



Outcomes of Eye Examination and Vision Screening in Term Infants Presenting to a Tertiary Hospital in Türkiye

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Abstract

Objectives: Ophthalmic screening is an important part of the medical care of children as some eye abnormalities can lead to irreversible vision loss if not treated in the first few months or years of life. The aim of this study is to evaluate the outcomes of the ophthalmic screening program in term infants aged ≤ 1 year who presented to a tertiary hospital in Türkiye.

Materials and Methods: The records of 1,035 infants ≤ 1 year old who underwent ophthalmic screening between November 2019 and February 2022 were reviewed retrospectively. Demographic and medical details, parental complaints about the infants' eyes, family history of ocular, adnexal, and systemic pathologies, light reactions, red reflex test results, eye movements, blink response to light, fixation and following, noticeable strabismus, conjunctivitis, epiphora, anterior segment and fundus pathologies, and treatments applied were recorded. The referring physician (family physician, pediatrician) and reason for reference were also noted.

Results: Abnormal ophthalmological findings were detected in 136 infants (13.14%). The most common finding was congenital nasolacrimal duct obstruction (72.05%), followed by strabismus (8.82%), ptosis (4.41%), absence of following (3.67%), congenital cataract (2.94%), hemangioma of the adnexa (2.94%), nystagmus (2.94%), albino fundus (1.47%), preretinal hemorrhage (1.47%), and coloboma of the iris and choroid (1.47%). We detected abnormal red reflex in 4 infants who were not referred for red reflex abnormality by the referring physician, while another 4 infants referred for red reflex abnormality had no pathology on ocular examinations including the red reflex test.

Conclusion: The importance of ophthalmic screening in infants is well appreciated but there are inadequacies in performing and interpreting the red reflex test among family physicians and pediatricians. Efforts should be directed at improving vision screening skills, especially red reflex testing.

Keywords: Congenital cataract, national vision screening program, red reflex, retinoblastoma

Introduction

In Türkiye, pediatric ophthalmic screening has been carried out since its adoption in 2019 by the Ministry of Health. The screening aims to determine the risk factors that threaten the healthy growth and development of the eye and visual system as well as to identify infants and children with insufficient vision in the early period.^{1,2} It is an important part of the medical care of children because some eye abnormalities can lead to irreversible vision loss if not treated in the first few months or years of life. The American Academy of Ophthalmology and the American Academy of Pediatrics recommend visual assessment from birth and during all routine check-ups. Moreover, children who miss screening at the recommended time should be screened as soon as they are noticed.³ According to the circular issued by the General Directorate of Public Health, infants should be referred to an ophthalmologist immediately if they have any ocular problems detected in the screening examination, have known risk factors (prematurity; cerebral palsy; Down syndrome; family history of strabismus, amblyopia, and refractive error ≥ 5 diopters; metabolic disease; sensorineural hearing loss, especially Refsum disease; family history of congenital/infantile glaucoma and cataract; history of craniofacial abnormality, ptosis, hemangioma, and nasolacrimal duct pathology), or if the family suspect the child has an eye pathology.⁴ In Türkiye, ophthalmic screening by family physicians is recommended at 0-3 months, 36-48 months, and in the first year of primary school.² Ophthalmic screening at 0-3 months includes questioning about ocular and systemic risk factors and examination of the adnexa, light responses, and red reflex test.

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In this study we aimed to evaluate the reasons for referrals to ophthalmologists, examination results, and ocular problems in term infants ≤ 1 year of age referred to our clinic in accordance with the ophthalmic screening program.

Materials and Methods

The medical records of infants ≤ 1 year age who underwent ophthalmic screening between November 2019 and February 2022 were reviewed retrospectively. A total of 1,035 infants were included in the study. Infants referred for retinopathy of prematurity (ROP) were excluded.

Sex, date of birth, gestational age at birth, examination date, chronological age at examination, concomitant diseases, medications used, history of incubator care, parental complaints about the infant's eyes or vision (if any), family history of ocular, adnexal, and systemic pathologies, pupillary light responses, red reflex test results, eye movements, blink response to light flash, fixation and following, noticeable strabismus, conjunctivitis, epiphora, and anterior segment and fundus pathologies were recorded. Red reflex examinations were performed with a direct ophthalmoscope. Fundoscopic examinations were done after pupil dilation. Ophthalmologic examinations were performed by four ophthalmologists trained in pediatric retinal examination.

The referring physician (family physician, pediatrician) and reasons for referral (history of Lowe syndrome in brother, abnormal red reflex test, Horner syndrome, absence of following, albinism, and hemangioma of the adnexa) were recorded. Patients were divided into three groups according to their chronological age at the time of examination: 0-3 months, 3-6 months, and 6-12 months. The ocular pathologies, treatment applied, and follow-up examinations were noted.

The study was carried out in accordance with the principles of the Declaration of Helsinki and approved by the Local Institutional Ethics Committee of University of Health Sciences Türkiye, Kartal Dr. Lütfi Kırdar City Hospital (decision no: 2022/514/224/16; date: 27.04.2022). Signed informed consent was obtained from the parents of the infants.

Results

The study sample comprised 1,035 infants, including 502 girls and 533 boys. The mean gestational age at birth was 38.87 ± 1.24 weeks (range, 35-42 weeks). The mean age at the time of examination was 83.60 ± 58.81 days (range, 7-364 days).

Sixty infants had accompanying systemic diseases, 5 infants had coronavirus disease-2019, 195 infants had a history of incubator care, and 17 infants used medication. The demographic and clinical features of the patients are summarized in [Table 1](#).

Referrals were made by family physicians for 319 infants (30.82%) and by a pediatrician for 569 infants (54.98%). For 109 infants (10.53%), clear information could not be obtained from the family about which physician referred the infant for examination. The parents of 38 infants (3.67%) self-referred for ophthalmologic examination because they noticed an eye problem in their infants. These problems included

facial asymmetry, conjunctivitis, tearing, strabismus, ptosis, nystagmus, leukocoria, family history of cataract, strabismus, and heterochromia.

Of the 1,035 infants, 136 (13.14%) had abnormal ophthalmological findings. Congenital nasolacrimal duct obstruction (CNLDO) was the most common finding and was detected in at least one eye of 98 infants. This accounted for 72.05% of all abnormalities and 9.46% of all screened infants. Strabismus was the second most common finding, detected in 12 infants (11 with esotropia, 1 with exotropia), accounting for 8.82% of all abnormalities and 1.15% of all screened infants. Other abnormalities were ptosis (6; 4.41%), absence of following (5; 3.67%), congenital cataract (4; 2.94%), hemangioma of the adnexa (4; 2.94%), nystagmus (4; 2.94%), albino fundus (2; 1.47%), preretinal hemorrhage (2; 1.47%), coloboma of iris and choroid (2; 1.47%), heterochromia (1; 0.73%), corneal opacity (1; 0.73%), chalazion (1; 0.73%), retinal white changes (1; 0.73%), corneal dystrophy (1; 0.73%), intraocular tumor

Table 1. Demographic and clinical features of the infants screened in our clinic

Sex, n (%)	
Male	533 (51.50%)
Female	502 (48.50%)
Gestational age at birth, mean (SD) [range], weeks	38.87 (1.24) [35-42]
Age at screening, mean (SD) [range], days	83.60 (58.81) [7-364]
Comorbidity, n (%)	
Congenital hypothyroidism	60 (5.79%)
Hearing loss	10 (0.96%)
Anal atresia	2 (0.19%)
Diabetes mellitus	1 (0.09%)
Epilepsy	1 (0.09%)
Renal pathologies	3 (0.28%)
Cardiac disease	9 (0.86%)
Hip dislocation	8 (0.77%)
Hydrocephaly	1 (0.09%)
Scaphocephaly	2 (0.19%)
Trigonocephaly	1 (0.09%)
Microcephaly	1 (0.09%)
Systemic toxoplasmosis	1 (0.09%)
Systemic cytomegalovirus infection (West syndrome)	1 (0.09%)
COVID-19 infection	5 (0.48%)
Lowe syndrome	1 (0.09%)
Rett syndrome	1 (0.09%)
Sandhoff disease	1 (0.09%)
Joubert syndrome	1 (0.09%)
Down syndrome	1 (0.09%)
Horner syndrome	2 (0.19%)
Brachial plexus damage	2 (0.19%)
Biotinidase deficiency	1 (0.09%)
Corpus callosum agenesis	1 (0.09%)
Cleft lip and palate	1 (0.09%)
Cleft lip	1 (0.09%)
History of incubator care, n (%)	195 (18.84%)
Medication use, n (%)	17 (1.64%)
n: Number of patients, SD: Standard deviation, COVID-19: Coronavirus disease-2019	

(1; 0.73%), facial asymmetry (1; 0.73%), conjunctivitis (1; 0.73%), cherry red spot (1; 0.73%), megalocornea (1; 0.73%), optic disc hypoplasia (1; 0.73%), and suspected congenital glaucoma (1; 0.73%). Eleven infants had more than one ocular pathology.

Intervention was required in 7 infants, which accounted for 5.14% of all abnormalities and 0.67% of all infants screened. Two infants had cataract surgery, 1 infant with corneal opacity had corneal debridement, 1 infant with intraocular tumor diagnosed as retinoblastoma received intravitreal chemotherapy, and 1 infant with CNLDO in the left eye underwent probing while another infant with CNLDO underwent bilateral probing and right balloon dilatation. In addition, 1 infant with corneal dystrophy underwent ocular examination under general anesthesia. Five infants were given spectacles and occlusion therapy was prescribed for at least one eye as needed. Once the ophthalmologic diagnosis was made, 4 infants were referred to a pediatric neurologist for testing for possible systemic disease. Demographic data of the patients with ocular abnormality and the interventions done are summarized in [Table 2](#).

Discussion

Although infant ophthalmic examinations are routinely performed in Türkiye as part of the national screening program, there are no studies evaluating the results of this program in term infants. To the best of our knowledge, our study is the first to present the ophthalmologic examination results obtained within the scope of the National Ophthalmic Screening program in our country.

We divided our patients into three groups according to their chronological age at the time of examination: 0-3, 3-6, and 6-12 months. The vast majority of the infants screened were 0-3 months of age. This could be attributable to parents understanding the importance of the screening program and showing good compliance, as well as to effective implementation of the screening program in our country.

In the literature there are various publications regarding the results of neonatal eye screening, with abnormality rates ranging from 4.7% to 41.2%.^{5,6,7,8} Our data revealed that a significant proportion (13.14%) of infants younger than 1 year of age exhibited ocular abnormalities. Similar rates were reported by Jac-

Table 2. Demographic and clinical data of the patients with ocular abnormality and interventions done

Ocular diagnosis	Frequency in screened infants, n (%)	Mean age \pm SD (range), days	Previous ophthalmological examination, n	Intervention (surgical/medical/follow-up) n
Cataract	4 (0.38%)	57.25 \pm 46.97 (7-111)	1	2/0/4
Dacryocystitis	98 (72.05%)	80.61 \pm 51.61 (27-346)	22	2/22/98
Esotropia	11 (1.06%)	159.63 \pm 71.71 (43-278)	7	0/6/11
Exotropia	1 (0.09%)	140	1	0/1/1
Retinal white changes	1 (0.09%)	82	0	0/0/1
Ptosis	6 (0.57%)	108.50 \pm 109.31 (52-279)	3	0/0/6
Hemangioma on the lids	4 (0.38%)	63.50 \pm 28.89 (33-102)	2	0/0/4
Conjunctivitis	1 (0.09%)	115	0	0/1/1
Optic disc hypoplasia	1 (0.09%)	177	0	0/0/1
Corneal dystrophy	1 (0.09%)	99	1	0/1/1
No following	5 (0.48%)	110.60 \pm 63.46 (70-209)	3	0/0/5
Facial asymmetry	1 (0.09%)	118	0	0/0/1
Suspect congenital glaucoma	1 (0.09%)	113	0	0/0/1
Iris and choroid coloboma	2 (0.19%)	144.00 \pm 26.87 (125-163)	0	0/0/2
Megalocornea	1 (0.09%)	288	0	0/0/1
Corneal opacity	1 (0.09%)	244	1	1/1/1
Nystagmus	4 (0.38%)	156.25 \pm 72.18 (70-242)	3	0/1/4
Chalazion	1 (0.09%)	85	1	0/1/1
Albino fundus	2 (0.19%)	189.00 \pm 74.95 (136-242)	2	0/1/2
Heterochromia	1 (0.09%)	98	1	0/0/1
Preretinal hemorrhage	2 (0.19%)	39.50 \pm 9.19 (33-46)	0	0/0/2
Cherry red spot	1 (0.09%)	60	1	0/0/1
No red reflex/exophytic mass in fundus	1 (0.09%)	87	1	0/1/1

n: Number of patients, SD: Standard deviation

Okereke et al.⁹ (15.5%) and Goyal et al.⁶ (14.93%). In contrast, Li et al.⁵ and Ma et al.⁸ found higher rates of ocular abnormality (24.4% and 41.2%, respectively), while Vinekar et al.⁷ reported a lower rate of 4.7%. These studies included heterogeneous groups in terms of chronological age at examination, which may explain the variable results. All the authors performed dilated fundus examination with digital widefield retinal imaging, whereas Jac-Okereke et al.⁹ did not perform fundus examination during vision screening. We included only term infants in our study, excluded preterm infants referred for ROP examination, and performed dilated fundus examination on all infants.

CNLDO, the most common ocular abnormality in our study, was detected in 98 infants and was bilateral in 36 infants. The disease accounted for 9.46% of 1,035 infants screened and 72.05% of 136 infants with ocular abnormality. In 8 infants, CNLDO was accompanied by preretinal hemorrhage, eyelid hemangioma, strabismus, coloboma of the iris and choroid, suspected congenital glaucoma, absence of following, optic disc hypoplasia, nystagmus, or sutural cataract. The prevalence of CNLDO in our study seems higher than reported in previous vision screening studies. Ma et al.⁸ detected CNLDO in 4 (0.8%) of 481 infants. Jac-Okereke et al.⁹ examined 142 infants up to 12 months old and detected CNLDO in 14%. CNLDO occurs in 5-20% of newborns and often resolves spontaneously or with conservative treatment, although persistent cases require surgical treatment.^{10,11,12} Medical treatment consists of massaging the nasolacrimal sac and applying topical antibiotics when discharge is present. We explained lacrimal sac massage to the parents of all infants with CNLDO, assuming they either did not know it or had not performed it effectively. We prescribed topical antibiotics to the infants as needed. Two infants (2.04%) underwent surgical treatment, but we cannot say whether the massage actually improved the recovery rate because we are not certain how parents performed the massage.

Strabismus, the second most common finding in our study, was detected in 12 infants, accounting for 8.82% of all abnormalities and 1.15% of all screened infants. None of our patients required strabismus surgery. They were prescribed spectacles and occlusion therapy as needed.

Ptosis is another ocular abnormality which we encountered frequently. Treatment of congenital ptosis is indicated when the upper eyelid obscures the visual axis, causing stimulus deprivation, or induces astigmatism that is amblyogenic.¹³ In our study, none of the 6 patients with congenital ptosis required surgical intervention.

Tang et al.¹⁴ examined 199,851 neonates by RetCam imaging and found that approximately 9% (18,198) of the infants had abnormal findings, with retinal hemorrhage being the most common (12,810, 70.39%). Callaway et al.¹⁵ reported the prevalence of fundus hemorrhages as 20.3%. In the study by Ma et al.⁸, retinal white areas were the most common abnormality (42.9% of abnormalities and 17.7% of all screened infants), followed by retinal hemorrhage (16.2% of abnormalities and 6.7% of all screened infants). In our study, only 2 infants had preretinal hemorrhages, accounting for

1.47% of all abnormalities and 0.19% of all screened infants. Retinal hemorrhages in infancy are often self-limited and resolve quickly with no effect on visual development, although some can persist and have a long-term impact on vision.¹⁶ Some authors hypothesize that any obstruction of the visual axis that persists for a sufficient period can induce amblyopia that appears later in life without ophthalmoscopic findings, referred to as idiopathic.⁵ Therefore, detection of retinal hemorrhages may be a warning sign that should prompt vigilance for visual problems that may appear later in life. This is only possible with dilated fundus examination. However, to advocate for detailed fundus examinations in every newborn would have staffing, economic, and logistic implications. Each country must evaluate and implement the details of the ophthalmological examinations to be carried out within the scope of national eye screening programs.

Red reflex testing is a simple screening test that can be performed by anyone and enables early diagnosis of vision-threatening and sometimes life-threatening ocular pathologies such as cataract and retinoblastoma.^{3,17,18,19} Although the definitive diagnosis of these diseases is made by dilated fundus examination, the red reflex test helps identify patients who need a detailed fundus examination. In our study, 4 infants (0.38%) had abnormal red reflex test, accounting for 2.94% of all abnormalities. However, none of these infants were referred for red reflex abnormality. In one of these infants, the reason for referral by their pediatrician was a history of Lowe syndrome in his brother. Three of four infants had cataract in at least one eye and two of them underwent cataract surgery. According to our study and studies concerning ophthalmic screening in infants, the red reflex test is sufficient for screening congenital cataract, especially those that require surgery.¹⁷ In Sweden, where routine eye screening in the maternity ward is recommended, congenital cataract is detected earlier than in Denmark, where screening is done at 5 weeks of age with a penlight.^{20,21} However, dilated ophthalmologic examination is needed to diagnose some cataracts. In our study, one infant with sutural cataract exhibited a normal red reflex and was diagnosed by dilated fundus examination.

Another disease that can be detected by red reflex examination is retinoblastoma.²² In our study, retinoblastoma was detected in the left eye of one infant who had an abnormal red reflex. She was 87 days old, giving the advantage of early intervention, and she was treated with intravitreal chemotherapy. It has been estimated that about 40% of infants with neonatal retinoblastoma have a positive family history, which itself should prompt referral for a pediatric ophthalmologic examination.²³ However, this infant did not have family history.

In Türkiye, there are no data regarding the ratio of congenital cataract and retinoblastoma that are detected by red reflex examination. Gürsel Özkurt et al.²⁴ questioned family physicians about their knowledge and practice of red reflex screening. They reported that 12% of the respondents had never heard of the red reflex test, 12% did not know how to use a direct ophthalmoscope, 33% knew this test should be performed in

every infant, 36% knew about but had never performed the test, and 16% performed it regularly.

In our study, of 997 infants (96.33%) referred by either a family physician or pediatrician, 4 were referred due to abnormal red reflex test. However, these infants exhibited normal red reflexes in our ophthalmologic examination, and two of them were diagnosed with conjunctivitis and CNLDO.

Our study demonstrates that the importance of the screening program is well understood among physicians and parents, but almost all of the screening tests are performed by ophthalmologists instead of primary care physicians. We examined the patients with an indirect ophthalmoscope, which is time-consuming compared to digital imaging systems. These differences influenced the results of our study. Our study also showed some differences in the diagnoses when compared with similar studies. The most common ocular abnormality in our study was CNLDO, while retinal pathologies were rare (8.08%). Although we excluded infants referred for ROP screening, we included all term infants, even those with infectious disease and signs of systemic disease.

Study Limitations

Our study has some limitations. It was a single-center study with a relatively small sample size. Although our sample size was adequate to detect common ocular pathologies, a larger sample is needed to detect rare ocular abnormalities. Further studies involving multiple centers with large samples and long follow-up times are needed to discuss the prevalence, etiology, clinical course, and prognosis of ocular abnormalities and document patterns of regression. Though our study did not aim to document follow-up results, the impact of the pathologies on quality of life and visual development could have provided additional information and allowed us to see the impact of ocular abnormalities on the visual system.

Conclusion

Our study showed that although the importance of red reflex test is well appreciated, there are inadequacies in performing and interpreting this test among family physicians and pediatricians. Thus, it is important to encourage these clinicians to perform all steps of the ophthalmic screening, and efforts should be directed at improving ophthalmic screening skills (particularly red reflex testing) among physicians who see children for infant and well-child visits. Another issue is whether dilated fundus examination should be performed even in healthy infants to detect certain pathologies. Although the exact answer of who should be examined in detail may vary according to each country's politics and patients, further studies will be more instructive. The outcomes would help in formatting guidelines for the scalability of the nationwide expansion of this program planned by the government.

Ethics

Ethics Committee Approval: Local Institutional Ethics Committee of University of Health Sciences Türkiye, Kartal Dr.

Lütfi Kırdar City Hospital (decision no: 2022/514/224/16; date: 27.04.2022).

Informed Consent: Signed informed consent was obtained from the parents of the infants.

Declarations

Authorship Contributions

Surgical and Medical Practices: N.Z.K., A.Y.K., Concept: N.Z.K., Design: N.Z.K., Data Collection or Processing: N.Z.K., A.T.K., S.Ö.Y., D.Ç.Y., Analysis or Interpretation: N.Z.K., R.D.G., B.T., Literature Search: N.Z.K., R.D.G., Writing: N.Z.K.

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