Optic Disc Atrophy in Children and Neuroimaging Findings

Thammanoon Surachatkumtonekul, M.D., Suchada Kampanartsanyakorn, M.D., Piyada Rutvisuttinunt, M.D.

Department of Ophthalmology, Faculty of Medicine Siriraj Hospital, Mahidol University, Bangkok 10700, Thailand.

Presented at the 34th Annual Meeting of the European Paediatric Ophthalmological Society, Leuven, Belgium, October 23-25, 2008.

ABSTRACT

Objective: To study the correlation of neuroimaging findings in children who presented with optic disc atrophy.
Methods: Retrospective analysis of medical records of patients aged less than 17 years who presented with optic disc atrophy with neuroimaging examinations (magnetic resonance imaging or computed tomography) performed between January 1998 and December 2007 were included in the study.
Results: Eighty cases were included for analysis. There were abnormal neuroimaging findings in 54 cases (67.50%) and normal findings in 26 cases (32.50%). Stratification of these patients in 3 age groups were shown: less than 6 years of age, between 6 and 12 years of age, and more than 12 years of age. In the less than 6 years of age group, central nervous system (CNS) malformations and hydrocephalus were the most common neuroimaging findings. In the 6 to 12 years age group, brain tumors were the most common findings. In the more than 12 years of age group, one case had hydrocephalus and another had encephalomalacia at the right temporal lobe.
Conclusion: Children who presented with optic disc atrophy need neuroimaging examination to find life- threatening causes. CNS malformations and hydrocephalus are common findings in children aged less than 6 years and CNS tumors are common in children aged up to 12 years, especially in age group 6-12 years.

Keywords: Optic atrophy, brain tumor, visual loss, central nervous system, children

Siriraj Med J 2010;62:116-118
E-journal: http://www.sirirajmedj.com

Optic atrophy is one of the important causes of childhood blindness and can be diagnosed by direct visualization. It may occur since birth or develop later. Optic atrophy is a morphologic change which results from loss of retinal nerve fibers.[1,2,3] It is characterized by reduction of nerve diameter with loss of axon and little or no gliosis. The disc remains its normal size and shows pale color. The pallor in optic atrophy results from thinning of the neural tissue of the optic disc and decreased transmission of light.[4,5] The cause of optic atrophy can be found by history taking, physical examination, and investigation such as visual field, neuroimaging, and blood chemistry. The authors were interested in the correlation between neuroimaging findings and childhood optic atrophy.

MATERIALS AND METHODS

Medical record charts between January 1998 and December 2007 were reviewed. Patients aged less than 17 years who presented with optic disc atrophy and had neuroimaging studies (magnetic resonance imaging or computed tomography) performed were included in the study. Patients with optic nerve hypoplasia, glaucomatous optic neuropathy, incomplete medical record or not performed neuroimaging studies were excluded from the study. Demographic data, visual acuities, ophthalmic examination, and neuroimaging findings were analysed. This study was approved by the Siriraj Ethics Committee (Si 432/2008).

RESULTS

Eighty cases of 169 cases met the criteria. Forty three cases (53.75%) had presented at the ophthalmology clinic by themselves with decreased vision 19 cases (44.19%), no eye contact 12 cases (27.91%), nystagmus 6 cases (13.95%), headache 3 cases (6.98%), strabismus 2 cases (4.65%) and diplopia 1 case (2.33%). Thirty seven cases (46.25%) were referred from pediatricians and neurosurgeons. Patients were aged from 3 days to 16 years. Forty four cases were male and 36 cases were female. Seventy seven cases had bilateral

Correspondence to: Thammanoon Surachatkumtonekul
E-mail: sitsr@mahidol.ac.th

TABLE 1. Neuroimaging findings in childhood optic atrophy.

Age (years)	Cases	Neuroimaging (cases)		
		Tumor	Non tumor	Normal
< 6	52 (M=31, F=21)	10	24	18
6-12	23 (M=11, F=12)	12	6	5
>12	5 (M=2, F=3)	0	2	3

M: male, F: female

optic disc atrophy. In the unilateral optic disc atrophy group, two cases were traumatic optic neuropathy and one case was pilocytic astrocytoma at the suprasellar region and hypothalamus. The initial visual acuities of these patients were in the range between no light perception and 6/6. We classified the patients in 3 groups according to age less than 6 years, 6 to 12 years, and more than 12 years. Fifty two cases of 80 cases (65%) were less than 6 years, twenty three cases (28.75%) were from 6 to 12 years and five cases (6.25%) were over 12 years. Twenty six patients (32.5%) with pale disc had normal neuroimaging findings, and 54 cases (67.5%) had abnormal neuroimaging findings. In the normal neuroimaging findings group, there were traumatic optic neuropathy, postinflammatory process, and unknown causes. Fifty four cases had abnormal neuroimaging findings, with central nervous system tumors in 22 cases (40.74%). Table 2 showed that there were craniopharyngiomas in 8 cases (36.36%) which was the most common brain tumor. In the non-tumor group, there were central nervous system malformations (CNS) and hydrocephalus 16 cases (Table 2). In the sub-group analysis, in the less than 6 years group, ten cases (19.23%) were brain tumors while other 24 cases (46.15%) were other brain abnormalities. There were 4 cases of craniopharyngioma which was the same amount as pilocytic astrocytoma. Central nervous system malformations such as schizencephaly, holocephaly, hydranencephaly, porencephaly, and absent cerebrum, were found in 8 cases and were the most common neuroimaging findings in the less than 6 years age group. Eighteen cases of 52 cases (34.62%) had normal neuroimaging findings (Table 1 and 3).

In the aged 6-12 years group, there were central nervous system (CNS) tumors in 12 out of 23 cases (52.17%) and 5 cases (21.74%) with optic disc atrophy had normal neuroimaging. More than 50% of children

TABLE 3. Causes of abnormal neuroimaging findings in patients aged less than 6 years.

Tumor	Cases	Non-tumor	Cases
Craniopharyngioma	4	CNS malformations	8
Pilocytivc astrocytoma	4	Hydrocephalus	6
Malignant mixed germ cell tumor	1	Encephalamalacia	3
Optic glioma	1	Delayed maturation of white matter	3
		Abnormal signal at frontal region, periventricular region	2
		Subarachnoid cyst and microcephaly	1
		Cerebral atrophy	1

CNS: central nervous system

TABLE 2. Causes of abnormal neuroimaging findings.

Tumor (22 cases)		Non-tumor (32 cases)	
Findings	Cases	Findings	Cases
Craniopharyngioma	8	CNS malformations	8
Pilocytic astrocytoma	6	Hydrocephalus	8
Pituitary adenoma	4	Encephalomalacia	4
Germinoma	2	Delayed maturation of white matter	3
Malignant mixed germ cell tumor	1	Increased signal at periventricular, thalamus, frontal area	3
Optic chiasmal glioma (hypothalamus)	1	Brain atrophy	2
		Subarachnoid cyst and microcephaly	1
		Brain swelling	1
		Toxic brain disorder	1
		Ischemic and destructive brain	1

CNS: central nervous system

in this age group who presented with optic atrophy had CNS tumors (Table 1 and 4).

In the over 12 years group, 3 cases (60%) had normal neuroimaging findings and 2 cases (40%) had abnormal neuroimaging findings. One case had hydrocephalus and another had encephalomalacia at the right temporal lobe. None had CNS tumor.

DISCUSSION

Optic atrophy is an important cause of visual loss in children. In the series by Repka and Miller,[6] the underlying causes of optic atrophy in children were tumor 29%, postinflammatory (meningitis, optic neuritis) 17%, trauma 11%, undetermined 11%, hereditary 9%, perinatal disease 9%, hydrocephalus 6%, neurodegenerative disease 5%, toxic or metabolic disease 1%, and miscellaneous 3%. Parikshit and Clare[7] reported that optic nerve disease was one of the causes of blindness in children. Its proportion was 6-25% compared to other parts of the eye ball. Neuroimaging studies of the optic nerve size in optic disc atrophy are less useful because the dimensions of the optic nerves may be either normal or reduced.[8] Optic disc atrophy not only demonstrates the anterior visual pathway pathology, but it also implies the central nervous system lesions too.

Fifty four out of eighty cases (67.5%) with optic disc atrophy had abnormal neoroimaging findings.

TABLE 4. Causes of abnormal neuroimaging findings in patients aged 6 -12 years.

Tumor	Cases	Non-tumor	Cases
Craniopharyngioma	4	Abnormal signal at thalamus (CMV encephalopathy)	1
Pituitary adenoma	4	Brain atrophy	1
Germinoma	2	Brain swelling	1
Pilocytic astrocytoma	2	Hydrocephalus	1
		Ischemic and destructive of brain	1
		Toxic brain disorder	1

CMV: cytomegalovirus

Twenty six cases (32.5%) had normal neuroimaging findings. In the normal neuroimaging findings group, there were traumatic optic neuropathy and postinflammatory process. We hypothesized that these normal findings may be from late neuroimaging studies and the active processes were resolved and some cases may have optic disc atrophy from metobolic disorders, hereditary causes, and medications. Twenty two out of fifty four cases (40.74%) had CNS tumors. Craniopharyngioma was the most common CNS tumor (8 cases). In the non-tumor finding, CNS malformations (schizencephaly, holocephaly, hydranencephaly, porencephaly, absent cerebrum) and hydrocephalus were the most abnormal findings (16 out of 32 cases).

In subgroup analysis, CNS tumors were found in children aged less than 6 years in 10 out of 22 cases (45.45%) and in aged 6-12 years in 12 out of 22 cases (54.55%), but there was no brain tumor in the aged over 12 years group. The mechanism of optic disc atrophy in CNS tumors may be optic nerve compression, infiltration, postpapilledema, toxic effect of chemotherapy such as vincristine,[9] paraneoplastic optic neuropathy, and radiation optic neuropathy. Mudgil and Repka reported the causes of optic atrophy in children under 10 years old. The most common cause was complications from premature birth 16% (most cases had intraventricular hemorrhage), CNS tumor 15% (pilocytic astrocytoma was most common) and hydrocephalus 10%.[10]

According to Denne, et al,[11] sequelae of premature birth, perinatal hypoxia and brain trauma were common causes of childhood optic atrophy. In our study, central nervous system (CNS) malformations and hydrocephalus were the most common causes of optic atrophy in children less than 6 years old which was similar to the Denne, et al[11] study. Our hypothesis is that 1: these patients may have optic disc atrophy from retrograde trans-synaptic degeneration from the brain injury in utero or their early infancy period, 2: after brain injury, an immature brain will have a liquefaction process and dilated ventricles, and 3: children with CNS malformations usually had delayed development, mental retardation or convulsions and were on anticonvulsion drugs, so the parents usually brought them to see the doctors early. The pathogenesis of optic disc atrophy in children with CNS malformations may be related with CNS infections, ischemia, trauma or metabolic disorders which interfere with neuronal development during the intrauterine period. Some children who used anticonvulsion drugs such as vigabatrin (Sabril) may have retinal atrophy and optic atrophy from the drug side effect.[12] However, the limitations of this study were loss of some important data and subjects of the less than 6 years group were much more than other groups.

CONCLUSION

Neuroimaging is an essential investigation in children with optic disc atrophy. CNS malformations and hydrocephalus were common findings in children aged less than 6 years. CNS tumors are common in children aged up to 12 years, especially in the age group 6-12 years.

ACKNOWLEDGEMENTS

The authors wish to thank all clinical staff in the radiology department for neuroimaging data.

REFERENCES

1. Miller NR. Optic atrophy. Walsh FB, Hoyt WF. Clinical Neuro-Ophthalmology. 4th ed. Baltimore, MD: Williams & Wilkins; 1982. p. 329-42.
2. Quigley HA, Davis EB, Anderson DR. Descending optic nerve degeneration in primates. Invest Ophthalmol Vis Sci 1977 Sep;16:841-9.
3. Sadun AA, Martone JF, Muci-Mendoza R, Reyes L, DuBois L, Silva JC, Roman G, Caballero B, et al. Epidemic optic neuropathy in Cuba. Eye findings. Arch Ophthalmol 1994 May;112(6):691-9.
4. Quigley HA, Anderson DR. The histologic basis of optic disk pallor in experimental optic atrophy. Am J Ophthalmol 1977 May;83(5):709-17.
5. Quigley HA, Hohman RM, Addicks EM. Quantitative study of optic nerve head capillaries in experimental optic disc pallor. Am J Ophthalmol 1982 Jun;93(6):689-99.
6. Repka MX, Miller NR. Optic atrophy in children. Am J Ophthalmol 1988 Aug;106:191-3.
7. Parik G, Clare G. Blindness in children: a worldwide perspective. Community Eye Health 2007 June;20(62):32-3.
8. Parravano JG, Toledo A, Kucharczyk W. Dimensions of the optic nerves, chiasm, and tracts. MR quantitative comparison between patients with optic atrophy and normal. J Comput Assist Tomogr 1993 Sep/Oct;17:688-90.
9. Shurin SB, Rekate HL, Annable W. Optic atrophy induced by vincristine. Pediatrics 1982 Aug;70:288-91.
10. Mudgil AV, Repka MX. Childhood optic atrophy. Clin Experiment Ophthalmol 2000 Feb;28(1):34-7.
11. Denne C, Käsmann-Kellner B, Ruprecht KW. Prevalence of optic atrophy and associated ocular and systemic diseases in a department of paediatric ophthalmology. Klin Monatsbl Augenheilkd 2003 Nov;220(11):767-73.
12. Buncic JR, Westall CA, Panton CM, Munn JR, MacKeen LD, Logan WJ. Characteristic retinal atrophy with secondary "inverse" optic atrophy identifies vigabatrin toxicity in children. Ophthalmol 2004 Oct;111(10):1935-42.