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CASE REPORT

Neurofibromatosis (NF1) Associated With Optic Nerve Pathway Glioma. –A Case Report

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ABSTRACT

Neurofibromatosis (NF) is a distinct condition with a multitude of clinical manifestations. It has been classified recently into two subtypes, NF1 and NF2. NF1 is an autosomal dominant disorder, affecting about 1 in every 3000-4000 persons from all races and from both the genders. We present here, a case of 36 year old male NF1, who was associated with multiple papulo-nodular lesions involving the left eye optic nerve, presenting as optic nerve pathway glioma.

Key words: Neurofibromatosis, Optic nerve pathway glioma, Multiplenodulopapular lesions.

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Introduction

Neurofibromatosis (NF) is a distinct condition with a multitude of clinical manifestations. It has been classified recently into two subtypes, NF1 and NF2. NF1 gene mutations of tumour suppressing genes code for the neurofibromatosis on chromosome 17q11.2 and neurofibromatosis type 2 (NF2) gene mutations of the tumour suppressor gene on chromosome 22q12.1 [1]. NF1 is an autosomal dominant disorder, affecting about 1 in every 3000-4000 persons from all races and from both the genders. About half of the cases are sporadic. It has been described as a syndrome like associated with hereditary aetiology and the sporadic cases represent apparently new mutations. We present here, a

case of NF1 associated with optic nerve involvement, presenting as optic nerve pathway glioma.

Case report

A 36 year old male presented with multiple skin papular lesions on the left side of the face [Table/Fig 1]. The right side of the face appeared to be normal. The patient was apparently alright before 3 years. To start with, he developed the lesion as a papular rash and the nodules grew rapidly in size and number within 3-4 months. The growth was not regular but was sporadic. His family history was negative for any such similar lesions or conditions.

[Table/Fig 1]: Multiple nodular lesions of neurofibromas on left side of face.



[Table/Fig 2]: - Left eye showing corneal opacity



He experienced severe headache since 6 months and complained of diminished vision. The patient was referred to an ophthalmologist and was examined for visual acuity. The findings revealed the pupils to be 6mm in the dark and 3mm in the light. There was no light

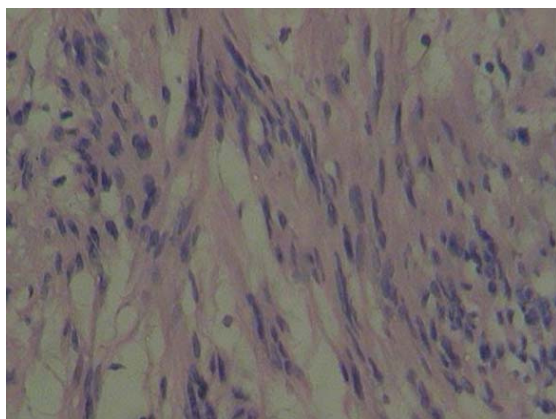
perception (NLP) and a slightly impaired upgaze was noted. The eyeball was protruded on the affected side. [Table/Fig 2]

A radiographical examination of PA skull showed an enlarged optic foramen and canal, suggesting optic nerve involvement too [Table/Fig 3]. A provisional diagnosis of optic nerve glioma associated with neurofibroma was made. There was a 1 x 0.5 cm single lesion on the tongue, which was excised and sent for histopathological examination. Microscopical findings showed characteristics of plexiform neurofibroma, which consisted of a tortuous arrangement of the hypertrophic nerve, which appeared as lobules or discrete bundles within the connective tissue. The elongated spindle tumour cells showed wavy nuclei and were aligned in a herring bone pattern at some places [Table/Fig 4]. This confirmed the diagnosis of neurofibroma. Clinico-pathological correlations suggested that the diagnosis was Neurofibromatosis 1 (NF1).

[Table/Fig 3]: Radiograph showing enlarged left optic canal.



[Table/Fig 4]: Histopathology showing connective tissue stroma with spindle cells having wavy nuclei arranged in fascicles and whorls.



Discussion

Neurofibromatosis is a rare condition which is characterised by hamartomas of neural crest origin, which are inherited as an autosomal dominant trait. In familial cases, the expressivity of NF1 is extremely variable, but there is high penetrance [2]. The genotype-phenotype correlations have not been established as yet. The NF1 gene was identified in 1990 on the long arm of chromosome 17q11.2 and it codes for the protein, neurofibromin. NF1 is a pleiotropic congenital multiple dysplasia syndrome [3]. NF1 has been described by some authors as a polyclonal hamartoma or as a hyperplastic proliferation of several Schwann cell types. Malignant transformation representing a further mutational event in a single cell, leading to monoclonal malignancy, has also been suggested [3].

Clinically, the cardinal features of NF1 are multifocal hyperplasia and neoplasia in the supportive tissue throughout the entire nervous system. The lesions of neurofibromatosis are multiple neurofibromas, presenting as nodules or papules to large masses. These usually begin during or after puberty and gradually increase through adulthood. Hormonal variation has been noted with the growth [4]. There is also a definite increased risk of systemic malignancies, including chronic myelomonocytic leukaemia (CML), neurofibrosarcoma, melanoma, non-Hodgkin's lymphoma and lymphoblastic leukaemia. Patients with NF1 are predisposed to develop malignant peripheral nerve sheath tumours and these tumours often result from the malignant transformation of pre-existing neurofibromas. Optic nerve gliomas are rare in the general population, but are common in NF1 patients.

Other systemic manifestations such as café-au-lait macules and Lisch nodules (iris hamartomas) and osseous lesions such as sphenoid wing dysplasia or thinning of the long bone cortex, with or without pseudoarthrosis and the deformity of the long bone also may be present [5],[6].

Optic nerve glioma (ONG) refers to a group of glial tumours involving the anterior portion of the optic pathway. ONGs occurring in adults are the much less common form. The present case was an uncommon occurrence in an adult. ONG falls into two major clinical-pathological subgroups [7]. These tumours are slowly progressive, with many of them restricted to the optic nerve. However, there is also a significant number of visual pathway gliomas which are isolated to the optic chiasma and the retrochiasmal tracts, sparing the optic nerve. When the tumour has no intracranial extension, the prognosis is excellent even when it is incompletely excised. When there is intracranial invasion, the prognosis is guarded. Most of these tumours have histological features as those of pilocytic astrocytomas. Spontaneous regression has been described in some of these tumours. Often, they behave in a malignant manner. Histologically, they are often anaplastic astrocytomas or glioblastomas [8].

In the present case, a young male patient with no familial history of NF, reported with multiple papulo-nodular lesions involving the left eye as well as the left nasal mucosa, with optic nerve glioma. The other clinical features like lisch nodules, osseous lesions, pigmentations, etc, were not found on examination. Histologically, the nodular lesions were confirmed to be neurofibroma, showing the proliferation of spindle cells, with a thin delicate wavy nucleus and the remnants of neurites with intertwining delicate connective tissue.

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