A CASE OF UNRECOGNIZED INTELLECTUAL DISABILITY AND AUTISM MISDIAGNOSED AS SCHIZOPHRENIA: ARE THERE LESSONS TO BE LEARNED?

ANNA M. PALUCKA, PH.D.,1,2 ELSPETH BRADLEY, M.D.2,3 & YONA LUNSKY, PH.D.1,2

1Department of Psychiatry, University of Toronto, ON, Canada
2Department of Psychiatry, University of Toronto, ON, Canada
3Surrey Place Centre, Toronto, ON, Canada

The present case illustrates the importance of ruling out developmental disorders before making a diagnosis of schizophrenia. The case demonstrates how an initial mind set can perpetuate the “reasonableness” of an inaccurate diagnosis (of schizophrenia), and points to the need for constant review of the psychiatric diagnosis as new information becomes available and as clinical concepts change with the infusion of new understanding.

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Schizophrenia is a serious mental illness characterized by “positive” symptoms such as delusions, hallucinations, and “negative” symptoms such as flattened affect and social withdrawal. DSM-IV-TR diagnostic criteria specify that schizophrenia should not be diagnosed if there is a history of any pervasive developmental disorder such as autistic disorder unless prominent delusions or hallucinations (positive symptoms) are also present (Criterion F). Autistic disorder (DSM-IV-TR)k also known as childhood autism (ICD-10 RDC),32 is the most severe of a spectrum of related developmental conditions (which include Asperger’s syndrome, atypical autism and pervasive developmental disorders not otherwise specified) and is defined behaviorally by impairments in socialization and communication, and by a lack of behavioral flexibility. Autism occurs with a frequency of around 1/1,0005 while the prevalence of the broader autism spectrum is estimated at 6/1,00028; autism has recently been estimated as present in 28% of the population with intellectual disabilities.7

As several symptoms of autism overlap with the symptoms of schizophrenia, failure to consider autism and the broader autism spectrum may result in the inappropriate assignment of a schizophrenia diagnosis, particularly in individuals with intellectual disabilities. Similarities between schizophrenia and autism have been explored: O’Dwyer25 concluded that there is a degree of overlap in the symptoms of these disorders in the areas of social dysfunction, qualitative impairment in social interaction and communication, as well as repetitive stereotyped behaviors. These symptoms represent the core symptoms of autistic disorder and the negative symptoms of schizophrenia. However, delusions and hallucinations, core symptoms of schizophrenia, are absent in autistic disorder. In reported cases of autism and coexisting schizophrenia, these core symptoms of schizophrenia are clearly present.10,23,25

Kontantareas and Hewitt16 found that one half of the individuals with autism in their study group met criteria for schizophrenia, disorganized type. While these individuals tended to show primarily negative symptoms, 5 out of 14 (36%) displayed positive features (as measured on the SAPS),2 mainly bizarre behavior, with one manifesting positive formal thought disorder. However, as Wing30 points out, it is particularly important in autism to evaluate any such “positive” symptoms against baseline of premorbid behaviors and thought patterns as persons with autism often have idiosyncratic perceptions and meaning attached to their experiences which may appear bizarre to others unless this historical and current meaning is understood. In the same vein, Howlin13 describes a 19-year-old student who developed symptoms of severe anxiety, including headaches and stomachaches shortly after starting college. Insisting that a “shark in his insides” caused his stomach pains, he was admitted to hospital and treated with medication for schizophrenia. His mother later explained that as a child he had a favorite book that explained illnesses in this way: a crocodile in the head giving people headaches, a monkey with a hammer
producing pains in the ears etc.; her explanation exposed the misdiagnosis of schizophrenia.

Current confusion between schizophrenia and autism diagnoses may also have arisen historically. Prior to the 1970’s the concept of “childhood schizophrenia” included autism and early onset schizophrenia. Work by Kolvin and colleagues14,15 provided clear diagnostic guidelines that distinguished between these two conditions, the most notable being that core features of autism can be identified before the age of three years while early onset schizophrenia in their sample did not occur before the age of 6-7 years. In 80% of their sample the onset of schizophrenia was after age 8 years. By age six, in normally developing children, cognitive and linguistic capacity are sufficient to provide an account of subjective experience (necessary in the evaluation of positive symptomatology). Those rare children who do develop childhood schizophrenia are more similar in their symptomatology to adult schizophrenia and are easily distinguished from children with autism.10 The high prevalence of co-existing intellectual disabilities with autism adds further complexity to these diagnostic issues. In terms of the diagnosis of schizophrenia in the population with intellectual disabilities, some studies have found a greater occurrence of positive symptoms3 (e.g., hallucinations and delusions), while others have found negative symptoms8 (e.g., affective flattening and avolition) to be more common. Partly in response to these disparate views, Grafmont et al.12 recommended extensive assessment, careful observation and a longitudinal approach to the diagnosis of psychosis and schizophrenia in persons with mild and borderline intellectual disabilities. Bouras et al.4 suggested that specialist knowledge of mental disorders in people with intellectual disabilities may be required to enable accurate diagnosis. Given the change in the diagnostic classification of “childhood schizophrenia” noted above, older adults, in the current population of persons with intellectual disabilities with a diagnosis of childhood schizophrenia, need to be carefully re-evaluated to determine whether their original symptoms were those of autism or early onset schizophrenia.

Case Report

Summary of initial psychiatric assessment (as reported in the hospital chart):

Mr. A, an 18-year-old grade twelve student in a special education class, was referred to an outpatient psychiatrist by his school due to concerns regarding his interpersonal behavior: his refusal to listen, tendency to ignore others, his avoidant, repetitive and immature behavior and difficulty maintaining a train of thought.

Developmental history provided by his mother indicated a slight delay in learning to talk. After coming to Canada at age 5, it took him six years and remedial language classes to gain working knowledge of English. Mother reported that he underwent psychological, medical and neurological assessments at age 8 and was given a diagnosis of minimal brain dysfunction but was described as “otherwise normal.” She also stated that he had not changed since that assessment. She reported that he was very shy, introverted, did not socialize outside of his family and even at home, did not play with other children or participate in sport activities at school or extracurricular events, and behaved younger than his age. He had fears that the Devil would punish him because he was bad as a child, and that he might die of a heart attack at an early age or be struck by lightning when he walked outside in the rain.

On mental status examination, Mr. A spoke with a low tone voice; his speech was rambling and pressured and often unintelligible as he garbled most words. He exhibited echolalia, echopraxia, inappropriate grimacing and poor fine motor coordination. Circumstantial thinking and intermittent thought blocking were noted. He denied visual or auditory hallucinations but stated that from time to time while alone he heard voices of the Devil, experienced both outside, and echoing inside his head, reminding him how bad he had been. Based on his vocabulary, his intelligence was assessed to be average.

The diagnostic impression at the time was to rule out 1) schizophrenic disorder, disorganized type; 2) schizophrenic disorder, simple type; and 3) personality disorder with inadequate, immature, dependant and narcissistic traits.

Lesson 1: The initial diagnostic formulation at that time failed to rule out developmental disorders, particularly intellectual disabilities, despite cues suggesting longstanding learning difficulties (slow acquisition of English, report of prior assessments and diagnosis of minimal brain dysfunction, placement in special education). In addition, this assessment failed to get an extensive early developmental history.
Further assessments (also as reported in the hospital chart):

In the course of the next seven months, Mr. A underwent a number of psychological and psychiatric assessments and consultations including an inpatient admission all leading to the solidification of his diagnosis of schizophrenia.

Psychological consultation was requested to aid in clarifying the diagnosis and was completed a few weeks after the initial assessment. It indicated “clear cognitive difficulties” hypothesized to be long standing but likely in the low-average range prior to psychosis. Personality assessment revealed a “regressed individual, very frightened, isolated and fearful of sickness and death without coping resources whose difficulty reached psychotic proportions” and antipsychotic medication was recommended.

Two months after the initial psychiatric assessment, Mr. A was brought to hospital by his mother, presenting with extreme anxiety associated with fears of dying of a stroke or heart attack and the belief that his heart had a defective valve. He admitted to hearing voices but was unable to provide any details. No delusional content was noted. The diagnostic impression was low average intelligence and acute schizophrenic illness; he was hospitalized and treated with antipsychotic medication. During this admission, the neuropsychologist reviewed prior childhood assessments unavailable at the time of the earlier assessments, and noted that Mr. A had previously been diagnosed with “educable retardation” and had been described as displaying “autistic-like” behaviors (rocking and hands flapping) as a child. His current intellectual functioning was assessed by the neuropsychologist to be in the range of intellectual disabilities. There was no evidence of bizarre speech or peculiar verbalizations but he lacked spontaneity in interaction during testing. It was concluded that, despite psychotropic medication, psychosis along with diffuse cognitive deficits continued to contribute to his severe impairments.

At discharge, Mr. A was diagnosed with intellectual disability, schizophrenia, and mitral valve prolapse (initially diagnosed four years earlier) and was referred for services (day program) for people with intellectual disabilities.

Lesson 2: Although the presence of longstanding intellectual delay had by this time been recognized, the cues suggesting the possibility of an autistic disorder (autistic-like behaviors, longstanding nature of social difficulties) were missed. Furthermore, clinicians failed to revise the initial diagnosis of schizophrenia made in the context of average IQ despite the finding of intellectual disability and absence of delusional thinking.

Fifteen years later... (and involvement of present authors):

Mr. A’s next significant contact with psychiatry was at age 33, when due to an incident of serious aggression at home, a consultation from the specialized dual diagnosis (dual diagnosis is defined as intellectual disabilities and psychiatric or behavioral disorder) program in which the present authors worked, was sought. At the time of referral, he had continued to live at home and had been attending a day program with a developmental sector agency for fifteen years.

In our formulation, we re-emphasized that his mild to moderate intellectual disability had its origin at an early age rather than attributing it to functional deterioration due to schizophrenia.

Early childhood history was reviewed with mother. Mother reported no prenatal or postnatal complications. Medical consultation was sought when he was 1-1/2 years old because he “took a long time to do things” but no opinion was offered. Her recall was that there were no delays in reaching developmental milestones: he walked by 11 months, spoke single words by first year and spoke well by 2-3 years, and was toilet trained by 3-1/2 years of age.

Review of records indicated that after coming to Canada at age 5, he entered a senior kindergarten program from which he was shortly suspended and referred for an assessment because of “shy, withdrawn behavior in class, little socialization skills, destroying things he made and bizarre behavior such as hand flapping.” He was diagnosed as “mentally retarded with atypical features.” He attended primary special education classes and benefitted from individualized programs. At 12, he was recommended for placement in level II vocational school but this was rejected because of his persistent fear of traveling alone and need for continuing transportation by a special school bus. At 14, when re-assessed for a high school placement, he was found to exhibit the same social and emotional problem behaviors as in the past. On cognitive measures, his receptive language was in the 4-1/2 year old range, his reading and spelling skills were at grade 6 and arithmetic skills were at
grade 3 level. His performance on Visual Motor Integration Test corresponded to age equivalent of 7-4 years.

Further inquiry suggested difficulties in all areas associated with autistic disorder present from a very early age: pervasive lack of interest in peer relationships, lack of eye contact, inappropriate emotional responses, no interactive or pretend play, and hours spent performing repetitive activities. He whirled and turned in circles, rocked (even in bed), banged his head against the wall or his knuckles against each other, made high-pitched sounds and sniffed objects and people. When older, he developed an intense preoccupation with bicycles and buses (having 10 at home and calling the bicycle store daily). Based on this history he was diagnosed for the first time as having autistic disorder. Previous clinical symptomatology was reviewed in the context of both his autistic disorder and mild to moderate intellectual disabilities. There was no evidence for a diagnosis of schizophrenia. His “delusional” talk was construed as very concrete interpretations/processing of real life events with significant emotive meaning to him. For example, his considerable anxiety and preoccupation with his heart’s “defective valve” represented his incomplete understanding of the implications of his mitral valve prolapse diagnosis, specifically the fact that there were no significant health risks associated with his condition. This understanding might have been further compromised by the fact that one of his relatives died of a heart condition. Similarly, his concerns about the Devil (as reported during early psychiatric assessments) were clearly related to his exposure to and understanding of religious concepts and his experience of himself as “being bad” and “going to hell” as a result. Consequently, the diagnosis of schizophrenia was removed.

**Discussion**

History taking is critically important in psychiatric assessment because “diagnosis relies on knowing the course and context within which the symptoms developed.”19 (p.31)

In the case of Mr. A, the developmental perspective was missed and the longstanding nature of the difficulties and behaviors were not given consideration in the initial diagnostic formulation. In addition, the impact of his level of intellectual functioning on his presentation was not taken into account. Mental status examination in the generic mental health system usually assumes that the individual possesses normal intellectual ability and is able to understand the contextual or implied nature of the questions. However, individuals with a cognitive disability may not understand or appreciate the contextual aspects of even simple questions and their responses may be misinterpreted as indicative of serious psychopathology.18 Furthermore, “bizarre” thoughts or odd obsessions which would suggest a presence of delusional thought disorder in a normally functioning person are fairly typical manifestations in autism.30

Mr. A’s initial diagnosis of schizophrenia, although provisional, appears to have biased subsequent clinicians’ diagnostic formulations even after the intellectual level had been elucidated through extensive neuropsychological testing and review of childhood records. No change in his behavior and personality over the years and the absence of deterioration in his social/adaptive functioning should have raised suspicion as to the accuracy of a schizophrenia diagnosis.21

Given the overlap between some of the symptoms of autism spectrum disorders and schizophrenia23 and the diagnostic issues that may arise,5 as well as much higher prevalence estimates for the broad category of autism spectrum disorders than was once thought,6,11,28 some clients with autism are likely to be misdiagnosed as having schizophrenia.24,26 There is also evidence that autism is underdiagnosed both in clinical and population based samples which may contribute to diagnostic errors: one review of outpatient clients receiving psychiatric services revealed that 3% (N=17) had previously undiagnosed autism spectrum disorder and for 5 of these (29%) the registered psychiatric diagnosis was schizophrenia;22 a Canadian study of prevalence of autism in teenagers with intellectual disabilities revealed that less than half of those who met diagnostic criteria for autism had previously been diagnosed as such.7 Prevalence figures for the co-occurrence of autism spectrum disorder and schizophrenia are reported to be low —0.6%.29 However, prevalence rates for intellectual disability and the co-occurrence of schizophrenia vary between 1.3 and 3.7%.8 At this time it is unclear to what extent this latter variability in prevalence rates may reflect misdiagnosis of schizophrenia in persons with intellectual disabilities and in particular those who have both autism spectrum disorder and intellectual disabilities.
The present case highlights the need to assess specifically for intellectual disabilities and autism spectrum disorders in adult clients before a diagnosis of schizophrenia is confirmed. Such assessments should include systematic enquiry about early development and behavior, cognitive and functional capacity prior to the onset of the symptoms of concern (with review of psychological and school reports as a necessary part of the clinical assessment). Screens for autism in previously undiagnosed adults are now available, and standardized systematic approaches for the assessment of autism in children and young adults and the wider spectrum of social and language impairments across all levels of functioning and ages are available. These latter approaches not only elicit important client information about co-existing autism (and the wider spectrum of autism symptoms and behaviors) but also provide excellent clinical learning experiences about autism for clinicians and trainees new to the area. Without specific enquiry in areas associated with the core features of autism, a generic developmental history may not elicit all the relevant behaviors and the diagnosis will be missed. If necessary, consultation from clinicians experienced in intellectual disabilities and psychiatric or mental health issues should be sought before making a co-morbid diagnosis of schizophrenia in anyone who appears low functioning.

References


Correspondence: Anna M. Palucka, Ph.D., Dual Diagnosis Program, Centre for Addiction and Mental Health, 1001 Queen St. West, Toronto, ON Canada M6J 1H4; tel.: 416-535-8501 ex. 3174; fax: 416-583-1268; email: anna_palucka@camh.net.