

Dual Diagnosis: Down's syndrome and Autism Spectrum Disorder
A Review of the Literature

Written by Erin L. Evans

Consultant Intern Psychologist

Disability Services Australia: Specialist Intervention Services

Introduction

In the past there has been a widely believed misconception that individuals with intellectual disabilities do not have the cognitive capacity to experience, much less--have the same emotional variability and mental health problems as the general population. In contrast, relatively current studies report an alarmingly higher prevalence rate of psychiatric disorders in adults with intellectual disabilities compared to their "neuro-typical" peers (Pasher, 2003; Deb, Thomas, & Bright, 2001).

Of these cases Down Syndrome (DS) is the most common genetic cause of intellectual disability accounting for approximately 15-30% of cases. DS results from a 47th chromosome and affects approximately 1 in 600 to 1000 births. Like other syndromes and disorders the behavioural characteristics of DS demonstrates both group similarities and marked individual variation (Capone et al, 2005, Prasher, 1995). Such variation is further emphasised by the presence of co-morbid psychiatric disorders secondary to an intellectual disability. In a study of 193 children and adolescents with Down syndrome 73 (38%) were found to have co-morbid psychiatric disorders (Gath and Gumley, 1986). Another study at a similar time estimated that 25% of individuals with Down syndrome experienced psychiatric difficulties (Lund, 1988).

DS has been associated with higher prevalence of co-morbid psychiatric disorders including, Depression, Anxiety, Obsessive Compulsive Disorder, and Dementia of the Alzheimer's type (DAT) (Prasher, 1995). In addition to these, there has been growing discussion on the co-morbidity of DS and Autism Spectrum Disorders (ASD) (Capone et al, 2001). DS and ASD are both among the more common forms of developmental disabilities typically observed in children (Coleman, 1986).

ASD represents a 'spectrum' of psychological conditions characterised by widespread varying abnormalities across three domains including 1) social interactions and communication, 2) restricted interests, and 3) repetitive behaviour. ASD is an umbrella term for 3 main conditions depending on the individual's behavioural sequelae which includes Autism, Asperger's Syndrome, and Pervasive Developmental Disorder not otherwise specified (PDD-NOS). For the purpose of the current paper ASD will be used to discuss the core group of Autism characterised by possessing an intellectual disability of some degree.

Although the co-morbid presentation of ASD is relatively rare compared to other psychiatric co-morbidities in DS; The prognostic significance carries significant weight in valuable information for caregivers, medical professionals and educators alike in establishing appropriate early intervention programs to maximising the individuals quality of life..

The current paper will review the literature on the dual diagnosis of DS and ASD. Specifically, prevalence; barriers to diagnosis and clinical delineation/neurobehavioral phenotypes will be discussed.

, Phenotype: An observable characteristic or trait of an organism. Phenotypes result from the expression of an organism's genes as well as the influence of environmental factors and possible interactions between the two.

Prevalence

Prevalence rates of DS-ASD appear to fluctuate in research largely due to the varied diagnostic criteria utilised (Vatter, 1999). Available research estimates the prevalence of DS-ASD to range from 1 to 10%. A study by Myers and Pueshchel (1991) surveyed 497 children and adults with Down syndrome as a part of an outpatient study concluding an autism prevalence rate of 1.2% in both children and adults with Down syndrome compared to a study which reported 9% of the sample of children with DS met full criteria for autism (Turk, 1992). Howard et al (2006) reported autism is 10 times more prevalent in children with Down's syndrome compared to the general population which was conservative to Capone's (2001) estimated prevalence of 5 to 7% compared to the prevalence of autism in the general population of (approximately 0.4%). Additionally, research supports a greater prevalence in males compared to autism in the general population.

Barriers to Valid and Reliable Diagnosis

There is significant support for early intervention of ASD particularly when dual diagnosed with Down syndrome. Dual diagnosis is critically important when these two conditions co-exist as ASD, although secondary, takes precedence in terms of educational and management needs. A dual diagnosis of autism where appropriate, enables further specialised services and support; which would otherwise not have been made available the individual and the key caregivers involved. There are arrays of factors which impact negatively on widespread valid and reliable diagnoses of the diagnosis of ASD in individuals with Down syndrome; such as:

- Some professionals do not want to burden the family with an additional diagnosis combined with the general stigma of psychiatric illness.
- An overshadowing effect of the intellectual disability obscuring the psychiatric illness; combined with poor understanding of the typical DS developmental profile which is essential to identify any developmental abnormalities (Pary, 1997)
- Stereotypes regarding the personality of DS as resulting in sociable and happy and empathetic individuals despite their reduced cognitive capacity. Such ill founded assumptions are dangerous and are akin with previously discussed assumption that individuals with intellectual disability do not experience the same emotional variability as their 'neuro-typical' peers (Nordhauser, 1997)
- The nature of ASD as a spectrum disorder which requires qualitative clinical discretion to apply a diagnosis unlike stringent blood tests or genetic screening as with DS.
- An issue of early onset development observations required in the diagnosis of ASD is often missed by caregivers who are heavily focused on the primary disorder.

Clinical Delineation and Behavioural Phenotypes

Diagnostic Criteria

Vatter (1998) provided a comprehensive overview of the diagnostic criteria for Autism as per the Diagnostic and Statistical Manual, Version 4 (DSM-IV). Vatter commented on each diagnostic criteria of its relevance to the typical behavioural characteristic of DS-alone. *Please see the Appendix to review this reproduced table in full.*

In summary, the diagnostic criteria for DS-ASD are (Coleman, 1986):

- A Trisomy 21 chromosomal karyotype (or one of the other less common chromosomal variants including Mosaicism, and Translocation Down syndrome.

AND; have two or more of the following:

- Significant disturbance of social relatedness
- Repetitive routines and insisting the sameness
- Unusual, inconsistent sensory responsiveness
- Despite the individual's intellectual disability, they may possess a special ability at an excelled level to the general level of functioning.

Importance was emphasised on observing the above behavioural features over a long period of times to rule out any confounding medical conditions/differential diagnoses. Patterson (1999) reported a comparatively higher frequency of seizure disorder in DS-ASD individuals than the general population.

Behavioural Characteristics

Based on clinical experience and an extensive review of current literature, Capone (2001) further clarified two distinct patterns of ASD onset in children with Down syndrome. The following was reproduced from the original article:

Type 1: Characterised by atypical behaviours early during infancy or toddler years

The following behaviours may be observed;

- Repetitive motor behaviours such as hand flapping or oral fixation.
- Fascination with staring at lights, ceiling fans or fingers
- Extreme food refusal
- Receptive language problems (poor understanding and use of gestures) possibly giving impression that the child does not hear
- Spoken language may be highly repetitive or absent

Medical conditions may be present:

- Seizures, dysfunctional swallowing, nystagmus (constant moving of the eyes) or severe hypotonia (low muscle tone) with a delay in acquiring motor skills

Type 2: Typically older children with ASD onset occurring between 3-7years

- The following behaviours may be observed;
- Tend to experience dramatic loss or plateau in their acquisition and use of language and social attending skills
- This development regression may be followed by excessive
 - Irritability
 - Anxiety
 - Onset of repetitive behaviours
- Follows otherwise typical course of early development for children with downs syndrome

Capone discussed that although young children may demonstrate one or two of the above mentioned characteristics however this does not equate to a dual diagnosis of DS-ASD. It is recommended the individual be closely monitored and seek formal assessment. Appropriate intervention strategies based on professional and informed knowledge of the individual.

In contrast to Capone (2001) whom explored the inter diagnostic behavioural characteristics of the ASD onset, Carter et al (2006) attempted to clarify the characteristic behavioural phenotypes of DS-ASD to better understand to the neuro-anatomical underpinnings of the disorders both singularly and co-morbidly. The Aberrant Behaviour Checklist and Autism Checklist were used to measure such differences. Results suggested individuals with co-morbid DS-ASD possess a distinctive set of aberrant behaviours marked by characteristic odd/bizarre stereotypic behaviour, anxiety, and social withdrawal compared to those with DS alone.

Furthermore, research suggests compared to individuals with DS-alone, those with DS-ASD tend to possess a significantly lower cognitive capacity (Carter et al, 2007), demonstrate more profound communication delay (Nordhauser, 1997), and are generally slower to master more advance motor skills (Winders, 1999), such as walking down stairs or alternate feet jumping however observed nil regression in overall gross motor development between DS-ASD and DS-alone diagnoses. Winders discussed two distinct types of temperaments regarding how individuals tend to react when learning new motor skills:

- *'Motor Driven'* describes individuals who move quickly and like to take risks physically;
- *'Observers'* which describes individuals who prefer to be stationary, observe their environment and like to be careful and in control.

Winders concluded support that DS-ASD individuals tend to be of the 'Observer' temperament style.

Neuroanatomical characteristics of DS-ASD

In addition to observable behavioural manifestations characteristic of DS-ASD, there is increasing evidence of observable neuroanatomical differences (structural/activity levels) (Carter et al, 2008; Capone, 2001). Carter et al (2008) recently published a new study which extended on the previous research into the neuro-behavioural phenotypes of DS-ASD and DS-alone. Carter et al (2008) found evidence of hyperplasia (propagation) of white matter in the cerebellum and brainstem compared to DS-alone. In addition Carter et al found this enlargement of white matter was positively associated with severity of stereotypy's characteristic of DS-ASD. Accelerated brain growth in children with DS-ASD between the ages 2 to 5 years was also observed. These neuroanatomical changes appear consistent with Capone's account of ASD onset in children with Down syndrome.

Conclusion

There appears to be a slow but steady increase of research into the dual diagnosis intellectual disability and psychiatric illness particularly in the co-morbidity of Down syndrome and Autism Spectrum disorder. DS-ASD was not so long ago viewed as being incompatible. Capone (2001) expressed the need for research to move beyond mere descriptive accounts of the dual diagnosis. Both advocacy and informative studies such as those presented by Carter et al and Capone et al have benchmarked the research as possessing characteristic behavioural and neuroanatomical differences which distinguish it from Down syndrome- or ASD-alone. Growing support that similar neurological functions underlie both DS and ASD is evident in literature.

Future studies may further distinguish between the clinical differences of Down Syndrome Subtypes as explored by Prasher (1995) extending this research into the realm of dual diagnosis of DS subtypes and ASD. Improvements in identifying behavioural and neurological characteristics may lead to increased ability to identify the presence of abnormalities sooner and to maximise early intervention outcomes, explore environment and family risk factors and assist in the development of new treatment therapies to improve development and inform therapeutic best practice.

References

- Capone, G. (2001) Down's syndrome and autistic spectrum disorder: a look at what we know. *Down's Syndrome Association Journal*, 97, 6-13.
- Carter, J. C., Capone, G. T., and Kaufmann, W. E. (2008). Neuroanatomical correlates of autism and stereotypy in children with down syndrome. *Neuro Report*, 19, 6, pp. 653-656.
- Carter, J. C., Capone, G. T., Gray, R. M., Cox, C. S., Kaufmann, W. E. (2006). Autistic-spectrum disorders in down syndrome: further delineation and distinction from other behavioural abnormalities. *American Journal of Medical Genetic Neuropsychiatry Part B*.
- Deb, S., Thomas, M., & Bright, C. (2001). Mental disorder in adults with intellectual disability: Prevalence of functional psychiatric illness among a community-based population aged between 16 and 64 years. *Journal of Intellectual Disability Research*, 45, 495 -505.
- Gath, A., Gumley, D. (1986). Behaviour problems in retarded children with special reference to Down's syndrome. *British Journal of Psychiatry*, 149, 156-161.
- Howard, J., Molloy, C., Patterson, B., Hickey, F., Castillo, H. (2006). *Age of development regression in children with autism and with and without down syndrome*. Kentucky Junior Academy of Science Psychology Undergraduate Research Competition.
- Howlin, P., Wing, L., Gould, J. (1995). The recognition of autism in children with Down syndrome: implications for intervention and some speculations about pathology. *Dev Med and Ch Neurol*. 37, 406-414
- Lund, J. (1985). The prevalence of psychiatric morbidity in mentally retarded adults. *Acta Psychiatrica Scandinavica*, 72, 563-570.
- Myers B. A, Pueschel S. M. (1991). Psychiatric disorders in a population with down syndrome. *Journal of Nervous & Mental Disease*; 179: 609-13.
- Nordhauser, M. (1997). Down syndrome and pervasive developmental disorders. *Down Syndrome Aim High Newsletter, News and Notes*.
- Pary, R. J. (1997). "Comorbidity of Down syndrome and autism", *The Habilitative Mental Healthcare Newsletter*, 16(1).
- Patterson, B. (1999) Dual Diagnosis: The importance of Diagnosis and Treatment. *Disability Solutions*, 3, 5/6.
- Prasher V P. (1995). Prevalence of psychiatric disorders in adults with down syndrome. *European Journal of Psychiatry*; 9: 77-82.

Prasher, V. P. (2003). Psychiatric morbidity in adults with down syndrome. *Psychiatry*, 2, 8.

Pueschel SM. Towards optimal mental health of persons with down syndrome. *Down Syndrome Research and Practice*. 1998;5(1);43-46.

Turk, J. (1992). Children with Down's Syndrome and Fragile X syndrome: A comparative study. Society for study of Behavioural Phenotypes: 2nd Symposium Abstracts, Oxford: SSSP

Vatter, G. (1998). Diagnosis of autism in children with down syndrome. *Riverbend Down Syndrome Parent Support Group Journal*. 26, p 7. At: www.altonweb.com/cs.downsyndrome/autism.html

Winders, P. C. (1999) Language and communications: The key is effectiveness. *Disability Solutions*, 3, 5/6.

Appendix

The following extract was obtained from Vatter (1998) which describes the diagnostic criteria of Autism of DSM-IV. Vatter commented on whether each trait was normal or abnormal for normal DS alone presentation:

A. A total of six (or more) items from (1), (2), or (3), with at least two from (1) and one from each from (2) and (3).	
DSM-IV description	Normal or Abnormal in DS alone
1. Qualitative impairment in social interaction, as manifested by at least two of the following:	
a. Marked impairment in the use of multiple non-verbal behaviours such as eye to eye gaze, facial expression, body postures and gestures to regulate social interaction.	<i>Not normally seen in Down syndrome, more indicative of autism.</i>
b. Failure to develop peer relationships appropriate to developmental level.	<i>Not normally seen in Down syndrome, more indicative of autism.</i>
c. A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by lack of showing, bringing, or pointing out objects of interest).	<i>Somewhat true in Down syndrome but much more pronounced in autism.</i>
d. Lack of social or emotional reciprocity.	<i>Somewhat true in Down syndrome but much more pronounced in autism.</i>
2. Qualitative impairments in communication as manifested by at least one of the following:	
a. Delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime).	<i>Not normally seen in Down syndrome.</i>
b. In individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others.	<i>Normally seen in Down syndrome, although not to the extent seen in autism.</i>
c. Stereotyped and repetitive use of language or idiosyncratic language.	<i>Seen somewhat in Down syndrome but not to the extent seen in autism.</i>
d. Lack of varied, spontaneous make-believe play or social initiative play appropriate to developmental level.	<i>Not normally seen in Down syndrome.</i>
3. Restricted, repetitive, and stereotyped patterns of behaviour, interests, and activities, as manifested by at least one of the following:	
a. Encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal in either in intensity or focus.	<i>Not normally seen in Down syndrome.</i>
b. Apparently inflexible adherence to specific, non functional routines or rituals	<i>Not normally seen in Down syndrome.</i>
c. Stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting or complex whole body movements).	<i>Not normally seen in Down syndrome.</i>

d. Persistent preoccupation with parts of objects.	<i>Not normally seen in Down syndrome.</i>
--	--

B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) Social interaction, (2) Language as used in social communication, or (3) Symbolic or imaginative play.

Children with Down syndrome will normally display some deficits in these traits depending on the severity of mental retardation. Since this is not entirely unexpected, it is usually ignored in the records and not so noted. Therefore when autism is diagnosed at a later age and the records or memory is searched for this evidence it is either not found in the official records of the child or it is forgotten about because it was not deemed all that unusual at the time. The diagnostician may then discard the possible diagnosis of autism because the appearance is given (falsely) that these traits were not present prior to 36 months of age. Because of this quirk in the diagnosis, there probably are many persons with Down syndrome with autism but it is not so diagnosed.

C. The disturbance is not better counted for by Rhetts Disorder or Childhood Disintegrative Disorder.