale arthrodesis, matig verlies van cervicale flexie, en matige tot uitgesproken beperking van de cervicale rotatie. De auteurs bespreken de incidentie en de verscheidenheid van halswervelkolom-afwijkingen bij neurofibromatosis, alsook de resultaten bekomen bij behandeling.

RÉSUMÉ

L. M. VERAS, J. CASTELLANOS, G. RAMÍREZ, A. VALER, J. CASAMITJANA, F. GONZÁLEZ. *Instabilité atlanto-axiale secondairea à la neurofibromatose: Présentation d'un cas.*

La neurofibromatose est une maladie génétique dominante autosomique, caractérisée par des taches café-au-lait, des neurofibromes et diverses atteintes osseuses. Les déformations du rachis sont fréquentes. L'atteinte du rachis cervical a été étudiée moins fréquemment. Le cas d'un patient de 16 ans présentant une neurofibromatose, avec douleur cervicale sans symptômes neurologiques est présenté. Les radiographies, le CT-scan et la RMN ont montré la présence d'une cyphose cervicale, d'une instabilité occipito-axiale et d'une instabilité atlanto-axiale avec subluxation. Une arthrodèse postérieure occipito-C2 a été réalisée après mise en place d'une veste halo. Les résultats à quatre ans ont été bons avec fusion occipito-axiale, perte modérée de flexion du rachis cervical et réduction modérée à importante de la mobilité en rotation du rachis cervical. L'incidence et les types d'atteintes du rachis cervical dans la neurofibromatose sont discutés, ainsi que le résultat obtenu par le traitement.