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The Ophthalmic Anomalies in Children with Down Syndrome in Split-Dalmatian County

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A B S T R A C T

Our aim was to present the ophthalmic anomalies in patients with Down syndrome in Split-Dalmatia County born from 1992 until 2009 year. It was a cross-sectional study. 153 children with Down syndrome aged 0–18 years from the Split-Dalmatia County were examined. One hundred twelve participants were borne in Split,13 in Vrgorac,16 in Makarska, 12 in Sinj. All enrolled children underwent a complete ophthalmological examination (anterior segment, ocular motility, refractive status, fundus, measuring intraocular pressure (IOP). Of 89.5% percent of responders with refractive errors, 48.1% had myopia, 35.0% had hypermetropia, astygamtism in 16.7%, 28.7% strabismus, nystagmus (8.4%), cataracts (1.3%), glaucoma (1.9%), supernumerary optic disc vessels (24.1%) and keratoconus (1.3%). Conclusion: In patients with Down syndrome the prevalence of refractive errors (myopia prevalence), as well as other ophthalmological diseases was determined.

Key words: *Down syndrome, eye manifestations, children, caucasian race, Croatia*

Introduction

Down syndrome is the most frequent chromosomal anomaly found in live-births, total incidence in general population is 1 *per* 600–800 births, rising with the mother's age. The major clinical features of Down syndrome are found on the head: brachycephaly and microcephaly, flat faces with hypertelorism, depressed nasal bridge, flat occiput, and broad short neck, epicantus, protrusion of the tongue (*lingua scrotalis* or *lingua geographica*), small deformed auricles. Other organs can be also affected: hypothyreosis, congenital heart defects, duodenal atresia, Hirschsprung disease and mental retardation (mild to moderate) with delaye speech development. The children are short in stature, prone to obesity, as well as to diabetes and other autoimmune diseases as celiakia and Hashimoto thyroiditis[1–3].

Ophtalmic anomalies are frequent too. Hypertelorism, epicanthus, Brushfield's spots (multiple white spots on the iris), cataract are the most common in Down syndrome[1–3].

Accentuated epicanthal folds and narrow and tilted eyelid apertures, vary according to race. Reported incidence of various signs of Down's syndrome, such as refractive anomalies, strabismus, keratoconus (which increases with patients age), cataract, and Brushfield's spots vary according to different authors[4–8].

Supernumerary optic disc vessels which go over the edge of the optic disc are found in 11–38% of cases. Inflammations of the conjunctiva, eyelids, and the lacrymal ducts are frequent too[4–8].

The aim of this study is to compare the ophthalmological anomalies and diseases in persons with Down syndrome born from 1992 until 2009 in the Split-Dalmatian County with results reported from other authors (Table 1). Having this analisys we can better organize our ophtalmologycal outpatient clinics for children and people having Down syndrome in Croatia.

Matherials and Methods

153 children with Down syndrome have been examined. The number of children with Down syndrome have

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TABLE 1
A PARALLEL SURVEY OF OUR RESEARCH AND THOSE PERFORMED BY OTHER AUTHORS

Study	Karlica et al. 2009 N%	Shapiro and France 1985 N%	Caputo et al. 1989 N%	Berk et al. 1996 N%	Da Cunha and Moreira N%
Number of examinees	153	53	187	55	152
Country	Republic of Croatia, Split	USA	USA	Turkey	Brazil
Age span	0–18	7–36	0–26	0–25	0–18
Epicantus	64 (41.8)	–	–	13 (24)	92 (61)
Refraction errors	137 (89.5)	47 (88)	122 (65)	55 (100)	149 (98)
Hypermetropia	48 (35.0)	17 (36)	39 (32)	24 (44)	39 (26)
Myopia	66 (48.1)	18 (38)	42 (34)	7 (12)	19 (13)
Astigmatism	23 (16.7)	12 (26)	41 (34)	24 (44)	91 (61)
Emetropia	–	–	–	–	–
Nistagmus	13 (8.4)	5 (9)	55 (29)	7 (13)	28 (18)
Strabismus	44 (28.7)	22 (96)	107 (57)	12 (22)	57 (38)
Divergent	8 (18.1)	–	4 (4)	0	0
Convergent	36 (81.9)	22 (96)	97 (91)	11 (92)	51 (90)
Cataract	11 (7.1)	7 (13)	21 (11)	11 (20)	20 (13)
Keratoconus	2 (1.3)	8 (15)	–	–	–
Superfluous blood vessels of the retina	37 (24.1)	–	–	21 (38)	42 (28)
Glaucoma	3 (1.9)	–	10 (5)	–	–

been obtained from Down syndrome Association Split--21. Total number of Down syndrome patient is 208 in the area of the Split-Dalmatian County.

Following tests have been performed:

1. ocular motility testing and determining the angle of squinting with sinoptophore
2. biomycroscopic examination of the anterior eye segment
3. retinoscopy (mydriasis with 1% Homatropin sol.)
4. indirect ophthalmoscopy
5. measurement of the intraocular pressure (non contact tonometry (NCT)

Entire study was planned and done according to the Declaration of Helsinki. All the data have been recorded using computer program Excell (Office 2003, Microsoft Co., USA).

The data pertaining to the number of children with Down syndrome have been obtained from Down syndrome Association Split-21, The Laboratory for Human Genetics, Department of Medical Genetics at the Paediatrics Clinic of the University Clinical Center Split, while the number of live-born children has been provided by The Institute of Public Health of the Split-Dalmatian County.

Results

We examined 153 patients with Down syndrome (there are 208 patients with Down syndrome in whole Split--Dalmatian County), aged between 0–18 years. Mean age was 11.7±3.2. Ninety patients were females, and 63 were males. All were Caucasians, Croats, from Split-Dalmatian County. We divided them in four groups according to

TABLE 2
RATIO OF CHILDREN BORN WITH THE DOWN SYNDROME AND THE LIVE-BIRTHS *PER* YEAR

Year	Live-births	Down syndrome	Ratio between DS and live-births
1992	5.810	15	1:387
1993	5.861	10	1:586
1994	5.746	6	1:957
1995	5.677	18	1:315
1996	5.831	7	1:833
1997	6.106	16	1:381
1998	5.440	10	1:544
1999	5.293	16	1:330
2000	5.204	17	1:306
2001	4.805	7	1:686
2002	4.741	11	1:431
2003	4.593	14	1:328
2004	4.962	7	1:708
2005	4.970	7	1:710
2006	4.906	9	1:545
2007	4.883	13	1:375
2008	4.910	12	1:409
2009	5.082	13	1:446

age. First group comprised patients aged between 0–5 years, second from 6–10 years, third from 11–14 years and fourth group from 15–18 years.

Ophthalmological examinations were performed in all patients.

We compared our results with results reported from other authors (Table 1).

Tables 3, 4, 5 and 6 show the distribution of refractive status (hypermetropia, myopia, astygmatismus) according to age and the amount of refractive status.

Discussion

Refractive anomalies in patients with Down syndrome are very common and their incidence vary from 65–100%[1–3]. One study which examined 140 children found that 137 (98%) had refractive anomalies, and 54% had mixed refractive anomaly on both eyes[4,5].

TABLE 3
DISTRIBUTION OF HYPERMETROPY IN CHILDREN WITH DOWN'S SYNDROME (N=137)

	Diopters (Dph)			
Age	0–1	1–2	>2	Total, n (%)
0–5	9	7	5	21 (15.3)
6–10	12	2	0	14 (10.2)
11–14	6	1	0	7 (5.1)
15–18	6	0	0	6 (4.37)
Total, n (%)	33	10	5	48 (35.0)

TABLE 4
DISTRIBUTION OF MYOPIA IN CHILDREN WITH DOWN'S SYNDROME (N=137)

	Diopters (Dph)			
Age	0–1	1–2	>2	Total, n (%)
0–5	5	0	0	5 (3,64)
6–10	11	3	1	15 (10.9)
11–14	14	8	0	22 (16.0)
15–18	5	19	0	24 (17.5)
Total, n (%)	35	30	1	48.04%

TABLE 5
DISTRIBUTION OF ASTYGMATISMUS IN CHILDREN WITH DOWN'S SYNDROME (N=137)

	Diopters (Dph)			
Age	0–1	1–2	>2	Total, n (%)
0–5	5	3	0	8 (5.8)
6–10	5	0	0	5 (3.6)
11–14	3	1	0	4 (2.9)
15–18	6	0	0	6 (4.37)
Total, n (%)	19	4	0	16.7%

TABLE 6
DISTRIBUTION OF EMETROPY IN CHILDREN WITH DOWN'S SYNDROME (N=153)

	Diopter (Dph)	
Age		Total n (%)
0–5	1	0.7
6–10	9	5.8
11–14	2	1.3
15–17	4	2.8
Total n (%)	16	10.6%

Strabismus may occur in 20–57% of patients with Down syndrome. Esotropia was found in 90–96%, while exotropia and hypertropia were found substantially less frequently (0–8%)[1–3]. Increased incidence of exotropia, found by Kim et al., could be caused by the fact that most of his patients were Asian who are more prone to develop exotropia than Caucasians and Africans wich more frequently develop esotropia[4].

Patients with Down syndrome frequently present with cataract (3–20%). Cataractss formation in patients with Down syndrome occur mostly between 12 and 15 years[1–3].

Nistagmus in patients with Down syndrome is tightly connected with refractive anomalies and lens changes. Keratoconus can occur in 0–15% of those patients and its incidence increases proportionally with age. First sign of keratoconus is the irregularity and discontinuity of retinoscopic reflex[1–3].

Glaucoma may develop in 1–5% of patients with Down syndrome, and most frequently it develops during childhood. That is the reason why the children has to be checked regularly with special attention on development of corneal edema, megalocornea, high intraocular pressure and optic disc pit progression[1–3].

Supernumerary optic disc vessels may be found in 11–38% of patients with Down syndrome[1–3].

A number of authors have reported so far ophthalmological anomalies in children with the Down syndrome[4–9]. Our results showed that the frequency of ophtalmological anomalies depended upon the age of the subjects. Our paper is particularly focused on the analysis of refraction errors in patients with the Down syndrome. Refractive error was found with retinoscopy in 137 (89.5%) of examined patients, while 16 (10.6%) did not have any refractive anomaly.

Hypermetropia was found in 48 (35.0%) of patients, myopia in 66 (48.1%) and astygmatisms in 23 (16.7%). A number of authors have so far dealt with refraction errors in children with the Down syndrome. The results differ between particular cases. As has been established by Little et al., in a group of 29 children with Down syndrome, there seems to exist a prevalence of hypermetropical astigmatism with relation to the control group[10] while Stewart et al. seem to have found a prevalence of hypermetropia in a group of 27 children[11]. A prevalence

of hypermetropia has been also established by Akinici et al. in a group of 77 children[12].

In our patients myopia was the most frequent refractive anomaly, except from group of children with age between 0–5 years who had most frequently hypermetropia.

We agree with some authors that the precise cause of high prevalence of myopia in children with Down's syndrome is still not clear. The reasons could be longer axial length of the eye, what should be still investigated. The reason is also lower cooperation with the patient and the difficult medical visit and examination.

We have also found other ophthalmologic changes in children with the Down syndrome, there by establishing a higher occurrence of epicanthus, strabismus, nistagamus, glaucoma, keratocunus, supernumerary optic disc vessels and cataract.

Conclusion

In children with Down syndrome ophthalmologycal anomalies are to be more frequently encountered, i.e. refractive errors – hypermetropia, myopia, astigmatisms (89.5%), epicantus (41.8%), strabisms (28.7%, of which 81.9% had esophoria and 18.1% exophoria), nistagmus (8.4%), cataracts (1.3%), glaucoma (1.9%), supernumerary optic disc vessels (24.1%) and keratoconus (1.3%). By examining the refraction errors in 153 children with the Down syndrome we have also found a prevalence of myopia (except in children aged 0–5). This analisys shows that is important to organize our ophtalmologycal outpatient clinics for children and people with Down syndrome in Croatia.

In order to avoid complications which leads to uncorrigable refractive errors (visual impairment, strabisms), elevated intraocular pressure and cataract, it is necessary to do ophthalmological examination as soon as it is possible (before 6 months of age). In children, the refractive error must be detected as early as possible, prescribe glasses, in the case of amblyopia ortoptoptic conducted exercises, and operational corrections of strabisms, cataracts and glaucoma. Only regular inspections and timely intervention can prevent the consequences that would occur.

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OFTALMOLOŠKE PROMJENE U DJECE S DOWNOVIM SINDROMOM U SPLITSKO-DALMATINSKOJ ŽUPANIJI

SAŽETAK

Cilj nam je bio prikazati oftalmološke promjena u djece s Downovom sindormu u Splitsko-dalmatinskoj županiji u razdoblju od 1992. do 2009. godine. Ovo istraživanje uključuje 153 djece iz Splitsko-dalmatinske županije s Down sindromom u dobi od 0–18 godina. Svim ispitanicima je učinjen kompletan oftalmološki pregled (prednji segment, bulbomotorika, refrakcijske greške – miopija, hiperemtropija i astigmatizam, fundus, mjerenje intraokularnog tlaka). U 89,5% ispitanika pronađena refrakcijska pogreška, 48,1% je imalo miopiju, 35,0% hipermetropiju, 16,7% astigamtizam, 28,7% strabizam, 8,4% nistagmus, 1,3% kataraktu, 1,9% glaukom, 24,1% prekobrojne krvne žile optičkog diska i keratokonus (1,3%). U osoba s Down sindromom utvrđena je veća učestalost refrakcijskih pogrešaka (posebice miopija), kao i drugih oftalmoloških bolesti.