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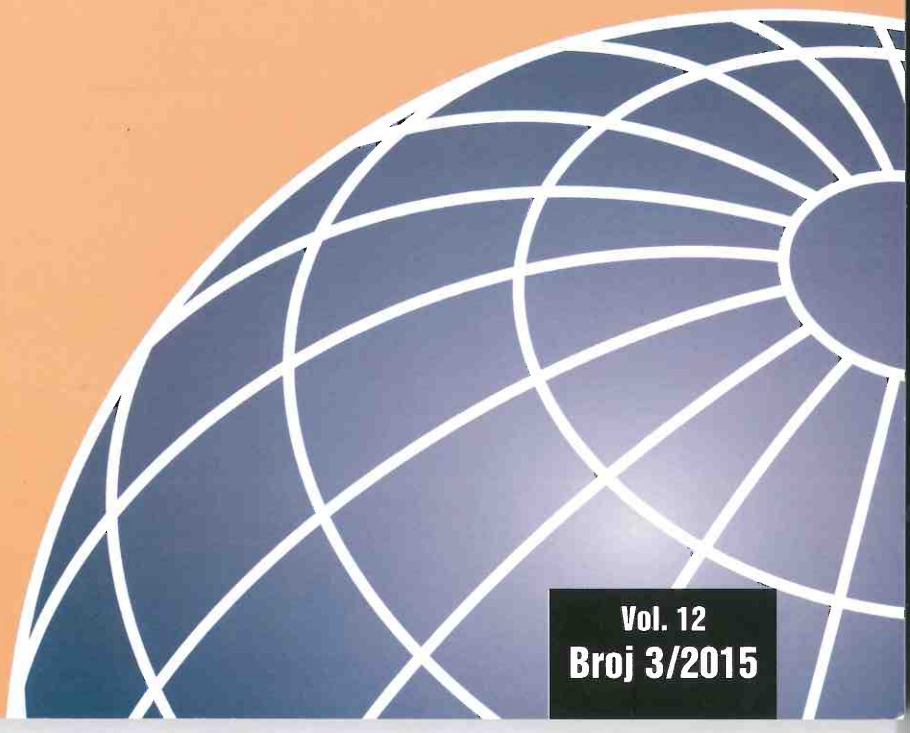
ZA JUGOISTOČNU EVROPU



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KONGENITALNA KATARAKTA KOD ADOLESCENTA SA DAUNOVIM SINDROMOM

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APSTRAKT

Cilj: Obostrana kongenitalna katarakta kod DS (Daunov sindrom) je redak nalaz, i cilj našeg rada bio je prikazivanje retkog slučaja obostrano operisane katarakte povezane sa visokom miopijom kod adolescenta sa DS.

Prikaz slučaja: Kod adolescenta sa DS, uzrasta 30.6 godina, muški pol bila je prisutna katarakta na oba oka, koja je bila operisana na desnom oku 1990. godine sa metodom ekstrakapsularne katarakte ekstrakcijom bez ugradnje sočiva (Aphakia operata), dok je levo oko bilo operisano 2002. godine sa metodom fakoemlzifikacije i ugradnjom prednjokomornog sočiva (Phaco +AOL). Kod pacijenta bilo je utvrđeno prisustvo stečene ezotropije sa sledećim strabološkim nalazom: u primarnom položaju OS u eso 30 pdp na daljinu i blizinu, Cover-test pokazao je prisustvo spontanog alternansa, motilitet je bio uredan, punctum proximum convergenciae (PPC) pokazalo je uredne vrednosti 3-5 cm. U odnosu na stereovid koji je bio ispitan stereotestom Lang 1, pacijent nije sarađivao. U odnosu na prisustvo nistagmusa, ispitanik je pokazao prisustvo horizontalnog senzorielnog nistagmusa. U odnosu na vidnu oštrinu i kolorni vid, nisu dobijeni podaci zbog nesarađivanja ispitanika.

Zaključak: Veliki broj studija utvrdilo je da su oftalmološka stanja, kao sto su katarakta i glaukom, potencijalno devastirajući za vidni potencijal ispitanika sa DS, i potrebni su rutinski oftalmološki scrining pregledi.

Ključne reči: Daunov sindrom, kongenitalna katarakta, abnormalnosti sočiva, vizuelni – scrining.

UVOD

Daunov sindrom (DS) predstavlja kongenitalnu anomaliju uzrokovanu trisomijom, kompletnom ili parcijalnom, hromozoma broj 21. DS predstavlja najčešći uzrok za mentalnu retardaciju, kao i uzrok za brojne sistemske i oftalmološke promene. Oftalmološke promene kod DS prvi put su opisane u kasnom periodu 19 veka (1). Sa sledećim istraživanjima utvrđeno je da brojne oftalmološke promene kod DS imaju značajni uticaj na vizuelni potencijal ove specijalne populacije subjekata. Od ukupno 23 istraživačke studije, 4 navode ukupnu prevalenciju oftalmoloških poremećaja kod DS. Prevalencija ukupnih oftalmoloških poremećaja kod DS rangirana je u vrednostima od 46% (2) i 61% (3) i 91% (4) do 100% (5). Najčešće oftalmološke promene kod DS su sledeće: kosa postavljenost palpebralnih fisura, epiblpeharon, visok procenat prisutnih refraktivnih grešaka posebno hipermetropije kao i prisustvo strabizma. Flokularna zamućenja sočiva su karakteristični nalaz DS, za čiju prevalenciju ne postoje podaci u objavljenim studijama. Obostrana kongenitalna katarakta kod DS je retki nalaz i cilj našeg rada je bio prikazati retki slučaj obostrane operisane kongenitalne katarakte povezne sa visokom miopijom.

PRIKAZ SLUČAJA

Adolescent sa Daunovim sindromom, uzrasta od 30.6 godina, muški pol. Dijagnoza DS bila je postavljena na osnovu citogenetskog nalaza, kao i prisustva kliničkih simptoma. U odnosu na povezane sistemske promene kod ispitanika bio je prisutan Daunov sindrom – povezana stanja – protokol prema Van Cleeve-u (6,7). Analizirani su: kardiološki defekti, audiološke anomalije, dentalni deformiteti, tiroidne abnormalnosti, dermatološke promene, gastro-intestinalni poremećaji, mišićno-koštane promene, kao i neuro-psihijatrijske promene. Kod ispitanika je utvrđeno prisustvo gotskog nepca (od dentalnih deformiteta) i prisustvo dermatitisa (od dermatoloških promena). Ispitanik nije pokazao prisustvo obezitasa i TT je iznosila 55 kg. Starost majke kada je dobila dete iznosila je 38 godina, dok oca 46 godina.

U protokolu Daunov sindroma – oftalmološke promene, pojedinačno po grupama bile su analizirane: promene prednjeg okularnog segmenta, promene srednjeg okularnog segmenta, kao i promene zadnjeg okularnog segmenta. U promenama prednjeg okularnog segmenta analizirano je: iskošenje palpebralnih fisura, suženje palpebralnih fisura, epicanthus,epiblepharon, ptosis palpebrae, conjunctivitis, blepharitis, blepharoconjuctivitis, opstrukcija suznih puteva i keratokonus. Niti jedna od navedenih karakteristika, nije bila prisutna kod našeg ispitanika. U promenama srednjeg okularnog segmenta, analizirani su: prisustvo flokularnih opa-

citeta sočiva, prisustvo kongenitalne katarakte, iris abnormalnosti, prisustvo Brushfled-ovih tačaka, boja irisa, kao i prisustvo glaukoma. Kod ispitanika bila je prisutna kongenitalna katarakta oba oka, koja je bila operisana, na desnom oku 1990. godine metodom ekstrakapsularne katarakt ekstrakcije bez ugradnje sočiva (Aphakia operata), a na levom oku operisana 2002 godine metodom fakoemulzifikacije i ugradnja prednjokomornog sočiva (Phaco + AOL). Boja oba irisa ispitanika bila je smeđa. U analizi zadnjeg okularnog segmenta bili su prisutni: povećan broj retinalnih krvnih sudova preko papile očnog živca (PNO), hipoplazija PNO, atrofija PNO. Kod ispitanika sa metodom indirektne oftalmoskopije pomoću Volk 90 lupe, utvrđene su miopične degenerativne promene peripapilarno.

U protokolu Daunovog sindroma -strabizam, analizirano je prisustvo rizičnih faktora: pre-natalnih, peri-natalnih, post-natalnih, težina pri rođenju i gestaciska starost. Težina pri rođenju ispitanika iznosila je 3500 gr., gestaciska starost 40 gestaciskih nedelja. Ispitanik je pokazao prisustvo peri-natalnog rizik-faktora (sectio cesarea). Pojedinačno, analizirani su prisustvo strabizma i veličina ugla krivljenja. Utvrđena je stečena ezotropija sa sledećim strabološkim nalazom: u primarnom položaju OS u eso 30 pdp na daljinu i blizinu, Cover- test je pokazao prisustvo nesigurnog spontanog alternansa, motilitet je bio uredan, punctum proximum convergenciae (PPC) pokazao je uredne vrednosti 3-5 cm. U odnosu na stereovid, koji je bio ispitan Lang 1-stereotestom, nismo dobili rezultat zbog nesarađivanja ispitanika. U odnosu na prisustvo nistagmusa, pacijent je pokazao prisustvo senzorielnog horizontalnog nistagmusa. U odnosu na određivanje vidne oštrine kolornog vida, nisu bili dobijeni podaci zbog nesarađivanja pacijenta.

Kod ispitanika bilo je sprovedeno ispitivanje pomoću Carl Zeiss IOL Master-aparatom, i na desno i na levo oko. Dodatno bilo je sprovedeno ehografsko ispitivanje (Quantel medical) oba očna bulbusa, pri čemu je bila utvrđena miopična konfiguracija očnog bulbusa obostrano, sa diskretnim opacitetima staklastog tela, isto obostrano. Sa ozbirom na miopiju i ambliopiju desnog oka, utvrđeno da ne bi došlo do znatnog poboljšanja vidne oštrine, pa prema nalazima nije preporučena sekundarna ugradnja sočiva. Na levom oku bila je prisutna početna sekundarna fibroza zadnje lentalne kapsule, pa je bila preporučena YAG-laser disrupcija.

Ispitanik je imao sledeće dijagnoze: 1) Aphakia operata OD (ECCE)
2) Pseudophakia OS (Phaco + AOL)
3) Myopia alta degenerativa OU
4) Amblyopia gravis OU

DISKUSIJA

Creavin AL (8) u svom revijalnom radu analizira ukupno 23 studije za oftalmološke promene kod dece sa DS uzrasta od 0-16 godina. Osam od 18 publikovanih studija koje su proučavale prevalenciju katarakte kod DS, utvrdili su da prevalencija iznosi 5% ili manje. Dodatnih 8 studija utvrdile su da prevalencija katarakte iznosi od 6% do 15%. Jedino 2 studije, utvrdile su da prevalencija katarakte kod DS iznosi od 20% do 37%. Caputo AR et al. (9) utvrdio je prisustvo bilateralne lentalne subluksacije, ali nije pokazao prevalenciju. Veliki broj studija utvrdio je da oftalmološka stanja, kao što su katarakta i glaukom, su potencijalno devastirajuć za vidni potencijal ispitanika sa DS i neophodni su rutinski oftalmološki skrining – pregledi. Zbog visoke frekvencije okularne patologije kod DS, norveška grupa oftalmologa-strabologa (10), daje sledeće preporuke za kontinuiran vizuelni-skrining kod osoba sa DS, u vremenskom periodu od rađanja do zrelog doba:

1) Neonatalni oftalmološki pregled (pregled u prvom mesecu posle rađanja): pri ovom pregledu utvrđuje se prisustvo kongenitalne katarakte, glaukoma i stenoze suznih puteva.
2) Oftalmološki pregled na jednogodišnjem uzrastiu: pri ovom pregledu utvrđuju se promene na prednjem, srednjem i zadnjem okularnom segmentu, vrši se ortoptička procena, kao i određivanje refraktivnog statusa.
3) Oftlamolški pregled na 2-3 godišnjem uzrastu: pri ovom pregledu utvrđuju se promene prednjeg, srednjeg i zadnjeg okularnog segmenta, vrši se ortoptička procena, kao i određivanje refraktivnog statusa.
4) Oftalmološki pregled na 5-6 godišnjem uzrastu: pri ovom pregledu utvrđuju se promene prednjeg, srednjeg i zadnjeg okularnog segmenta, vrši se ortoptička procena i određivanje refraktivnog statusa.
5) Oftalmološki pregledi na svakih 5 godina: pri ovom pregledu utvrđuju se promene prednjeg, srednjeg i zadnjeg okularnog segmenta, kao i određivanje refraktivnog statusa.

U slučaju pozitivnog nalaza postojanja refraktivne anomalije, deficientne akomodacije, strabizam), frekvenciju oftalmoloških pregleda potrebno je da odredi oftalmolog.

Rana identifikacija i korekcija oftalmoloških poremećaja kod osoba sa DS, dovodi do poboljšanja razvojnog i funkcionalnog ishoda posebno u dečjem periodu kod osoba sa DS, što je od neprocenjivog značaja u edukativnom i u procesu socijalizacije osoba sa DS.

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CONGENITAL CATARACT IN ADOLESCENT WITH DOWN SYNDROME

ABSTRACT

Aim: Bilateral congenital cataract in Down syndrome (DS) is a rare finding and purpose of our work was to present a rare case of bilateral operated congenital cataract associated with high myopia in adolescent with DS.

Case report: Adolescent with Down syndrome, aged 30.6 year male, was examined in University Eye Clinic „Svjetlost" in Zagreb, Republic of Croatia, 2007. In examinee it was present congenital cataract in both eyes, right eye was operated in 1990 with the method of Extracapsular cataract extraction without intraocular lens (Aphakia operata), and the left eye was operated in 2002 with the method of Phacoemulsification and implatantation of anterior intraocular lens (Phaco + AOL). The patient showed presence of acquired esotropia with the following strabological finding: in the primary position in OS eso 30 pdp on distance and near, Cover-test showed the presence of uncertain spontaneous alternans, motility was regular, punctum proximum convergenciae (PPC) showed regular values of 3-5 cm. The stereoacuity was tested with the stereotest Lang 1, but the patient

does not cooperate. The examinee showed the presence of sensorial horizontal nystagmus. Visual acuity and color vision was not obtained because of lack of cooperation.

Conclusion: A large number of studies determined that ophthalmic manifestation as cataract and glaucoma are potentially devastating for the visual potential in the patients with DS, and they need regular ophthalmic-screening examinations.

Key words: Down syndrome, congenital cataracts, lens abnormalities, visual screening.

INTRODUCTION

Down syndrome (DS) represents congenital genetic disorder caused by complete or partial trisomy of hormosom number 21. DS is wide frequent cause of mental reatradation, as well cause for multiple systemic and ophthalmic disorders. Ocular disorders in DS are first described in the period of 19 centery(1). Next research confirmed that multiple opthalmological disorders in DS had significant impact on visual capacity of DS special population. There are 23 research studies that analised ophthalmic manifestations in DS, but only 4 studies refers the exact prevalence of all ophthalmic manifestations. The prevalence of all ophthalmic manifestations in DS is ranged in values from 46%(2) and 61%(3) and 91%(4) to 100%(5). Most frequent ophthalmic features in DS are: mongoloid presentation of palpebral fissures, epiblepharon, high percentage of refractive errors, especially hyperopa, as well as strabismus.Flocular opacitates of the lens are characteristic finding in DS, which prevalence is not reffered in present studies. Bilateral congenital cataract in DS is rare finding, and our case report presentation aimed to study rare case of bilateral operated congenital cataract associated with high myopia.

CASE REPORT PRESENTATION

Adolescent with Down syndrome, age 30.6 years, male, was examined in University Eye Clinic „Svjetlost" in capital Zagreb, R. Croatia, year 2007. The diagnosis of DS was confirmed on cytogentic finding as well on presence of clinical features. Considering associated systemic disorders, special Down syndrome-associated conditions protocol according Van Cleeve (6,7) was fulfilled. Following conditions were analysed: cardiac defects, audiological anomalies, dental deformitates, thyroid abnormalities, dermatological changes, gastro-intestinal disorders, muscle-osseal changes as well neuro-psychiatric conditions. The examinee did not show obesitas and body weight was 55 kg.The age of the mother of the examinee when she delivered the child was 38 years, the age of the father 46 years.

In the Down syndrome-ophthalmological manifestations protocol, following findings were analysed: findings of anterior ocular segment, findings of medial ocular segment and findings of posterior ocular segment. In anteri-

or ocular segment, following findings were analysed: mongoloid presentation of palpebral fissures, narrowing of palpabral fissures, epicanthus, epiblepharon, ptosis palpaebrae, conjunctivitis, blepharitis, blepharoconjuctivitis, lacrimal duct stenosis and keratoconus. None of these findings were present in the examinee. In medial ocular segment following findings were analysed: flocular lental opacitates, presence of congenital cataract, iris abnormalities, presence of Brushfiled spots, colour of the iris and presence of glaucoma. In the examinee congenital cataract of both eyes was present, operated on the right eye 1990 year with the method of Extracapsular cataract exatraction with no IOL implant (Aphakia operata), and operated on the left eye 2002 year with the method of Phacoemulsification and implant of anterior chamber lens (Phaco +AOL). The colour of the iris in the examinee was brown on both eyes. In the posterior ocular segment following findings were analysed: higher number of vessels over papilla nervi optici (PNO), hypoplasie of PNO, atrophy of PNO. In the examinee with the method of indirect opthalmosopy with Volk 90 lens peripapillar myopic degenerative changes were confirmed.

In the Down syndrome- strabismus protocol, presence of risk factors was analysed: pre-natal, peri-natal, post-natal risk factors, as well birth weight and gestational age. Birth weight of examinee was 3500 gr., gestational age was 40 gestational weeks. Th examinee showed presence of perinatal risk factor (sectio cesarea). Separate, presence of strabismus and the angle of deviation were analysed. The examinee showed presence of acquired esotropia with following strabismological finding: in primary position OS was in eso 30 pdp on distance and near, Cover-test showed uncertain spontaneous alternans, motility was regullar, punctum proximum convergenciae (PPC) was regullar 3-5 cm. Considering stereoacuity which was tested with Lang 1 stereotest, the patient did not cooperate.Considering presence of nystagmus, the examinee showed presence of sensorial horizontal nystagmus. Considering the visual acuity and color vision,also the patient did not cooperate.

In the patient examination with Carl Zeiss IOL Master was made, on the right and on the left eye. Also echografic examination with Quantel medical on the both eyes was made, and myopic configuration of the bulbus of both eyes was determinate, with discrete vitreal opacitates also in both eyes. Considering the myopia and amblyopia on the right eye it was determinate that it should not be achieved better outcome, so it was not recommended implantation of intraocular lens.On the left eye incipent secundar fibrosis on the posterior lental membrane was determinate, so it was reccomended YAG laser disrupction.

The patient had following diagnoses:

1) Aphakia operata OU (ECCE)
2) Pseudophakia OS (Phaco +IOL)
3) Myopia alta degnerativa OU
4) Amblyopia gravis OU

DISCUSSION

Creavin AL(8) in his review article analysed 23 studies considering ophthalmological manifestations in children with DS on age of 0-16 years. Eight from 18 published studies,analysed the prevalence of cataract in DS, and determine that the prevalence was 5% or less. Additional 8 studies determined that prevalence was between 6% and 15%. Only 2 studies determined that prevalence of cataract in DS was between 20% and 37%. Caputo AR et al. (9) determined bilateral lental subluxation, but did not present the prevalence.A large number of studies determined that ophthalmic conditions, as cataract and glaucoma, are potential devastating on visual capacity on the DS exeminees and required regullar ophthalmic- screening examinations. Because of high frequency of ocular pathology in DS, norweg group of ophthalmologist-strabologist (10) are giving following recommendations for regullar visual screening in individuals with DS, form birth to adult age :

1) Neonatal ophthalmological examination (examination in first month after birth): in this examination presence of congenital cataract, glaucoma or lacrimal duct stenosis is determined.
2) Ophthalmological examination on age of 1 year: in this examination changes in anterior, medial and posterior ocular segment are determined, as well ortoptic evaluation and refractive state.
3) Ophthalmological examination on age of 2-3 years: in this examination changes in anterior, medial and posterior ocular segment are determined, as well ortoptic evaluation and refractive state.
4) Ophthalmological examination on age of 5-6 years : in this examination changes in anterior, medial and posterior ocular segment are determined, as well ortoptic evaluation and refractive state.
5) Ophthalmological examination on each 5 years : in this examination changes in anterior, medial and posterior ocular segment are determined, as well refractive state.

In the cases of positive finding (presence of refractive error, deficient accommodation, strabismus), the frequency of ophthalmological examinations should be frequent as determined individual from the responsible ophthalmologist.

Early identification and correction of ocular disoreders in individuals with DS, leads to better developing and functional outcome especially in the childhood of children with DS, which fact is crucial in education and process of socialisation of individuals with DS.