CASE REPORT

A rare occurrence and management of familial schwannomatosis

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drreddybanda@rediffmail.com**SUMMARY**

We are presenting a familial schwannomatosis without the features of neurofibromatosis (NF). We retrospectively reviewed the hospital charts, radiology films, operative notes and pathology slides of the patient. There was a family history of schwannomatosis. The patient had contrast-enhanced MRI, which was negative for vestibular schwannomas. The patient underwent surgical excision of symptomatic lesions. Histopathology confirmed these lesions as schwannomas consisting of areas of Antoni A and B, and immunohistochemical study was positive for S-100 protein. We recommend surgery for symptomatic lesions. Asymptomatic tumours can be monitored. Regular follow-up is essential as they may develop fresh lesions at any time. The relevant literature is discussed.

BACKGROUND

Schwannomas are benign, slow growing, encapsulated tumour of peripheral nerve; they are very homogenous tumours and consist of only Schwann cells. They develop on the outside of the nerve, but may push it aside or against adjacent structures causing damage.¹ Most schwannomas occur as solitary lesions. However, schwannomatosis is uncommon. Patients with schwannomatosis may develop multiple schwannomas on cranial, spinal and peripheral nerves, but they do not develop vestibular schwannomas (VS). They do not develop other tumours such as meningiomas, ependymomas or astrocytomas.² Patients with schwannomatosis usually present with pain,³ whereas NF-2 patients present with neurological deficits.² Molecular and genetic analysis also suggests that schwannomatosis is a distinct genetic and clinical syndrome.⁴ We present a rare case of familial schwannomatosis with a review of literature.

CASE PRESENTATION

A 19-year-old boy presented to surgery opd with complaints of swellings over the back, chin and posterior aspect of right knee region of 9 years, 5 years and 9 months duration, respectively, and abdominal pain of 15 days duration. Five years back, the patient was operated for a swelling in the back, which was associated with paraesthesia in both lower limbs; 1.5 years later, he developed pain and paraesthesia in the left lower limb for which he was operated and was diagnosed to be having L2 schwannoma. His father had undergone surgery for spinal tumour in the cervical region twice 25 years and 6 months back, both of which have been histopathologically proven to be schwannoma. On

physical examination, three tender swellings palpated over chin, left infra scapular area and right popliteal fossa ranging from 2×2 to 5×5 cm with smooth surface, well-defined borders, not adherent to skin or fascia (figures 1 and 2). On palpating swelling in popliteal fossa, the patient complains of severe pain radiating along the distribution of posterior tibial nerve; on palpation of the chin and infra scapular swelling, the patient developed radiating pain along the mental nerve and the intercostal nerve, respectively. There was no palpable mass in the abdomen; digital rectal examination revealed a firm tender mass in the left lateral wall, with smooth surface, well-defined borders and free from rectal mucosa, and was associated with pain radiating to left lower limb on palpation. Cutaneous examination did not show any café au lait spots. Ophthalmological examination did not show any lenticular opacity. Radiological investigation failed to show any intracranial tumour. Laparotomy was done and the swelling in the retroperitoneal region of the pelvis enucleated; other swellings were enucleated in the same sitting (figure 3). Histologically, all the tissues obtained at biopsy revealed schwannomas consisting of areas of Antoni A and B (figure 4). Immunohistochemical study revealed that most cells in both the Antoni A and B areas reacted strongly for S-100 protein. Postoperatively, the patient developed mild tingling sensation in the right leg and mild abdominal discomfort which improved gradually.

INVESTIGATIONS

Radiological investigations—MRI.

DIFFERENTIAL DIAGNOSIS

Neurofibromatosis.

Figure 1 Swellings over back.

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Figure 2 Swellings over popliteal fossa.

TREATMENT

Symptomatic schwannomas are removed surgically. Asymptomatic tumours can be monitored conservatively with serial MRI studies, usually at yearly intervals.

OUTCOME AND FOLLOW-UP

Schwannomatosis is a disorder distinct from neurofibromatosis. Most symptomatic schwannomas can be removed surgically. Asymptomatic tumours can be monitored conservatively with serial MRI studies, usually at a yearly interval. It is important to consider genetic testing. Molecular diagnosis rules out NF-2 and appropriate genetic counselling can be offered. Patients are advised to monitor themselves for any new neurological problems. Regular and long-term follow-up is essential as they may develop fresh lesions at any time.

DISCUSSION

Multiple cutaneous schwannomas were first described by Shisiba in 1984, who coined the term neurilemmomatosis for this disease as a distinct clinic-pathological entity.⁵ Several articles subsequently reported examples of multiple cutaneous schwannomas.^{6–7} Many of these earlier reports included patients with acoustic neuromas in addition to cutaneous schwannomas.^{5–7} Michael *et al*² have described that all patients with definite or possible schwannomatosis must not fulfil any of the existing sets of diagnostic criteria for NF-2, have no evidence of VS on high-quality MRI and no first-degree relative with NF-2; definite schwannomatosis should have either age above 30 years

Figure 4 Histopathology slide confirming diagnosis.

and two or more non-intradermal schwannomas, at least one with histological confirmation or one pathologically confirmed schwannoma plus a first-degree relative who meets the above criteria.

In our case, the patient's father has two histologically confirmed non-intradermal schwannoma and the patient has multiple spinal and peripheral schwannomas with histological confirmation and without any intracranial tumour on radiological investigation; hence, we believe that this is a rare case of familial schwannomatosis. Schwannomatosis is by itself a rare disease; it can be either familial or sporadic—the sporadic is as common as NF-2, but the familial schwannomatosis is a very rare condition.⁵ The surgical treatment of schwannomas should be reserved for only symptomatic tumours.⁶ In our case, the schwannomas at all the locations were symptomatic, and hence they were excised.

Learning points

- The presence of non-intradermal schwannomas in a patient with family history of schwannomas; we should think of rare condition called familial schwannomatosis and rule out NF-2.
- MRI should be done in all such cases to rule out intracranial tumours.
- Symptomatic tumours should be excised.

Contributors All the authors have made significant contribution for the publication.

Competing interests None.

Patient consent Obtained.

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Figure 3 Excised specimen.

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