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Congenital Blindness and Visual Impairment Cause Infection or Non Infection

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ABSTRACT

Introduction: Authors are from reference documentation to archive at Organization of Federation of blind and visually impaired in central Serbia (Kragujevac), by retrograde analysis, of 2007-2012, comprehend two groups by etiology-clinical characteristics of congenital blindness and visually impaired, caused infection or non infection example. **Aim:** to analyze relationship between infectious and non infectious of congenital blindness and visually impaired in our referent region and compare with world references. **Material/methods:** With 6-years analysis included the most frequency cases of congenital blindness and visually impaired in two groups, according to presence or absence infectious causes. From infectious causes of congenital blindness and visually impaired are included: CMV - infection, congenital rubella syndrome, congenital toxoplasmosis, congenital syphilis and rare mixed syndrome. From non infectious causes are included: retinitis pigmentosa, retinopathy prematurity, primary congenital glaucoma, Leber's congenital amaurosis and rare syndrome. **Results:** From total number of registered blind and visually impaired - 1308 (100%), over the last 6 years, the registration was 349 (26.68%) with congenital blindness and visually impaired. From recorder with the number of the most common congenital blindness and visually impaired-194 (55.59%) with infections cause, and 155 (44.41%) non infection cause. **Conclusion:** Congenital blindness has shown permanent increase in past 6 years, in group with infectious and with non infectious causes. Congenital blindness and visually impaired of the most common etiology among registered members of our association in Kragujevac is subject of our correlation and global trends mentioned observation of these diseases.

Key words: congenital blindness; infectious causal; noninfectious causal

1. INTRODUCTION

The authors are from reference documentation to archive at Organization of Federation of the blind and visually impaired in central Serbia (Kragujevac)-referent and tertiary level health care system, by retrograde analyses, by 2007-2012 year, comprehend two groups by etiology-clinical characteristics of the most common congenital blindness and visually impaired caused infection or non infection example.

Material /methods

With 6-years analysis included the cases of the most common congenital blindness divided in two groups, according to the presence or absence infectious causes are the same quantitatively and qualitatively correlation.

From infectious causes of congenital blindness are included: CMV - infection, congenital rubella syndrome (with congenital cataract), congenital toxoplasmosis, congenital syphilis and rare mixed syndrome (anophthalmia, microphthalmia, etc).

From non infectious causes included the following: retinitis pigmentosa, retinopathy of prematurity, primary congenital glaucoma, Leber's hereditary optic neuropathy, congenital strabismus and rare mixed syndrome (congeni-

tal strabismus, etc).

Our records of congenital blindness and visual impairment included all patients, and the neonate and adult patients of usually processed the clinical pathology by the past 6 years.

Statistical analysis of quantitative data, including descriptive statistics, parametric and non parametric comparisons was performed for variables. Frequency analysis was performed by the chi square test. All P values were two sided, and P values less than 0.05 were considered statistically significant. Statistical analyses were by SPSS, versus 19.0.

2. RESULTS

From total number of registered blind and visually impaired - 1308 (100%), over the last 6 years, our registration was 349 (26.68%) with congenital blindness and visually impaired. From recorder with the number of the most common congenital blindness and visually impaired-194 (55.59%) with infections cause, and 155 (44.41%) with non infection cause. The authors analyzed the following by each year and report (Table 1, 2).

In 2007, the authors observation compared noninfec-

2007 year		2008 year		2009 year		2010 year		2011 year		2012 year													
noninfectious causa ↓	infectious causa ↓	noninfectious causa ↓	infectious causa ↓	noninfectious causa ↓	infectious causa ↓	noninfectious causa ↓	infectious causa ↓	noninfectious causa ↓	infectious causa ↓	noninfectious causa ↓	infectious causa ↓												
retinitis pigmentosa1	10	CMV-infection	13	retinitis pigmentosa	12	CMV-infection	10	retinitis pigmentosa	14	CMV-infection	15	retinitis pigmentosa	6	CMV-infection	14	retinitis pigmentosa	7	CMV-infection	17				
fibroplasia retrolent.2	13	rubela syndrome	9	fibroplasia retrolent.	10	rubela syndrome	12	fibroplasia retrolent.	11	rubela syndrome	7	fibroplasia retrolent.	5	rubela syndrome	8	fibroplasia retrolent.	6	rubela syndrome	9	fibroplasia retrolent.	5	rubela syndrome	0
congenital glaucoma3	5	toxoplasmosis	10	congenital glaucoma	4	toxoplasmosis	6	congenital glaucoma	6	toxoplasmosis	8	congenital glaucoma	4	toxoplasmosis	14	congenital glaucoma	5	toxoplasmosis	10	congenital glaucoma	7	toxoplasmosis	12
Leber amaurosis4	3	siphylis	2	Leber amaurosis	2	siphylis	3	Leber amaurosis	4	siphylis	1	Leber amaurosis	3	siphylis	0	Leber amaurosis	1	siphylis	0	Leber amaurosis	6	siphylis	0
Σ	31	Σ	34	Σ	28	Σ	31	Σ	31	Σ	30	Σ	22	Σ	36	Σ	18	Σ	34	Σ	25	Σ	29

Table 1. Congenital blindness and visually impaired by 6 years divided of noninfectious and infectious causa

tious and infectious causes of congenital blindness and visual impaired, so the noninfectious was 31 (47.69%) and infectious 34 (52.31%), which isn't statistical significant ($\chi^2=0.138$; $p=0.710$), i.e., authors do not show any dominance, nor noninfectious, nor infectious causes of this retrospective study (Table 1, 2).

During 2008 observations, the authors were compared noninfectious and infectious causes of congenital blindness and visual impaired, so the noninfectious was 28 (47.46%), and infectious–31 (52.54%), which isn't statistical statement significant ($\chi^2=0.153$; $p=0.696$), i.e., the authors don't show dominance of the most common causes (Table 1, 2).

During 2009 observations, authors were compared noninfectious and infectious causes, so noninfectious was 31 (50.82%) and infectious–30 (49.18%), which isn't statistical significant ($\chi^2=0.016$; $p=0.898$), i.e., authors don't show dominance nor noninfectious, nor infectious causes (Table 1, 2).

In 2010 during the observation, authors compared same causes, so the noninfectious was 22 (37.93%) and infectious–36 (62.07%), which isn't statistical significant ($\chi^2=3.379$; $p=0.066$), i.e., authors don't show dominance nor noninfectious, nor infectious causes of our retrospective study (Table 1, 2).

During 2011 observations, authors compared the same causes, so the noninfectious was 18 (34.62%) and infectious 34 (65.38%) with statistical statement significant ($\chi^2=4.923$; $p=0.027$), i.e., authors show domination of the infectious causes in 2011 years.

In 2012 year, during our observation and compared noninfectious and infectious cause, so as the noninfectious was 25 (46.30%) and infectious–29 (53.70%), which is not statistical significant ($\chi^2=0.296$; $p=0.586$) we don't show dominance nor infectious, nor noninfectious causes in retrospective study by 2012 (Table 1, 2).

Analysis of noninfectious causes–retinitis pigmentosa during 6 years, show the greatest number, in 2008 with 12 (21.82%) of the 55 (100%), a similar quantitative values of this disease and the remaining years of copyright monitoring, which isn't statistical significant ($\chi^2=2.709$; $p=0.745$) (Table 1, 2).

Analysis of noninfectious causes – fibroplasia retrolentalis, during 6 years, show the greatest number in 2007 with 13 (26.00%), of total number–50, (100%), a similar quantitative values of this disease and the remaining years of copyright monitoring (2008–20%, 2009–22%, 2010–10%, 2011–12%, 2012–10%), which isn't statistical

	Most infectious causa, 2007-2012, Σ	Most noninfectious causa, 2007-2012, Σ	
CMV-infection	83 42.78%	retinitis pigmentosa	55 35.48%
rubella syndrome	45 23.20%	fibroplasia retrolentalis	50 32.26%
toxoplasmosis	60 30.93%	congenital glaucoma	31 20.00%
syphilis	6 3.09%	Leber's amaurosis	19 12.26%
	Σ 194 55.59%	Σ 155 44.41%	349

Table 2. The most infectious and noninfectious causa of congenital blindness and visually impaired by 6 years

significant ($\chi^2=7.120$; $p=0.212$), based on and why the not more to infer that the number of ill, meaningful changes during the years observation (Table 1, 2).

Analysis of noninfectious causes–primary congenital glaucoma, during 6 years, show the greatest number in 2012 with 7 (22.58%) of total–31 (100 %), a similar quantitative values of this disease and the remaining years of copyright monitoring, which isn't statistical significant ($\chi^2=1.323$; $p=0.933$) based on and why the not to infer that the number of patients diagnosed with this disease meaningful changes (Table 1, 2).

Analysis of noninfectious causes – Leber's congenital amaurosis, during 6 years, show the greatest number in 2012 with 6 (31.58%) of total number–19 (100%), a similar, and quantitative values of this disease and the remaining years of copyright observation, which is not statistical significant ($\chi^2=4.684$; $p=0.456$) based on and why not to infer, that number of patients diagnosed with this disease, meaningful changes in observation for years, but there are genetic theory which give etiological explanation (Table 1, 2).

Analysis this infectious causes–CMV, during 6 years, show the greatest number in 2012 with 17 (20.48%) of total–83 (100%), a similar and quantitative values of this disease, and in all remaining years of copyright observation, which is not statistical significant ($\chi^2=1.940$; $p=0.857$) based on and why not to infer that number of patients diagnosed with this disease for years meaningful changes in observation, although could anticipate as an viral epidemic and today as mixed viral infection in general (Table 1, 2).

Analysis of infectious causes–rubella viral syndrome, during 6 years, shows the greatest number in 2008 with 12 (26.67%) of total–45 (100%). In 2012 year is not any causes and similar and quantitative values of this disease were, and the remaining years of copyright observation, which isn't statistical significant ($\chi^2=8.873$; $p=0.817$) based on and why cannot conclude that number of patients diagnosed with this disease for years meaningful changes in observa-

tion, except in 2012 (Table 1, 2).

Analysis of infectious causes—*Toxoplasmosis*, during 6 years, show greatest number in 2010 of 14 (23.33%) of total—60 (100%), a similar and quantitative values of this disease were, and remaining years of copyright observation, which is not statistical significant

($\chi^2=4.000$; $p=0.549$) based on and why could not conclude that number of patients diagnosed with this disease for years meaningful changes in observation (Table 1, 2).

Analysis of infectious causes—*sypphilis*, during 6 years, show greatest number in 2008 with 3 (50.00%) of total—6 (100%), a similar and quantitative values of this disease were, and the remaining years of copyright monitoring, that isn't statistical significant ($\chi^2=0.000$; $p=0.877$), by Fisher's exact-test upon which cannot be to infer that number of patients diagnosed with this disease for years meaningful changes in observation, i.e., that the disease has been nearly eradicated, and that rare and poor appears (Table 1, 2).

Analysis by sex and into age population, by 6 years of observation, is not show, because random sample population of neonate to geriatric population.

4. DISCUSSION

Serbia has about 12000 blind and visually impaired et now. Over 50% are older than 60 years. Our region belongs to the medium developed in terms of tertiary health care of the patients. We showed (21 March 2012) the number 1308 (100%) of blind and visually impaired of whom and with congenital blindness—349 (26.68%). The Organization of Federation of the blind and visually impaired is a good concept in our referent regional associations with the reference data of these diseases (www.imenik.rs; www.savezslepih.org.rs).

Retinitis pigmentosa comprises the complex group of inherited dystrophies characterized by progressive degeneration and dysfunction of retina; and may occur alone or as part of syndrome; and may be inherited as dominant, recessive, or X-linked trait, or occur sporadically (1). NYX gene mutations were more frequent cause of CSNBX than CACNA1F gene mutations in the 11 British families studied. As evidence of functioning rod system was identified in the majority of subjects tested, the clinical phenotypes “complete” and “incomplete”(not correlate with genotype) [1]. The term “retinitis” is misnomer, since the pathogenesis is not inflammatory. A family history of retinitis pigmentosa is present in about 70 percent of patients (1, 2). The vision problems associated with this condition are congenital, which means they are present from birth. Patients have vision problems, including loss of sharpness (reduced acuity—detects light, color), severe nearsightedness (high myopia) and in voluntary movements of the eyes (nystagmus, strabismus, ptosis) (3). Sectoral changes have been observed in autosomal dominant retinitis pigmentosa and in females heterozygous for X-linked retinitis pigmentosa (3).

The worldwide prevalence is estimated at 1 in 4000 to 5000. About 100,000 patients are affected in the United States. Special consideration is given to unusual inheritance patterns. The aggregate carrier frequency for reces-

sive retinitis pigmentosa alleles may be as high as 10% (4).

Retinopathy of prematurity (ROP) is one of the three leading causes of legal blindness in infants in developed countries. During the last decades, international and national guidelines for ROP screening have been continually updated. The present article surveys ROP classification, current national and international guidelines and new aspects of ROP screening (5). ROP is an emerging cause of blindness. Analyses of the incidence and risk factors for ROP from a secondary care center indicate that the incidence (19.7%) is higher than that noted in tertiary care centers (6).

Primary congenital glaucoma is characterized by elevated intraocular pressure, enlargement of the globe (buphthalmos), edema, opacification of cornea with rupture of Descemet's membrane and progressive glaucomatous optic atrophy. Typically, the diagnosis is made before first year of life. Depending on when treatment is instituted, visual acuity may be reduced and/or visual fields may be restricted. In untreated cases, blindness invariably occurs (7). Genetic heterogeneity is the hallmark of all glaucoma and multiple chromosomal loci have been linked to the disease, but only a few genes have been characterized as myocilin (MYOC), optineurin (OPTN), WDR36 and neurotrophin 4 (NTF4) in primary open angle glaucoma, and CYP1B 1, LTBP2 in congenital and developmental glaucoma (7).

Leber's congenital amaurosis, an severe dystrophy of retina, typically becomes evident before first year and later. Visual function is usually poor and often accompanied by nystagmus, sluggish or near absent pupilar responses, high hyperopia and keratoconus (8). Leber's congenital amaurosis occurs in 2 to 3 per 100,000 newborns.

It is one of the most common causes of blindness in infants (9). The reported incidence of mental retardation in Leber's congenital amaurosis has varied from 10% to 87% (10).

Cytomegalovirus (CMV) is the leading cause of congenital viral infection, with a range of incidence between 0.5-3% of live births worldwide. Approximately 30% of maternal infections during pregnancy can result in congenital infection. Neonates with congenital or natal cytomegalovirus infection can be asymptomatic and infection can also be transmitted post natal from mother to infant by breastfeeding (11). CMV infection is ranked as one of the most common infections in adults, with the serology positive rates ranging from 60-99% globally. Congenital cytomegalovirus infection is also a cause of non hereditary congenital sensory-neural hearing loss (11, 12). The epidemiology of CMV varies widely in different populations but, wherever tested to date, congenital CMV is a major cause (20-25 %) of severe neurologic and ophthalmologic deafness, often with delayed onset (11, 12).

Congenital rubella syndrome (CRS) can lead to deafness, and cataracts, and variety of the other permanent manifestations. In developing countries, the burden of CRS has been assessed as follows: by surveillance of CRS; by surveillance of acquired rubella; by age stratified serology surveys; by serology surveys documenting the rubella susceptibility of women of childbearing age (13). The

chance of embryonic infection decreases in the second semester only to increase again in the third trimester, as known (14). When the embryo is infected early in the first trimester it does not appear to have any conventional immunological response to prevent spread of the virus (15). Yet it has been suggested that only 1 in 10^3 to 10^5 of its cells become infected (16).

Congenital toxoplasmosis is group of symptoms that occur when unborn baby is infected with parasite *Toxoplasma gondii*. The developing baby can become infected with toxoplasmosis if mother becomes infected with toxoplasmosis during pregnancy. The infection may spread to the developing baby during pregnancy itself, or during labor or delivery. Up to half of the developing babies who become infected with toxoplasmosis during pregnancy a born early (prematurely). Congenital toxoplasmosis can damage the baby's eyes, nervous system, skin, ears. Often, there are signs of infection in the baby at birth (17). Anti-toxoplasma IgG, IgM, IgA-antibody and IgG avidity were assessed using ELISA. About 85% of female population of Chandigarh is susceptible to toxoplasmosis infection and thus should be specifically educated about prevention of this infection during pregnancy (17).

Congenital syphilis is a severe, disabling and often life threatening infection seen in infants (eyes). The pregnant mother who has syphilis can spread the disease through the placenta to the unborn infant. Congenital syphilis is caused by bacterium *Treponema pallidum*, which is passed from mother to child during fetal development or at birth. Nearly half of all children infected with syphilis while they are in the womb die shortly before or after birth. Despite the fact that this disease can be cured with antibiotics if caught early, rising rates of syphilis among pregnant women in the United States have increased the number of infants born with congenital syphilis (18).

Congenital fibrosis of extraocular muscles (CFEOM) refers to at least seven genetically defined strabismus syndromes: CFEOM1A, CFEOM1B, CFEOM2, CFEOM3A, CFEOM3B, CFEOM3C, and Tukel syndrome, characterized by congenital non progressive ophthalmoplegia with or without ptosis. Individuals with CFEOM3A may also have intellectual disability, social disability, facial weakness, and/or a progressive axonal peripheral neuropathy (Charcot-Marie-Tooth disease). Individuals with CFEOM3C also have intellectual disability and facial dysmorphism reminiscent of Albright hereditary osteodystrophy like syndrome. Individuals with Tukel syndrome also have postaxial oligodactyly or oligosyndactyly of the hands, etc (19). A congenital cataract is a clouding of the lens of the eye that is present at birth. Unlike most cataracts, which occur with age, congenital cataracts are present at birth.

Congenital cataracts are rare. Congenital cataracts often occur as part of the following birth defects: chondrodysplasia syndrome, congenital rubella, Down syndrome, ectodermal dysplasia syndrome, familial congenital cataracts, galactosemia, Lowe syndrome, Marinesco-Sjogren syndrome, Pierre-Robin syndrome, etc. Patching to force the child to use the weaker eye is often needed to prevent amblyopia. The infant may need to be treated for the in-

herited disorder that is causing the cataracts (20).

3. CONCLUSION

Congenital blindness has shown permanent increase in the past 6 years, both in group with infectious, and in group with non infectious causes. Congenital blindness and visually impaired of the most common etiology-clinical the features, and among registered members of our association in central Serbia (Kragujevac) is the subject of our correlation and global trends mentioned observation of the congenital diseases.

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