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Testicular teratoma and peripheral neurofibromatosis

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Summary: Peripheral neurofibromatosis (Von Recklinghausen's disease) has been previously reported in association with a number of tumours and recently with aqueductal stenosis. We report a case which had both aqueductal stenosis and a testicular teratoma, which has not previously been reported in this condition.

Introduction

Peripheral neurofibromatosis (Von Recklinghausen's disease) is an autosomal dominant condition that occurs with a frequency of 1:3000 live births,[1] and is characterized by the presence of cutaneous neurofibromas, café au lait spots of the skin and pigmented iris hamartomas (Lisch nodules).[2] It has been associated with a number of malignant neoplasms, both of neural and non-neural origin.[3-5]

We report a case of testicular teratoma occurring in a patient with peripheral neurofibromatosis, which to the best of our knowledge is the first reported instance of such an association.

Case report

A 23 year old man presented with a 1 month history of pain and swelling of his left testicle. He was known to suffer from peripheral neurofibromatosis (as does his mother and brother) and past history included a left inguinal herniorrhaphy when 5 years old and right orchidopexy for maldescent and insertion of Spitz Holter valve for hydrocephalus secondary to aqueductal stenosis when aged 10.

On examination the right testis was normal but the left testis was enlarged and tender with no palpable lymphadenopathy. There were also multiple café au lait spots and cutaneous neurofibromas. Tumour markers at this time revealed an elevated alpha fetoprotein of 192 μg/l (normal $<$ 10 μg/l) and beta human chorionic gonadotrophin of 140 IU/l (normal $<$ 5 IU/l).

He underwent a left orchidectomy and histology revealed a predominantly undifferentiated teratoma forming tubular and papillary structures with some organoid areas and squamous differentiation (Figure 1). Immunocytochemistry revealed occasional areas staining for alpha-fetoprotein. Adjacent testicular tissue showed tubular atrophy with Leydig cell prominence.

Discussion

We report a case of peripheral neurofibromatosis in association with testicular teratoma. Our patient

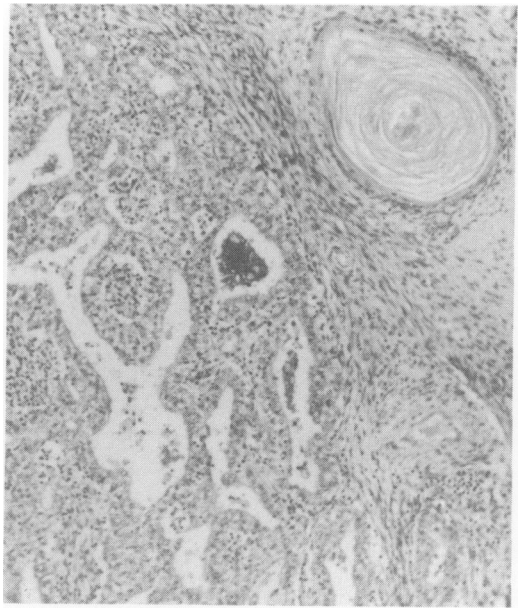

Figure 1 Section showing undifferentiated teratoma and an area of squamous differentiation (× 200).

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also had aqueductal stenosis and hydrocephalus which is a recently described complication of neurofibromatosis.[6]

Neurofibromatosis appears to predispose to a number of tumours particularly gliomas and meningiomas involving the central nervous system.[5] Other associated tumours include phaeochromocytoma,[7] neuroblastoma,[8] gastrointestinal carcinoid,[9] Wilms tumour,[10] rhabdomyosarcoma,[11] carcinoma of the colon,[12] and adenocarcinoma of the pancreas.[13]

The incidence of testicular teratoma is 1.2–2:100,000,[14,15] so that the likelihood of this occurring by chance in association with peripheral neurofibromatosis is very low ($3.6–6 \times 10^{-8}$). We therefore believe that this case may represent part of the increased incidence of malignancy found in patients with peripheral neurofibromatosis.

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