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

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 (DiGuiseppi 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.

DiGuiseppi 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

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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