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Development and characteristics of children with Usher syndrome and CHARGE syndrome

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ABSTRACT

Objective: Individuals with Usher syndrome or CHARGE syndrome are faced with a number of difficulties concerning hearing, vision, balance, and language development. The aim of the study is to describe the developmental characteristics of children with Usher syndrome and CHARGE syndrome, respectively.
Method: Data about the developmental characteristics of 26 children with Usher syndrome and 17 children with CHARGE syndrome was obtained. Associations between deafblindness (dual sensory loss), motor development (age of walking), language abilities, and intellectual outcome of these children were explored for each group independently.
Results: Both groups of children face a number of difficulties associated with vision, hearing, language, balance and intellectual outcome. Intellectual disability and/or language delay was found among 42% of the children with Usher syndrome and among 82% of the children with CHARGE syndrome. Intellectual disability was associated with language delay and age of walking for both groups.
Conclusions: Even though Usher and CHARGE are two different genetic syndromes, both groups are challenged with a number of similar developmental delays. Clinicians need to be aware of several developmental issues in order to offer adequate support to children with Usher or CHARGE syndrome.

1. Introduction

CHARGE syndrome is a multiple congenital condition. Some of the common features are coloboma, heart defects, atresia choanae, retarded growth and development, genital hypoplasia, vestibular malfunction, and ear abnormalities/deafness [1]. Individuals with CHARGE syndrome have cranial nerve abnormalities that may affect vestibular functioning, swallowing, sense of smell, facial palsy, ocular motor control and sensorineural hearing [2]. CHARGE syndrome is particularly challenging during the first years of life because of the numerous physical abnormalities, as well as several severe medical issues for example feeding problems and heart defects. Mental and/or behavioural problems may also accompany CHARGE syndrome. About 50% of the children with CHARGE syndrome were concluded as having good intellectual outcome, 25% moderate, and 25% very poor intellectual outcome [3,4]. Behaviour associated with autism, attention deficit disorder, obsessive–compulsive disorder, tic disorder, and deafblindness has been described as being specific CHARGE characteristics [5–12].

Usher syndrome is characterized by deafness and a gradual loss of vision. The hearing loss is sensorineural, whereas the vision loss is associated with retinitis pigmentosa (RP), a degeneration of the retinal cells. Three subtypes of Usher syndrome have been found [13]. People with Usher I are congenitally deaf and start to lose vision early in childhood. They also face balance difficulties due to problems in the vestibular system. Individuals with Usher II also experience hearing loss but are not profoundly deaf. They have no noticeable problems with balance. Individuals with Usher syndrome III are not congenitally deaf, but a gradual loss of hearing and vision is experienced. Some individuals with Usher III experience balance difficulties where as others do not [14,15]. Deviant behavioural characteristics have also been reported among individuals with Usher syndrome as well as a higher prevalence of psychosis (schizophrenic type picture) [16–18]. Several case studies have been published on the subject [19–21]. Intellectual disability [22], anorexia nervosa [23], and ADHD [19] have also been discussed or reported in some case-studies. Stress, anxiety, social isolation and depression are other mental health related issues reported to be associated with Usher syndrome and the loss of hearing and vision [24,25].

Usher and CHARGE syndromes are two different conditions and therefore cannot be used in a direct comparison. As they do share numerous developmental characteristics, it may be interesting in the same study to analyse the two groups individually. Both Usher

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and CHARGE syndromes affect balance, hearing and vision, and are the leading causes of acquired and congenital deafblindness, respectively. Deafblindness and balance difficulties may both be associated with the reported developmental delays.

Deafblindness: The most significant ramification of deafblindness is a severe deficiency in communication and language development [26] which may have a profound impact on a person's overall development. Dual sensory impairment has been found to be associated with different mental and behavioural problems [27,28]. Salem-Hartshorne and Jacob [29] have found a negative relationship between the degree of deafblindness and adaptive behaviour among children with CHARGE syndrome. Similar findings have also been reported by Hartshorne and Cypher [30]. Raqbi et al. [4] found that bilateral coloboma was the strongest prognostic factor for low intellectual outcome in children with CHARGE syndrome.

Balance: Because of balance difficulties children with Usher or CHARGE syndrome often learn to walk at a later age. Age of walking among children with CHARGE syndrome has, in some studies, been found to be associated with developmental outcome. Late age of walking has been shown to be associated with poor adaptive behaviour skills [11], increased autistic-like behaviour [5], sleep problems [31] and the need for psychotropic medication [32]. Thelin and Fussner [33] found in a study involving 28 children and young adults with CHARGE syndrome using a questionnaire form and follow up interviews, that the ability to walk independently was one significant factor in the development of symbolic language. Successful management of hearing loss and communication therapy initiated by three years of age was also found to be significantly related to the development of symbolic language. No studies were found investigating the association between age of walking and hindered development among children with Usher syndrome.

Too little is known about the developmental characteristics and possible associations between them for either children with Usher or CHARGE syndrome. Some studies (those introduced above) have been conducted involving children with CHARGE syndrome, but no studies could be found concerning developmental characteristics of children with Usher syndrome. The aim of this study is to describe developmental characteristics of children with Usher or CHARGE syndrome, respectively, and to explore possible associations between deafblindness, balance/motor problems, language delay and intellectual outcome.

2. Method

2.1. Participants

All children (0–18 years of age) known to have Usher or CHARGE syndrome by the Center for Deafblindness and Hearing Impairment, located in Aalborg, Denmark, were included in the study. The centre offers services for all children with Usher or CHARGE syndrome in Denmark. This amounted to 19 children with CHARGE syndrome and 26 children with Usher syndrome. All diagnoses were confirmed by clinical diagnostic procedures and in some cases, gene tests.

The children with Usher syndrome were between 3 and 17 years of age (M = 11 years, SD = 4.2). Twelve were boys. Usher type I was diagnosed in 20 cases, type II in 4 cases, there were no participants with type III. The remaining 2 cases could not be categorized but Usher type III was excluded because of stationary congenital hearing loss. Children with CHARGE syndrome were between 0 and 15 years of age. Two of the children were below 3 years of age from whom only information about vision and hearing was included in the study. For the remaining 17 children the median age was 9 years (SD = 4.2, range 3–15 years). Six were boys.

2.2. Method and procedure

Data was obtained from the medical case records gathered in a questionnaire form completed by deafblind consultants from the Center for Deafblindness and Hearing Impairment in collaboration with the parents of the children involved. The consultants had close contact to the children and their families and were all experts in children with dual sensory loss. Informed consent was obtained from parents or legal guardians. All agreed to participate. The protocol was approved by the Center for Deafblindness and Hearing Impairment. Data included:

Deafblindness (dual sensory impairment). Hearing impairment was categorized into four categories: (1) deaf (>80 dBHL), (2) severe (61–80 dBHL), (3) moderate (41–60 dBHL) or (4) no hearing impairment (<41 dBHL). Vision impairment was similarly categorized into four categories: (1) blind (worse than 1/60 or visual field $< 6^\circ$), (2) severe visual impairment (worse than 6/60 and equal to or better than 1/60 or visual field 6–10°), (3) moderate visual impairment (worse than 6/18 and equal to or better than 6/60 or visual field 11–90°), or (4) no visual impairment (equal to or better than 6/18 or visual field >90). Hearing impairment was assessed by an otolaryngologist and vision impairment by an ophthalmologist, to ensure data validity. A simple sum score of hearing and vision impairment (range 2–8) was chosen to score the degree of deafblindness.

Language delay. Because of the children's vision and hearing impairments it was not possible to find any formal test for the evaluation of language acquisition. The deafblind consultants were skilled in estimating language delay of children with dual sensory loss using informal procedures [34]. A single item scale including three categories for language abilities were used. (1) Severe language delay, (2) moderate language delay, (3) little or no language delay.

Motor development. The age when the child started to walk (three steps alone without support) was obtained as an indicator of early motor development.

Intellectual outcome. Information about diagnosed intellectual disability was obtained: no intellectual disability (IQ > 69), mild intellectual disability (IQ 50–69), or moderate to profound intellectual disability (IQ < 50). Intellectual disability was assessed by psychologists with professional experience in administering formal tests to children with dual sensory loss. Leiter-R [35] or another non-verbal instrument appropriate for use with children with hearing impairments and/or a non-visual instrument appropriate for use with children with visual impairments [36] was employed in all cases.

Additional information. Information about age, gender, language modality (oral language, sign-language, tactile language, or preverbal communication), type of school/institution (school for deaf, blind or deafblind, special school not specialized for sensory loss, or mainstream school), number of hospitalizations (a lot, some, or no/few), sleep problems (a lot, some, or no/few), and balance problems (a lot, some, or no/few) were also included.

Descriptive statistics was conducted to describe the developmental characteristics of children with Usher or CHARGE syndrome. To explore the possible associations between deafblindness, balance/motor problems, language delay and intellectual outcome non-parametric bivariate correlation tests (Kendall's tau-b) were used. A p-value of 0.05 was used. Statistics were calculated in SPSS 17.0.