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Original article

Six cases of sporadic schwannomatosis: Topographic distribution and outcomes of peripheral nerve tumors

Six cas de schwannomatose sporadique : répartition topographique et devenir des tumeurs nerveuses périphériques

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A B S T R A C T

The diagnosis of schwannomatosis is often overestimated and is based on the existence of multiple peripheral nerve tumors composed exclusively of schwannomas, in the absence of clinical signs of neurofibromatosis type 2 (NF2). Sporadic forms are much more frequent than familial forms. The objective of this study was to describe the distribution of peripheral nerve tumors and investigate the outcomes of schwannomas in the context of sporadic schwannomatosis. We conducted a retrospective study of patients who fulfilled clinical diagnostic criteria for sporadic schwannomatosis. Six patients were reviewed with a mean follow-up of 38.5 months (27–60 months). Patients' demographic, clinical, radiographic, and pathologic data were extracted. All patients underwent slit-lamp examination, enhanced brain magnetic resonance imaging (MRI) and a spinal MRI. Enucleation that preserved nerve continuity was performed in symptomatic patients. On average, patients were 36 years of age at the time of diagnosis with no sex predominance. The topographic distribution of the peripheral nerve tumors was always unilateral and most frequently targeted the upper limb. In four cases, the tumors involved the same peripheral nerve exclusively. The average number of nerve tumors observed per patient was 4.7 (2–8). The outcome after enucleation was marked by the systematic appearance of new tumors. After enucleation, no recurrence or malignant transformation was observed at the final follow-up. There was no transition to a NF2 configuration. The absence of neurofibroma and NF2 criteria makes schwannomatosis a diagnosis of exclusion. While a good prognosis can be expected following enucleation, two risks related to neurofibromatosis type 3 (NF3) are worth monitoring: the transition to NF2, particularly in young patients, and the appearance of new tumors.

Mots clés :
Schwannomes multiples
Schwannomatose
IRM

R É S U M É

Le diagnostic de schwannomatose est souvent porté par excès. Il est basé sur l'existence de tumeurs nerveuses périphériques multiples composées exclusivement de schwannomes, sans signe clinique de neurofibromatose neuro-acoustique (NF2). Les formes sporadiques sont beaucoup plus fréquentes que les formes familiales. Le but de cette étude était d'analyser la localisation des tumeurs nerveuses périphériques et leur devenir dans le cadre des schwannomatoses sporadiques. Six patients répondant aux critères diagnostiques de schwannomatose sporadique ont été rétrospectivement revus avec un recul moyen de 38,5 mois (27–60 mois). L'âge, le sexe, l'examen clinique, les examens complémentaires et l'histologie ont été analysés. Tous les patients ont eu un examen à la lampe à fente et une imagerie par résonance magnétique (IRM) des méats acoustiques internes et de la moelle spinale. En cas de tumeur nerveuse symptomatique, une énucléation a été réalisée, préservant la continuité nerveuse. L'âge moyen des patients était de 36 ans au moment du diagnostic sans prédominance de sexe. La distribution des

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tumeurs nerveuses périphériques était toujours unilatérale et intéressait le plus souvent le membre supérieur. Dans 4 cas, les tumeurs intéressaient un même nerf. Le nombre moyen de tumeur par patient était de 4,7 (2–8). Après énucléation, de nouvelles tumeurs sont apparues systématiquement : aucune récidive ou transformation maligne n'a été observée au dernier recul, ni forme de passage vers une neurofibromatose de type 2. L'absence de neurofibrome ou de critères de NF2 font de la schwannomatose un diagnostic d'élimination. Le pronostic est favorable après énucléation : les deux risques qui nécessitent une surveillance sont la possible transformation en NF2 et l'apparition de nouvelles tumeurs.

1. Introduction

Three major forms of neurofibromatosis (NF) are recognized as distinct entities based on their genetic origin and pathogenesis: type 1 (NF1), type 2 (NF2), and schwannomatosis or NF3. The type of NF is determined by a questionnaire, clinical examination and several systematic investigations, including cranial magnetic resonance imaging (MRI) (to detect possible vestibular schwannoma), MRI of the spine (to identify possible ependymomas, astrocytomas or meningiomas) and slit-lamp examination (to detect Lisch nodules pathognomonic of NF1 [1] or posterior cataract, which is present in 80% of NF2 cases [2]). The diagnosis criteria of schwannomatosis have evolved since it was described by Jacoby et al. [3]. Schwannomatosis can be distinguished from NF2 in most patients, but some patients meet the diagnostic criteria for both. Diagnostic criteria for "definite" schwannomatosis have been proposed in a consensus statement on schwannomatosis [4] and were revised in 2006 [5]. There are two types of schwannomatosis: sporadic and familial [6]. The majority (85%) of schwannomatosis cases are sporadic [6–8].

This study aimed to describe the distribution of peripheral nerve tumors and investigate outcomes of schwannomas in the context of sporadic schwannomatosis.

2. Material and method

We conducted a retrospective review (1995 to 2015) of patients who fulfilled clinical diagnostic criteria for sporadic schwannomatosis (Table 1). Six patients were identified who met the clinical criteria for schwannomatosis and reviewed after a mean follow-up of 38.5 ± 10.9 months (range 27–60 months).

These six patients were part of a cohort of 99 patients treated for a single peripheral nerve tumor (82 cases with 49 solitary schwannomas) or NF (17 cases, with six NF1 and five NF2) at the Claude-Bernard University in Paris and the Latour Hospital in Geneva [9,10]. Patients' demographic, clinical, radiographic, and pathologic data were extracted with attention to age at onset, gender, number and location of tumors, tumor on one side or bilateral locations, spinal location, ophthalmologic evaluation, family history, and other hallmarks of NF1 or NF2. Investigations for peripheral nerve tumors consisted of ultrasonography (US), magnetic resonance imaging (MRI) (T1, T2, short-TI inversion recovery [STIR] and gadolinium-enhanced sequences) and/or computerized tomography (CT) scans. All patients had a slit-lamp examination, which was negative for Lisch nodules (pathognomonic of NF1) in every patient, an enhanced brain MRI, which revealed no evidence of vestibular tumor (NF2) in any patient, and a spinal MRI, which did not detect any ependymoma, astrocytoma or meningioma. The schwannomas were treated in the same manner as an isolated lesion. Enucleation that preserved nerve continuity was performed in case of nerve compression symptoms (pain, paresthesia, neurological deficit). Outcomes of multiple schwannomas were studied (i.e., recurrence, new tumors, malignant transformation, appearance of a vestibular schwannoma).

Institutional review board approval was obtained for this study. Written informed consent and permission to use medical records, investigations and photographs in the study was obtained from participants when they presented for surgery (ethics statement).

3. Results

Of the 61 patients with schwannomas (49 single schwannomas, one case of NF1, five cases of NF2, six cases of NF3), 6 presented with schwannomatosis. The diagnosis of schwannomatosis was based on the existence of peripheral nerve tumors composed exclusively of schwannomas.

3.1. Patient demographics, presentation and physical examination

3.1.1. Patient age at diagnosis and gender

On average, patients were 36 ± 4.4 years at the time of diagnosis (range 31–45 years). Of the six patients who met the clinical criteria for schwannomatosis, three were male and three were female.

3.1.2. Presentation and physical examination

The time between the onset of symptoms and diagnosis of schwannomatosis varied between 2 months and 6 years, with an average of 29.8 ± 19.4 months. In three cases, the disease appeared with one or several painless lumps whose volume increased; in one case, the disease was discovered intraoperatively (patient 1). In two cases, the appearance of other tumors after enucleation of a schwannoma led to the diagnosis. None of the six patients had a positive first-degree relative with diagnosis of schwannomatosis or NF2. There were no clinical findings relating to Von Recklinghausen disease or NF2 in the patients.

3.2. Peripheral nerve tumors

The topographic distribution of peripheral nerve tumors was always unilateral and most frequently targeted the upper limb (four cases of segmental schwannomatosis). The tumors involved the same peripheral nerve exclusively in four cases (with trunk and branches of the ulnar nerve in three cases) (Table 2, Figs. 1–4). The average number of nerve tumors (confirmed by histology) per patient was 4.7 (2–8). From a total of 28 exclusive schwannomas, 19 were discovered because of a lump with a positive Tinel sign (superficial tumors). In four cases, nerve tumors developed as single tumors in nerves or branches and in two cases, as multiple lesions close to each other that made the nerve appear as a rosary (patients 1 and 5). MRI allowed confirmation of the diagnosis of a deep tumor on a nerve. The clinical signs linked to the nerve tumors were identical to those linked to isolated tumors. No neurological deficit was observed. No spinal tumors were found.

On average, 2.8 (1–8) schwannomas were enucleated surgically. The average size of schwannomas was 2.5 cm in diameter (0.4–9 cm). After surgery, neurological deficit was frequent but, in general, transient and clinical symptoms disappeared quickly in the month following surgery.