

BLINDNESS AND AUTISM SPECTRUM DISORDERS

Abstract

Autism spectrum disorder (ASD) is common in children with blindness. Differentiating between autistic-like features related to blindness and "true" autistic features can be very difficult, and demands clinical experience of children with blindness and their typical development, as well as properly adapted assessment tools. The purpose of this study was to present an overview of the characteristics of blindness and ASD. A literature review was performed according to PRISMA criteria. Publications from ERIC, EBSCO-Host, Science-Direct, PROQUEST, SCOPUS, and Google Scholar have been analysed.

Recent research has indicated that the prevalence of ASD may be higher in individuals with visual impairments (48%) than in the general population (1.5%), and the risk of ASD increases with the severity of visual impairment. Also, research has shown that certain aetiologies of blindness seem to have especially high comorbidity with ASD, mainly optic nerve hypoplasia, retinopathy of prematurity, and anophthalmia.

Keywords: BLINDNESS, AUTISM SPECTRUM DISORDERS, PREVALENCE, INSTRUCTIONAL PROGRAMME

Introduction

The most commonly reported developmental disorder in children with blindness is autism spectrum disorder (ASD). During the past few decades, awareness of the high prevalence of ASD in those children has increased. ASD is a neurodevelopmental disorder characterized by deficits regarding social interaction and communication, as well as restricted and repetitive behaviours and interests (American Psychiatric Association, 2013). Blindness in itself has a significant impact on a child's development, and if the child has additional disabilities the situation is even more challenging. Congenitally blind children are generally reported to be at risk for serious behavioural and psychological problems, such as withdrawal, isolation, and ASD (Warren, 1984).

The first report of ASD in children with blindness appeared in more than 60 years ago. Namely, similarities between symptoms described in children with congenital blindness and ASD were first observed by Keeler in 1958. He described five cases from 60 premature babies completely or partially blind from birth, which presented autistic behaviours. Based on his observations Keeler made a hypothesis that children with limited sight or completely blind

from birth, emotionally neglected, and with brain damage are more likely to show autistic traits. Also, he firstly suggested that children with blindness show behaviours, such as social isolation, non-functional play with toys, frequent use of imitations, and formulaic speech or stereotyped behaviours, which were similar to those shown by children with ASD. Moreover, among those features were delayed and reversed use of personal pronouns, high rate of imitative speech or echolalia, frequent use of verbal routines and formulaic speech, an abundance of egocentric speech, scarcity, or absence of initiations of conversations, and difficulties with the use of symbolic play.

Many young children with blindness, also, display autistic-like features (ALC), sometimes referred to as "blindisms", like eye-poking, rocking or hand-flapping, as well as delayed social interaction and communication skills (McHugh and Lieberman, 2003), but these symptoms do not necessarily mean that the child has ASD. However, in some cases, the symptoms are more pronounced and cannot be explained only by the blindness, and it may be that the child meets the criteria for ASD.

Methodology

The purpose of this study was to present an overview of the characteristics of blindness and ASD. A literature review was performed according to PRISMA criteria. Publications from ERIC, EBSCO-Host, Science-Direct, PROQUEST, SCOPUS, and Google Scholar have been analysed. The inclusion and exclusion criteria were introduced and based on an analysis of the titles and abstracts related to the prevalence and diagnostic criteria of ASD in children with blindness. Evidence relating to the prevalence of ASD in children with blindness comes from intensive studies of groups of relatively small numbers of blind children, sometimes with specific diagnoses.

Results

Table 1 is describing the similarities in behaviours of children who have ASD and blindness (ASDB) and children who only have ASD, highlighting the critical areas associated with ASD.

Table 1. Similarities behaviours

Critical areas	Children with ASDB/ASD
Peer interactions including play behaviours, particularly pretend play	Both groups of children tend to lack spontaneous and imaginative play, they enjoy playing repetitively and frequently, and they do not use toys for their intended purpose; they also display little social curiosity and prefer to spend time alone rather than with others (Frith, 2003; Gense and Gense, 2002).

Joint attention and social referencing behaviours	Children who are blind and ASD and children who are sighted and ASD experience difficulties (Frith, 2003; Loots et al., 2003; Recchia, 1997).
Language and communication skills	Both these groups of children frequently exhibit echolalia and display problems with pragmatic features of conversation like topic switching and initiating conversations (Frith, 2003; Gense and Gense, 2002).
Stereotypes	Stereotypes are restricted, repetitive and stereotyped behaviours associated with children who are blind and ASD and children who are sighted and ASD (Frith, 2003; Gense and Gense, 2002). These can be motor movements, thought processes and/or elaborate routines of behaviour (Frith, 2003).

The evidence analysed suggests that certain impairments in social interaction and communication, as well as the presentation of stereotyped and repetitive behaviours, are common to both the visually impaired and those with ASD. This kind of overlapping behaviours, often referred as "autistic-like features", "autistic tendencies" or "blindisms" create a conflict between professionals of whether they can be considered part of the core diagnostic criteria for ASD in this population (Hobson and Bishop, 2003).

Differentiating between autistic-like features related to blindness and "true" autistic features can be very difficult, and demands clinical experience of children with blindness and their typical development, as well as properly adapted assessment tools (Williams et al., 2014; Matsuba, 2014). Lack of experience of the typical development patterns in children with blindness could lead to either giving the child an incorrect diagnosis or overlooking possible co-existing ASD. Either way, the child and their family are at risk of not receiving adequate support.

The similarity of these behaviours, coupled with the absence of ASD diagnostic measures designed for use with people who are blind or visually impaired, complicates the diagnosis of ASD in children who are visually impaired. Namely, the assessment of ASD in blind children is a delicate process. The most common methods for assessment of autistic behaviour include several items dependant on vision. Furthermore, no agreement has been reached as to distinguishing blindisms (stereotype, repetitive behaviour commonly found in blind children) from ASD.

The evidence relating to the prevalence of ASD in children with blindness comes from intensive studies of groups of relatively small numbers of blind children, sometimes with specific diagnoses (Table 2).

Table 2. Prevalence of ASD in children with blindness

Author	N	Prevalence of ASD/specific diagnosis
de Verdier, Ek, Löfgren, & Fernal, 2017	150	47/150 (31%) had been diagnosed with ASD; The most common causes of blindness in the ASD population were retinopathy of prematurity (ROP) in 21 children (37%), optic nerve hypoplasia (ONH) in 16 children (28%)
Jure, Pogonza, & Rapin, 2016	38	50 % overall, 65 % with ROP, and 72 % with congenital blindness (CB)
Jutley-Neilson et al., 2013	42	23/42 (54,8%) of children with ONH met diagnostic criteria for ASD
Parr, Dale, Shaffer, & Salt, 2010	83	31% of children with ONH had a clinical diagnosis of ASD
Fazi et al., 2007	24	4/28 (14,3%) of children with Leber's congenital amaurosis had a mild/moderate ASD
Johansson et al., 2006	31	10/31 (32,3%) of children with CHARGE (coloboma); Five of the 31 met diagnostic criteria for ASD and five for an autistic-like condition (ALC)
Ek, Fernal, & Jacobson, 2005	13	6/13 (46%) children with ONH, ALC was found in another three
Smith et al., 2005	13	ASD symptoms were found to be moderate to strong in 6/13 (46,2%) children with CHARGE (coloboma)

Four studies (de Verdier, Ek, Löfgren, & Fernal, 2017; Jutley-Neilson et al., 2013; Parr, Dale, Shaffer, & Salt, 2010; Ek, Fernal, & Jacobson, 2005) found a broad range of impairments common in ASD (total 40%) particularly social interaction, communication, and repetitive behaviour.

Characteristics of ASD that have been observed clinically in children with ONH include: repetitive behaviour; echolalia; “sing-song” language tone; difficulty engaging in the give and take of interactions; obsessions; self-stimulatory behaviours; hypersensitivity to certain sounds, textures, tastes, and smells; and other behaviours that are qualitatively different and result in more functional impairment than the behaviours seen in children with other types of visual impairments.

According to de Verdier et al. (2017), at least one-third of all children with blindness have been reported to meet the diagnostic criteria for ASD. Other studies have suggested that ASD prevalence is as high as 50% in congenitally blind children (Jure, Pogonza, & Rapin, 2016) compared with 1.0%-1.5% in the general population (Brugha et al., 2016). In preterm children with blindness due to ROP or ONH, the number has been described to be even higher, with three quarters having a major neurological impairment and nearly two-thirds having ASD.

Children with blindness and ASD have complex needs, and the environment has a great responsibility to interpret and meet these needs to promote development and learning. For many of these children, the surrounding world can be very confusing. A constant mix of sensory stimuli can impede learning (Gense and Gense 2005). The child's multiple processing problems can also create conflicts regarding the choice of pedagogical approach in the teaching situation (Jordan 2005).

Professionals need to explain to parents of children with blindness affected by an ASD that it is not a "disease" but define a particular group of behavioural symptoms of variable severity with differing pathophysiologies depending on its cause. They need to emphasize that ASD in blindness has special characteristics and that, all else being equal, the outlook for abatement of autistic symptoms may be somewhat better, long term, in some children with blindness than sighted children.

Conclusion

There is a long tradition of research into the presence of ASD and autistic-like clinical features among children with blindness. Keeler (1958) described five preschool children with ASD who had been born prematurely and were totally or near-totally blind from retrorenal fibroplasia arising from neonatal oxygen administration, and Chase (1972) reported that according to parent report, there was a gradient of 'autistic symptomatology' among 263 individuals with retrorenal fibroplasia (also Ek, Fernell, Jacobson, & Gillberg, 1998). Recent research has indicated that the prevalence of ASD may be higher in individuals with visual impairments (11.6%) than in the general population (0.6%); and the risk of ASD increases with the severity of visual impairment. Also, research has shown that certain aetiologies of blindness seem to have especially high comorbidity with ASD, mainly optic nerve hypoplasia, retinopathy of prematurity and anophthalmia. Nonetheless, some researchers have proposed that the connection between ASD and blindness is cognitive.

It is, of course, essential to identify and validate ASD diagnostic tools, so that clinicians can accurately diagnose ASD in children with blindness and initiate appropriate interventions. Overall, few studies are presently available to assess the potential of incorporating tactile symbols into the picture exchange communication system (PECS) for children with ASD and blindness.

References:

- AMERICAN PSYCHIATRIC ASSOCIATION (APA). (2013). *Diagnostic and statistical manual of mental disorders* (DSM-5) (5th edn.). Washington, DC: American Psychiatric Association.
- BRUGHA, T. S., SPIERS, N., BANKART, J., COOPER, S. A., McMANUS, S., SCOTT, F. J., AND TYRER, F. (2016). Epidemiology of autism in adults across age groups and ability levels. *British Journal of Psychiatry*, 209(6), pp. 498–503.
- DE VERDIER, K., EK, U., LÖFGREN, S., AND FERNELL, E. (2017). Children with blindness—major causes, developmental outcomes and implications for habilitation and educational support: A two-decade, Swedish population-based study. *Acta Ophthalmologica*, 96(3), pp. 295–300.
- EK, U., FERNELL, E., JACOBSON, L., AND GILLBERG, C. (1998). Relationship between blindness due to retinopathy of prematurity and autistic spectrum disorders: a population based study. *Developmental Medicine & Child Neurology*, 40, pp. 297–301.
- FAZZI, E., ROSSI, M., SIGNORINI, S., ROSSI, G., BIANCHI, P. E., AND LANZI, G. (2007). Leber's congenital amaurosis: is there an autistic component? *Developmental Medicine and Child Neurology*, 49, pp. 503–507.
- FRITH, U. (2003). *Autism: Explaining the Enigma* (2nd edn). Melbourne, Victoria: Blackwell Publishing.
- GENSE, M.H. AND GENSE, D.J. (2002). *Autism Spectrum Disorder in Learners with Blindness/Visual Impairments*. Washington, DC: Heldref Publications.
- GENSE, M. H., AND GENSE, D. J. (2005). *Autism spectrum disorders and visual impairment. Meeting students' learning needs*. New York, NY: AFB Press, American Foundation for the Blind.
- HOBSON, R. P., AND BISHOP, M. (2003). The pathogenesis of autism: insights from congenital blindness. *Philosophical Transactions of the Royal Society B: Biological Sciences*, 358, pp. 335–344.
- JOHANSSON, M., RASTAM, M., BILLSTEDT, E., DANIELSSON, S., STROMLAND, K., MILLER, M., ET AL. (2006). Autism spectrum disorders and underlying brain pathology in CHARGE association. *Developmental Medicine and Child Neurology*, 48, pp. 40–50.
- JORDAN, R. (2005). *Educational implications of autism and visual impairment*. In L. Pring (Ed.), *Autism and blindness: Research and reflections* (pp. 142–157). London: Whurr Publishers Ltd.
- JURE, R., POGONZA, R., AND RAPIN, I. (2016). Autism spectrum disorders (ASD) in blind children: Very high prevalence, potentially better outlook. *Journal of Autism and Developmental Disorder*, 46(3), pp. 749–759.
- JUTLEY-NEILSON, J., HARRIS, G., AND KIRK, J. (2013). The identification and measurement of autistic features in children with septo-optic dysplasia,

- optic nerve hypoplasia and isolated hypopituitarism. *Research in Developmental Disabilities*, 34, pp. 4310–4318.
- KEELER, W. R. (1958). *Autistic patterns and defective communication in blind children with retrothalamic fibroplasia*. Psychopathology of communication. New York: Grune & Stratton.
- LOOTS, G., DEVISE, I. AND SERMIJN, J. (2003). The interaction between mothers and their visually impaired infants: an intersubjective developmental perspective. *Journal of Visual Impairment and Blindness*, 97(7), pp. 403–418.
- MATSUBA, C. A. (2014). Assessment of autism in children with visual impairment. *Developmental Medicine & Child Neurology*, 56(1), pp. 8–9.
- MCHUGH, E., AND PYFER, J. (1999). The development of rocking among children who are blind. *Journal of Visual Impairment and Blindness*, 93(2), pp. 82–96.
- PARR, J. R., DALE, N. J., SHAFFER, L. M., AND SALT, A. T. (2010). Social communication difficulties and autism spectrum disorder in young children with optic nerve hypoplasia and/or septo-optic dysplasia. *Developmental Medicine & Child Neurology*, 52(10), pp. 917–921.
- RECCHIA, S.L. (1997). *Establishing intersubjective experience: developmental challenges for young blind children with congenital blindness and autism and their caregivers*, in V. Lewis and G.M. Collis (eds) *Blindness and Psychological Development*, pp. 116–29. Leicester: Psychological Society.
- SMITH, I. M., NICHOLS, S. L., ISSEKUTZ, K., AND BLAKE, K. (2005). Behavioral profiles and symptoms of autism in CHARGE syndrome. *American Journal of Medical Genetics*, 133A, pp. 248–256.
- WARREN, D. H. (1984). *Blindness and Early Childhood Development*. American Foundation for the Blind, New York, NY.
- WILLIAMS, M. E., FINK, C., ZAMORA, I., AND BORCHERT, M. (2014). Autism assessment in children with optic nerve hypoplasia and other vision impairments. *Developmental Medicine & Child Neurology*, 56(1), pp. 66–72.