

Introduction

Torticollis arises from various causes, including musculoskeletal and ophthalmic conditions. Ocular torticollis, often caused by 4th cranial nerve palsy, is frequently misdiagnosed, leading to complications like facial asymmetry. Since standard eye exams are challenging for infants and in primary care, we previously developed a 5-item parent-reported questionnaire. This study aims to validate this tool in a new cohort to confirm its reliability as an effective screening method for ocular torticollis.

Methods

This retrospective cohort study included the patients > 12 months with abnormal head posture who completed a 5-item questionnaire and underwent comprehensive ophthalmic examination. Diagnostic accuracy of the questionnaire was assessed. Age-stratified analyses were conducted to compare diagnostic value across developmental stages. A weighted scoring model was also developed using logistic regression.

Results

A total of 88 children were included, and ocular torticollis was diagnosed in 25 (28.5%) patients. Superior oblique palsy was the most common cause (68.0%), followed by combined horizontal and vertical strabismus, amblyopia, Brown syndrome, and third cranial nerve palsy (Table 1). The mean questionnaire score was significantly higher in the ocular group than in the non-ocular group (3.1 ± 0.8 vs. 1.9 ± 0.8 , $p < 0.001$). Age-stratified analysis revealed statistically significant score differences in the 25-60 and > 60-month groups ($p < 0.001$, 0.009 , respectively), but not in the 12-24-month group ($p = 0.10$) (Table 2). The area under the curve for the total and weighted scores in the overall cohort were 0.839 and 0.880, respectively (Table 3).

Table 1. Clinical characteristic of patients with ocular and non-ocular group

Characteristic	Overall (n=88)	Non-ocular (n=63)	Ocular (n=25)	p-value
Sex				0.6
Male	53 (60.2)	39 (73.6)	24 (26.4)	
Female	35 (39.8)	24 (68.6)	11 (31.4)	
Preterm				>0.9
No	84 (95.5)	60 (71.4)	24 (28.6)	
Yes	4 (4.5)	3 (75.0)	1 (25.0)	
Systemic comorbidities				0.2
No	85 (96.6)	62 (73.9)	23 (27.1)	
Yes	3 (3.4)	1 (33.3)	2 (66.7)	
Age (months)	32.3 ± 22.9	28.5 ± 22.2	42.1 ± 22.2	<0.001
Age stratification				0.002
12-24 months	47 (53.4)	41 (87.2)	6 (12.8)	
25-60 months	29 (33.0)	15 (51.7)	14 (48.3)	
> 60 months	12 (13.6)	7 (58.3)	5 (41.7)	

Data are presented as mean ± standard deviation or n (%)

* Fisher's exact test; Wilcoxon rank sum test

Table 2. Five-item questionnaire result in the age-stratified group

Item	Total			12-24 months			25-60 months			> 60 months		
	Non-ocular (N=63)	Ocular (N=25)	p-value	Non-ocular (N=41)	Ocular (N=6)	p-value	Non-ocular (N=15)	Ocular (N=14)	p-value	Non-ocular (N=7)	Ocular (N=5)	p-value
Q1			>0.9			>0.9			0.5			0.6
No	7(77.8)	2(22.2)		4(100.0)	0(0.0)		0(0.0)	1(100.0)		3(75.0)	1(25.0)	
Yes	56(70.9)	23(29.1)		37(86.0)	6(14.0)		15(53.6)	13(46.4)		4(50.0)	4(50.0)	
Q2			0.006			>0.9			0.4			0.5
No	38(84.4)	7(15.6)		31(86.1)	5(13.9)		5(71.4)	2(28.6)		2(100.0)	0(0.0)	
Yes	25(58.1)	18(41.9)		10(90.9)	1(9.1)		10(45.5)	12(54.5)		5(50.0)	5(50.0)	
Q3			0.1			0.7			0.7			<0.9
No	27(81.8)	6(18.2)		20(90.9)	2(9.1)		4(66.7)	2(33.3)		3(60.0)	2(40.0)	
Yes	36(65.5)	19(34.5)		21(84.0)	4(16.0)		11(47.8)	12(52.2)		4(57.1)	3(42.9)	
Q4			0.069			0.074			>0.9			0.2
No	58(75.3)	19(24.7)		39(90.7)	4(9.3)		12(50.0)	12(50.0)		7(70.0)	3(30.0)	
Yes	5(45.5)	6(54.5)		2(50.0)	2(50.0)		3(60.0)	2(40.0)		0(0.0)	2(100.0)	
Q5			<0.001			>0.9			<0.001			0.2
No	63(82.9)	13(17.1)		41(87.2)	6(12.8)		15(78.9)	4(21.1)		7(70.0)	3(30.0)	
Yes	0(0.0)	12(100.0)		0	0		0(0.0)	10(100.0)		0(0.0)	2(100.0)	
Score	1.9 ± 0.8	3.1 ± 0.8	<0.001	1.7 ± 0.7	2.2 ± 0.4	0.1	2.6 ± 0.5	3.5 ± 0.7	<0.001	1.9 ± 0.9	3.2 ± 0.4	0.009

Data are presented as mean ± standard deviation or n (%)

* Pearson's Chi-squared test; Fisher's exact test; Wilcoxon rank sum test

Table 3. Diagnostic performance of the questionnaire by age group

Subgroup	Variable	AUC (95% CI)	Delong's test for AUC comparison	Optimal cutoff					
				Threshold	Accuracy	Sensitivity	Specificity	PPV	NPV
Overall	Score	0.839 (0.755-0.924)	0.114	2.50	77.3 (68.2-86.4)	76.0 (60.0-92.0)	77.8 (66.7-87.3)	57.6 (46.2-71.4)	89.1 (82.1-96.2)
	Weighted Score	0.880 (0.799-0.960)		0.15	78.4 (69.3-86.4)	84.0 (68.0-96.0)	76.2 (65.1-85.7)	58.3 (47.7-70.6)	92.3 (85.5-98.1)
Age ≤ 24 months	Score	0.687 (0.552-0.822)	0.409	1.50	44.7 (31.9-57.4)	100.0 (100.0-100.0)	36.6 (22.0-51.2)	18.8 (15.8-23.1)	100.0 (100.0-100.0)
	Weighted Score	0.746 (0.564-0.928)		0.07	44.7 (31.9-57.4)	100.0 (100.0-100.0)	36.6 (22.0-51.2)	18.8 (15.8-23.1)	100.0 (100.0-100.0)
Age >24-≤ 60 months	Score	0.836 (0.701-0.970)	0.055	3.50	79.3 (65.5-89.7)	57.1 (28.6-78.6)	100.0 (100.0-100.0)	100.0 (100.0-100.0)	71.4 (60.0-83.3)
	Weighted Score	0.957 (0.905-1.000)		0.67	89.7 (79.3-100.0)	78.6 (57.1-100.0)	100.0 (100.0-100.0)	100.0 (100.0-100.0)	83.3 (71.4-100.0)
Age > 60 months	Score	0.943 (0.827-1.000)	0.394	2.50	91.7 (75.0-100.0)	100.0 (100.0-100.0)	85.7 (57.1-100.0)	83.3 (62.5-100.0)	100.0 (100.0-100.0)
	Weighted Score	0.971 (0.906-1.000)		0.33	91.7 (75.0-100.0)	100.0 (100.0-100.0)	85.7 (57.1-100.0)	83.3 (62.5-100.0)	100.0 (100.0-100.0)

AUC, area under the curve; CI, confidence interval; PPV, positive predictive value; NPV, negative predictive value.

Conclusion

The parent reported questionnaire demonstrated good diagnostic utility for discriminating ocular torticollis, especially in children older than 24 months. This tool may facilitate early identification of ocular torticollis in clinical setting where ophthalmologic evaluation is not readily accessible.