A phenomenological approach to diagnosing psychosis in autism spectrum disorder and intellectual disability: a case series

Rahul Rai, Samuel Tromans, Chaya Kapugama, Verity Chester, Ignatius Gunaratna, Peter Langdon and Regi T. Alexander

Abstract

Purpose – The diagnosis of psychosis in individuals with autism spectrum disorder (ASD) poses a unique clinical challenge. The presence of intellectual disability (ID) further complicates the diagnostic picture. Reliable and timely diagnosis of psychosis in such individuals minimises the duration of untreated psychotic symptoms and the subsequent impact on the quality of life of the patients concerned. The paper aims to discuss this issue.

Design/methodology/approach – The authors present four patients with psychosis, ASD and ID, who have received care within forensic mental health and ID settings. These examples demonstrate the interaction between these conditions, as well as issues pertaining to diagnosis and management.

Findings – In all four patients, sustained use of antipsychotic medication was objectively associated with an improvement in psychotic symptoms and quality of life. In instances where autistic phenomena were accentuated upon development of psychosis, such features returned to the baseline levels evident prior to the onset of psychosis.

Practical implications – The discussion and related case examples could improve the understanding of the possibility of psychosis in individuals with ASD and ID, and increase awareness of this diagnostic possibility among healthcare professionals.

Originality/value – This is the first published case series illustrating the challenges of diagnosing psychosis in individuals with ASD and ID.

Keywords Mental health, Learning disability, Mental disorder, Schizophrenia, Autism spectrum condition, Comorbidity

Paper type Case study

Introduction

Autism spectrum disorders (ASD) and psychotic disorders have historically been considered as related diagnostic entities (Sugranyes et al., 2011). However, the nature of this relationship has been the subject of extensive debate (Padgett et al., 2010), alternating between the view that ASD is an early manifestation of childhood schizophrenia on the one hand (Kanner, 1949), and that people with ASD could not be diagnosed with schizophrenia on the other. It is now widely believed that ASD and psychosis are two distinct clinical entities. Kolvin et al. (1971) highlighted that children whose abnormal behaviours were apparent before the age of three years fitted with Kanner’s description of “early infantile autism” (Kanner, 1968). In contrast, those children whose development was essentially normal until school years, but then later developed hallucinations, delusions or other behavioural abnormalities, were felt to be more in keeping with a diagnosis of schizophrenia.
A number of studies have examined whether schizophrenia is significantly more prevalent amongst people with ASD, in comparison to the general population (Billstedt et al., 2005; Ghaziuddin et al., 1998; Volkmar and Cohen, 1991). A recent systematic review examined the rates of psychosis in individuals with ASD, reporting a prevalence which ranged from 0 to 53 per cent (Padgett et al., 2010). However, the authors noted marked differences and heterogeneity in terms of the methodological approaches of the included studies, which precluded a meaningful pooling of the findings. For example, the prevalence of psychosis in adults with ASD differed greatly between studies. Joshi et al. (2013) found a lifetime prevalence of 13 per cent and point prevalence of 8 per cent of psychosis in adults with ASD, whereas a study by Volkmar and Cohen (1991) found only 0.6 per cent from a sample of 163 patients with ASD. Stahlberg et al. (2004) reported that 7.8 per cent of participants with diagnosed ASD had comorbid schizophrenia, which is higher than the 1 per cent prevalence of schizophrenia in the general population (McGrath et al., 2004). A preliminary study reported a prevalence of 2.4-5.3 per cent across three secure hospitals, depending on whether equivocal cases were considered within the estimate (Hare et al., 1999). A meta-analysis highlighted that childhood-onset schizophrenia is preceded by and comorbid with ASD in 30-50 per cent of cases (Rapoport et al., 2009). Likewise, it has been noted that in a population of people with childhood-onset schizophrenia, 25 per cent met the criteria for childhood ASD (Sporn et al., 2004).

Furthermore, Larson et al. (2017) conducted a study whereby 116 individuals with ASD and psychosis were compared with a group with psychosis only. They found that a diagnosis of atypical psychosis was more likely in individuals with ASD relative to those with psychosis only, who were more likely to receive a diagnosis of schizophrenia. This suggests that there may be a specific manifestation of ASD linked to comorbid psychosis, yielding an atypical clinical picture, especially with regard to affective disturbance.

Phenomenological overlap is a likely contributory factor to the wide variability in the reported rates of schizophrenia in people with ASD (Skokauskas and Gallagher, 2010). The neurodevelopmental hypothesis of schizophrenia suggests that its origins in adolescence are partially explained by the consequences of events in early development (Owen et al., 2011). Evidence suggesting a genetic overlap between neurodevelopmental disorders such as ASD, attention deficit-hyperactivity disorder (ADHD), bipolar affective disorder and major depressive disorder are emerging (Cross-DisorderGroup of the Psychiatric Genomics, 2013). Such findings suggest viewing these functional psychoses as a group of related and overlapping syndromes, with origins partially based in the early developmental period.

**Diagnostic issues**

The diagnosis of a psychotic disorder in an individual with ASD poses a clinical challenge. In fact, numerous cases of misdiagnosis of psychosis and schizophrenia have been reported, especially in instances where the ASD was not previously diagnosed (Tantam, 2003). Through their ASD presentation alone, some people with ASD may meet the criteria for schizophrenia on clinical interview schedules, such as the Structured Clinical Interview for the Diagnostic and Statistical Manual of Mental Disorders (Konstantareas and Hewitt, 2001). More specifically, the negative symptoms of schizophrenia have an overlap with the symptoms of ASD (Frith and Frith, 1991). Carpenter (2007) reported that it is relatively common for more able individuals with ASD to be erroneously diagnosed with schizophrenia, and cited examples of how some common features of ASD may be misinterpreted, as summarised in Table I.

Conversely, there is also a risk that the diagnosis of schizophrenia can be missed among those diagnosed with ASD. It is therefore essential to carefully elicit the content and form of the person’s thoughts, which will help identify a change from the usual preoccupations/meanings attached to these thoughts. A thorough exploration of phenomenology can minimise the risk of psychosis being missed in people with ASD.

The presence of intellectual disability (ID) adds a further layer of complexity to the aforementioned difficulties of misdiagnosing ASD as schizophrenia, or vice versa, as well as establishing cases where there is a dual diagnosis of ASD and schizophrenia. This is further discussed in Box 1 (Fletcher et al., 2007). Such cases are rarely described within the literature, and thus there is a limited understanding
of the prevalence of coexisting ASD and schizophrenia within the ID population. Esan et al. (2015) assessed a sample of 138 patients treated over 6 years within an inpatient forensic ID service, finding that 6 of these patients had both ASD and schizophrenia. However, it is difficult to make assumptions of the representativeness of this single study (Esan et al., 2015).

It has been suggested that the reliability of diagnosing psychosis in people with ASD who also have ID is poorer than in those in the general population (Dossetor, 2007). This is unsurprising given that there is no consensus on the best way to assess psychopathology in adults with ASD and ID (Underwood et al., 2010). There is no guidance for the clinician on the diagnosis of psychosis in ASD in the 10th revision of the International Statistical Classification of Diseases and Related Health Problems (ICD-10) (World Health Organisation, 1992). The 5th edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-V) specifies that in individuals with a history of ASD, the diagnosis of schizophrenia can only be made if there is a presence of hallucinations and delusions for at least one-month duration (American Psychiatric Association, 2013). However, this seeming lack of guidance and literature on psychosis and ASD should not prevent the consideration of the diagnosis in patients where such clinical suspicion exists. The failure to correctly identify psychosis in individuals with ASD would mean a lack of provision of antipsychotic treatments, prolonging psychotic experiences and further compromising quality of life. Conversely, the failure to identify ASD in individuals with comorbid psychosis would lead to individuals receiving inadequate treatment for their actual clinical needs (Larson et al., 2017).

In this case series, we present four patients with mild ID who illustrate the clinical challenges of differentiating between diagnoses of ASD and psychosis, or may indeed have both conditions, with the overall aim of improving recognition, understanding of the interface between these conditions, as well as issues pertaining to diagnosis and management.
Method

Design

In this case series, four patients with psychosis, ASD and ID, who have received care within forensic mental health and ID settings, are presented.

Procedure

Clinicians working within two forensic ID services (one national health service and one independent sector) in the East of England were invited to submit cases to the project, and provided with the procedure to adhere to. The clinicians were asked to provide key information which included the apparent age of onset of psychosis, relevant family history, clinical diagnoses according to diagnostic manuals, any diagnostic tools to assist in their decision making, evidence of impairment due to symptoms and the patient’s response to described treatment. All participants had diagnoses of ID and ASD according to established diagnostic criteria, made by consultant psychiatrists during their care.

Ethical considerations

Case studies are presented according to principles established by the British Medical Journal Ethics Committee (British Medical Journal, 2018). These include obtaining informed consent and removal of patient identifying details. Informed consent was obtained from all patients included as case studies within the paper.

Case Series

Four case studies are presented (cases A-D). Cases A and B are described in Tables II and III, and cases C and D are described in Tables IV and V.

Table II Case study A

<table>
<thead>
<tr>
<th>Age of onset of psychosis</th>
<th>20 years</th>
</tr>
</thead>
</table>

Family history  
A has a family history of schizophrenia in his brother, anxiety and depression in his mother, and harmful use of alcohol in his father

Diagnoses  
Mild intellectual disability (F70.1)  
Autism spectrum disorder (F84.0)  
Schizoaffective disorder (F25.1)

Diagnostic tools used  
Harmful use of alcohol, cannabis and solvents (F10.1, 12.1 and 18.1) though currently abstinent in a forensic setting  
WAIS-III (Wechsler, 1997): full scale intelligence quotient = 61  
ICD-10 (consensus of diagnosis established between two psychiatrists) (World Health Organization, 1992)

Brief description  
A is a 33-year-old male with mild ID. He experienced sexual abuse during his childhood. Though he attended mainstream school, he struggled with lessons and left school aged 14 years, with no formal qualifications. He was described as a loner, having only one close friend. After leaving school, he began work at an animal shelter. He struggled to understand instructions, often interpreting them in a concrete manner. In his teens, A felt that he was homosexual, and also developed a sexual interest in animals. He fantasised about the latter, and occasionally acted on such fantasies, though after such encounters, he would ruminate and become distressed. At around 20 years of age, A started hearing voices commanding him to carry out bizarre sexual acts with animals. He firmly believed that if he did not comply with these commands, either he or his mother would be killed by the voices. A also experienced unpleasant bodily sensations, and was convinced that the individuals responsible for the voices were imposing these experiences on his body. Along with these symptoms, he had episodes of mood disturbances alternating between low mood and elation/irritability, lasting for few weeks at a time.

Evidence of impairment due to symptoms  
Subjective distress  
Risks of self-harm (alcohol abuse)  
Risks of sexually inappropriate behaviours (sexual acts against animals)  
Vulnerability (deterioration in level of functioning related to onset of symptoms)

Response to treatment  
While on antipsychotic medication, the intensity of A’s hallucinatory experiences reduced considerably, alongside a subsequent reduction in distress, and sexually inappropriate behaviour towards animals
Discussion

This paper has described the phenomenological approach to diagnosing psychosis in individuals with both ID and ASD. The paper has a number of drawbacks, including all of the associated difficulties with case studies, such as limited generalisability. Furthermore, all of the relevant cases identified during this study were male, and as such, future research on women with this combination of diagnostic comorbidity is required. Nevertheless, the paper has a number of implications. While there is a growing clinical ability in professionals to recognise and diagnose ASD, and an increasing recognition that mental disorder is more prevalent in people with ID, the differentiation between, and diagnosis of psychosis in ASD remains a clinical conundrum, with further complexity if ID is also present.

Clinical training should place appropriate emphasis on the ability to understand, describe and document the interface between ASD and psychosis. More research, including qualitative research, is needed to support the development and evaluation of reliable diagnostic criteria, and subsequent tools to support clinical practice. There are a number of issues currently affecting practice in this area. There appears to be a reluctance to diagnose psychosis (when clinically appropriate) which poses difficulty in early recognition and treatment (Larson et al., 2017). A delayed diagnosis, or a diagnosis not being made, is likely to result in a poor treatment response, as well as a need for higher doses of antipsychotics. Symptoms not appropriately treated may result in risky behaviour, subjective distress, mistrust in professionals and a poorer quality of life. In some cases, delayed diagnosis may increase the likelihood of being admitted to inpatient or secure placements.

There is currently a lack of guidance from diagnostic criteria, and, subsequently, tools to assist with the diagnosis of ASD and psychosis. As such, the clinical approach must be based on a phenomenological assessment, including differentiating objective reality, the “normal alternate reality” of autism and the “loss of reality contact” observed in psychosis, from one another.
The value of establishing a good picture of premorbid and baseline level of functioning, obtaining information from multiple reliable and consistent sources, conducting detailed observations in various settings and contexts, and seeking another opinion where possible to enhance reliability cannot be overemphasised. Until better tools and criteria are developed, a judicious clinical approach may separate reliable early diagnosis and treatment with therapeutic benefit, from progressive deterioration and therapeutic nihilism.

All the cases described in our series had a mild level of ID, and were capable of adequate communication and engaged well, allowing independent assessment of their mental states by at least two psychiatrists. The qualitative change in behaviour, new-onset symptoms including hallucinations and delusions, as well as the alteration or modulation of pre-existing autistic phenomena coupled with a decline in general functioning all supported the diagnosis of a psychotic illness. While a treatment response cannot be considered a diagnostic test, the treatment with antipsychotic medication alleviated the new-onset symptoms with minimal effects on pre-existing autistic symptoms.

The patient described in the first case had a pre-existing sexual interest in animals. With the onset of psychosis, the rationale behind committing sexual acts to animals was to prevent harm to himself and his mother by a persecutor. The accompanying distress was reduced after commencement of antipsychotic medications. In the second case, the onset of psychosis correlated with a clear accentuation of autistic phenomena, as well as the development of clear-cut delusions and affective symptoms. With treatment, the autistic symptoms returned back to baseline levels.

### Table IV  Case study C

<table>
<thead>
<tr>
<th>Age of onset of psychosis</th>
<th>24 years</th>
</tr>
</thead>
<tbody>
<tr>
<td>Family history</td>
<td>C has a family history of schizophrenia in a second degree relative</td>
</tr>
<tr>
<td>Diagnoses</td>
<td>ICD-10 (World Health Organization, 1992)</td>
</tr>
<tr>
<td></td>
<td>Mild intellectual disability (F70.1)</td>
</tr>
<tr>
<td></td>
<td>Autism spectrum disorder (F84.0)</td>
</tr>
<tr>
<td></td>
<td>Schizophrenia (F20)</td>
</tr>
<tr>
<td>Diagnostic tools used</td>
<td>Psychometric assessment when aged 18 years, as well as ICD-10 (World Health Organization, 1992) criteria used by experienced psychiatrists</td>
</tr>
<tr>
<td>Brief description</td>
<td>C is a 32-year-old male with mild ID. With regard to his early development, his speech was delayed, and he received speech and language therapy input. During his childhood, he was also noticed to have flapping hand movements as well as jerking movements of his trunk. Additionally, he had ritualistic behaviours and difficulty in adjusting to change.</td>
</tr>
<tr>
<td></td>
<td>C attended special educational needs schooling followed by a brief period in college. He was employed once, at a shop briefly, but this was terminated due to his difficulties in coping with changes within this setting</td>
</tr>
<tr>
<td></td>
<td>Aged 22 years, C was detained under the Mental Health Act after he presented with weight loss, self-neglect and disengagement from his carers, refusing them entry into his flat. Two years later, he was hospitalised for the second time with gradual deterioration and self-neglect over several months. During this time, he held a firm belief that he was both infected with HIV and had leukaemia. He also believed that he was an internationally renowned surfer and a football player for Manchester United, married to a famous actress. He was convinced that his leg was sewn back after a shark attack and that his wife had been killed during this same attack. He was non-compliant with his medication throughout this period</td>
</tr>
<tr>
<td></td>
<td>C was later transferred to a rehabilitation unit, where he remained for around a year before being discharged into supported living in the community. He stayed in this placement for one year. He was then placed in a different flat in order to support him living more independently. C was ambivalent about this change and after a few months, he developed suicidal thoughts and was non-compliant with his medication regime. He seemed to settle with repeated reassurances, and continued to routinely visit his parents every week for a meal. During one of these visits, C stabbed his mother in the head, with the intention of murdering her. In subsequent interviews, he said that the thoughts of killing his mother started around the time of moving accommodation. These thoughts intensified when his belongings were moved to the new flat. C said “it seemed so final”, “I just couldn’t take it any more” and “I just had to get it done and she (his mother) was the right person”. He also stated “they are messing with my head”</td>
</tr>
<tr>
<td>Evidence of impairment due to symptoms</td>
<td>Subjective distress</td>
</tr>
<tr>
<td></td>
<td>Risks of harm to family (related to delusions)</td>
</tr>
<tr>
<td></td>
<td>Vulnerability (deterioration in level of functioning related to onset of symptoms)</td>
</tr>
<tr>
<td>Response to treatment</td>
<td>Following the assault on his mother, C was recommenced on antipsychotic medication. At this point, his mental state improved, and he did not display any further aggression or self-neglect</td>
</tr>
</tbody>
</table>

The value of establishing a good picture of premorbid and baseline level of functioning, obtaining information from multiple reliable and consistent sources, conducting detailed observations in various settings and contexts, and seeking another opinion where possible to enhance reliability cannot be overemphasised. Until better tools and criteria are developed, a judicious clinical approach may separate reliable early diagnosis and treatment with therapeutic benefit, from progressive deterioration and therapeutic nihilism.

All the cases described in our series had a mild level of ID, and were capable of adequate communication and engaged well, allowing independent assessment of their mental states by at least two psychiatrists. The qualitative change in behaviour, new-onset symptoms including hallucinations and delusions, as well as the alteration or modulation of pre-existing autistic phenomena coupled with a decline in general functioning all supported the diagnosis of a psychotic illness. While a treatment response cannot be considered a diagnostic test, the treatment with antipsychotic medication alleviated the new-onset symptoms with minimal effects on pre-existing autistic symptoms.

The patient described in the first case had a pre-existing sexual interest in animals. With the onset of psychosis, the rationale behind committing sexual acts to animals was to prevent harm to himself and his mother by a persecutor. The accompanying distress was reduced after commencement of antipsychotic medications. In the second case, the onset of psychosis correlated with a clear accentuation of autistic phenomena, as well as the development of clear-cut delusions and affective symptoms. With treatment, the autistic symptoms returned back to baseline levels.
Similarly, in the third case, there was a clear onset of new symptoms during episodes of psychosis. Circumstances leading to the committing of a criminal offence may have been attributable to a combination of autistic and psychotic phenomenology. The patient appeared to have developed persecutory ideations towards his mother in the context of being destabilised by changing placement. In the last case of the series, the onset of psychosis was mistakenly attributed to ASD for a period of time. The clear delusional content and perceptual disturbances were eventually identified whilst the patient was in hospital following detailed psychiatric assessments and longitudinal observations.

In all of these cases, the diagnostic difficulty lay in determining if the patients’ preoccupations, thoughts and behaviours were qualitatively different from the features of ASD. For example, while the “content” of the psychopathology was similar to that of ASD, the “form” was different. Features suggestive of a comorbid psychosis include the degree of distress, an increase in the intensity and frequency of preoccupations, the appearance of morbid themes, new behaviours and a change in functioning. A careful exploration of phenomenology to differentiate between the content and form of thoughts is required.

Regarding management, it is important for clinicians to develop a clinically sensible, sensitive and balanced approach to ensure people with ASD with possible development of a comorbid psychotic disorder access the principles of early recognition, intervention and treatment. Psychosis diagnosed in a person with ASD should be treated in the same way as psychosis affecting any other individual. This includes prescribing adequate doses of antipsychotic medication, after ensuring clarity around the target symptoms that need to be addressed.

Table V  Case study D

<table>
<thead>
<tr>
<th>Age of onset of psychosis</th>
<th>15 years</th>
</tr>
</thead>
<tbody>
<tr>
<td>Family history</td>
<td>D had no known family history of mental illness or intellectual disability</td>
</tr>
</tbody>
</table>
| Diagnoses                 | ICD-10 (World Health Organization, 1992)  
Mild intellectual disability (F70.1)  
Autism Spectrum Disorder (F84.0)  
Hyperkinetic Conduct Disorder (F90.1)  
Paranoid schizophrenia (F20.0) |
| Diagnostic tools used     | Schedules for Clinical Assessment in Neuropsychiatry (SCAN) (World Health Organization, 1994) |
| Brief description         | D is a 32-year-old male with mild ID, ASD and paranoid schizophrenia. His family reported that his early developmental milestones were delayed, particularly his speech. He attended special educational needs schooling and came to the attention of mental health services at around the age of 5 years. He was diagnosed with ADHD and conduct disorder during his childhood. Later on, he was diagnosed with paranoid schizophrenia during his adolescent years, at a time when he had been experiencing difficulties in affect regulation, an intense preoccupation in death, poor relationships with his peers and a history of fire setting. D was first admitted to hospital at the age of 15 years and had multiple subsequent readmissions. Most readmissions were due to relapses resulting from non-compliance with medication, as well as alcohol and illicit substance abuse. When initially admitted to hospital, D would typically report hearing the tweets of birds, with the conviction that they were directed specifically to him. He believed that such experiences represented warnings that people intended to harm him. D had previously worn a stab proof vest and carried a knife in order to protect himself from such perceived harm. D also complained that he was able to see small lights on walls and lights coming through windows, which he interpreted as an indication that there were men outside using torches, awaiting an opportunity to murder him. He additionally reported that women were communicating to him through the television to warn him of forthcoming danger. D specifically mentioned a well-known British television presenter doing this repeatedly. D also reported other firmly held beliefs consistent with a clinical picture of Capgras syndrome, stating that his family, and the nursing staff, had been taken and replaced by clones, who were intent on murdering him. He believed that they were taking exclusively about him into a recording device. He believed staff were “wearing wires” and spying on him and planned to murder him. D also disclosed that he thought these individuals were part of the mafia. |
| Evidence of impairment due to symptoms | Subjective distress  
Risks of harm to others (related to hallucinations/delusions)  
Risks of property damage/ fire setting (related to hallucinations/delusions)  
Risks of substance misuse (as a means of managing his distressing experiences)  
Vulnerability (deterioration in level of functioning related to onset of symptoms) |
| Response to treatment     | D initially responded well to Olanzapine but would frequently stop taking oral medication. His periods of non-compliance correlated with increases in illicit drug and alcohol abuse. Commencement of Olanzapine in depot form led to sustained symptomatic improvement |
Non-pharmacological treatments, such as interventions to address maladaptive assumptions, the use of distraction or masking techniques to treat auditory hallucinations (Nelson et al., 1991) and interventions to reduce expressed emotions (Anderson and Adams, 1996) may also be used as adjunctive measures.

It should also be recognised that the combination of ASD and psychosis may have an impact on the length of stay in hospitals. Hare et al. (1999) reported that people with ASD stayed in hospital settings for an average of 8.5 years, which is 2-3 years longer than other patients. This may be because people with ASD pose unique challenges in terms of management, treatment and eventual placement (Alexander et al., 2016). When a comorbid psychotic disorder is present, this may potentially increase the length of stay even further. However, it is likely that improving the recognition of the needs of patients with comorbid ASD and psychosis will begin to improve the effectiveness of treatment in this area.

A key area of relevance is risk management of people with ASD and comorbid psychosis. Some of the features of ASD, such as a lack of understanding of social norms, concrete interpretation of rules, misinterpretations of others’ intentions, difficulties in expressing emotions and pursuit of special interests with morbid/unusual qualities, may pose additional risks on patients when they are diagnosed with psychosis, potentially making them even more prone to offending. Future research should seek to clarify these issues, in order to more effectively assess and treat patients with complex presentations.

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