Thai J Ophthalmol 2025;39(1):21-31.

Original Article/นิพนธ์ต้นฉบับ

Clinical Characteristics, Diagnosis, and Treatments of Pediatric Nystagmus

Pittaya Phamonvaechavan, MD[1], Rawi Jongpipatchai, MD[1], Supathida Jiamsawad, MD[1]

Abstract

Objective: To report the clinical characteristics, types, causes, and management of pediatric nystagmus diagnosed over a 15-year period.

Patients and Methods: A retrospective study was conducted on pediatric patients under the age of 18 years old who were diagnosed with nystagmus at Siriraj Hospital from 2005 to 2019. The patients' demographic data, characteristics, causes, and treatments of nystagmus were collected.

Results: A total of 328 patients were enrolled. The median age at diagnosis was 1.8 years, with 180 male patients (54.9%). The most prevalent cause was nystagmus associated with retinal or optic nerve abnormalities (42.4%), followed by idiopathic infantile nystagmus syndrome (INS, 17.7%), and nystagmus associated with anterior segment abnormalities (11.6%). Idiopathic INS was more common in Down's syndrome (37.5%) than in non-Down's syndrome patients (14.3%). Among the electroretinogram (ERG) results for nystagmus associated with retinal or optic nerve abnormalities, 83.7% were abnormal. Treatments varied by cause, with isolated refractive correction being the most common (40.5%). Nystagmus from central nervous system (CNS) tumors had the highest rate of surgical treatments (85.7%). while nystagmus associated with anterior segment abnormalities often required multimodal treatments (34.2%).

Conclusion: This study offers insights into the characteristics, prevalence, and treatment of pediatric nystagmus within a tertiary care hospital in Thailand. The primary cause identified was nystagmus associated with retinal or optic nerve abnormalities. ERG is recommended in uncertain diagnosis cases or when retinal function is questionable. Treatment modalities are determined by the underlying causes of nystagmus, with refractive correction being the most commonly employed approach across all causes.

Keywords: eye movement disorders; congenital nystagmus; idiopathic infantile nystagmus; electroretinogram; infantile nystagmus; pediatric ophthalmology

[1]*Department of Opthalmology, Faculty of Medicine Siriraj Hospital, Mahidol University, Bangkok 10700, Thailand*

Introduction

Nystagmus stands as a significant pediatric eye problem, denoting an involuntary oscillation of eye movement affecting either one or both eyes. It can be a pendular or jerk type and manifest at any stage of life. Some patients present as congenital or infantile nystagmus in early childhood, while others may experience its onset at a later time (acquired nystagmus). Prior investigations have estimated the prevalence of childhood nystagmus to range between 1 in 1,000 and 1 in 500,000 cases, predominantly reported in European nations and certain Asian populations.[1-6]

This condition can be categorized into physiological or pathological nystagmus. Pathological nystagmus, in particular, originates from a range of abnormalities within the visual pathway. These encompass instances of nystagmus associated with anterior segment abnormalities, retina or optic nerve abnormalities, neurological pathologies, and the presence of central nervous system (CNS) tumors. These structural irregularities significantly impair the eyes' ability to focus on visual stimuli. Furthermore, a connection has been established between nystagmus and various developmental disorders, including but not limited to Down's syndrome, Noonan syndrome, fetal alcohol syndrome, and septo-optic dysplasia (de Morsier syndrome)[7,8]. Studies have indicated nystagmus prevalence in Down's syndrome, ranging from 5% to 30%[8-10]. Despite the generally benign nature of childhood nystagmus and its usual lack of oscillopsia[11], it requires thorough neurological scrutiny and comprehensive imaging workups[5]. Acquired nystagmus often necessitates additional investigations, such as neuroimaging techniques, to rule out severe or life-threatening etiologies.

Treatment modalities for nystagmus depend on the distinctive pathology unique to each patient, offering a spectrum of interventions, including glasses, eye drops, oral medications, or surgical procedures. Ocular surgery primarily aims for two objectives. First is to enhance vision and diminish the severity of nystagmus, which is often accomplished through procedures such as cataract surgery. Second is to correct the null point of nystagmus to mitigate abnormal head postures[12,13].

Hence, the ability to distinguish between various types of pediatric nystagmus becomes crucial, given the distinct etiologies and corresponding management approaches associated with each type. This study aimed to investigate the clinical characteristics, causes, and treatment modalities of pediatric nystagmus in patients at Siriraj Hospital in Thailand, specifically focusing on cases diagnosed over a 15-year period through the retrospective analysis of medical records of individuals aged 18 years old or younger.

Patients and Methods

This investigation received ethical endorsement from the Human Research Protection Unit, Faculty of Medicine Siriraj Hospital, Mahidol University, in strict adherence with the Declaration of Helsinki guidelines. The research scrutinized the medical records of individuals under 18 years old diagnosed with nystagmus at Siriraj Hospital, a tertiary care institution, from January 2005 to December 2019. Patients with physiologic nystagmus and those subsequently diagnosed with saccadic eye movement disorders or other abnormal eye movements were excluded.

Comprehensive data were collected from the date of the initial diagnosis to the most recent visit in 2019 at the end of the study period. The recorded demographic variables encompassed sex, onset of the disease, age at diagnosis, underlying diseases, family history of nystagmus, presence of genetic disorders, visual acuity (VA), nystagmus characteristics, causes, and treatment modalities. The causes of nystagmus were categorized into idiopathic infantile nystagmus syndrome

(INS), nystagmus associated with anterior segment abnormalities, retinal or optic nerve abnormalities, neurological diseases, tumors of the central nervous system (CNS), and latent or manifest latent nystagmus (LN/MLN). Treatment options for nystagmus were systematically classified into three primary categories: surgery, medication, and refractive correction. Surgery embraced various operative interventions, such as neurologic and ocular procedures; while medication referred to pharmaceutical interventions, and refractive correction involved the application of glasses, prisms, and contact lenses. All data were analyzed using SPSS (Statistical Package for Social Science) version 18. The interpretation of the collected data relied on descriptive analysis.

Results

In total, 328 patients were included in this study, with 21 patients excluded due to missing exact nystagmus onset. Consequently, age at diagnosis was employed for enrollment, resulting in a median age at diagnosis of 1.8 (0.5, 7.0) years old, and a male prevalence of 54.9%. The majority of patients (91.8%) displayed binocular involvement, as depicted in Table 1. Neurological conditions were identified as the underlying cause in approximately 25.3% of patients, including nystagmus patients diagnosed with CNS tumors, other defined neurological disorders, cases with no definitive diagnosis, and LN/MLN.

Table 1 Baseline characteristics of the 328 enrolled patients

Characteristics	n (%)
Sex	
Male	180 (54.9)
Female	148 (45.1)
Age year, Median (IQR)	
at diagnosis	1.8 (0.5, 7.0)
at onset	0.5 (0.2, 2.5)
Underlying disease	
No known underlying disease	196 (59.8)
Down's syndrome	48 (14.6)
Neurological disease	83 (25.3)
Others	1 (0.3)
Eye involvement	
Monocular	27 (8.2)
Binocular	301 (91.8)
Family history of nystagmus, (n = 150)	
Absent	130 (86.7)
Present	20 (13.3)

**Abbreviation: IQR, interquartile range.

As listed in Table 2, the causes of nystagmus were as follows (in descending order): nystagmus associated with retinal or optic nerve abnormalities (139, 42.4%), idiopathic INS (58, 17.7%), no definitive diagnosis (43, 13.1%), nystagmus associated with anterior segment abnormalities (38, 11.6%), neurological disease (25, 7.6%), LN/MLN (18, 5.5%), and CNS tumor (7, 2.1%). Across all nystagmus causes, a prevailing trend indicated horizontal nystagmus was the most common directional manifestation, while vertical nystagmus was least presented. Additionally, the pediatric nystagmus characteristics in this study were recorded in 97 patients (29.6%), comprising jerk nystagmus (62, 63.9%), pendular nystagmus (30, 30.9%), and a combination of jerk and pendular nystagmus (5, 5.2%). Abnormal head positions were identified in 84 subjects (25.6%), with head turn being the most frequent position (57.1%).

Table 2 Age at diagnosis, nystagmus direction, and ERG in different causes of nystagmus.

	Causes of nystagmus (n = 328 patients)						
	Anterior segment abnormalities	**Retina or optic nerve abnormalities**	**Idiopathic INS**	**LN/MLN**	**CNS tumor**	**Neurologic**	**No definite diagnosis**
n (%)	38 (11.6)	139 (42.4)	58 (17.7)	18 (5.5)	7 (2.1)	25 (7.6)	43 (13.1)
Age, Median (IQR)*	1.4 (0.4, 6.1)	1.3 (0.4, 4.0)	2.5 (0.7, 7.7)	3.8 (1.4, 9.3)	12.0 (4.7, 14.7)	1.3 (0.7, 12.8)	2.0 (0.5, 8.0)
Nystagmus directions (%)							
n (%)	18 (47.4)	93 (66.9)	49 (84.5)	7 (38.9)	4 (57.1)	14 (56.0)	25 (58.1)
Horizontal	77.8	78.5	89.8	100.0	100.0	50.0	84.0
Vertical	5.6	4.3	2.0	0.0	0.0	14.3	4.0
Torsional	5.6	4.3	4.1	0.0	0.0	14.3	8.0
Mixed	11.1	12.9	4.1	0.0	0.0	21.4	4.0
ERG							
Abnormal, n (%)	0	43 (60.6)	16 (22.5)	0	0	6 (8.5)	6 (8.5)
ERG, n (%valid)	0	36 (83.7)	1 (6.3)	0	0	0	0
Abnormal head position, n (%)	4 (10.5)	30 (21.6)	29 (50.0)	4 (22.2)	2 (28.6)	7 (28.0)	8 (18.6)

*Age at diagnosis (year).

Abbreviation: INS; infantile nystagmus syndrome, CNS; central nervous system, IQR; interquartile range, ERG; electroretinogram, LN/MLN; latent or manifest latent nystagmus.

Regarding VA, among the cases where VA measurement was possible, nystagmus associated with CNS tumor exhibited the highest VA, where 7 out of 14 eyes (50%) had an optimal VA defined as 20/20 to 20/40.The majority of eyes with VA equal to or worse than 20/200 were nystagmus associated with anterior segment abnormalities (63.3%) and retina or optic nerve abnormalities (60.8%). Additionally, 68.9% of idiopathic INS patients demonstrated VA superior to finger counting. On the contrary, in situations where VA measurement was not possible, Idiopathic INS (97.1%), anterior segment abnormalities (84.6%), and LN/MLN (80.0%) demonstrated the highest three proportions among patients that were able to fix and follow. The

lowest assessment of infantile VA, characterized by neither fixed nor follow responses, was most prominent in nystagmus resulting from neurological diseases at 40.9%, affecting 9 out of 22 cases. (Table 5)

In terms of the results of the investigations performed (Table 2), 37 out of 71 patients, who underwent electroretinogram (ERG) (52.1%), exhibited abnormal findings. Among patients with abnormal ERG results, 97.3% were diagnosed with nystagmus from retina or optic nerve abnormalities, in which 36 out of all 43 ERGs performed in this group (83.7%) were abnormal. Additionally, Down's syndrome was diagnosed in 14.6% of the 328 patients. Table 3 shows a higher prevalence of idiopathic INS and no definite diagnosis in Down's syndrome (37.5% and 37.5%) compared to non-Down's syndrome patients (14.3% and 8.9%, respectively). Furthermore, an increased occurrence of LN/MLN was found in Down's syndrome individuals (10.4%), as opposed to the comparatively lower rate of 4.6% in the non-Down's syndrome counterparts.

Table 3 Causes of nystagmus in Down's syndrome and non-Down's syndrome patients (total = 328 patients)

Causes of nystagmus	Number (%)	
	Non-Down's syndrome n = 280	Down's syndrome n = 48
Anterior segment abnormalities	36 (12.9)	2 (4.2)
Retina or optic nerve abnormalities	134 (47.9)	5 (10.4)
Idiopathic INS	40 (14.3)	18 (37.5)
LN/MLN	13 (4.6)	5 (10.4)
CNS tumor	7 (2.5)	0
Neurologic	25 (8.9)	0
No definite diagnosis	25 (8.9)	18 (37.5)

INS = infantile nystagmus syndrome, CNS = central nervous system, LN/MLN = latent nystagmus or manifest latent nystagmus.

Regarding the treatments in general, the nystagmus patients required refractive correction (64.6%), surgery (28.7%), and medication (16.5%). Overall, isolated refractive correction was the most common treatment modality employed in 40.5% of cases in this series (Table 4). Also, surgical intervention was most frequently performed in patients with nystagmus caused by CNS tumors (6 of 7, 85.7%), followed by those with nystagmus associated with anterior segment abnormalities (28 of 38, 73.7%). Patients with nystagmus associated with anterior segment abnormalities most often required multimodal treatments (34.2%), including surgical, medical, and refractive interventions, while those with nystagmus of no definitive diagnosis mostly required no definitive treatment.

Table 4 Treatments of different causes of nystagmus

	Causes of nystagmus (n = 328 patients)							
	Overall	**Anterior segment abnormalities**	**Retina or optic nerve abnormalities**	**Idiopathic INS**	**LN/MLN**	**CNS tumor**	**Neurologic**	**No definite diagnosis**
n (%)	328 (100)	38 (11.6)	139 (42.4)	58 (17.7)	18 (5.5)	7(2.1)	25 (7.6)	43(13.1)
Single treatment								
Surgery	19 (5.8)	4 (10.5)	3 (2.2)	1 (1.7)	4 (22.2)	3(42.9)	2 (8.0)	2(4.7)
Medication	7 (2.1)	1 (2.6)	2 (1.4)	2 (3.4)	1 (5.6)	01(4.0)	0	
RC	133 (40.5)	4 (10.5)	70 (50.4)	29 (50.0)	8 (44.4)	08(32.0)	14 (32.6)	
Combined treatments								
Surgery & Medication	7 (2.1)	1 (2.6)	2 (1.4)	1 (1.7)	0 2(28.6)	01(2.3)		
Surgery & RC	39 (11.9)	10 (26.3)	12 (8.6)	8 (13.8)	1 (5.6)	1(14.3)	3 (12.0)	4(9.3)
Medication & RC	11 (3.4)	2 (5.3)	5 (3.6)	1 (1.7)	1 (5.6)	00 2	(4.7)	
Medication, typo , Surgery & RC	29 (8.8)	13 (34.2)	10 (7.2)	3 (5.2)	2 (11.1)	00 1	(2.3)	
No definite treatment	83 (25.3)	3 (7.9)	35 (25.2)	13 (22.4)	1 (5.6)	1(14.3)	11 (44.0)	19 (44.2)

INS = infantile nystagmus syndrome, CNS = central nervous system, LN/MLN = latent or manifest latent nystagmus, RC = refractive correction.

Table 5 Visual acuity of nystagmus categorized by causes

	Causes of nystagmus (n = 656 eyes) , n (%)						
	Anterior segment abnormalities	**Retina or optic nerve abnormalities**	**Idiopathic INS**	**LN/MLN**	**CNS tumor**	**Neurologic**	**No definite diagnosis**
Total (eye), n (%)	76 (11.6)	278 (42.4)	116 (17.7)	36 (5.5)	14 (2.1)	50 (7.6)	86 (13.1)
Measurable VA							
n (eyes)	49	189	80	26	14	28	47
VA > 20 /40	4 (8.2)	6 (3.2)	8 (10.0)	5 (19.2)	7 (50.0)	7 (25.0)	6 (12.8)
20/40 ≥ VA > 20/100	3 (6.1)	32 (16.9)	32 (40.0)	11 (42.3)	1 (7.1)	11 (39.3)	28 (59.6)
20/100 ≥ VA > 20/200	11(22.4)	36 (19.0)	20 (25.0)	4 (15.4)	1 (7.1)	3 (10.7)	8 (17.0)
20/200 ≥ VA > CF	17(34.7)	83 (43.9)	20 (25.0)	6 (23.1)	4 (28.6)	6 (21.4)	3 (6.4)
Count fingers	1 (2.0)	13 (6.9)	0	0	0	1 (3.6)	0
Hand motion	4 (8.2)	7 (3.7)	0	0	0	0	1 (2.1)
Light projection	3 (6.1)	2 (1.1)	0	0	0	0	0
Light perception	3 (6.1)	5 (2.6)	0	0	1 (7.1)	0	1 (2.1)
No light perception	3 (6.1)	5 (2.6)	0	0	0	0	0
Infantile VA							
n (eyes)	26	87	34	10	0	22	36
Fixed and followed	22 (84.6)	51 (58.6)	33 (97.1)	8 (80.0)	0	13 (59.1)	26 (72.2)
Fixed, not followed	0	4 (4.6)	0	2 (20.0)	0	0	2 (5.6)
Not fixed nor fellowed	4 (15.4)	32 (36.8)	1 (2.9)	0	0	9 (40.9)	8 (22.2)
Phthisis bulbi							
n (eyes)	1	2	2	0	0	0	3

INS = infantile nystagmus syndrome, CNS = central nervous system, LN/MLN = latent nystagmus or manifest latent nystagmus; VA = visual acuity ; CF = counting finger.

Discussion

This study endeavored to delineate the clinical characteristics, types, and treatments of nystagmus in individuals below the age of 18, with a focus on subdividing the cases based on their causes. While the most predominant cause identified was nystagmus associated with retinal or optic nerve abnormalities, supporting an earlier study[14], our results revealed notable deviations in the prevalence rates of the other nystagmus subcategories. Specifically, our study identified the most common causes in decreasing order as: nystagmus associated with retinal or optic nerve abnormalities (42.4%), idiopathic INS (17.7%), and nystagmus associated with anterior segment abnormalities (11.6%). In contrast, a study by Nash et al. in 2017 reported different leading causes, with nystagmus associated with retinal or optic nerve abnormalities at 32.4%, idiopathic INS at 31.1%, and manifest latent nystagmus (MLN) at 24%[14]. The differing prevalence of nystagmus subtypes between our study and Nash et al. may be attributed to differences in study design and population source, with our data derived from a tertiary referral center and theirs from a community-based cohort. Additionally, our study was conducted in an Asian-based population, in contrast to the predominantly White population in Olmsted County, which may contribute to differences in the distribution of underlying etiologies.[14]

Our findings regarding the direction of nystagmus revealed that horizontal oscillation was the most prevalent characteristic, consistent with those of previous research[15]. Additionally, various forms of nystagmus, such as down-beat, torsional, periodic alternating, and see-saw nystagmus, may be indicative of diseases affecting the central connections of the vestibular system[15,16]. Interestingly, while vertical nystagmus typically raises concerns on brainstem nuclei and pathways that mediate vertical eye movement[17], all patients with CNS tumors in our study exhibited horizontal rather than vertical nystagmus. Notably, among the nine patients who presented with vertical nystagmus, 4 (44.4%) were ultimately diagnosed with nystagmus associated with retinal or optic nerve abnormalities. This finding underscores the need to consider other ocular causes in the differential diagnosis of vertical nystagmus, despite its traditional association with CNS pathology.

An assessment of diagnostic modalities indicated that ERG is preferable when the diagnosis is unclear or the clinical presentations suggest retinal dystrophy. Among our patients, 21.6% had ERG investigated. Our study showed that 83.7% of the ERGs performed in cases associated with retinal or optic nerve abnormalities yielded abnormal results, supporting the recommendation for ERG to evaluate retinal function in patients with normal structural retinal examinations. Without ERG investigation, those 36 patients might possibly be misdiagnosed as idiopathic INS. Prior research also showed a high incidence of congenital achromatopsia (29%) diagnosed by ERG, which otherwise might be undiagnosed with early onset nystagmus.[18] Therefore, it is important to consider performing ERG where the diagnosis is uncertain. In our study, one (6.7%) idiopathic INS case had an abnormal ERG result as mildly decreased retinal function, which was not correlated with any specific retinal degeneration or any other photoreceptor disease.

A significant observation pertained to Down's syndrome, a genetic disorder associated with nystagmus. Down's syndrome is the most commonly identified genetic disorder affecting multiple organ systems, including ocular and visual abnormalities, and has a reported association with nystagmus[19,20]. Our study reports a higher prevalence of Down's syndrome (37.5%) compared to previous research (9.7%), and idiopathic nystagmus was found more frequently in

Down's syndrome patients than those without Down's syndrome. Additionally, our result showed a higher LN/MLN in Down's syndrome (10.4%) than in the non-Down's syndrome population (4.6%), supporting that MLN is usually associated with Down's syndrome[21]. LN/MLN is typically associated with binocular fusion dysfunction resulting from childhood strabismus[22]. Subgroup analysis further corroborated our findings, revealing that 83.3% of our LN/MLN patients also had a history of strabismus, thereby reinforcing the connection between LN/MLN and strabismus.

Measurable VA assessments highlight that all idiopathic INS and LN/MLN cases in our study exhibited a VA better than counting fingers. This finding may be attributed to the preservation of normal anterior and posterior segment structures in these patients, which likely contributes to the relatively better VA observed in both idiopathic INS and LN/MLN. Fu et al. reported the LogMAR VA of idiopathic INS patients aged more than 2 years old as 0.29 ± 0.25[17], corroborating the trend of a VA superior to finger counting. However, our investigation yielded a mean LogMAR VA of idiopathic INS patients aged more than 2 years old of 0.68 ± 0.36. This discrepancy in visual acuity might be attributed to differences in research methodology. Fu et al. primarily calculated the value from binocular VA with some monocular VA of the better eye in strabismic patients. Conversely, our mean VA was derived from isolated monocular VA assessments. Nonetheless, despite the different VA calculation method, both our research and that of Fu et al.[17] emphasize the trend in which idiopathic INS patients show VA better than finger counting. Therefore, further investigation should be considered in patients who are suspected of having idiopathic INS with poor VA at initial presentation.

In discussing the treatment approaches, it is crucial to note that the plan for addressing nystagmus is specific to its etiology. Nystagmus from CNS tumors (85.7%) and anterior segment abnormalities (73.7%) are the two leading causes involving surgery in the treatment plan. The specified surgical procedures encompass brain surgery for CNS tumors, cataract surgery for lens abnormalities, and muscle surgery designed to adjust the null point for individuals experiencing abnormal head positions. Beyond surgical options, alternative choices, such as medications and refractive corrections, are available for managing nystagmus. It is noteworthy that refractive correction plays a substantial role in the overall correction of nystagmus across various causes. Specifically, approximately 40% of all patients underwent isolated refractive correction, and 24% more had refractive correction integrated into their comprehensive treatment plan. This highlights the significance of considering multiple modalities to ensure a thorough and effective approach to addressing nystagmus based on its underlying causes.

Despite these findings, it is crucial to acknowledge certain limitations of our study, including the inadequate sample size, recruitment methods, and missing data due to the retrospective nature of the study. We mainly retrieved the data from patients who were diagnosed with nystagmus. However, latent nystagmus (LN), usually found related to infantile strabismus, and manifest latent nystagmus (MLN) are also commonly associated with strabismus[9,10]. Therefore, some LN/MLN cases who presented with strabismus were not included in this study, resulting in much lower number of LN/MLN cases in our study compared to those in previous research[11]. Also, our research was a retrospective study, in which data were tracked back to the patient's first visit, some as far back as 18 years, with some data such as details of nystagmus characteristics and treatments missing. Additionally, there were 43 (13.1%) patients in the No definitive diagnosis group due to an unclear diagnosis determination in their medical records. However, the No definite diagnosis

group showed some similarities with the idiopathic INS group, such as the distribution of treatment choices (Table 4) and VA (Table 5), suggesting that some data regarding the distribution of nystagmus by etiology may be underestimated relative to their true prevalence. Lastly, the data in this study were collected in a tertiary care hospital, in which cases were referred from other local hospitals. Some patients might have gone unreported at their primary care hospitals, so our prevalence rates might deviate from the actual ones in the population.

Conclusions

In conclusion, this study provides valuable insights into the clinical characteristics, prevalence, and treatments of pediatric nystagmus in a tertiary care hospital in Thailand. The most common type was nystagmus associated with retinal or optic nerve abnormalities. In cases where the cause of nystagmus is unclear, especially questionable idiopathic INS, ERG investigation is recommended for diagnosis confirmation. Treatments including surgery, medication, and refractive correction depend on the causes of nystagmus, with refractive correction was the most common treatment modality used across all nystagmus causes. This research contributes valuable information to the understanding of pediatric nystagmus, prompting further exploration and refinement of the diagnostic and treatment approaches.

Conflict of Interest

The authors declare no conflict of interest.

References

1. Ehrt O. Infantile and acquired nystagmus in childhood. Eur J Paediatr Neurol. 2012;16(6):567-72.
2. Forssman B, Ringner B. Prevalence and inheritance of congenital nystagmus in a Swedish population. Ann Hum Genet. 1971;35(2):139-47.
3. He J, Lu L, Zou H, He X, Li Q, Wang W, et al. Prevalence and causes of visual impairment and rate of wearing spectacles in schools for children of migrant workers in Shanghai, China. BMC Public Health. 2014;14:1312.
4. Norn MS. Congenital idiopathic nystagmus. Incidence and occupational prognosis. Acta Ophthalmol (Copenh). 1964;42(4):889-96.
5. Sarvananthan N, Surendran M, Roberts EO, Jain S, Thomas S, Shah N, et al. The prevalence of nystagmus: the Leicestershire nystagmus survey. Invest Ophthalmol Vis Sci. 2009;50(11):5201-6.
6. Stewart-Brown S, Haslum M. Partial sight and blindness in children of the 1970 birth cohort at 10 years of age. J Epidemiol Community Health. 1988;42:17-23.
7. Bertsch M, Floyd M, Kehoe T, Pfeifer W, Drack AV. The clinical evaluation of infantile nystagmus: What to do first and why. Ophthalmic Genet. 2017;38(1):22-33.
8. Da Cunha RP, De Castro Moreira JB. Ocular Findings in Down's Syndrome. Am J Ophthalmol. 1996;122(2):236-44.
9. Makateb A, Hashemi H, Farahi A, Mehravaran S, Khabazkhoob M, Asgari S. Ocular alignment, media, and eyelid disorders in Down syndrome. Strabismus. 2020;28(1):42-8.
10. Wagner RS, Caputo AR, Reynolds RD. Nystagmus in down syndrome. Ophthalmology. 1990;97(11):1439-44.
11. Penix K, Swanson MW, DeCarlo DK. Nystagmus in pediatric patients: interventions and patient-focused perspectives. Clin Ophthalmol. 2015;9:1527-36.
12. Dell'Osso LF, Flynn JT. Congenital nystagmus surgery. A quantitative evaluation of the effects. Arch Ophthalmol (Chicago, Ill : 1960). 1979;97(3):462-9.
13. Flynn JT, Dell'Osso LF. The effects of congenital nystagmus surgery. Ophthalmology. 1979;86(8):1414-27.
14. Nash DL, Diehl NN, Mohney BG. Incidence and Types of Pediatric Nystagmus. Am J Ophthalmol. 2017;182:31-4.
15. Abadi RV, Bjerre A. Motor and sensory characteristics of infantile nystagmus. Br J Ophthalmol. 2002;86(10):1152-60.

16.Leigh RJ, David SZ. Nystagmus Due to Vestibular Imbalance. 5th ed. Oxford, UK: Oxford University Press; 2005.
17. Fu VL, Bilonick RA, Felius J, Hertle RW, Birch EE. Visual acuity development of children with infantile nystagmus syndrome. Invest Ophthalmol Vis Sci. 2011;52(3):1404-11.
18. Good PA, Searle AE, Campbell S, Crews SJ. Value of the ERG in congenital nystagmus. Br J Ophthalmol. 1989;73(7):512-5.
19. Oster-Granite ML, Parisi MA, Abbeduto L, Berlin DS, Bodine C, Bynum D, et al. Down syndrome: national conference on patient registries, research databases, and biobanks. Mol Genet Metab. 2011;104(1-2):13-22.
20. Weijerman ME, de Winter JP. Clinical practice. The care of children with Down syndrome. Eur J Pediatr. 2010;169(12):1445-52.
21. Papageorgiou E, McLean RJ, Gottlob I. Nystagmus in childhood. Pediatr Neonatol. 2014;55(5):341-51.
22. Kushner BJ. Infantile uniocular blindness with bilateral nystagmus. A syndrome. Arch Ophthalmol (Chicago, Ill : 1960). 1995;113(10):1298-300.

การศึกษาวิเคราะห์ลักษณะทางคลินิก การวินิจฉัย และการรักษาของโรคตากระตุกในเด็ก

Pittaya Phamonvaechavan, MD[1]
พิทยา ภมรเวชวรรณ, พ.บ.[1]

Rawi Jongpipatchai, MD[1]
รวิ จงพิพัฒน์ชัย, พ.บ.[1]

Supathida Jiamsawad, MD[1]
ศุภธิดา เจียมสวัสดิ์, พ.บ.[1]

บทคัดย่อ:

วัตถุประสงค์: เพื่อรายงานลักษณะทางคลินิก ประเภท สาเหตุ และการรักษาของโรคตากระตุกในเด็กที่ได้รับการวินิจฉัยในช่วงเวลา 15 ปี

วิธีดำเนินการวิจัย: การศึกษาย้อนหลังของข้อมูลประชากร ลักษณะ สาเหตุ และการรักษาของผู้ป่วยที่ได้รับการวินิจฉัยว่าเป็นโรคตากระตุกก่อนอายุ 18 ปี ที่โรงพยาบาลศิริราช ระหว่างปี 2548 ถึง 2562

ผลการศึกษา: จำนวนผู้ป่วยในโครงการวิจัยทั้งหมด 328 คน มีอายุมัธยฐานขณะวินิจฉัยที่ 1.8 ปี โดยมีผู้ป่วยเพศชาย 180 ราย (54.9%) สาเหตุที่พบมากที่สุดคือโรคตากระตุกที่เกี่ยวข้องกับความผิดปกติของจอตาหรือเส้นประสาทตา (42.4%) ตามด้วยโรคตากระตุกที่ไม่ทราบสาเหตุในทารก (17.7%) และโรคตากระตุกที่เกี่ยวข้องกับความผิดปกติของส่วนหน้าของดวงตา (11.6%) โดยโรคตากระตุกที่ไม่ทราบสาเหตุในทารกสามารถพบได้บ่อยในผู้ป่วยที่เป็นกลุ่มอาการดาวน์ (37.5%) มากกว่าผู้ป่วยที่ไม่เป็นกลุ่มอาการดาวน์ (14.3%) จากการวิจัยพบว่า 83.7% ของผู้ป่วยโรคตากระตุกที่เกี่ยวข้องกับความผิดปกติของจอตาหรือเส้นประสาทตาพบมีผลความผิดปกติของคลื่นไฟฟ้าจอตา (electroretinogram; ERG) การรักษาโรคตากระตุกขึ้นอยู่กับสาเหตุ โดยการแก้ไขความผิดปกติของค่าสายตาเพียงอย่างเดียวเป็นวิธีที่ใช้บ่อยที่สุด (40.5%) โรคตากระตุกจากเนื้องอกในระบบประสาทส่วนกลาง (central nervous system; CNS) มีอัตราการรักษาด้วยการผ่าตัดสูงที่สุด (85.7%) ในขณะที่โรคตากระตุกที่เกี่ยวข้องกับความผิดปกติของส่วนหน้าของดวงตามักต้องการการรักษาหลายวิธีร่วมกัน (34.2%)

สรุป: การศึกษานี้ให้ข้อมูลเชิงลึกเกี่ยวกับลักษณะ ความชุก และการรักษาโรคตากระตุกในเด็กภายในโรงพยาบาลตติยภูมิในประเทศไทย สาเหตุหลักคือโรคตากระตุกที่เกี่ยวข้องกับความผิดปกติของจอตาหรือเส้นประสาทตา แนะนำให้ทำการตรวจ ERG ในกรณีที่การวินิจฉัยไม่ชัดเจนหรือมีความสงสัยเกี่ยวกับการทำงานของจอตา การรักษาขึ้นอยู่กับสาเหตุของโรคตากระตุก โดยการแก้ไขสายตาเป็นวิธีที่ใช้บ่อยที่สุดในทุกสาเหตุ

คำสำคัญ: ความผิดปกติของการเคลื่อนไหวของตา, ตากระตุกตั้งแต่กำเนิด, ตากระตุกในทารกที่ไม่ทราบสาเหตุ, คลื่นไฟฟ้าจอตา, ตากระตุกในทารก, จักษุวิทยาเด็ก

[1] *ภาควิชาจักษุวิทยา ตึกสยามินทร์ ชั้น 14 คณะแพทยศาสตร์ศิริราชพยาบาล มหาวิทยาลัยมหิดล*

Footnotes and Financial Disclosures
Originally receive: 7/8/2024
Final revision: 19/5/2025
Accepted: 3/6/2025

Corresponding author: Pittaya Phamonvaechavan; E-mail: pittaya2002@gmail.com; Tel: 02-419-8037, 081-682-1744

Financial Disclosure(s)
All authors declare that they have no financial disclosures.