Professional article/Pregledni prispevek

ECHOGRAPHIC PICTURE OF OPTIC NERVE GLIOMA IN NEUROFIBROMATOSIS TYPE-1

EHOGRAFSKA SLIKA GLIOMA VIDNEGA ŽIVCA PRI NEVROFIBROMATOZI TIP 1

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Arrived 2001-03-15, accepted 2001-05-14; ZDRAV VESTN 2002; 71: Supl. II: 79–81

Key words: *echography; optic nerve glioma; neurofibromatosis type-1*

Abstract – Background. *Authors want to present echographic picture of orbital part of low-grade pilocytic astrocytoma involving the optic nerve and/or chiasm and optic tract (optic pathway glioma or visual pathway glioma).*

Methods. *4 children with neurofibromatosis type-1 complicated with optic pathway glioma diagnosed earlier with magnetic resonance were examined by ultrasound.*
Standardised A-scan technique was used for optic nerve width measurement. The 30° test and B-scan (axial, transverse and longitudinal sections) of both eyes and orbits were performed as well.

Results. *The optic nerve diameter in our cases ranged from 4.48 to 8.5 mm. Two children had the left side optic pathway glioma, one boy had the right side optic pathway glioma and in one tumour was bilateral. The transversal section of the nerve revealed dark oval and in more perpendicular sections round void of the nerve. As the beam is swept towards the orbital apex void becomes more fusiform. The nerve and its sheaths are markedly widened. An abnormal increase in reflectivity and irregularity of the spike's pattern is exhibited as well. No calcification along the sheaths is noticed. The transverse section of the tumour demonstrated an »inverse doughnut« sign. The outer whiter outline of the widened sheaths surrounds an inner darker circle. The longitudinal section revealed the optic nerve head continuing into the widened optic nerve. The 30° test was negative. The differential diagnosis of meningeoma, optic neuritis and orbital cysticercosis should be considered.*

Conclusions. *Ultrasound as a cheap, safe, easily repeatable imaging method should become a method of choice for screening optic nerve tumours in neurofibromatosis type-1, especially in children, as well as for follow-up after treatment.*

Ključne besede: *ehografija; gliom optičnega živca; nevrofibromatoza tip 1*

Izvleček – Izhodišča. *Avtorji želijo predstaviti ehografsko sliko orbitalnega dela nizko razvojnega pilocitičnega astrocitoma, ki vključuje vidni živec in/ali hiazem in očesni trakt (gliom optične poti oziroma gliom vidne poti).*

Metode. *Z ultrazvokom so pregledali štiri otroke z nevrofibromatozo tip-1 z dodatno komplikacijo glioma očesne poti, ki je bil pred tem diagnosticiran z magnetno resonanco.*
Za merjenje širine vidnega živca so uporabili standardizirano tehniko A-scan. Prav tako so opravili 30° test in B-scan (osni, prečni in vzdolžni prerez) obeh očes in očesne votline.

Rezultati. *Premer vidnega živca je segal od 4,48 do 8,5 mm. Otroka sta imela gliom leve strani optične poti, en deček je imel gliom desne strani optične poti, v enem primeru pa je bil tumor obojestranski. Prečni prerez živca je odkril temno ovalno vrzel živca, pravokotni prerez pa okroglo vrzel živca. Z usmeritvijo žarka proti vrhu očesne votline je postala vrzel bolj vretenasta. Živec in njegove ovojnice so postale izrazito razširjene. Prav tako je bilo razvidno tudi nenormalno povečanje pri reflektivnosti in nepravilnosti vzorca. Vzdolž ovojnic ni bilo zaznavne kalcifikacije. Prečni prerez tumorja je kazal znak »obrnjenega krofa«. Zunanji belkasti obris razširjene ovojnice je obkrožal notranji temnejši krog. Vzdolžni prerez je razkrival nadaljevanje glave vidnega živca v razširjeni vidni živec. 30° test je bil negativen. Potrebno je upoštevati diferencialno diagnozo meningoma, vnetje vidnega živca in cisticerkozo očesne votline.*

Zaključki. *Ultrazvok kot poceni, varna in lahko ponovljiva metoda slikanja bi morala postati metoda prvega izbora za presejanje tumorjev vidnega živca pri nevrofibromatozi tip-1, posebno pri otrocih, kakor tudi za opazovanje po zdravljenju.*

Introduction

Neurofibromatosis (NF), also known as von Recklinghausen's disease is one of four major disorders designated phakomatoses (1). Persons with NF manifest characteristic lesion composed of melanocytes or neuroglial cells (1). There are at least two genetically distinct forms of neurofibromatosis, type-1 (NF-1), also referred to as peripheral neurofibromatosis and type-2, central form of the disease (1, 2). Both forms are familial disorders that show autosomal dominant inheritance with very high penetrability. Currently NF-1 is diagnosed if two or more criteria from the group of seven (3) are met. Low-grade

pilocytic astrocytoma involving the optic nerve and/or chiasm and optic tract (optic pathway glioma, OPG or visual pathway glioma, VPG) is found in 15% of unselected NF-1 patients if computed tomography (CT) or magnetic resonance imaging (MRI) are performed (1, 4). Symptomatic OPG occur in 1–5% of persons with NF-1. 9.5–62% of patients with OPG have NF-1 (5–8). CT and MRI are indispensable in detecting OPG of the orbital apex, optic chiasma and optic tract (1, 7). However echography is a cheap, safe, repetable, reliable method of detecting an optic nerve tumour. The purpose of this paper is to present echographic characteristics of optic nerve glioma in its retrobulbar portion.

Methods

4 patients with NF-1 complicated with optic glioma diagnosed earlier with MRI were examined with ultrasound. Standardised A-scan technique was used for optic nerve width measurement (9, 10). The 30° test and B-scan (axial, transverse and longitudinal sections) were performed as well.

Results

A 16-year-old boy had isolated the left optic nerve glioma, a 12-year-old boy the right optic tract, right side of the chiasm and right optic nerve glioma, a 9-year-old girl presented with MRI characteristics of the left side optic glioma involving optic nerve, chiasm and hypothalamus and a 12-year-old boy the right optic nerve, chiasm, hypothalamus and the ventral part of the third ventricle glioma. The optic nerve diameter in our cases was 4.48–8.5 mm. The transversal section of the nerve revealed a dark oval and in more perpendicular sections round void of the nerve. As the beam is swept towards the orbital apex void becomes more fusiform. The nerve and its sheaths are markedly widened. An abnormal increase in reflectivity and irregularity of the spike's pattern is exhibited as well. No calcification along the sheaths is noticed. The transverse section of the tumour demonstrated an »inverse doughnut« sign. The outer whiter outline of the widened sheaths surrounds an inner darker circle. This is due to the characteristic tumour growth pattern. The longitudinal section revealed the optic nerve head continuing into the widened optic nerve. The 30° test was negative.

Discussion

The abnormal proliferation of supporting neuroglial cells, fibrillary astrocyte, has been termed glioma. The shape of the tumour is determined by its environment (7), so it is elongated or hair-like (pilocytic). The patohistological characteristic of low-grade pilocytic astrocytoma are Rosenthal degenerative bodies (7). It may arise anywhere in the central nervous system including the optic nerve. The presenting pattern of the tumour is usually either protrusion, change in visual acuity and/or disturbance in ocular motility (7). The course, treatment and prognosis differ depending on their location along the »nerve tract« (7). Henderson (7) documented 34 histologically verified cases of glioma affecting the intraorbital portion of the optic nerve in their 40-year study. It is 2.4% of total number of their cases of orbital tumours recorded in that period. However the percentage would have been greater if they have encountered those patients with assumed glioma on the bases of imaging studies but not surgically verified. It was the fifth most common primary orbital tumour and the second most frequent orbital tumour in children. 68% of them were seen in the first decade of life. It would be very instructive if this study has shown the percentage of cases with optic nerve gliomas having been diagnosed NF-1.

Optic gliomas in NF-1 that become symptomatic do so before the age of 10, usually after a brief period of enlargement (1). Than they may enter the quiescent phase even without treatment. If confined to the optic nerve at the time of clinical presentation do extend into chiasm but very rarely develop an extradural extension or distant metastasis. Mortality is nil. A complete or subtotal excision is recommended through transfrontal approach (1). Those primarily involving chiasm produce a bilateral visual loss and hypothalamic dysfunction (precocious puberty, hypopituitarism, hydrocephalus) (1, 11). Mortality is 50%. Megavolt radiation therapy (5000 cGy) may retard or reverse progression in many cases (8, 12). In order to avoid complications of brain radiation in early childhood chemotherapy has recently been investigated as an alternative method (1, 13). The 9-year-old girl in our study was presented with symptoms of precocious puberty and bilateral visual decrease.

Magnetic imaging is the principal method of the OPG diagnosis. Alone or combined with computed tomography it provides the OPG diagnosis with a high degree of certainty. The OPG orbital portion shows cylindrical or fusiform enlargement (1). »A relatively narrow central core usually differs in CT density (higher) or MRI intensity (higher with T1 weighting, lower with T2 weighting) from surrounding tissue because of the characteristic growth pattern of optic nerve glioma in NF-1: most cellular proliferation occurs in the perineural intradural space (arachnoidal gliomatosis), associated with production of abundant mucionous material that gives this tissue the signal characteristic of water« (1). Increased length of the intraorbital optic nerve results in its sinuousness, »kinking«, bending or buckling within the confined orbital space. It gives an impression of lobulated, constricted or discontinued optic nerve axial images. The OPG is well marginated due to dural covering (7). The usefulness of MRI is in the study of the intracanalicular portion of the optic nerve. However, it is less efficient for the orbital optic nerve due to similarity of signal intensities of the nerve and fat that is nowadays gradually overcome with fat suppression techniques. MRI either alone or combined with computed tomography does not have 100% accuracy. The most difficult is the differential diagnosis of optic nerve glioma and some meningeomas especially in the in-between groups (older children and adolescents) (14, 15). Imaging features helpful in distinguishing optic nerve glioma from meningeoma are widening of the bony optic canal, rare calcific-like densities, absence of sclerosis in the surrounding bone, prevalence of cystic degeneration in long-standing gliomas, little or no contrast enhancement (7), different density of the glioma core vs. surrounding tissue and »kinking« of the nerve (1). Finally, although both tumours occur with increased frequency in NF-1 than general population, meningeoma is less common and appears rarely in childhood (7).

MRI is a rather expensive and complicated method to be used in screening for intraorbital optic nerve gliomas in children with NF-1. CT, although cheaper, is potentially dangerous, especially to the screened population. To the best of our knowledge this is the first try to give ultrasound characteristics of intraorbital optic nerve glioma in children with NF-1.

In performing ultrasound in children with NF-1 it is important to exclude meningeoma, optic neuritis and orbital cysticercosis. Optic nerve glioma is fusiform, not round massive lesion as meningeoma. It usually has no calcification. Furthermore, meningeoma is less common than glioma in NF-1 and comes in older group of patients than glioma. A sudden visual loss, widened optic nerve sheaths with the positive 30° test and »doughnut sign« highly suggest optic neuritis. Cysticercosis is an infestation by cysticercus cellulosae, the larval form of pork tapeworm, Tenia solium. It should be kept in mind in the differential diagnosis for children in endemic areas with poor hygiene and lack of health awareness, such as in India. A man

becomes the accidental intermediate host by consuming food and water contaminated with eggs of the tapeworm. Common sites of encystment include subcutaneous tissue 24.5%, brain 13.6% and eyes 12.8% (16). Optic nerve cysticercosis may mimic an optic nerve tumour like glioma (17), presenting with a diminished vision and field loss. Common presenting signs are papiloedema, papillitis and papillary involvement. An association with systemic cysticercosis is rare (17–19). A positive ELISA test and anticysticercus antibodies may be rarely found (20). Ultrasound has proved to be an affective alternative to magnetic resonance and computed tomography for orbital cysticercosis (21, 22). Ultrasonography shows »a cystic lesion just behind the globe with an echodense, curvilinear highly reflective structure suggestive of a scolex. The cyst had induced pericystic inflammation seen as finely scattered echodensities in all the ill defined echolucent area surrounding the cyst« (21).

Conclusions

The OPG is among the most characteristic and potentially serious complication of NF-1 (1). Magnetic resonance is a powerful method for its diagnosis, however its availability and cost play the major role in search for another screening method for the intraorbital OPG. Computed tomography, due to its potentially dangerous complications, especially in children, should be reserved for questionable cases, when optic nerve glioma must be distinguished from meningeoma and when surgery is planned. Ultrasound as a cheap, safe, easily repeatable imaging method should become a method of choice for screening for optic nerve glioma in NF-1, especially in children as well as for follow-up after treatment. This paper, for the first time, brings ultrasonic characteristics of the OPG orbital part.

References

1. Basic and Clinical Science Course 1996–1997. Pediatric ophthalmology and strabismus. American Academy of Ophthalmology 1996: 169–75.
2. Gutmann DH. Recent insights into neurofibromatosis type 1: clear genetic progress. Arch Neurol 1998; 55 (6): 778–80.
3. National Institute of Health Concensus Development Conference Statement on Neurofibromatosis. Arch Neurol 1988; 45: 575–80.
4. Kuenzle C, Wissert M, Roulet E, Bode H, Schfer S, Huisman T, Landau K, Boltshauser E. Follow-up of optic pathway gliomas in children with neurofibromatosis type 1. Neuropediatrics 1994; 25 (6): 295–300.
5. Yanoff M, Davis RL, Zimmerman LE. Juvenile pilocytic astrocytoma (glioma) of optic nerve. Clinicopathologic study of 63 cases. In: Jakobiec FA ed. Ocular and adnexal tumors. Birmingham: Aesculapius Publishing Co., 1978: 685–707.
6. Lewis RA, Gerson LP, Axelson KA et al. Von Recklinghausen neurofibromatosis II. Incidence of optic gliomata. Ophthalmology 1984; 91: 929–35.
7. Henderson JW. Orbital tumors. 3rd ed. New York: Raven Press, 1994: 244–52.
8. Shuper A, Horev G, Kornereich L, Michowiz S, Weitz R, Zaizov R, Cohen IJ. Visual pathway glioma: an erratic tumour with therapeutic dilemmas. Arch Dis Childhood 1997; 76 (3): 259–63.
9. Guthoff R. Ultrasound in ophthalmologic diagnosis. New York: Thieme Medical Publishers, Inc., 1991.
10. Atta HR. Ophthalmic ultrasound. A practical guide. New York-Edinburgh-London-Madrid-Melbourne-San Francisco-Tokyo: Churchill Livingstone, 1996: 105–10.
11. Cnossen MH, Stam EN, Cooiman et al. Endocrinologic disorders and optic pathway gliomas in children with neurofibromatosis type-1. Pediatrics 1997; 100 (4): 667–70.
12. Adams C, Fletcher WA, Myles ST. Chiasmal glioma in neurofibromatosis type 1 with severe visual loss regained with radiation. Ped Neurol 1997; 17 (1): 80–2.
13. Packer RJ, Sutton LN, Bilaniuk LT et al. Treatment of chiasmatic-hypothalamic gliomas of childhood with chemotherapy: an update. Ann Neurol 1988; 23: 79–85.
14. Spencer WH. Tumors of the optic nerve. In: Spencer WH ed. Ophthalmic pathology. 3rd ed. Philadelphia: WB Saunders Co., 1986: 2429–51.
15. Wright JE, McNab AA, McDonald WI. Optic nerve glioma and the management of optic nerve tumours in the young. Br J Ophthalmol 1989; 73: 967–74.
16. Malik SRK, Gupta AK, Choudhry S. Ocular cysticercosis. Am J Ophthalmol 1968; 66: 1168–71.
17. Bosquet CF, Dufour TFL, Derome PCE. Retrobulbar optic nerve cysticercosis – case report. J Neuro Surg 1996; 84: 293–6.
18. Madan VS, Dhamija RM, Gill HS, Boparai MS, Souza PD, Sanchetee PC, Bhardwaj JR. Optic nerve cysticercosis – a case report. J Neurol Neurosurg Psychiatry 1991; 54: 470–1.
19. Bawa YS, Wahi PL. Cysticercus cellulosae of optic disc with generalized cysticercosis. Br J Ophthalmol 1962; 46: 753–5.
20. Cardenas F, Quiroz H, Plancarte A, Meza A, Dalma A, Flisser A. Taenia solium ocular cysticercosis-findings in 30 cases. Ann Ophthalmol 1992; 24: 25–8.
21. Murthy H, Kumar A, Verma L. Orbital cysticercosis – an ultrasound diagnosis. Acta Ophthalmol (Copenh) 1990; 68: 612–4.
22. Atul K, Kumar TH, Mallika G, Sandip M. Socio-demographic trends in ocular cysticercosis. Acta Ophthalmol Scand 1995; 73: 438–41.