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REVIEW PAPER

Autism and Visual Impairment: a Review of the Literature

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Abstract
Purpose This review presents the evidence on the prevalence of visual impairments in children and adults with Autism Spectrum Disorder (ASD), and the similar behavioural traits associated with both visual impairment and autism.
Method A systematic literature review was conducted using online databases.
Results Seven studies explored the incidence of visual impairments in people with ASD and found a higher incidence of strabismus (squint) (8.3%) than in a comparable child population (1.5 to 5.3%). Eleven studies identified behavioural traits common to both autistic and visually impaired populations. The majority were small-scale screening studies using varied methodologies, constituting an emerging field of research.
Conclusion Further large-scale, multicentre studies are required to accurately identify prevalence rates of ophthalmic conditions in people with ASD. There is a small but evolving evidence base that establishes behavioural and linguistic traits common to both visual impairment and autism.

Keywords Visual impairment · Sight loss · Autism · ASD

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Introduction

Autism is a neural developmental disability associated with impairments in communication and social interaction as well as restricted or repetitive interests and behaviours (American Psychiatric Association 2000, 2013). People with autism also frequently experience difficulties in sensory processing (American Psychiatric Association 2013). Communication and interaction difficulties may include delayed language development or a lack of spoken language and impairments in non-verbal communication such as limited eye contact, reduced facial expression and limited expressive gestures. Additionally, individuals with autism often have difficulty processing non-literal and pragmatic elements of speech (Andrews and Wyver 2005; Bowler 2007; Carvill 2001). Repetitive, stereotypical behaviours may include eye pressing, hand flicking, light gazing and rocking (Turner 1999). Similar repetitive behaviours have been evidenced in congenitally blind children, particularly eye poking, rubbing and rocking (Hobson et al. 1999; Kaplan et al. 1999; Scharre and Creedon 1992; Turner 1999). Equally present in both groups are the verbal traits of echolalia and pronominal reversal (Turner 1999). Moreover, children with severe visual impairments suffer from deficits in social and relational development similar to that observed in children with autism (Hobson et al. 1999).

Research indicates visual impairment is frequently undetected amongst children, adults and older people with intellectual disabilities and related conditions that are often referred to collectively as "complex needs" (Emerson and Robertson 2011; van Splunder et al. 2006). This population includes individuals with both autism and intellectual disabilities. Visual impairment is characteristically under-diagnosed in people with complex needs as the signs can be difficult to delineate. Diagnostic overshadowing where one condition may mask another is common in people with intellectual disabilities (Brown et al. 1997; Cass 1998; Turner 1999).

Undetected visual impairments are likely to impact quality of life, stress levels, receptiveness to new information and therefore the behaviour of individuals with autism. Awareness of sight loss and visual impairment should, therefore, be of concern to those providing care and support for people on the autism spectrum.

The present review focuses on the incidence of ophthalmic conditions in people with ASD. To the best of our knowledge, no previous systematic review has been published to-date on this issue. It is anticipated this review will identify gaps in knowledge which can be used to inform further practice, education and research in this field. It should be noted that the review deals exclusively with ophthalmic conditions; for a review of broader vision and visual processing issues, see Simmons et al. (2009).

Aim and Scope of the Review

The review aims to draw together existing knowledge of visual impairment and autism to establish the evidence base to answer two questions:

1. What is the prevalence of visual impairments in people with Autism Spectrum Disorder established in the scientific literature?
2. What behavioural traits are established as being associated with both Autism Spectrum Disorder and visual impairment?

Methods

The literature search was carried out between April and June 2015. A review of online relevant databases was conducted and this included the following: CINHAL, MEDLINE, Psychinfo, Pubmed, OVID, ERIC and EMBASE databases. Primary search terms included combinations the following key words; "autism", "autism spectrum disorder", "blindness", "sight loss" and "visual impairment".

Definitions of Autism and Visual Impairment

Autism is defined by the American Psychiatric Association's Diagnostic and Statistical Manual of Mental Disorders (DSM-IV-TR, DSM-V) (American Psychiatric Association 2000, 2013), the International Classification of Diseases 10 (ICD 10) (World Health Organisation) 2014 or recognised using ASD diagnostic tools such as the Childhood Autism Rating Scale (CARS) developed by Eric Schopler, Robert J. Reichier and Barbara Rochen Renner, and the Autism Behaviour Checklist (ABC) developed by Krug, Arick, and Almond to identify intellectually disabled individuals with high incidences of behaviour associated with autism.

Visual impairment is defined by a range of terms including "blindness", defined by the World Health Organisation (WHO) as vision of <3/60, that is being unable to see at 3 m what someone with typical vision can see at 60 m within a <5° field.

Inclusion and Exclusion Criteria

The review was restricted to papers published from 2000-2015. The timeline was selected by researchers to confine the review to recent literature.

The review inclusion and exclusion criteria were as follows:

- Type of paper: Restricted to primary research, published in English in peer-reviewed journals.
- Study Design: For question 1, only studies using quantitative research methods were included. For question 2, both quantitative and qualitative studies were included.
- For question 1, only studies which stated a measure of the prevalence of visual impairment in people with ASD and provided quantitative/statistical estimate of the prevalence were included. For question 2, only studies which directly discussed behavioural indicators of autism as well as typical behaviour associated with visual impairment were included.

Criteria for Inclusion of Autistic Research Samples

- Population: Studies were included that used the DSM-IV-TR criteria (American Psychiatric Association 2000), the International Classification of Diseases 10 (ICD 10) (World Health Organisation 2014) or recognised ASD diagnostic tools (in this case, the CARS and ABC tools) to diagnose or identify individuals as being on the autism spectrum.

Criteria for Inclusion of Visually Impaired Research Samples

- Studies were included which noted specific ophthalmic disorders or conditions such as strabismus (squint) and refractive errors (long- and short-sightedness) in participants.
- No restrictions were placed on the severity of visual impairment type. No restrictions were placed on the age, ethnicity or socioeconomic status of research subjects.

Quality Assessment

The quality of the studies was assessed using a standardised tool, *the Quality Assessment Tool for Quantitative Studies* (Thomas et al. 2004). The tool consists of nine components: (1) target population selection bias, (2) study design, (3) confounders, (4) blinding, (5) data collection methods and validity, (6) withdrawals and dropouts, (7) intervention integrity, (8) analysis and (9) global ratings. For this review, the fourth, seventh and eight components were not applicable as included studies were cross-sectional or case controlled and therefore did not test an intervention. Included studies were then rated strong, moderate or weak according to the remaining defined criteria outlined in the assessment tool. The component ratings were used to obtain an overall rating. A study was given a global rating of "strong" when there were no weak component ratings, "moderate" when there was one weak rating and "weak" when there were two or more weak component ratings (Thomas et al. 2004).

Synthesis

A narrative synthesis approach was adopted to integrate the data extracted from the qualifying studies. This approach synthesises findings from multiple studies through an examination of the texts to summarise and explain the findings (Popay et al. 2006). The following key data was extracted and collated from all the included papers: sample size; participant characteristics including gender and age, prevalence rates of coinciding visual impairment and autism; types of ophthalmic condition identified in samples; information on the methods of autism diagnosis; and behavioural traits associated with both autism and visual impairment observed in subjects.

Results

The combined searches yielded 252 citations (see Fig. 1). A total of 222 studies were excluded that did not meet the inclusion criteria or were duplicates, books or policy documents. At stage II, 52 abstracts were reviewed and a further 28 were excluded. At stage III, 24 papers were retained for secondary evaluation where a further six were excluded as they did not meet the inclusion criteria for at least one of the following reasons:

- Not primary research (× 5)
- Not within the scope of review questions (× 1)

The remaining 18 full text papers were retrieved for detailed review. Seven studies provided data on the prevalence of visual impairments in people with ASD, and 11 provided data on the behavioural traits common to both visual impairment and autism.

The evidence included in this review predominantly involves screening studies with only six studies employing a control group (Dammeyer 2014; Gal et al. 2008; Hartshorne et al. 2005; Hobson and Lee 2010; Hoevenaars-van den Boom et al. 2009; Milne et al. 2009). While the lack of control groups limits the robustness of the evidence, the screening studies provide quality information on the aetiology of a condition. Data from screening studies convey cumulative incidences of features and symptoms that indicate a relationship to a given condition (Mann 2003).

The Prevalence of Visual Impairment in People with ASD

The evidence relating to the prevalence of sight loss in people with ASD since 2000 is limited to seven studies (Black et al. 2013; Ezegwui et al. 2014; Ikeda et al. 2013; Kabatas et al. 2015; Kielinen et al. 2004; Milne et al. 2009; Mukaddes et al. 2007). Table 1 details the quality of the evidence based on the criteria proposed in the *Quality Assessment Tool for Quantitative Studies* (Thomas et al. 2004). Six studies were rated as moderate and one as strong according to the above criteria. The moderate ratings were attributed as the papers lacked information on the study design and provided no information on control for confounding variables or dropout rates.

Of the seven studies, one cross-sectional study explored the range of medical disorders in people with autism (Kielinen et al. 2004), five studies screened ASD participants for the presence of ophthalmic conditions (Black et al. 2013; Ezegwui et al. 2014; Kabatas et al. 2015; Ikeda et al. 2013; Milne et al. 2009) while one further study alternated the perspective to examine a visually impaired cohort for ASD (Mukaddes et al. 2007). Table 2 details the participant demographics, study design and findings from the studies included in the review.

Sample size varied across the studies; of the six screening studies, four included large samples which were, respectively, 324 (Kabatas et al. 2015), 257 (Mukaddes et al. 2007), 187 (Kielinen et al. 2004), and 154 (Ikeda et al. 2013). Three were smaller scale studies; Milne et al. (2009) had 51 participants. Black et al. (2013) included 44 subjects and Ezegwui et al. (2014) had 18 subjects.

All seven studies examined child/adolescent subjects with an age range from 1 to 18 years of age. Additionally, Black et al. (2013) included young adults up to the age of 22 but the distribution of ages within the sample was not specified. That all the qualifying studies included young samples means that the incidence of age-related visual impairments remains to be established in the autistic population.

Male predominance has been reported in autism (Fombonne et al. 2011), and in six of the seven studies, male

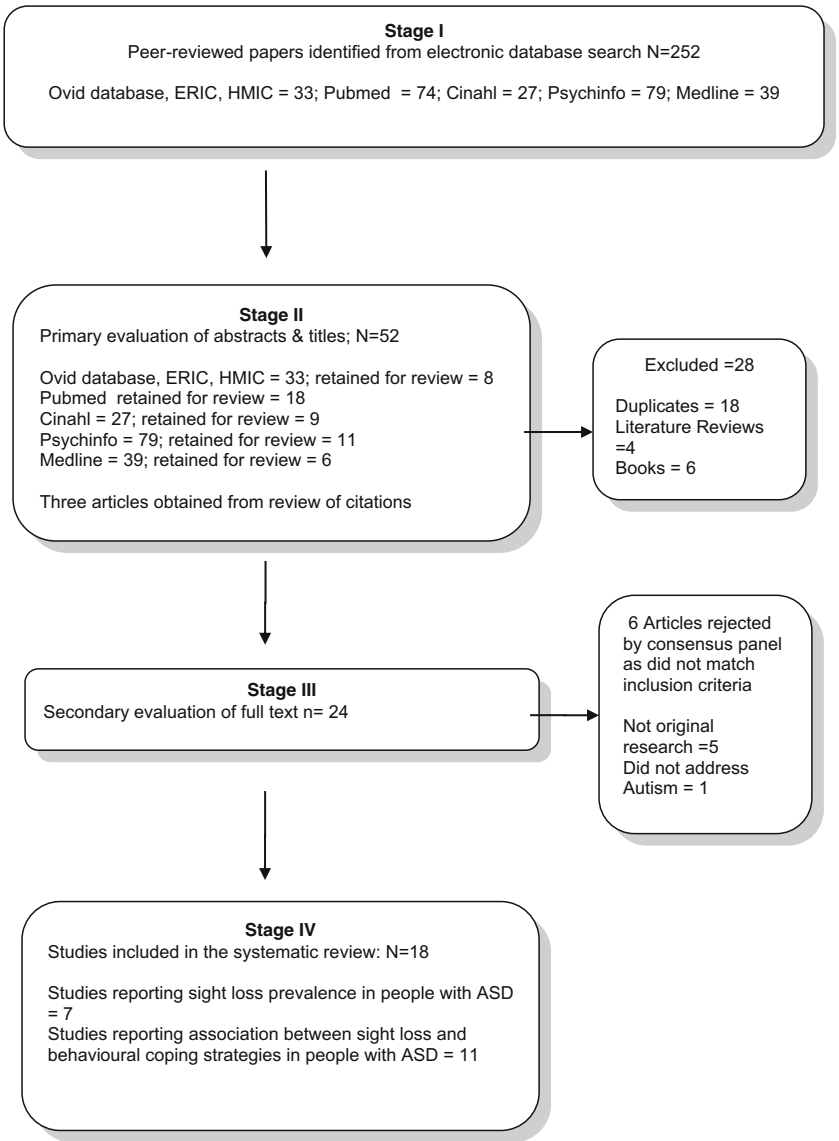

Fig. 1 Flow diagram of data retrieved at each stage of the review

subjects largely dominated. The study by Mukaddes et al. (2007) was 70% male, Milne et al. (2009) had 86% male participants in the ASD group, Ikeda et al. (2013) had 79% males, Black et al. (2013) had 75% male subjects, Ezegwui et al. (2014), a 72% male sample, and the sample in Kabatas et al. (2015) was 82% male. Kielinen et al. (2004) did not specify gender division. The gender balance of the included research samples reflects the male majority in the currently diagnosed autistic population. However, it may be noted that current research relating to possible sex differences in the autism phenotype suggests that females with autism may be under- or mis-diagnosed (Lai et al. 2015; Van Wijngaarden-Cremers et al. 2014) and that the sex ratio of males and females diagnosed with autism may change over time.

As noted above, the screening studies either examined individuals diagnosed with autism and assessed their vision or looked at a visually impaired cohort and assessed them for autism. This is a potentially rigorous study design; however, lack of detail regarding selection criteria and specificity of diagnoses limits the strength of the findings. For example, Milne et al. (2009) screened the vision in children and adolescents with ASD ($n = 51$) and a typically developing non-matched control group ($n = 44$), and noted, in the limitations, that not all participants completed all tasks and IQ data were not completed for some participants. Similarly, Ikeda et al. (2013) examined an identified autistic population from a specific clinic and retrospectively examined records for ophthalmic conditions. However, only 38% of the sample had complete eye examinations.

Table 3 documents the prevalence of refractive errors and strabismus found in each of the studied groups. As the table shows, Kabatas et al. (2015) found that 26.9% of their participants had refractive errors and 8.6% strabismus (squint); Black et al. (2013) found 27% of their sample to have

Table 1 Quality assessment of seven studies reporting the prevalence of VI in people with autism

	Study	Component rating					Global rating
		Representativeness	Study design	Confounders	Validity	Dropout	
1	Black et al. (2013)	Moderate	Moderate	Weak	Moderate	Moderate	Moderate
2	Ezegwui et al. (2014)	Moderate	Moderate	Weak	Moderate	Moderate	Moderate
3	Kabatas et al. (2015)	Moderate	Moderate	Weak	Moderate	Moderate	Moderate
4	Kielinen et al. (2004)	Strong	Moderate	Moderate	Strong	Weak	Moderate
5	Ikeda et al. (2013)	Strong	Moderate	Moderate	Moderate	Weak	Moderate
6	Milne et al. (2009)	Moderate	Moderate	Moderate	Strong	Moderate	Strong
7	Mukaddes et al. (2007)	Moderate	Moderate	Moderate	Strong	Weak	Moderate

refractive errors and 41% strabismus. Ikeda et al. (2013) identified 28.7% with refractive errors and 20.7% with strabismus. Milne et al. (2009) found 31.4% with refractive errors and 10.6% with strabismus. Kielinen et al. (2004) noted that 22.9% had refractive errors. The study of visually impaired individuals found 11% of their sample to meet the diagnostic criteria for autism (Mukaddes et al. 2007).

Summary

Overall, evidence regarding the prevalence of ophthalmic conditions in people with ASD is limited to seven studies with six of the seven achieving a moderate global quality rating. Combining the aggregate evidence from six of the seven studies suggest a refractive error in the childhood ASD populations studied at 22.9–32.7%, which is comparable with general childhood refractive error rates in 6–7 year olds at 29%, and 32.3% in 12–13 year olds (O'Donoghue 2010). Estimates of childhood strabismus in the UK is 1.5 to 5.3% (Pathai et al. 2010), and in the multi-ethnic refractive error study included here, the rate is lower at 0.14% (Xiao et al. 2015). However, the incidence of strabismus amongst the combined samples included in this review is higher at 8.3% (Friedman et al. 2009; O'Donoghue et al. 2010; Pathai et al. 2010; Xiao et al. 2015). The seventh study reviewed here examined a congenitally blind population for autism and found 11.6% with the condition, which is over ten times higher than the estimated prevalence rates for the UK (Baron-Cohen et al. 2009). The moderate global quality rating of these studies would, however, suggest that evidence is not adequate to accurately indicate a definitive prevalence rate of ophthalmic conditions in people with Autism Spectrum Disorder. The main limitations of the reviewed research include the lack of details in selection criteria and dropout rates, compliance with testing procedures and variation in testing procedures.

What Behavioural Traits Are Established as Being Associated with Both Autism Spectrum Disorder and Visual Impairment?

The 11 studies which reported common behaviours associated with both visual impairment and ASD (Table 4) were of varying quality. The majority of studies ($n = 8$) achieved a moderate rating as no information was presented on control for confounding variables such as non- matched controls in the analysis. Two studies (Fazzi et al. 2007 and Gal et al. 2008) were considered weak as, additionally, they did not give information on dropout rates.

Of the 11 studies, 6 were screening studies assessing congenitally visually impaired groups for autism (Dammeyer 2014; Fazzi et al. 2007; Hartshorne et al. 2005; Johansson et al. 2006; Smith et al. 2005), 3 were observational screening studies (Gal et al. 2008; Hoevenaars-van den Boom et al. 2009; Williams et al. 2014), 1 was a longitudinal case study which tracked development in congenitally blind children (Hobson et al. 1999), 1 was cross sectional study (Jutley-Neilson et al. 2013), and 1 was retrospective case note review (Parr et al. 2010).

Most were based on child and adolescent populations with four having an extended age range to include adults up to the age of 33 (Hartshorne et al. 2005; Hoevenaars-van den Boom et al. 2009; Johansson et al. 2006; Smith et al. 2005), giving a participant age range from 1 month to 33 years of age across

Table 2: Prevalence of sight loss in people with ASD

Author	Participants' characteristics Sample size Country Gender Age Ethnicity Ophthalmic conditions (OC) ASD diagnosis (ASD)	Study Design/Aim Primary study aim Directly/indirectly addressed sight loss in people with autism	How prevalence was measured	Study Findings	Limitations Study design Sampling Measures
1. Black et al. (2013)	**Sample size:** 44 **Country:** US **Gender:** 33 males, 11 females **Age:** 2 to 20 **Ethnicity:** Non stated **Ophthalmic conditions (OC):** Refractive error, strabismus, amblyopia, anisometropia **ASD diagnosis (ASD):** Not defined	Retrospective record review to determine the incidence of ophthalmic disorders in autistic children	Ophthalmic record review: VA = optotype testing & CSM. Refractive error = cytoplegic refraction by Titmus, fusion by Worth 4 Dot.	12 (27%), refractive error. 18 (41%) strabismus, 11% amblyopia and 7% anisometropia were found to be higher among patients with ASD	Retrospective chart review, lack of details on selection criteria Small sample size
2. Ezegwui et al. (2014)	**Sample size:** 18 **Country:** Nigeria **Gender:** 13 males, 5 females **Age**: 5-15 yrs, mean 10.28 **Ethnicity:** not stated **Ophthalmic conditions (OC):** refractive errors - astigmatism **ASD diagnosis (ASD):** DSM - IV	Screening study Aim: to identify refractive errors in children with autism in a developing country	Ophthalmic testing: Visual Acuity using LEA symbols. Hirschberg Test. Cycloplegic refraction (n=15)	Significant refractive error (33.3%), mainly astigmatism (22.2%)	Small scale screening study, not all participants completed tests
3. Kabatas et al. (2015)	**Sample size:** 324 **Country:** Turkey **Gender:** 267 male, 57 female **Age:** 18 months - 17 years; mean = 5 **Ethnicity:** Non stated **Ophthalmic conditions (OC):** refractive errors, strabismus **ASD diagnosis (ASD)**:No details	Retrospective record review to determine the incidence of ophthalmic conditions in autistic children	Ophthalmic record review of an identified autistic population.	Ophthalmic pathology noted in 26.9% of patients, of which 22% had significant refractive errors, and 8.6% had strabismus	Retrospective chart review No visual acuity testing on 91% of patients
4. Kielinen et al. (2004)	***Sample size***: 187 with autism 36 had mild VI correctable to >0.1<1 7 not correctable to >0.1 blind. ***Country:*** Finland ***Gender***: Not stated ***Age***: 3-18 years, mean 8yrs 11mths ***Ethnicity***: Not stated ***OC***: Not stated ***ASD***: DSM-IV	Retrospective review Survey to assess the association other medical disorders in children with autism).	Medical record review	187 children diagnosed with ASD, 36 had correctable refractive errors, 7 = non correctable (VI = correctable to >0.1<1Not correctable to >0.1 blind) Autism: DSM-IV	Retrospective case review. No demographic details other than class
5. Ikeda et al. (2013)	**Sample size**: 154 **Country**: US **Gender**: 122 male & 32 female **Age**: (1-12 years)	Retrospective record review to determine the incidence of ophthalmic disorders in autistic children	Ophthalmic record review of an identified autistic population.	57% had an eye related problem, 32 had strabismus, 16 had amblyopia with strabismus, 44 had significant refractive errors	Retrospective chart review (only 38% had full

Table 2: (continued)

Author	Participants' characteristics Sample size Country Gender Age Ethnicity Ophthalmic conditions (OC) ASD diagnosis (ASD)	Study Design/Aim Primary study aim Directly/indirectly addressed sight loss in people with autism	How prevalence was measured	Study Findings	Limitations Study design Sampling Measures
	Ethnicity: 115 white Caucasian, 29 black, 6 Asian/pacific islanders, 2 middle eastern **Ophthalmic conditions (OC)**: Strabismus, amblyopia, refractive error **ASD diagnosis (ASD):** Children with a diagnosis of autism - no criteria stated				ophthalmic examination) No information on ASD diagnosis criteria No comparison group
6. Milne et al. (2009)	***Sample size***: 95 (ASD group 51, TD group =44) ***Country:*** UK ***Gender***: ASD group 44 males, 7 females; TD group 13 males, 31 female ***Age***: 8-18 years ***Ethnicity***: Not stated ***OC***: Strabismus & nystagmus ***ASD***: DSM-IV - CARS	Screening study to asses visual function in autistic and non-autistic individuals	Screening study to assess visual function in children with ASD and typically developing control group	16 (31.4%) had abnormal vision. ASD group had poorer visual acuity in both left & right eyes compared to TD group. 5 (10.6%) had strabismus	IQ data not collected for all participants, not all participants completed all tasks
7. Mukkades et al. (2007)	***Sample size***: 257 ***Country***: Turkey ***Gender***: 77 girls and 180 boys ***Age***: 7-18 yrs (12.08 ±2.85) ***Ethnicity***: not stated ***OC***: congenitally blind children ***ASD***: DSM-IV	Prospective screening study to assess the prevalence and associated risk factors of autism in a sample of visually impaired children and adolescents	Vision: ICD-10, visual acuity assessed using Snellen E chart Autism: DSM-IV CARS & ABC checklist with vision questions omitted	30 of the 257 (11.6%) found to be autistic, a higher incidence than previous research	Screening study CARS & ABC checklist with vision questions omitted

the studies. Overall, there were slightly more males (368) than females (305) in the included studies. Most were small screening studies with only two studies having samples sizes larger than 100 participants (Gal et al. 2008; Hartshorne et al. 2005). Table 5 details the participant demographics, study design and findings from the studies included in the review.

The studies in this review are predominately small screening studies comparing common behaviours in both autistic and visually impaired populations (Dammeyer 2014; Fazzi et al. 2007; Hobson and Lee 2010; Hoevenaars-van den Boom et al. 2009; Johansson et al. 2006; Jutley-Neilson et al. 2013; Parr et al. 2010; Williams et al. 2014). Three of these studies explore autistic behaviours in people with optic nerve hypoplasia (ONH) and Septo-optic dysplasia (SOD) (Jutley-Neilson et al. 2013; Parr et al. 2010; Williams et al. 2014). Two studies examined deaf/blind populations (Dammeyer 2014; Hoevenaars-van den Boom et al. 2009). Four studies examined specific genetic disorders associated with a visual impairment, including three studies on CHARGE, a syndrome effecting different parts of the body that can include coloboma, heart defect, atresia choanae (also known as choanal atresia), retarded growth and development, genital abnormality, and ear abnormality (Johansson et al. 2006; Smith et al. 2005; Williams et al. 2014), and one on Leber's Amaurosis (Fazzi et al. 2007). One seminal study from 1999 (Hobson et al. 1999) reassessed the same group 8 years later for the presence of autism (Hobson and Lee 2010). The final study examined behavioural indicators of autism commonly found in children with congenital visual impairments (Gal et al. 2008).

The majority of studies in this review used the ABC checklist (Dammeyer 2014; Hartshorne et al. 2005) or CARS (Fazzi et al. 2007; Gal et al. 2008; Hobson and Lee 2010) or both (Johansson et al. 2006) to identify autism in visually impaired individuals. It should be noted, however, that these measures have a visual response component which is omitted when testing visually impaired individuals and therefore risks the reliability of the findings. The other studies (Jutley-Neilson et al. 2013; Parr et al. 2010) used the Vineland Adaptive Behaviour Scales (VABS) and Social Communication Questionnaire (SCQ), which were adapted to assess visually impaired individuals (Jutley-Neilson et al. 2013). Adaption again undermines the validity of the measure. Two final studies (Hoevenaars-van den Boom et al. 2009; Williams et al. 2014) used observational measures which were developed to detect autism in the visually impaired. These studies were conducted on small samples of <15 participants, which undermines the reliability of the findings. This further highlights the need for a measure that is responsive enough to differentiate between stereotypical behaviour stemming from neurodevelopmental impairment such as autism and those behaviours resulting from visual impairment (Cass 1998; Gal et al. 2008).

The evidence suggests that autistic traits such as limited social interaction and communication, and certain restrictive, repetitive behaviours such as eye poking, pressing, rocking are equally present in children who are congenitally blind (Fazzi et al. 2007; Hobson and Lee 2010; Parr et al. 2010). Noted developmental delay in congenitally blind children can also be observed in traits such as a lack of symbolic play, limited social interaction, and linguistic traits such as echolalia and pronominal reversal, but these may reduce with age (Fazzi et al. 2007; Hobson and Lee 2010; Williams et al. 2014).

Differentiating the causation of common behaviours as stemming from either visual impairment or from a neural developmental condition is not straightforward in deaf/blind and intellectually impaired population (Smith et al. 2005; Hartshoren et al. 2005; Johansson et al. 2006). One study (Hoevenaars-van den Boom et al. 2009) suggested it is possible to differentiate the impairments in social interaction, communication and language caused by sensory impairment from those of autism through close observational assessment (Hoevenaars-van den Boom et al. 2009); however, the small sample size and testing of a developed observational tool limit the generalisability of the findings.

Table 3 Incidence of refractive errors and strabismus (cross eyes)

	Listed in year order	Sample size (*n*)	*N* (%) refractive errors	*N* (%) strabismus
1	Black et al. (2013)	44	12 (27%)	18 (41%)
2	Ezegwui et al. (2014)	18	6 (33.3%)	–
3	Kabatas et al. (2015)	324	73 (22.5%)	28 (8.6%)
4	Kielinen et al. (2004)	187	43 (22.9%)	–
5	Ikeda et al. (2013)	154	44 (28.75%)	32 (20.7%)
6	Milne et al. (2009)	51	16 (31.4%)	5 (10.6%)
7	Mukaddes et al. (2007)	257	30 (11.6%)[a]	3 (15%)[b]
	Total	1035	237.9 (22.98%)	86 (8.3%)

– = not reported

[a] Congenitally blind children tested for autism

[b] Of the 20 who had autism 3 had strabismus

Table 4 Quality assessment of the eleven studies reporting ASD behaviours in people with visual impairments

	Study	Component rating					Global rating
		Representativeness	Study design	Confounders	Validity	Dropout	
1	Dammeyer (2014)	Moderate	Moderate	Weak	Strong	Moderate	Moderate
2	Fazzi et al. (2007)	Strong	Moderate	Weak	Strong	Weak	Weak
3	Gal et al. (2008)	Moderate	Moderate	Weak	Strong	Weak	Weak
4	Hartshorne et al. (2005)	Strong	Moderate	Weak	Strong	Weak	Weak
5	Hobson and Lee (2010)	Moderate	Moderate	Moderate	Strong	Strong	Strong
6	Hoevenaars et al. (2009)	Strong	Moderate	Weak	Strong	Strong	Moderate
7	Johansson et al. (2006)	Moderate	Moderate	Weak	Strong	Moderate	Moderate
8	Jutley-Neilson et al. (2013)	Strong	Moderate	Weak	Strong	Strong	Moderate
9	Parr et al. (2010)	Strong	Moderate	Moderate	Strong	Weak	Moderate
10	Smith et al. (2005)	Strong	Moderate	Moderate	Strong	Weak	Moderate
11	Williams et al. (2014)	Strong	Moderate	Moderate	Strong	Weak	Moderate

Two studies (Parr et al. 2010; Jutley-Neilson et al. 2013) found a broad range of impairments common in autism equally present participants with ONH and SOD (30 and 33%, respectively) particularly social interaction, communication, and repetitive behaviour. Williams et al. (2014) suggest that presentations of autistic behavioural traits may lessen with age in this group; however, findings were based on a small sample of nine.

Summary

The majority of studies on the similarity between visual impairment and autistic traits were small screening studies, focused on specific visually impaired populations. The evidence suggests that autistic traits such as limited communication and social interaction, in conjunction with repetitive, restrictive behaviours are also evident in children who are congenitally blind; however, the presence of these traits does not necessarily indicate a broad enough range of impairments to warrant an autism diagnosis.

The measures used to define autism have not been systematically tested on a visually impaired population, and the omission of visual components in the standardised autism diagnostic measures undermines their validity and reliability.

Discussion

Summary of Findings and Limitations

This review has comprehensively examined ophthalmic conditions in people with Autism Spectrum Disorders. Collating the evidence from six of the seven prevalence studies suggests a refractive error rate in the childhood ASD population studied at 22.9–32.7%, which is comparable with general childhood refractive error rates in 6–7 year olds at 29%, and 32.3% in 12–13 year olds (O'Donoghue 2010). Estimates of childhood strabismus in the UK is 1.5 to 5.3% (Pathai et al. 2010), but in the evidence collated in this review, the incidence of strabismus amongst autistic participants is higher at 8.3% (Friedman et al. 2009; O'Donoghue et al. 2010; Pathai et al. 2010; Xiao et al. 2015). The single study examining a congenitally blind population for autism found 11.6% meeting the criteria for a diagnosis of ASD, far higher than the estimated prevalence rates of around 1% for the UK (Baron-Cohen et al. 2009). It should be noted that the review evidence is predominately based on child and adolescent populations. There were no studies examining ophthalmic conditions and adult autistic populations who are more at risk of age-related visual impairments.

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 Autism Spectrum Disorder. This research field is dominated by small screening studies on populations with specific congenital visual impairments, which limits the generalisability of the findings. There is a need for a validated measure to assess for ASD which has been tested on the visually impaired populations and is not undermined by the exclusion of a visual response (Hartshorne et al. 2005; Johansson et al. 2006; Gal et al. 2008; Hobson and Lee 2010; Dammeyer 2014). Additionally, there is some evidence that retesting in older children and adolescents is appropriate for the severely visually impaired as difficulties with communication and social interaction may be due to developmental delay and reduce with age (Fazzi et al. 2007; Hobson and Lee 2010; Williams et al. 2014). It should also be noted that available evidence lacks information on participant

Table 5: Summary of studies reporting the association between sight loss and autistic behavioural presentations

Author	Participants' characteristics Sample size Country Gender Age Ethnicity Ophthalmic Condition (OC) Behavioural Traits (BT)	Aims Study Design	Study Findings	Limitations Design Sampling Measures
1. Dammeyer (2014)	***Sample size***:71 (two groups, deafblind with autism & deafblind without autism) ***Country***: Denmark ***Gender***: 35 males, 36 females ***Age*****:** 3-17 yr olds (mean 11.3 yrs, SD=4.5) ***Ethnicity****:* Not stated ***OC***: None stated but visual acuity defined **:** Blind = vision worse than 1/60. Residual vision = vision equal to or better than 1/60 **BT**: social interaction, communication. Restricted & repetitive behaviour	Case control Aim to examine symptoms of autism among children with congenital deaf-blindness using the ABC checklist	Children with congenital deaf/blindness found to have symptoms of autism	Screening study. Difference in sample size in methods (71)and analysis (69) no explanation for dropout
2. Fazzi et al. (2007)	***Sample size***: 24 ***Country***: Italy ***Gender***: 13 males, 11 females ***Age***: 2-11yrs ***Ethnicity***: 23 white European, 1 Asian ***OC***: Leber's Congenital Amaurosis ***BT***: eye pressing/poking Repetitive hand/finger movements (fluttering), Manipulation of objects, Head/body rocking, sniffing, crying/moaning, rubbing movements, light gazing, banging objects, echolalia, jumping, grimacing	Screening study Assessment of stereotypical behaviours was based on direct observations & parental observations CARS	20 children were non autistic (scores less than 28), four presented mild/moderate autism (scores between 28-35) Delay in symbolic play, language, social interaction. Eye poking, eye pressing not indicative of autism	Small screening Study No control group
3. Gal et al. (2008)	***Sample size***: 221 (5 groups) Typical children = 30, ID=29,VI=50, Hearing Imp= 51, Autism= 56 Sum = 216 ***Country***: Israel ***Gender***: 129 males, 92 females ***Age***: 6-13 yrs (mean=9.4, SD=1.81) ***Ethnicity***: not stated ***OC***: binocular visual acuity <0.3 ***BT***: stereotyped movements i.e. rhythmic and repetitive	Assess stereotypical movements associated with Autism in other impairment disorders through an observational study 35-60 min observations Autism diagnosed using: CARS & DSM-IV	Autism group exceeded others in self injurious behaviours 15 stereotyped movements (SM), VI 5 SM and hearing group on 1 SM Common behaviours = repetitive manipulation of objects, pace/move repetitively, touch/body clothing repetitively, pull hair, pinch	Inconsistency in sample reporting. Difficult to assess analyses as not enough information on the scoring of measures used
4. Hartshorne et al. (2005)	***Sample size***: 160 with CHARGE ***Country***: US (from 32 states) ***Gender***: 85 male & 75 female	To examine the autistic-like behaviours of children with CHARGE to see if they are similar to, or distinct from, those who are	Children with CHARGE frequently demonstrate behaviours that are autistic-like; repetitive movements, withdrawal, tantrums	Screening study ABC checklist not standardised for sensory impaired.

Table 5: (continued)

	Age: 3 to 33 yrs, mean 10.9 SD =5.6 ***Ethnicity***: 93% Caucasian, ***OC***: CHARGE - 85% Coloboma ***BT***: repetitive movements (rocking, spinning, flapping. Withdrawal	diagnosed with autism and deaf/blindness. ABC checklist		Self-report survey completed by parents. No details given on comparison groups autistic & deaf/blind
5. Hobson and Lee (2010)	***Sample size***: 9 congenitally blind children ***Country***: UK ***Gender***: 4 males and 5 females ***Age***: 3 to 9 years ***Ethnicity***: not stated ***OC***: Congenitally blind, condition not defined ***BT***: echolalia repetitive, restrictive movements, lack of imaginative play	Longitudinal study to assess whether 9 congenitally blind children continued to demonstrate symptoms of autism CARS	At time 1(T1) 9 congenitally blind children met the diagnostic criteria for autism DSM-III-R at time 2, 8 years later only one of the 9 met the criteria for autism	Small screening study n=9
6. Hoevenaars--van den Boom et al. (2009)	***Sample Size***: 10, 5 with deafblindness & autism & 5 deablindness ***Country***: The Netherlands ***Gender***: 5 male, 5 female ***Age***: 7-28 yrs (mean age with autism= 18yrs, without autism = 15yrs) **OC**: Vision: <0.3 **BT**: Autism diagnosed using clinical observation and CARS	Distinguish specific behaviours by which people with dual diagnosis of deaf/blindness & autism can be identified/or not (O-ADB) Observation of characteristics of autism in persons with deaf-blindness	All deaf/blind participants showed impairments in social interaction, communication & language. Found large "overlap" in overt behaviours between people with deaf/blindness and persons with autism	Small screening study. No validation information on developed observational measurement.
7. Johansson et al. (2006)	***Sample Size***: 31 diagnosed with CHARGE ***Country***: Sweden ***Gender***: 15 males; 16 females ***Age***: 1month to 31years of age, mean age 8y 11mths **OC**: Coloboma **BT**: None stated	Aim: to use current standardised autism diagnostic instruments to analyse the relation between CHARGE and ASDs ABC & CARS	Five of the 31 met diagnostic criteria for ASD and five for an autistic-like condition, and seven for autistic traits ABC & CARS DSM-IV checklist for autistic disorder	Small screening study
Author	**Participants' characteristics**	**Aims**	**Study Findings**	**Limitations**
8. Jutley-Neilson et al. (2013)	***Sample***: 42 ***Country***: UK ***Gender***: 24 male, 18 female ***Age***: mean 9.1 SD=3.1 (3-16 yrs) ***OC***: Septo-optic dysplasia (SOD) = 28, and Optic nerve Hypoplasia (ONH) = 14 ***BT***: repetitive, restrictive behaviours, communication, reciprocal social interaction	Cross-sectional study; Aim: to "highlight" the occurrence of autism in children with SOD & ONH VABS & ABC	Using the SGQ 23 children met the cut-off point for ASD and 9 met the cut-off point for autism. The SCQ lost its sensitivity and specificity in children with greater visual loss	Small cross sectional study
9. Parr et al. (2010)	***Sample size***:83 ***Country***: UK ***Gender***: 45 females, 38 males ***Age***: mean age 3y 5 m; range =10m-6y 10m. ***Ethnicity***: Not stated	Retrospective case note review of clinical behaviour reports of social, communication and repetitive behaviours in children with ONH, SOD ICD-10	58% of children had one Social, communicative and/or repetitive or restricted behavioural difficulties (SCRR) and 31% had a diagnosis of ASD	Case note review No standardised assessment of autism

Table 5: (continued)

	OC: ONH, SOD ***BT***: social, communication, repetitive, restrictive behaviours (not defined)			
10. Smith et al. (2005)	***Sample size***: 13 ***Country***: Canada ***Gender***: 8 males & 5 females ***Age***: 2yrs 9mths to 24 yrs, mean age 9 years ***Ethnicity***: Not stated ***OC***: CHARGE ***BT***: low adaptive behavioural skills, motor impairments	Screening study to assess behavioural profiles associated with CHARGE	ASD symptoms were found to be moderate to strong in six of the ten children who were above the age of 4-5 years.	Small screening study n=13
11. Williams et al. (2014)	***Sample size***: 9 children ***Country***: US ***Gender***: 5 males, 4 females ***Age***: 5-9 years. ***Ethnicity***: Not stated ***OC***: ONH, SOD ***BT***: social, communication and repetitive behaviours	Aim: to examine the utility of standard autism diagnostic measures in 9 children with severe vision loss ADOS = Autism Diagnostic Observation schedule	Found the modified autism measures (ADOS and ADI-R) demonstrated good agreement with clinical diagnoses	Small screening study n=9, no matched control

demographics such as ethnicity, social class, IQ level or accounting for the wide age ranges in both of the subject group and control groups (if present) all of which limits the generalisabilty of the findings (Rogers and Ozonoff 2005).

Implications for Autism Support Services

Given the current reported difficulties in differentiating whether certain developmental and behavioural features are caused by ophthalmic visual impairment or other neurological differences, it is important that individuals diagnosed with autism access regular optometry screening so that ophthalmic conditions can be either diagnosed or ruled out as being associated with an individual's day-today functioning.

Professionals supporting people with autism, particularly individuals with complex needs and limited verbal communication, need to recognise the potential for visual impairments amongst those that they support. An awareness of the similarity in traits means that autism support practitioners must be wary of attributing all impaired communication or stereotypy to an individual's autism diagnosis without considering the possibility of coincidental visual impairment—a phenomenon known as "diagnostic overshadowing" (Hepburn et al. 2014; NICE 2012).

Undiagnosed visual impairment is likely to severely impact quality of life. There is a need therefore for education and training that equip autism support practitioners with the awareness and skills to identify potential visual impairment, to refer individuals to optometry professionals if necessary, and to make necessary adjustments to service environments and support practices for individuals identified as having a visual impairment. Specialist autism services and optometry services are well placed to work collaboratively in order to develop eye care pathways as a means to promote equity of access to optometry assessment and to ensure autism-friendly optometry services (NICE 2012; Venkat et al. 2012). Successful optometry processes are likely to require the presence of carers or autism practitioners with knowledge of an individual that they support and skills to address issues such as increased anxiety when attending appointments for optometry assessment (Chiri and Warfield 2011). Optometrists may also need to undertake reasonable adjustments to diagnostic processes that enable access for assessment for people with autism (Coulter 2009; Turner and Robinson 2011).

Directions for Future Research

In systematically identifying and reviewing the literature relating to visual impairments in people with autism, it is evident that further research is required to investigate the prevalence of co-occurring autism and visual impairment. Researchers should also seek to ascertain whether it is possible to develop screening and diagnostic processes that can differentiate where common behavioural traits stem from visual impairment and where such traits

are rooted in other, neurodevelopmental differences that underlie Autism Spectrum Disorder. This may be achieved through standardised autism diagnostic measures that incorporate components for the assessment of sensory-impaired individuals. Further research is needed that provides detailed demographics of the subject groups under observation and the exact diagnoses of the visual impairment. It is hoped that the findings from this systematic review will raise awareness of the extent to which visual impairment may be experienced by people with autism, and this in turn will inform professional practice ensuring that individuals with autism and visual impairment receive appropriate diagnosis and support.

Compliance with Ethical Standards

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