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Etiology of childhood blindness in Izmir, Turkey

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PURPOSE. *To identify the anatomic sites and the etiology of childhood blindness and to discern treatable and preventable causes.*
METHODS. *The records of 998 patients seen in the authors' pediatric ophthalmology unit between June 1998 and May 2002 were examined retrospectively. A total of 148 patients who had visual impairment and blindness according to World Health Organization criteria were included in the study. They are classified according to the etiology based on time of insult and the anatomic site of visual loss.*
RESULTS. *The most common anatomic site of visual loss was retina, with a rate of 25.0%. The etiology according to the time of insult was unknown in 45.2% of the patients, of whom 20.2% had cataract. Genetic disorders were responsible in 25.0% of the patients. In 69.6% of the patients, the causes of visual impairment were considered either preventable or treatable, including cataract, retinopathy of prematurity, genetic disorders, and refractive errors.*
CONCLUSIONS. *A high percentage of our patients had avoidable causes of childhood blindness. Genetic counseling services, as well as national screening programs for amblyopia, red fundus reflex, and retinopathy of prematurity, should be established. (Eur J Ophthalmol 2004; 14: 531-7)*

KEY WORDS. *Childhood blindness, Congenital cataract, Retinopathy of prematurity*

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INTRODUCTION

For a child who is born blind or becomes blind later in childhood, the loss of productivity and the number of years of disability are greater than for an adult who becomes blind. Many causes of childhood blindness are also causes of childhood mortality, such as prematurity, measles, vitamin A deficiency, and meningitis, so the control of blindness is closely related to the child's survival (1). Moreover, these children require special education and rehabilitation programs in order to have an independent life. These requirements involve responsibilities and additional costs for society.

In order to set priorities for prevention programs, baseline data about the major causes of childhood blindness are required. Our aim was to identify the etiology and the anatomic sites of childhood blindness and to discern preventable and treatable causes.

MATERIALS AND METHODS

The records of 998 patients seen between June 1998 and May 2002 in our Paediatric Ophthalmology and Strabismus Unit were examined retrospectively. Best-corrected visual acuity was measured using the Snellen chart or Teller acuity cards where possible. Binocular fixation pat-

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tern (central, steady, maintained) was taken into account in patients whose visual acuity could not be measured. Signs of low vision such as nystagmus, ocular bobbing, or oculodigital massage were noted.

All patients had anterior segment examination by a penlight, hand biomicroscope, or biomicroscope according to age. The pupils were dilated with tropicamide 1% and cyclopentolate 1%. The posterior segment was examined by an indirect ophthalmoscope and by ultrasonography when required. The cycloplegic refractions and deviations were noted.

According to World Health Organization categories (2), patients who had vision less than 0.3 by Snellen chart in their better eye were included in the study (Tab. I). Patients who had low vision according to age by Teller cards, who had no central fixation, and who had signs of very poor vision like searching nystagmus and oculodigital massage were also included. The last group had serious ocular pathologies in both eyes. The pathology that had the greatest effect on vision was taken into consideration in patients with multiple ocular pathologies. Patients who had low vision in one eye caused by trauma were excluded because of good vision in the fellow eye. Electrophysiologic tests could not be performed because related equipment was not available in our clinic at the time.

RESULTS

A total of 148 patients (14.8%) were included in the study, of whom 57 (38.5%) were female and 91 (61.4%) were male. The mean age of the children was 36.6 months with a range between 20 days and 194 months.

Visual acuity was measured by Snellen chart in 44 (29.7%) patients and by Teller cards in 42 (28.3%) patients. In 62 (41.9%) patients visual acuity could not be measured but they had nystagmus, oculodigital massage, loss of fixation, and ocular pathologies that cause severe visual impairment such as grade 5 retinopathy of prematurity (ROP) or optic atrophy.

TABLE I - WORLD HEALTH ORGANIZATION CRITERIA OF VISUAL IMPAIRMENT

WHO category	Visual acuity
No impairment	6/6–6/18
Moderate visual impairment	<6/18–6/60
Severe visual impairment	<6/60–3/60
Blind	<3/60–no perception of light
Could not be tested (believed blind)	

Blindness by anatomic site

Retina (25%) was the most frequently involved site with ROP (9.4%) as the major cause. The other causes were macular dystrophies (7.4%), albinism (4.0%), Leber congenital amaurosis (2.0%), toxoplasma retinochoroiditis (1.3%), and retinoblastoma (0.6%). Among the patients with lenticular disorders (22.9%), 20.1% had aphakia or pseudophakia following surgery of congenital cataract. Thirty-three (22.2%) patients had cortical visual impairment (CVI), which comprised mostly magnetic resonance imaging findings such as cortical atrophy, lissencephaly, and periventricular leukomalacia. Optic nerve atrophy and hypoplasia were present in 10.8% of the patients. Uveal disorders such as uveitis or coloboma (6.0%), refractive errors (4.7%), idiopathic nystagmus (3.3%), sclerocornea (2.0%), and glaucoma (0.6%) were the next in frequency (Tab. II).

Etiology according to the time of insult leading to visual impairment

Genetic disorders were identified in 25% of the patients, mostly Down syndrome (8.1%), albinism (4.0%), and Leber congenital amaurosis (2.0%). The leading disorder related to the prenatal/perinatal period was ROP (9.4%). Other disorders related to this period were hypoxia (4.7%), infection (2.7%), and occipital cortical atrophy (2.7%).Disorders related to the postnatal period were neoplasm (2.7%) and meningitis (1.3%). The time of insult was unknown in 45.2% of the patients of whom 20.2% had cataract, 5.4% had coloboma, and 4.7% had refractive errors.

Avoidable causes

Sixty-two (41.9%) patients had treatable causes of visual impairment including cataract (20.2%), lens subluxation (0.6%), ROP (9.4%), refractive errors (4.7%), hydrocephalus (1.3%), glaucoma, craniopharyngioma, and uveitis (2.0%). Forty-one (27.7%) patients had preventable causes such as prenatal/perinatal infections (2.7%) and genetic diseases (25%), although some of them are not yet preventable with genetic counseling (Tab. III).

Additional ocular findings included nystagmus in 61 (41.2%) patients and strabismus in 46 (31.0%) patients, of whom 30 had esotropia and 16 had exotropia. The most common associated systemic pathology was motor and/or mental retardation (12.8%, 19 patients). Ten patients each had one of the following additional findings: meningoencephalitis, epilepsy, cleft palate and lip, hydrocephalus, pulmonic stenosis and patent ductus arteriosus, facial asymmetry, multicystic kidney, esophageal atresia, cardiomegalia, and macroglossia (Tab. IV).

TABLE II - ANATOMIC SITE OF ABNORMALITY LEADING TO VISUAL IMPAIRMENT

Anatomic site	N	%
Retina	37	25.0
ROP	14	9.4
Macular dystrophy	11	7.4
Albinism	6	4.0
Leber congenital amaurosis	3	2.0
Toxoplasmic retinochoroiditis	2	1.3
Retinoblastoma	1	0.6
Lens	34	22.9
Aphakia	17	11.4
Pseudophakia	7	4.7
Aphakia and glaucoma	6	4.0
Cataract	3	2.0
Lens subluxation	1	0.6
Cortical	33	22.2
MRI findings*	15	10.1
MMR	13	8.7
Cerebral palsy	3	2.0
Infection	2	1.3
Optic nerve	16	10.8
Optic nerve hypoplasia	6	4.0
Primary optic atrophy	5	3.3
Secondary optic atrophy	5	3.3
Hydrocephalus	2	1.3
Craniopharyngioma	1	0.6
Optic glioma	1	0.6
Glioblastoma	1	0.6
Uvea	9	6.0
Coloboma	8	5.4
Uveitis	1	0.6
Cornea	3	2.0
Sclerocornea	3	2.0
Globe	2	1.3
Microphthalmos	2	1.3
Glaucoma	1	0.6
Buphthalmos	1	0.6
Others	12	8.1
Idiopathic nystagmus	5	3.3
High myopia	2	1.3
High hypermetropia	5	3.3

*Atrophy, leukomalacia, and lissencephaly.
ROP = Retinopathy of prematurity; MMR = Motor and mental retardation

DISCUSSION

Currently it is estimated that there are about 1.5 million blind children worldwide, and of the 500,000 children who become blind each year, 60% die from the diseases that

TABLE III - AVOIDABLE CAUSES OF VISUAL IMPAIRMENT

Causes	N	% of all children
Treatable	62	41.9
Cataract/subluxation	34	22.9
ROP	14	9.4
Refractive errors	7	4.7
Hydrocephalus	2	1.3
Meningitis	2	1.3
Glaucoma	1	0.6
Uveitis	1	0.6
Craniopharyngioma	1	0.6
Preventable	41	27.7
Genetic diseases	37	25.0
Prenatal/perinatal infections	4	2.7

ROP = Retinopathy of prematurity

TABLE IV - ADDITIONAL FINDINGS

Additional findings	N	% of all children
Nystagmus	61	41.2
Strabismus	46	31.0
Esotropia	30	20.2
Exotropia	16	10.8
Motor-mental retardation	19	12.8
Others*	10	6.7

*Meningoencephalitis, epilepsy, cleft palate and lip, hydrocephalus, pulmonic stenosis and patent ductus arteriosus, facial asymmetry, multicystic kidney, esophageal atresia, cardiomegalia, macroglossia

resulted in their blindness (3, 4). Besides the emotional, social, and economic costs of childhood blindness to the child, the family, and society, the causes of blindness are also related to childhood mortality. Another aspect that emphasizes the urgency of prevention of childhood blindness is that for normal visual maturation, clear images should be transmitted to the higher visual centers. In case of failure of normal visual maturation, vision cannot be corrected in adult life; therefore attempts should be made for early diagnosis and treatment (1). Causes and prevalence of childhood blindness vary according to geographic localization with a nearly tenfold range from 0.1/1,000 children in wealthy countries to 1.1/1,000 children in economically disadvantaged countries (5). It is thought that socioeconomic development and the level of health care services play a major role in the pattern of the causes. According to the World Health Organization, half of the 1.5 million blind children have avoidable causes, including measles, ophthalmia neonatorum, vitamin A deficiency, and harmful traditional eye treatments (3).

In Africa, corneal scarring is the main cause of childhood blindness, which could be secondary to vitamin A deficiency, malnutrition, or measles (6). Dandona et al reported that, in India, one third of blindness is due to refractive error and 16.6% is due to preventable causes such as vitamin A deficiency and amblyopia after cataract surgery (7). In a former study from India, the major causes were identified as hereditary macular degeneration, retinitis pigmentosa, and albinism (8).

In highly industrialized countries of Europe, the leading causes of childhood blindness are lesions of the central nervous system, congenital anomalies, and retinal disorders, while in the middle-income countries of Europe, congenital cataract, glaucoma, and mainly ROP are more common (9). Hereditary diseases are frequent in the eastern Mediterranean region because of the high rate of consanguinity (16 to 55%) (6).

As survival rates of preterm and low birthweight babies increase, an increasing number of infants will be at risk of developing ROP. According to Gilbert et al, the highest rates of childhood blindness due to ROP are seen in the middle-income countries, which have developing neonatal intensive care services but limited or no screening and treatment system for ROP (10). Blindness due to ROP is unusual in countries where neonatal care facilities are not available or in industrialized countries with established screening and treatment systems for ROP. In the two blind school studies performed in the 1980s in Turkey, one of which took place in the same area as our study, ROP was not a cause of childhood blindness (11, 12). Currently, with an ROP rate of 9.4%, Turkey is between the industrialized and middle-income countries; however, this is not a population-based study and therefore may not reflect the whole spectrum of visually impaired children in Turkey. The increasing number of *in vitro* fertilization clinics in our country is also important since infants conceived through fertility programs have a higher incidence of multiple births and prematurity and thus have a higher risk of ROP (13, 14). Because both the incidence and the severity of ROP decrease as birthweight and gestational age increase, attempts should be made to delay birth (15, 16). We believe that a national screening program for ROP should be established.

Congenital cataract was identified in 20.2% of the patients, of whom 70% had aphakia following cataract surgery. In the two former reports from the 1980s in Turkey, congenital cataract rates were 16.9% and 21.7% (11, 12). Compared to the other countries, congenital cataract seems to be a problem for Turkey. The rates of congenital cataract cases in different countries are summarized in Table V (4, 17-22). In order to compare the results of childhood blindness surveys in different countries, there should be an agreement on definitions and classifications (23, 24). The reason for the important role of congenital cataract is partly late diagnosis and treatment, as well as insufficient postoperative visual rehabilitation, which is as important as the operation itself. A screening program for red fundus reflex might be a simple but effective step in the early diagnosis of congenital cataract. According to Wirth et al, examination of the parents is also important in order to avoid unnecessary further investigations in hereditary cataract cases (25).

According to worldwide data from blind school studies, it is thought that hereditary eye diseases are more frequent in industrialized countries (6). In most countries, autosomal recessive diseases predominate (22–54%) and retinal dystrophies are common (42–80%) (26). The high rate of genetic diseases in our study is estimated to be related to the high rate of consanguinity in our country. The rates of genetic diseases as causes of childhood blindness in different countries are summarized in Table VI (4, 16-21, 27). Genetic counseling would be of great benefit to prevent genetic diseases. A detailed family history and examination of the family members are necessary to determine the mode of inheritance and to calculate the risks in subsequent pregnancies in order to inform the parents (6).

CVI has emerged as the leading cause of low vision in children in developed countries (28). Many children with CVI have additional neurologic problems such as cerebral palsy, mental retardation, epilepsy, learning problems, and meningomyelocele (29). Guzzetta et al reported that severe visual deficits are present in 7 to 9% of children with cerebral palsy (30). In the study of Huo et al, perinatal hypoxia was the most common cause of CVI and it is mentioned that the majority of children with CVI showed at least some recovery (28, 31).

According to Casteels et al, normal MRI correlates with better visual outcome regardless of the gestational age at which the insult occurred and the presence of infarcts or periventricular leukomalacia indicates that full visual recovery will not occur (32). In our study, 22.9% of the patients had CVI, which comprised mostly MRI findings such as cortical atrophy, lissencephaly, and periventricular leukomalacia. Perinatal hypoxia was found in 4.7% of the patients.

Worldwide, corneal scarring seems to be the most important cause of avoidable blindness (1). In our study, corneal scarring did not cause low vision, implying that primary health services work well and malnutrition is not a problem in Turkey. In the "Vision 2020—The right to sight" program, the World Health Organization stated the following targets to control childhood blindness: measures for disease control, human resources development, and appropriate technology and infrastructure development. The program identifies training of pediatric ophthalmologists in developing countries as one of the strategies to control childhood blindness (1, 33). In June 2002, the World Health Organization launched another project for the prevention of childhood blindness (34). A great international attempt has been

TABLE V - BLINDNESS BY ANATOMIC SITE IN DIFFERENT COUNTRIES (4, 17-22)

Region	N	Cornea	Lens	Retina	Opticnerve	Glaucoma	Others
PR of China (4)	1,131	4.4	18.8	24.9	13.6	9.0	29.3
Czech Republic (17)	229	1.8	8.7	54.2	15.3	—	20.0
Malaysia (18)	358	15.1	22.3	20.8	8.7	7.2	25.9
Nigeria (19)	140	21.4	31.4	7.9	7.2	9.3	22.8
Mongolia (20)	80	7.5	30.0	17.5	12.5	3.8	28.8
Ethiopia (21)	295	62.4*	9.2	2.4	9.8	1.7	14.5
Nordic (22)†	2,527	0.5	10.0	28.2	24.3	0.9	36.0
Turkey (current study)	148	2.0	22.9	25.0	10.8	0.6	38.7

Values are percentages
*Cornea/phthisis
†Denmark, Finland, Iceland, and Norway

TABLE VI - ETIOLOGY ACCORDING TO THE TIME OF INSULT IN DIFFERENT COUNTRIES (4, 17-21, 25)

Region	N	Genetic	Intrauterine	Perinatal	Childhood	Unknown
PR of China (4)	1,131	30.7	0.1	2.3	14.0	52.9
Czech Republic (17)	229	9.2	0.4	43.7	4.4	42.3
Malaysia (18)	358	29.5	4.5	9.0	7.8	49.1
Nigeria (19)	140	14.9	7.9	0	38.6	38.6
Mongolia (20)	80	30.0	0.0	6.3	16.3	47.5
Ethiopia (21)	295	3.4	0.7	1.0	49.8	45.1
Nordic (25)*	2,527	35.0	30.8	21.6	6.7	5.9
Atlanta/USA (16)	228	16.0	27.0	27.0	8.0	22.0
Turkey (current study)	148	25.0	2.7	22.9	4.0	45.2

Values are percentages.
*Denmark, Finland, Iceland, and Norway

made at implementing prevention programs.

In conclusion, 69.6% of our patients had visual impairment caused by potentially preventable or treatable diseases, including congenital cataract, ROP, and genetic diseases. Fundus examination is of great importance in the examination of children, since retina was the most involved anatomic site in our study. National screening programs for amblyopia, red fundus reflex, and ROP should be established. Genetic counseling and educating and informing both the parents and society are crucial. By cooperation of pediatricians and ophthalmologists as a team and the development of primary health services, early diagnosis and treatment are possible.

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