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Features of deaf-blindness and hearing and vision combined impairments

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Resumo

A surdocegueira causa um profundo impacto nos indivíduos por ela acometidos. Também representa um desafio para os profissionais envolvidos com os serviços audiológicos e oftalmológicos. O presente estudo é uma revisão da recente literatura nas áreas da surdo-cegueira e de outros comprometimentos combinados da audição e visão, menos profundos. A síndrome de Usher é a causa genética mais comumente encontrada e, portanto, sua identidade é aqui descrita. Treze outras síndromes raras que incluem a surdo-cegueira são mencionadas, mais superficialmente. Algumas infecções como a síndrome de rubéola, a toxoplasmose congênita e a infecção congênita por citomegalovirus, são causas importantes que devem ser reconhecidas. A síndrome do álcool fetal é muito provavelmente uma causa normalmente negligenciada. Finalmente, o grande grupo de pessoas idosas com comprometimentos combinados, é mencionado.

Palavras-chave: *surdo-cegueira; síndrome; deficiência visual; deficiência auditiva*

Abstract

Deaf-blindness has a profound impact on the afflicted individuals. It also represents a challenge for personnel working within Audiological and Ophthalmological services. The present communication is a review of recent scientific literature covering the areas of deaf-blindness and less profound combined impairments of hearing and vision. In many cases the deaf-blindness is congenital or acquired early in childhood. An important part of this paper deals with genetic deaf-blindness. Usher syndrome is the most common genetic cause, and this entity is described. Thirteen other rare syndromes that include deaf-blindness are mentioned superficially. Infectious causes, that

are important to recognise, are congenital rubella syndrome, congenital toxoplasmosis, and congenital cytomegalovirus infection. Fetal alcohol syndrome is a probably neglected cause. Finally, a large group of elderly persons with combined impairments is mentioned.

Key words: deaf-blind; combined impairments; visual impairment; hearing loss.

The group of elderly individuals with combined impairments is large, and it is relatively unknown for health workers involved in geriatrics, aural and visual rehabilitation.

The most common cause of deaf-blindness in childhood is Usher syndrome. It is an inherited, autosomal, recessive syndrome, characterised by hearing loss, visual impairment, and vestibular dysfunction is common.

Introduction

The combination of sensory dysfunction of both hearing and vision causes extremely pronounced functional deficit and handicap. Two distinct patients groups can be discerned. One is a relatively small group of severely impaired individuals with pronounced deficits, often total deafness and blindness. The impairments often occur as different congenital syndromes or with onset at birth or early in life. Many of these syndromes are extremely rare, but since it is of the utmost importance identify them, the diagnosis of deaf-blindness is a challenge for personnel working in pediatric audiology.

The prevalence of moderate to profound bilateral hearing loss or total deafness is 1 – 2 per 10 000 births. This figure is higher in developing countries, 2.7–10/1000. About ¼ of these children have more than one handicap.

About 6% of the entire group of children with hearing loss have visual impairment. Many of these children have syndromic hearing loss. There is a genetic origin in a majority of the cases in western countries. This group of children has attracted considerable scientific interest in recent years. For many of the syndromes the chromosomal loci, as well as genes and the proteins they are encoding, have been identified. There are also extrinsic causes of deaf-blindness, most often infections. Compared to developed countries, the situation is different in developing countries and countries in rapid industrialisation, where infections often cause deaf-blindness.

The other group consists of elderly individuals with combined impairments. The extent of each of the impairments is, in a majority of the cases, less pronounced than in the deaf-blind group. The group of elderly persons with combined impairments is large, and the group is relatively unknown for personnel belonging to the services of geriatrics, aural and visual rehabilitation.

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Genetic deaf-blindness Usher syndrome

The most common cause of deaf-blindness in childhood is Usher syndrome. It is an inherited, autosomal, recessive syndrome, characterised by hearing loss, visual impairment, and vestibular dysfunction is common. The hearing loss in Usher syndrome is a bilateral, cochlear impairment, and the visual impairment is caused by retinitis pigmentosa. Cataract is also often present in Usher syndrome. Up to half of a deaf-blind population has Usher syndrome. The prevalence of Usher syndrome in developed countries is 3 to more than 4 (2.4 –6.2) per 100 000 live births. Usher syndrome is heterogeneous, i.e. different mutated genes can cause the same phenotype (Keats & Savas, 2004). In recent years the knowledge of this genetic disorder has increased enormously. To date, 8 genes and 12 independent loci have been identified (Ahmed *et al*, 2003; Reiners *et al*, 2006). Three major clinical types of Usher syndrome have been distinguished.

Usher type I (USH1). The hearing loss is profound in USH1, and the vestibular function is severely impaired (Keats & Savas, 2004). The symptoms are apparent at birth. The retinitis pigmentosa comes somewhat later, and has a pre-pubertal onset, and a progressive development. The prevalence of USH1 is about 2 per 100 000 births in developed countries. Seven different USH1 loci have been identified (Usher type IA-IG). USH1B accounts for 50% or more, and USH1D for about 1/3 of USH1 subjects.

Usher type II (USH2). The hearing loss in cases with USH2 is congenital and moderate to severe, and is stable. The vestibular function is normal (Keats & Savas, 2004). Retinitis pigmentosa has an onset at puberty. Three loci have been identified (Usher type IIA-IIC). More than 70% of the cases are USH2A.

Usher type III (USH3). The hearing loss in USH3 is progressive, and there is a variable, progressive vestibular dysfunction (Sadhegi *et al*, 2005). Retinitis pigmentosa can be variable, and is diagnosed between the 2nd and 4th decade of life. Two subgroups have been identified, USH3A-B. In most countries USH3 is relatively uncommon with a prevalence of 2-4% of all Usher cases.

Other genetic deaf-blind syndromes

There is large number of rare genetic syndromes with deaf-blindness, and a majority of them have other abnormalities as well. Information about additional important manifestations of 13 syndromes is provided in table 1. Impairments of hearing and vision occur in high frequencies in these syndromes (but not necessarily in all cases). Hearing loss is in most cases of the sensorineural type. In syndromes including craniofacial anomalies conductive or mixed hearing loss are common.

Infectious diseases and other extrinsic causes of deaf-blindness Congenital rubella syndrome (CRS)

Pregnant women who acquire a rubella infection during the first trimesters of pregnancy, are at risk to give birth to children with CRS. The syndrome includes a variety of impairments: hearing

	ANOMALIES								
	CRAN.-FACIAL	STATURE, GROWTH	CARDIAC	HEPATIC	RENA.-UROGENIT.	ENDOCR.	NEUROL	MENT.-RETARD.	HYPOGONAD
Charge	X	X	X					X	X
Alström		X	X	X	X	X	X		
Refsum			X	X			X		
Norrie							X	X	
Alport					X				
Wolfram		X			X	X			X
Noonan	X	X	X	X					X
Cornelia de Lange	X	X	X			X		X	
Craniofacial dysostoses	X						X		
NF2							X		
Wolf-Hirsch-horn	X	X	X				X	X	
Marshall & Stickler	X	X							

Table 1. Genetic syndromes in which deaf-blindness or hearing loss and visual impairment are always, or often, present. The presence of other important manifestations, occurring in a considerable percentage of the cases, is marked with X. Stature, growth includes short stature and underweight, but also obesity in one instance, Alström syndrome. NF2: Neurofibromatosis of type 2. In NF2 multiple cranial and spinal schwannomas, and also other CNS-tumours are common. Craniofacial dysostoses include craniofacial microsomia, Goldenhar syndrome, and Treacher Collins syndrome. Facial nerve dysfunction can be present in this entity.

loss, visual impairment, neurological and neuropsychiatric disabilities, cardiac abnormalities, and patent ductus arteriosus. The hearing loss is of a cochlear type, and it is in most cases severe to profound. Different visual impairments can be diagnosed in CRS: cataract, retinitis, microphthalmos, and keratoconus. The incidence of CRS has varied in different countries at different times. In the early 70ies a vaccination program was introduced in western countries, and this programme has resulted in a considerable decrease of the incidence of CRS. Morzaria *et al* (2004) have performed a comprehensive review of 43 scientific studies in English literature to determine the frequency of moderate to profound bilateral hearing loss. In 1966 – 1989 CRS was identified as the cause of hearing loss in 5.8% of children with hearing loss. In 1990 – 2002 the corresponding figure was 1.3%.

The situation has improved dramatically in countries where the immunisation programme works, but in the developing world CRS is still one of the major causes of deaf-blindness. According to WHO (2000) more than 100 000 children are born each year with CRS. This problem has been described in scientific reports from e.g. Brazil. Bento *et al* (2005) studied children where their mothers had serologically verified rubella during the first two trimesters of pregnancy. According to ABR 29.5% of the children had abnormal hearing. The hearing loss was profound in 80% of the children, and moderate to severe in 20%. Andrade *et al* (2006) studied 60 women with rubella infections, 33 before 12 weeks of gestation. In this latter group deafness was ob-

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served in three children and retinopathy in one. de Nobrega *et al* (2005) stated that CRS remains as an important cause of hearing loss among children in Brazil.

Congenital toxoplasmosis

Toxoplasmosis is caused by a parasite, Toxoplasma gondii. Primary infection with Toxoplasma gondii in pregnant women has been estimated to 0.1-1% (Stray-Pedersen, 1993). In approximately 40% of the cases the infection is transmitted to the fetus, and the risk increases if the maternal infection occurs later in pregnancy. Infants with congenital toxoplasmosis is often asymptomatic at birth, but up to 85% will develop sequelae later on (Stray-Pedersen, 1993). The most common ophthalmologic complication is chorioretinitis. Other sequelae are mental retardation and hearing loss. In a large study from Saudi Arabia 70% of children with Toxoplasma gondii had bilateral sensorinerual hearing loss (al Muhaimeed, 1996). Treatment with pyrimethamine and sulfadiazine reduces the risk of long-term complications of congenital toxoplasmosis considerably (Garweg *et al*, 2005; McCleod *et al*, 2006).

Congenital cytomegalovirus (CMV) infection

CMV is a virus belonging to the herpetoviridae family. Congenital CMV infection occurs between 0.2-2.5% of live births (Ornoy & Diav-Citrin, 2006). There is a risk of neurodevelopmental damage caused by congenital CMV, and the most common handicaps are sensorineural hearing loss, progressive chorioretinitis, and mental retardation. In a review of the literature Fowler and Boppana (2006) concluded that congenital CMV infection significantly contributes to sensorineural hearing loss in 22-65% of symptomatic children, and in 6-23% of asymptomatic children. About half of the children with CMV-related hearing loss have progression of their hearing loss. Visual impairments are common in symptomatic congenital CMV infections (22%), and uncommon in asymptomatic patients (Coats *et al*, 2000). The visual impairments observed were optic atrophy, macular scars, cortical visual impairment, and strabismus. Congenital CMV infection thus represents a serious problem.

Other infections

Severe infections like purulent meningitis and septicaemia, can cause sensorineural hearing loss, and occasionally also visual impairment. Especially pneumococcal meningitis is a serious disease, still afflicted with a substantial mortality rate.

Children surviving pneumococcal meningitis have a high rate of long-term sequelae (Pikis *et al*, 1996). In their

study the prevalence of neurological deficits was 30%. Hearing loss was present in 17%, and visual impairment in 2% of their patients.

Fetal alcohol syndrome (FAS)

Alcohol intake during pregnancy can cause fetal alcohol syndrome (FAS). The syndrome includes a number of features: craniofacial abnormalities including short palpebral fissures, telecanthus, epicanthus, short stature, and mental retardation (Strömland, 2004). Children with FAS may have impaired vision and various ocular abnormalities. All parts of the eye may be affected. Visual function may be reduced to a moderate or severe degree. FAS is associated with different types of hearing disorders: variable conductive hearing loss in conjuncture to SOM, sensorineural hearing loss, developmental delayed auditory function, and central auditory processing disorder (Church & Abel, 1998). The total prevalence of moderate to profound hearing loss associated to alcohol is very small, 0.12 – 0.14% of children with hearing loss (Morzaria *et al*, 2004), but auditory dysfunction in FAS is probably a somewhat neglected feature of the syndrome.

Combined impairments of hearing and vision in old age

There are only few studies on concomitant hearing loss and visual impairment in old age. The high incidence of presbyacusis in old age, in combination with a relatively high incidence of visual impairments in above all very advanced age, is one important reason that many elderly persons have both impairments (Klein *et al*, 1998; Klein *et al*, 2001; Bergman & Rosenhall, 2001; Berry *et al*, 2004). According to one of these investigations, the incidence of moderate to severe impairment of both senses increased from 1 % or less at age 70, to 3-6% at age 81-82, and to 8-13% at age 88 (Bergman & Rosenhall, 2001). Combined dysfunction of these two sensory systems is deleterious for both communication and orientation. These patients are secluded from most social activities, and they are at risk for severe isolation. Even mild impairments affecting both senses might result in problems in every-day activities.

Discussion and conclusions

Habilitation/rehabilitation of deaf-blind persons, including those with remaining, but impaired sensory functions, is extremely important and also demanding. Surgical treatment is possible for many ophthalmologic conditions, as well as for many patients with conductive hearing loss.

Cochlear implants is of the utmost importance for many of deaf-blind individuals. Early identification and correct diagnostics are necessary for efficient programmes. Deaf-blindness in childhood has

changed over time. The importance of rubella immunization has already been mentioned. Admiraal and Huygen (2000) have studied deaf-blind pupils in an institute in Holland. In 1986-87 heredity was the cause of deaf-blindness in 16% of the cases. In 1998-99 the corresponding figure was 31%, and the authors emphasized the importance to recognise rare hereditary conditions. Rubella was the most common cause of deaf-blindness 20 years ago (73%). In 1998-99 the figure had diminished to 39%, a figure that is nevertheless high. They also observed an increase of perinatal factors from 2% twenty years ago to 11% in recent years. According to the authors, the reason for this increase seems to be a risk of severe handicaps in very low birth weight children, who now have much higher survival rates than earlier.

An early and correct management of deaf-blind individuals is the responsibility of many professions and specialities. Doctors and other specialists from the habilitation/ rehabilitation teams should cooperate closely to achieve the best possible results.

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