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Case report

Early spontaneous regression of a hypothalamic/chiasmatic mass in neurofibromatosis type 1: MR findings

G. Zuccoli[1], **F. Ferrozzi**[1], **M. Sigorini**[2], **R. Virdis**[2], **P. Bassi**[1], **M. Bellomi**[3]

[1] Department of Radiology, University of Parma, V. Gramsci, 14, I-43100 Parma, Italy
[2] Department of Paediatrics, University of Parma, V. Gramsci, 14, I-43100 Parma, Italy
[3] Division of Radiology, European Institute of Oncology, Milan, Italy

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Abstract. A patient with neurofibromatosis type 1 was found to have an enhancing mass in the hypothalamus and in the anterior optic pathway. A 3-month MR study showed a reduction in the size and enhancement of the mass. At a 9-month MR follow-up the mass disappeared and ceased to enhance. This report shows the unusual behaviour of a hypothalamic/chiasmatic mass confirming that in such asymptomatic cases the conservative management can be considered the treatment of choice.

Key words: Neurofibromatosis – Magnetic resonance – Brain – Nervous system tumours

Introduction

Neurofibromatosis type 1 (NF-1) is an autosomal dominant genetic disorder, but approximately half of the cases are spontaneous mutations. Central nervous system (CNS) lesions are a frequent MR finding in NF-1 patients. The behaviour of CNS-enhancing lesions in NF-1 patients is still under debate, since they can regress spontaneously [1, 2, 3]. A spontaneous regression of a mass involving the optic pathways has been suggested by Parazzini et al. to be dysplastic changes rather than neoplastic lesions [3]. Change in the signal intensity has been thought to represent an activity index of the lesion and a reduction in contrast enhancement has been considered as a possible consequence of the blood-brain barrier maturation [3]. In this case report an NF-1 patient showed an enhanced mass infiltrating the anterior optic pathways and the hypothalamus. At 9-month MR follow-up no evidence of the mass was seen.

Correspondence to: G. Zuccoli

Case report

A 3-year-old asymptomatic child was referred to our institution with the diagnosis of neurofibromatosis type 1 made at the age of 1 year, on the basis of the criteria of the National Institutes of Health (NIH) [4]. The patient underwent a baseline MR study of the brain. A lesion was visualized at the level of the hypothalamic region. The mass showed a T1-weighted hypointense signal (Fig. 1 a), a marked enhancement after contrast medium administration (Fig. 1 b) and a T2-weighted hyperintense signal (Fig. 1 c). The anterior optic pathways were enlarged at the right optic nerve level from the mid orbit back to the chiasm (Fig. 1 a) but unenhanced (not illustrated). Numerous T2-weighted high signal intensity small areas (not illustrated) were found in the basal ganglia and in the cerebellum. Three months later, the patient underwent a second MR examination which showed the partial spontaneous decrease of the hypothalamic lesion and of the anterior optic pathway involvement (not illustrated). The last follow-up study performed at the age of 3 years and 9 months showed a regression of the lesion in the hypothalamus with a bulky inferior border to the hypothalamus between the infundibulum and the mamillary bodies (Fig. 2 a). The chiasm and the right optic nerve appeared to be moderately reduced (Fig. 2 a) and they did not enhance (Fig. 2 b). Only small focal areas of T2 hyperintense signal were detected in the chiasmatic/hypothalamic region (Fig. 3 c). No significant modifications were described in the T2 hyperintense lesions found at the previous examinations at the basal ganglia and at the cerebellum levels. At the age of 4 years the child remains asymptomatic.

Discussion

Patient with neurofibromatosis type 1 are at increased risk of developing both benign and malignant tumours [5, 6]. The most frequent central nervous system (CNS)

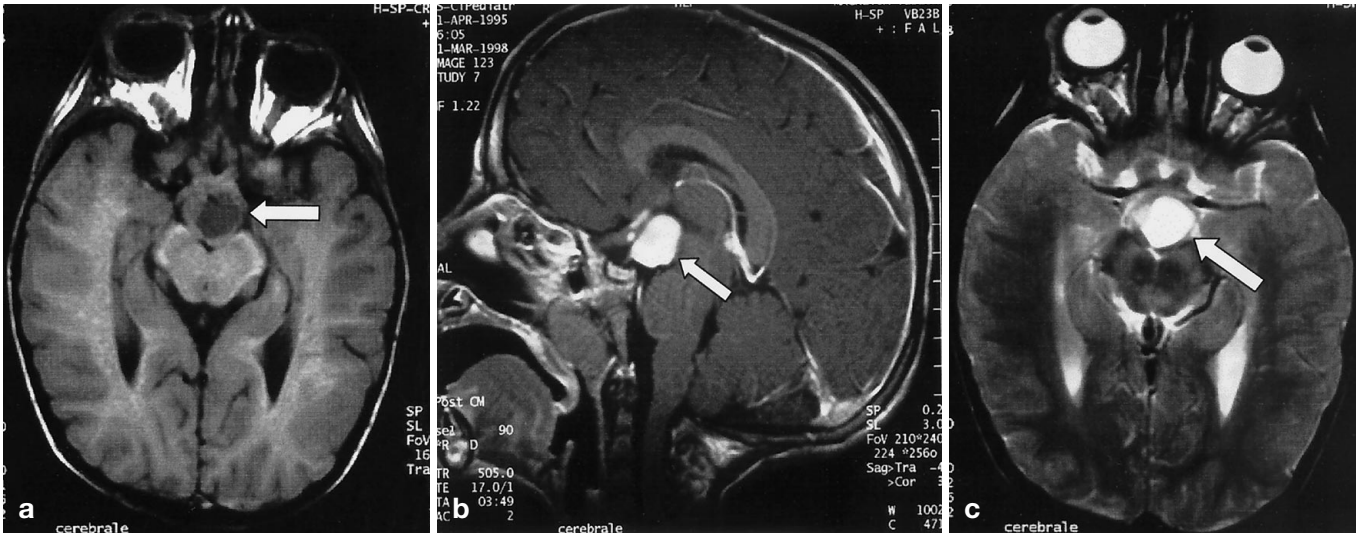
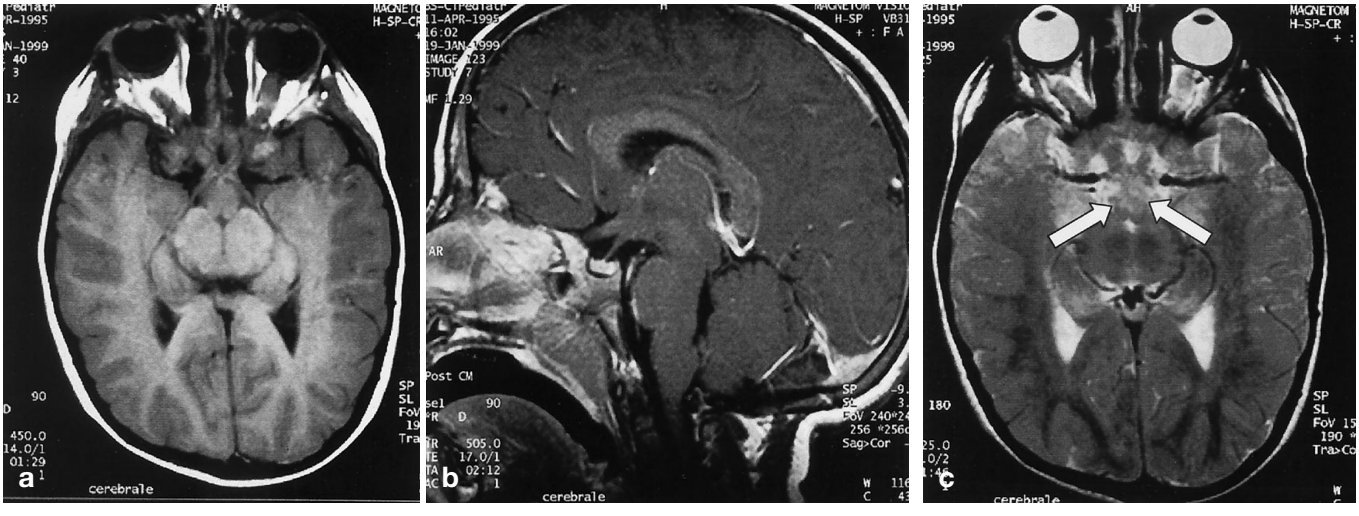


Fig. 1 a–c. A 3-year-old NF1 patient with an enhancing mass in the hypothalamus and in the anterior optic pathways. **a** Unenhanced axial T1-weighted image shows the hypointense mass in the hypothalamus *(arrow)*. **b** Contrast-enhanced sagittal T1-weighted image shows an enhancing mass involving the hypothalamic/chiasmatic region *(arrow)*. The optic nerve appears to be enlarged but unenhanced. **c** Axial T2-weighted image shows the hyperintense mass displacing the chiasma *(arrow)*

radiological findings in NF1 are high signal intensity foci on long-TR MR images. These MR features have been defined as hamartomas, heterotopias or areas of abnormal myelination [7, 8, 9, 10]. They do not enhance and do not have a mass effect [7], and correspond to a spongiotic change in the cerebral tissue [11]. Other frequent lesions of the CNS in NF1 are "gliomas" of the optic pathway and of the brain, astrocytomas and ependymomas [12]. The potential nature of these tumours is controversial. The benign nature of optic pathway tumours in NF1 patients is unrecognised, but it has been recently emphasised by some authors [3, 13, 14]. However, optic pathway gliomas have been included by the World Health Organization (WHO) and by some authors [15, 16] among the pilocystic astrocytomas. A spontaneous regression of an enhanced tectal mass over a 3-year follow-up period in a NF1 was thought to be a hamartoma [1] for its benign behaviour, and in one case a "hamartoma" was reported to enhance [2], but in these studies no biopsies were performed. Differentiating hamartomatous lesions from pilocytic astrocytomas may be difficult by means of MR scan only. In fact, an NF1 patient with various CNS lesions was found to have a histologically proved low-grade pilocytic astrocytoma in the hypothalamic region which spontaneously regressed over a 12-year period [17]. For these reasons non-aggressive treatment has been proposed by some authors in NF1 patients with CNS enhancing lesions [1, 3, 13]. When the "tumoral" regression is not complete, residual hyperin-

Fig. 2 a–c. At 9 months follow up the enhanced lesion disappeared. The hypointense transversal T1-weighted image mass was not visible, but the inferior border of the hypothalamus still remained enlarged showing mass effect. **a** The enlargement of the right optic nerve and the optic chiasm appeared to be moderately decreased. **b** The lack of enhancement of the mass was confirmed on the sagittal T1-weighted image after the contrast medium administration. **c** Small hyperintense foci could be observed in the hypothalamus on transversal T2-weighted image *(arrows)*



tense foci have been described on the long-TR images [3]. In the present case the mass of the chiasmatic/hypothalamic region regressed simultaneously with the optic nerve lesion and in a very short period of time. At the last follow-up residual T2-weighted hyperintense signal foci were visualized in the hypothalamus on the long-TR images. This confirmed a previously reported MR behaviour [3]. Our patient was asymptomatic and did not have any hypothalamic pituitary alterations. All these findings confirm the importance of a non-aggressive management of NF1 children with cerebral masses, even if a marked contrast enhancement is observed.

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