

**Critical Review:
What screening instruments are most effective for the dual diagnosis of Down syndrome and Autism Spectrum Disorders?**

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This critical review examines the effectiveness of various screening tools used for the dual diagnosis of Down syndrome (DS) and Autism Spectrum Disorder (ASD). Study designs consisted of qualitative research. Results of the studies reviewed provide suggestive to compelling evidence to support the use of the Aberrant Behaviour Checklist (ABC) to assist with a dual diagnosis of DS and ASD over other screening instruments. Recommendations for future research and clinical implications are discussed.

Introduction

Down syndrome (DS) is caused by the presence of an additional copy of chromosome 21 and is the most common chromosomal cause of intellectual disability (Moss & Howlin, 2009). Individuals with DS are typically described as having friendly, affectionate and extroverted personalities; however, evidence suggests that not all individuals with DS possess these personality traits (Reilly, 2009). Studies have found that 10 to 20 percent of individuals with DS have significant behavioural issues (Reilly, 2009).

Previously, the co-occurrence of DS and Autism Spectrum Disorder (ASD) was questioned due to the cognitive impairments in these individuals, yet recent studies have demonstrated that individuals with DS may also meet the diagnostic criteria for ASD. Prevalence rates of co-occurring DS and ASD range from 5 to 39 percent depending on the screening tool, standardized assessment measure and diagnostic criteria being applied (Moss & Howlin, 2009).

Much of the published research on screening for a dual diagnosis of DS and ASD have used questionnaires or checklists and are utilized by parents, caregivers, teachers, nurses and clinicians or any others who have knowledge and experience working with the individual being assessed. Screening instruments are intended to assist in the identification process of children who present with developmental delays or atypical behaviours and for whom a further assessment and possible diagnosis of an ASD may be warranted. Those who meet the cutoff criteria on a screening instrument would then be referred for a more intensive diagnostic evaluation.

These screening instruments tap into areas covering language delay, motor “clumsiness”, and friendships (Reilly, 2009). Such screening tools include The Autism Spectrum Screening Questionnaire (ASSQ), The Scale of Pervasive Developmental Disorder in

Mentally Retarded Persons (PDD-MRS), The Modified Checklist for Autism in Toddlers (MCHAT) and the Aberrant Behaviour Checklist (ABC) among others. There are many clinical implications of a dual diagnosis of DS and ASD. Early identification of ASD in individuals with DS may result in more individualized intervention plans and proper service delivery (Reilly, 2009). This paper aims to critically evaluate which screening tools are most accurate in assisting with the identification of a dual diagnosis of DS and ASD.

Objectives

The primary objective of this paper is to provide a critical evaluation of existing literature regarding the effectiveness of screening tools for the dual diagnosis of DS and ASD. The secondary objective is to offer recommendations for clinical practice and future research.

Methods

Search Strategy

Computerized databases including PubMed, Web of Knowledge and Scholars Portal were searched using the following terms: (Autism Spectrum Disorder) OR (ASD) AND (Down syndrome) AND (dual diagnosis) OR (comorbidity) OR (screening tool). The search was limited to articles written in English.

Selection Criteria

Studies selected for inclusion in this critical review were required to investigate the effectiveness of various screening tools used to assist with a dual diagnosis of ASD and DS/mental retardation. No limits were placed on the screening tool, study design, or outcome measures.

Data Collection

Results of the literature search yielded seven articles that achieved the aforementioned selection criteria for inclusion in this review. These included qualitative research designs. The following screening tools were reviewed: The Autism Spectrum Screening

Questionnaire (ASSQ), The Childhood Autism Rating Scale (CARS), The Scale of Pervasive Developmental Disorders for Mentally Retarded Persons (PDD-MRS), The Modified Checklist for Autism in Toddlers (MCHAT), The Social Communication Questionnaire (SCQ), and The Aberrant Behaviour Checklist (ABC).

Results

Autism Spectrum Screening Questionnaire (ASSQ)

The ASSQ is a 27-item screening instrument designed for use by parents and teachers to screen for social deficits associated with ASD (Kent et al., 1999). The checklist can be utilized for individuals between the ages of 6 and 17.

The Childhood Autism Rating Scale (CARS)

The CARS is a screening tool based on behavioural observation and interview and covers 14 domains generally affected in individuals with severe autism. The total score provides a rating in one of three categories; non-autistic, mild to moderately autistic or severely autistic. The CARS should not be used as a diagnostic tool for ASD, but can be useful when determining the severity of an ASD (Kent et al., 1999).

Kent et al. (1999) employed a qualitative research design using the ASSQ and CARS screening instruments to identify the comorbidity of ASD in a population of children with DS. The ASSQ and CARS were completed and a final diagnosis was made according to WHO's International Classification of Disease 10 (ICD-10) criteria following interview and observation. 33 of 58 identified participants completed the measures; four of whom received a diagnosis of an ASD. Forward stepwise logistic regression was utilized to determine which ASSQ items best predicted a diagnosis of ASD in the population with DS. The item "lives somewhat in a world of his own with restricted idiosyncratic intellectual interests" was the best predictor. Linear regression was also used to identify which items on the ASSQ best predicted severity of the ASD assessed by the CARS. Results indicated that "clumsy, ill-coordinated movements, lives in a world of his own with restricted idiosyncratic interests, invents idiosyncratic words and expressions and has a deviant style of gaze" were the best predictors of severity (Kent et al., 1999). Items pertaining to social withdrawal, restricted or repetitive interests, clumsiness and unusual eye contact were associated with an ASD. The overall correlation between the ASSQ and the CARS was good ($r=0.40$, $P<0.05$).

Appropriate statistical analyses were conducted to determine the items that best predicted ASD in the population of individuals with DS. Although it may be

beneficial to examine which items best predicted ASD in the DS population, sensitivity, specificity and validity measures of the screening tools were not evaluated. These measures would be beneficial for clinicians to consider before using these screening tools with individuals with DS. This sample size was small, which could also be considered a limitation to this study.

Although the results of this study identify specific characteristics that may be useful for dual diagnosis of ASD and DS, the overall results provide equivocal evidence for use of these screening tools in clinical practice due to the limitations discussed.

The Scale of Pervasive Developmental Disorder in Mentally Retarded Persons (PDD-MRS)

The PDD-MRS is a 12 item classification and screening instrument used for identification of ASD in persons with mental retardation (MR) from mild to profound levels (Kraijer & de Bildt, 2005). Items on the PDD-MRS were devised based on the DSM and ICD-10 criteria and it can be used for individuals aged 2 to 55 years.

Kraijer & de Bildt (2005) aimed to develop a screening tool to identify ASD in persons with MR. Participants includes 1230 subjects; including 254 individuals with DS. Appropriate statistical analyses were performed to determine the reliability and validity of the PDD-MRS. Internal consistency was determined for persons with functional speech ($n=658$) and for those without speech ($n=572$); Cronbach's alpha was found to be 0.86 and 0.8, respectively. Interrater reliability yielded Pearson's r values from 0.83-0.89. Sensitivity and specificity were also evaluated and were found to be 92.4 and 92.4, respectively. The concurrent and discriminative validity of the scale was deemed to be satisfactory. Overall, Kraijer & de Bildt (2005) concluded that the PDD-MRS is a reliable and valid instrument for screening PDD in individuals with MR.

The results of this study provide suggestive evidence that the PDD-MRS can be used as a valid and reliable screening instrument for the dual diagnosis of PDD in individuals with MR. Although a large sample size of individuals with MR was used, evidence for use with a dual diagnosis of DS and ASD is equivocal at this time, as results specific to the DS population were not discussed.

Modified Checklist for Autism in Toddlers (MCHAT)

The MCHAT is designed to screen for autism in toddlers 16 to 30 months of age. Parents respond to 23 yes/no items which focus on joint attention, social

orienting and imitation. Failing any three items or any two of six critical items indicates a positive screen. The authors of the MCHAT indicate that the instrument purposefully yields more false-positives.

The Social Communication Questionnaire (SCQ)

The SCQ is a 40 yes/no item screening tool based on the Autism Diagnostic Interview-Revised (ADI-R); a popular diagnostic tool for ASD in children aged four to five years old (DiGuseppi et al., 2010). Items on the SCQ related to social reciprocity, communication disturbance and repetitive behaviours. A score of 15 is used as the cutoff. The SCQ strongly discriminates between ASD and non-ASD individuals with sensitivity and specificity ranging from 0.85-0.88 and 0.72 to 0.75, respectively.

DiGuseppi et al. (2010) used the SCQ and the MCHAT screening tools to determine the prevalence of ASD and screening test characteristics in children with DS. A sample of 123 children with DS were screened using the MCHAT or SCQ as appropriate by age. This was followed by a comprehensive assessment using the Autism Diagnostic Schedule, Generic (ADOS-G) or the Autism Diagnostic Interview, Revised (ADI-R).

Results indicate that significantly more screen positive children than screen negative children were diagnosed with ASD or PDD-NOS. Results indicated that the MCHAT and SCQ were highly sensitive (87.5%) in identifying comorbidity, however false positives were also noted, as specificity was 49.9%. The authors noted that ASD prevalence rates increased with greater cognitive impairment. This limitation should be considered then utilizing these screening tools in the future. The authors acknowledged the high false positives rates and suggested that if these screening tools are to be implemented universally in the future, specific ASD screening procedures and improved diagnostic discrimination characteristics for children with DS should be considered (DiGuseppi et al., 2010).

Overall, research by DiGuseppi et al. (2010) provides equivocal evidence for the use of the MCHAT and SCQ to assist with the dual diagnosis of DS and ASD due to the high rate of false positives and suggested modifications needed in order to implement this screening tool with the DS population.

Aberrant Behaviour Checklist (ABC)

The ABC is one of the most commonly used tools for evaluating maladaptive behaviours in individuals with moderate to profound intellectual disabilities. It is a 58 item rating scale that assesses severity of maladaptive behaviours on five subscales: Irritability, Lethargy/Social Withdrawal, Stereotypy, Hyperactivity and

Inappropriate Speech (Ji et al., 2011). The ABC was used to determine criterion validity in a population of persons with DS (n= 159) with good results (Capone et al., 2005). Results of the literature search yielded the most articles pertaining to the ABC and its use with dually diagnosed populations.

Rojahn & Helsel (1991) investigated the appropriateness of the ABC for use with dually diagnosed individuals with MR and psychiatric disturbance. A sample of 204 patients from a child psychiatry unit were rated on the ABC twice daily by care staff. Internal consistency was found to be satisfactory with alpha coefficients ranging from 0.82-0.9. Interrater reliability varied between all subscales and was found to be relatively low (Pearson correlates between 0.39 and 0.61). Criterion validity was reported to be good. The ABC was sensitive to psychiatric diagnoses and age and the original 5-factor structure was found to be robust with congruence coefficients ranging from 0.80-0.89.

Rojahn & Helsel (1991) concluded that the ABC can be used with children and adolescents with MR and psychiatric impairments given that the factor structure was robust, the subscales were clinically meaningful and the internal consistency of the subscales was satisfactory. The Inappropriate Speech subscale appeared to contribute only a modest amount of information towards the dual diagnosis of ASD and MR or psychiatric disturbances, and it was suggested that with future revisions of the ABC, this subscale be refined.

The authors clearly discussed the objectives they wished to examine with their study. Appropriate tests were used to determine significance. This study also had a moderate sample size of dually diagnosed individuals. A limitation to the selected population is that all of the individuals were selected from a single child psychiatric unit and the participants did not consist solely of individuals with Down syndrome. The authors also discussed that they used an "unusual" method of measuring interrater reliability and indicated that the results may not be statistically valid. The authors clearly outlined the limitations of their research and presented appropriate causes for these limitations. Overall, this study provides equivocal evidence for clinicians when using the ABC to assist with a dual diagnosis of DS and ASD.

Marshburn & Aman (1992) investigated the use of the ABC in a sample of community children with MR. Teachers completed ratings on 666 students with developmental disabilities. A four-factor solution of the ABC was obtained and a factor corresponding to

Inappropriate Speech did not emerge. This finding is consistent with authors Rojahn & Helsel (1991). Congruence between the four derived factors and corresponding factors on the original five factor ABC was high with congruence coefficients ranging from 0.87 to 0.96. A series of ANOVAs were performed to evaluate the effect of subject characteristics on subscale scores.

The results of Marshburn & Aman's (1992) study are suggestive that a four factor analysis is best for dually diagnosed individuals with MR. This study utilized a large sample size. A limitation of this study is that the participants did not consist solely of individuals with Down syndrome. Exact numbers of individuals with Down syndrome was not able to be obtained. The authors present reasonable limitations to their study and possible causes for such limitations.

Results of this study provide equivocal evidence that the Aberrant Behaviour Checklist (ABC) can be applied to individuals with Down syndrome, however it is suggestive that the ABC is the most widely used screening instrument for individuals with MR. Statistical analysis of the four factor ABC reveal that it is a reliable and valid screening tool for the use of identifying a dual diagnosis for persons with MR.

Capone et al. (2005) aimed to determine the cognitive and behavioural characteristics of children with co-occurring DS and ASD. Participants included 61 individuals with DS and ASD, diagnosed according to DSM-IV criteria, 26 individuals with DS + Stereotypic Movement Disorder (SMD) and 44 typical DS controls without behaviour disorders. The study aimed to determine the accuracy of the ABC to characterize individuals with a dual diagnosis of DS + ASD and also aimed to test the hypothesis that the ABC could differentiate individuals with DS + ASD from their typical DS peers.

Parents completed the ABC and scores indicated that individuals with DS and ASD had significantly higher scores on all subscales than the other subgroups (Capone et al., 2005). Each of the subscales were highly significant in distinguishing between DS+ASD and typical groups. Appropriate tests were used to determine significance for each of the subscales of the ABC. Results indicated that the Lethargy and Stereotypy subscales of the ABC were highly significant in distinguishing DS+ASD from other subgroups. The Inappropriate Speech subscale was the only subscale that was not significantly different among groups. This is consistent with other researchers' findings (Rojahn & Helsel, 1991, Marshburn & Aman, 1992).

Although the sample size was relatively small, Capone et al. (2005) utilized appropriate participant groups to perform appropriate statistical analysis. The researchers excluded individuals whose behaviour could better be explained by a primary diagnosis of depression, obsessive-compulsive disorder etc., which could adversely affect their results. Overall, results indicate that there is a good correlation between the DSM-IV criteria for ASD and the ABC screening tool. This study also presents suggestive evidence that the ABC can distinguish ASD + DS from DS + other disorders and can identify which items best predicted DS and ASD compared to DS and other behavioural disorders. Research by Capone et al (2005) provides suggestive to compelling evidence that the ABC may be a useful tool for clinicians when screening for a dual diagnosis of DS and ASD as well as to rule out DS + other disorders.

Ji et al. (2011) employed a qualitative research design in order to determine whether a DSM diagnosis of ASD would be valid for individuals with DS due to a higher prevalence of cognitive impairments in this population. Participants included 293 individuals with DS who had attended the Kennedy Krieger Institute Down syndrome Clinic during a 16 year period. Clinical diagnoses and group assignments to DS + ASD, DS+ none, DS + Disruptive Behaviour Disorder (DBD) or DS + SMD were made by a single evaluator, based on DSM-IV criteria. Scores on the ABC were used to categorize the participants into three diagnostic groups: DS+ none, DS+ASD and DS+DBD. Based on the scores on the ABC, a sequential factor and cluster analyses were utilized. Results indicated that not only were researchers able to clearly identify which participants were characterized as DS+ASD, but also could distinguish between two other behavioural categories. The researchers concluded that despite the cognitive impairments of individuals with DS, DSM-based criteria of ASD are applicable to that population.

This study presented concrete research objectives and clearly discussed how they would be measured. Appropriate statistical analyses were also conducted. Authors validated their findings with DSM-IV criteria, which is the "gold standard" diagnostic tool for ASD in Canada. The use of a single evaluator positively impacted interrater reliability measures and is encouraged in the future. Results of this study provide suggestive to compelling evidence for the usage of the ABC for screening individuals for a dual diagnosis of DS and ASD and to rule out DS + other disorders.

Discussion

Overall, the research presented provides variable support for the use of screening tools to assist with dually diagnosing DS and ASD. The examined research consisted of qualitative design, which is considered a Level 4 in methodological design. This type of design is appropriate for the purpose of this research in developing standardized screening instruments.

Despite the limitations discussed with each research study, the majority of the literature reviewed suggests that the Aberrant Behaviour Checklist (ABC) is the best suited screening tool for use with this population. The evidence supports the use of the ABC over others due to its sound psychometric properties and demonstrated use with the DS population. Not only was the ABC able to identify individuals with DS and co-occurring ASD, it was also able to differentiate between a dual diagnosis of DS and other behaviour disorders. The ability to differentiate between these subgroups is beneficial for clinicians when planning assessment and intervention strategies.

Researchers continue to encourage cautionary measures when interpreting scores on screening instruments as the lower cognitive functioning of individuals with DS may influence their behavioural characteristics, leading to an over-identification of dually diagnosed individuals. Although screening tools and questionnaires can help guide parents and clinicians when making referrals for further testing, the “gold standard” as suggested by Reilly (2009) is a clinical diagnosis made by a multidisciplinary team of clinicians with experience working with both individuals with DS and individuals with ASD, utilizing the DSM-IV criteria for diagnosis.

Recommendations

- 1) Researchers should consider the use of a single rater for future research projects in order to increase interrater reliability measures of screening tools.
- 2) Future research should continue with a larger population sample in order to support the wider use of screening tools.
- 3) Research should continue regarding the use of a four-factor ABC screening tool, without the Inappropriate Speech subscale, for the use with individuals with DS.
- 4) Caution should be taken when interpreting results of screening tools due to the high rate of false positives.

- 5) Clinicians working with individuals with DS are encouraged to look for adverse behavioural characteristics and consider using screening tools to help identify co-occurring behavioural disorders, therefore facilitating greater client-centered practice.

Conclusions and Clinical Implications

Cautionary measures should be taken when interpreting current research due to the methodological limitations discussed. Future research has the potential to yield evidence that screening instruments can be effective tools for the dual diagnosis process of individuals with DS and ASD. Appropriate diagnosis of individuals leads to greater client-centered practice, therefore tailoring intervention strategies and specific goals to that individual.

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