HKJO Quiz

Answer

This patient has opticochiasmatic hypothalamic pilocystic astrocytoma and with the presence of cafe-au-lait spots, the diagnosis of neurofibromatosis 1 (NF-1) is made.

(Question on page 66)

Discussion

Meningioma usually occurs in middle-aged women and can be associated with calcification and hyperostosis. The tumor is more infiltrative. The involvement can be bilateral and very often the optic nerve is straight and may appear as negative image within the tumor, which is known as the tramtrack sign.

Differential 3 is excluded as no primary ocular tumor can be identified in the MRI examination.

Differential 4 is unlikely as lymphoma is uncommon is this age group. Moreover, it is uncommon for lymphoma to involve both optic nerves and the optic chiasma.

More than 50% of the optic nerve glioma occur in children less than 5 years old. The lesion has a well defined margin and the enhancement may not be homogenous which is related to mucin deposition.

Bilateral involvement strongly suggests neurofibromatosis.

Neurofibromatosis type 1 (Von Recklinghausen disease) is the most common phakomatoses.[1] The responsible gene is on the long arm of chromosome 17. It is autosomal dominant with high penetrance but variable expressivity.[2] Diagnosis is made when two or more of the following clinical features are present[3]:

1. **Six or more cafe-au-lait spots with size of 5 mm or larger**
2. **One plexiform neurofibroma or two or more neurofibromas of any type**
3. **Two or more pigmented iris harmatomas**
4. **Axillary or inguinal region freckling**
5. **Optic nerve glioma**
6. **First degree relative with NF-1**
7. **Presence of a characteristic bone lesion (e.g. dysplasia of the greater sphenoid wing, pseudarthrosis)**

Optic nerve glioma occurs in 5% to 15% of all NF-1 cases, although only about 25% of all patients with optic nerve gliomas have NF-1.[4] Optic nerve glioma commonly involves the chiasm, posterior involvement of optic tracts, lateral geniculate body and even optic radiation.[5] The posterior extent of the optic nerve glioma is best delineated by MRI. Most of these tumors are hypo to isointense on T1 weighted images and are usually hyperintense on T2 weighted images. Enhancement is variable, usually minimal or absent, but can be extremely marked and striking, as shown in this case.

References

1. *Osborn AG. Diagnostic neuroradiology. Patterson AS (ed).Mosby Year Book, St. Louis 1994 Chapter 5.*
2. *Smirniotopoulos JG, Murphy FM. The phakomatoses. AJNR 1992; 13:725-46.*
3. *National Institute of Health Consensus Development Conference: Neurofibromatosis Conference Statement. Arch Neurol 1988; 45:579-88.*
4. *Pont MS , Elster AD . Lesions of skin and brain. Modern image of the neurocutaneous syndromes. AJR 1992 ; 158:1193-203.*
5. *Menor F , Marti-Bonmati L, Mulas F et al. Imaging considerations of central nervous system manifestations in pediatric patients with neurofibromatosis type 1 . Pedi Radiol 1991; 21:389-94.*