A Patient with Optic Pathway Glioma, Scoliosis, Chiari Type I Malformation and Syringomyelia: Is it Neurofibromatosis Type 1?

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Summary

A 22 years old girl had features of optic pathway glioma, scoliosis, Chiari type 1 malformation and cervical syringomyelia. She had no cutaneous lesions. We considered this combination to be more than coincidental and argue in favour of considering the case as a variant form of Neurofibromatosis type 1. The relevant literature in favour of our contention has been reviewed.

Key words: Neurofibromatosis type I, Optic glioma, Chiari type 1 malformation, Syringomyelia.

Introduction

Neurofibromatosis (NF) type 1 is a dominantly inherited neurocutaneous syndrome due to a mutation in chromosome 17 (at 17q 11.2). The classic clinical triad includes café au lait spots, multiple neurofibromas and iris Lisch nodules. However several additional features have been described and include – axillary freckling, optic pathway gliomas, pseudoarthrosis of the tibia and other tubular bones, kyphoscoliosis, sphenoidal wing dysplasias, a variety of vascular dysplasias, learning disability, short stature, macrocephaly, genu varum/valgum, pectus excavatum, seizures, mental retardation, colonic hyperganglionosis, excessive pruritus, renal artery stenosis and renal and adrenal tumours. The disorder is highly variable in its manifestation. However, some cutaneous marker is almost always present. Optic pathway gliomas are present in about 15 percent of cases and these are not features of other types of neurofibromatosis. We describe here a case who had a combination of optic-pathway glioma, scoliosis, Chiari type 1 malformation and upper cervical cord syrinx but no skin lesions and argue considering it as a variant form of NF-1.

Case Report

SG, a 22 years old female presented with increasing numbness on the right side of face and upper right arm and occasional dizzy feeling for past two years. Enquiry revealed loss of vision in left eye since early childhood. Over the past few years, she also developed nasal regurgitation and slight hoarseness of voice though she had no dysphagia. There was no family history. In particular, none of her first degree relatives (parents and one brother) had any cutaneous lesions or any neurological problems.

Examination revealed a well built girl of 4 feet 10 inches height with obvious scoliosis and head tilt to the right. The speech was slightly hoarse and slurred as well. She had practically no vision in the left eye and VA in right eye was down to 6/36. Both optic discs, specially the left one, were very pale with clear margins. She had a partial right Horner’s syndrome with slight ptosis and miosis. She had a multidirectional nystagmus – horizontal gaze evoked as well as vertical. Sensations were impaired over right half of face down to C2 dermatone on the right. Palatal movements were restricted and tongue showed no atrophy. She had pyramidal signs in all the four limbs. Though she had scoliosis and head tilt, the neck was not short and she did not have a low hairline. She had no café au lait spots, cutaneous fibromas or axillary/groin freckling. Clinical impression was one a craniovertebral disease, possibly with Chiari malformation and syringobulbia.

Contrast MR scan of brain revealed the following features (Figs. 1a and b). A supresellar solid cystic lesion showing heterogeneous signal intensity and extending along left optic nerve (optic pathway glioma), (b) MR brain (T1 sequence) showing basilar invagination, Chiari malformation and upper cervical syringomyelia.

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The lesion also extended along the left optic nerve and left geniculate body and was partially encasing the circle of Willis. Focal areas of nodular enhancement were noted within the mass. Tonsillar ectopia 12 mm below foramen magnum was seen. Syringomyelic change was noted in the cervical cord from C3 downwards upto C6 level. Tip of the dens projected 11 mm above the Chamber lain’s line and the clivus angle was 130°. Rest of the brain appeared normal and there was no ventriculomegaly. The MR findings suggested optic pathway glioma along with Chiari 1 malformation, syringomyelia and basilar invagination. From the extent of the optic pathway tumor mass and the clinical pattern of visual loss (blind in left eye and VA impaired in right), it seemed highly likely that the tumor originated in the left optic nerve and subsequently involved the right visual pathway through the chiasm.

Discussion

Afifi et al. described two cases of NF-1 associated with ventriculomegaly and Chiari Type 1 malformation. Both cases had skeletal abnormalities at cervico-medullary junction. They suggested that Chiari type 1 malformation should be considered as a cause of non-tumoral ventricular enlargement in patients with NF-1. Dooley et al. reported one further case of association of Chiari type 1 malformation with NF-1 and commented that the former resulted from hypoplasia of the posterior fossa which may be a part of a variety of cerebral dysplasias seen in patients with NF-1. Battistella et al. added one further case to the literature and commented that the association seemed more than coincidental. They suggested that Chiari type 1 malformation be added to the list of CNS abnormalities encountered in patients with NF-1. More recently Fernandez et al. described another patient of NF-1 with syringomyelia, Chiari malformation and scoliosis and reviewed the reported cases.

Our patient had a combination of optic pathway glioma, scoliosis, Chiari type 1 anomaly and syringomyelia but no cutaneous marker for NF-1. As mentioned earlier, optic pathway gliomas occur in about 15% of cases with NF-1 but about of a third of all optic pathway gliomas have underlying neurofibromatosis (type 1). Considering all the aforementioned data, it seems likely that the associations noted in our patient are more than coincidental and we would argue in favour of a diagnosis of neurofibromatosis type 1 in our case even in absence of any cutaneous marker of the disease or a positive family history. Facility for molecular genetic confirmation of NF-1 was not available.

The diagnostic criteria for NF-1 had been established several years earlier. Of the seven features mentioned, three refer to cutaneous lesions and the rest include optic glioma, iris nodule, osseous lesions and family history. At least two of the features need to be present. It is certainly possible that a combination of optic pathway glioma with a non-cutaneous lesion like osseous defect would suffice for making a diagnosis of NF-1 even in absence of a classic cutaneous lesion. It is therefore also possible that such a diagnosis features like scoliosis and Chiari 1 anomaly, all of which have been described in patients with NF-1. One would wonder whether in the light of the recent reports (present one and others mentioned earlier) one needs to broaden the diagnostic criteria for NF-1.

References