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CASE REPORT

A case of mistaken diagnoses: diagnostic and management challenges in a case of adult autism spectrum disorder

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Autism spectrum disorder (ASD) is frequently identified in children but is often unrecognised in adults. ASD is characterised by difficulties in social interaction, communication and restricted interests, but other presentations are common, especially in adults. This report describes a 34-year-old man with a history of multiple psychiatric diagnoses including generalised anxiety disorder, major depressive disorder and panic disorder. He was diagnosed with ASD in his early 30s and engaged in a targeted treatment plan, including rationalisation of medications, supportive therapy and occupational therapy, which successfully facilitated discharge from mental health services. This case illustrates the atypical presentation of ASD in adults, which is diagnostically challenging. Such cases often present to community mental health services and may be misdiagnosed as treatment resistant cases of depressive, anxiety or personality disorders. Accurate diagnosis and targeted management is more likely to yield a successful outcome.

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Introduction

Neurodevelopmental disorders (NDD) are frequently identified in childhood but are often unrecognised in adults (Takara *et al*. 2015). NDDs constitute a group of conditions with an onset in the developmental period (American Psychiatric Association (APA), 2013). They include autism spectrum disorder (ASD), attention deficit hyperactivity disorder and specific disorders of areas of communication, motor function and sensory processing (APA, 2013). The prevalence of ASD is stable throughout lifetime and is estimated at 1% in the general population (Boilson *et al*. 2016), although some adults demonstrate compensatory strategies leading to reduced symptomatology later in life, for example, improved social skills (Hoekstra *et al*. 2007).

ASD consists of two diagnostic criteria: criterion A (social and communication deficits) and criterion B (restricted/repetitive behaviours and abnormal sensory sensitivity) (APA, 2013). Within these criteria exists a great deal of diversity. Clinical presentation is heterogeneous, with individuals often exhibiting signs of more than one NDD (Berenguer-Forner *et al*. 2015). Psychiatric co-morbidity is high, especially co-morbid mood and anxiety disorders (Vannucchi *et al*. 2014). Furthermore, the presentation of adult ASD may mimic other psychiatric disorders, such as schizophrenia, mood and anxiety disorders or personality disorders, which leads to misdiagnosis. Gender differences also pose challenges, with diagnosis often delayed or missed in women due to their distinct phenotype (greater capacity for traditional friendships, higher social motivation and fewer externalising behaviours) (Bargiela *et al*. 2016). Females with ASD score higher on friendship questionnaires than their male counterparts, which masks the criterion A deficits in Diagnostic and Statistical Manual of Mental Disorders, 5th Edition (DSM-5) (Head *et al*. 2014).

Development of care pathways for adult ASD has been outlined by National Institute for Health and Care Excellence (2016) and the need for such services was emphasised by the Health Service Executive (2012). Improving diagnostic expertise among adult psychiatrists has been identified as a necessary component of this, which has led to the Royal College of Psychiatrists providing resources for psychiatrists, such as *Diagnostic Interview Guide for the Assessment of Adults with Autism Spectrum Disorder* (Berney *et al*. 2011). The College of Psychiatrists of Ireland has taken steps towards provision of training by hosting autism seminars and the National Disability Association recently published guidelines for management of mental health difficulties in adults of normal intellect with ASD [National Disability Authority (NDA), 2017].

We present the case of a man diagnosed with ASD in adulthood, on a background of a 10-year history of

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atypical psychiatric presentations refractory to treatment in adult mental health services. This case was chosen from a university mental health service as it illustrates the interface between traditional adult mental illness and NDD. The University Mental Health Service is led by a consultant psychiatrist, who works along side occupational therapy and student counselling services. This case highlights the important role played by adult mental health services in managing the difficulties and co-morbidities associated with NDD in adults.

Summary of case

Mr X is a 34-year-old man who was diagnosed with ASD 5 years ago. He was referred to the University Mental Health Service during his first year of his postgraduate degree. Mr X described suffering with anxiety since childhood and becoming overwhelmed in stressful circumstances. He described himself as 'always feeling different from others' and recalled, in childhood, researching how to behave in social contexts. It was evident from his academic record that Mr X was highly intelligent. He excelled at school, attaining a score above 40 in the International Baccalaureate, and was academically successful at undergraduate and postgraduate levels. However, he described finding it difficult to know how to respond and behave in social situations. He perceived a high level of threat, feeling overwhelmed and paranoid in unfamiliar situations, and agitated in high levels of sensory exposure. This led to behaviour that was interpreted by others as bizarre.

Mr X had a history of attending mental health services since adolescence and had received multiple diagnoses including generalised anxiety disorder, social anxiety, emotionally unstable personality disorder, chronic dysthymia and psychosis. He had two involuntary admissions to psychiatric units, with discharge diagnoses of acute psychotic episodes.

Background and progress

Mr X provided discharge summaries from previous mental health services and a written biographical account of his previous history at the time of his initial assessment in the University Mental Health Service. According to these records, Mr X's early life was unremarkable. There was evidence of delayed speech but no engagement with speech therapy. He reported his mother to have been obsessional and overprotective. His younger brother was reported to suffer with behavioural difficulties and required additional support at school. Mr X described struggling socially in primary school and was bullied throughout, but he managed to make some friends in secondary school. He wrote that he played the role of 'the quirky guy making gaffs' which made him likeable. As a young adult, Mr X recalled questioning aspects of his identify, such as sexuality and gender. Mr X achieved an honours degree at undergraduate level and subsequently pursued postgraduate qualifications.

Mr X spent his adult life travelling, and attended psychiatrists in Ireland, Luxembourg, France, Canada and America. He described periods of low mood, emotional dysregulation, anxiety and paranoia. He had two involuntary admissions to psychiatric units with persecutory ideas and extreme agitation, which, according to clinical notes, resolved rapidly shortly after admission. He reported multiple overdoses. Mr X was prescribed a variety of psychotropic medications in an effort to treat his diverse presentations (anxiety, low mood, psychotic symptoms) including haloperidol, olanzapine, risperidone, quetiapine, fluoxetine, sertraline, nortipytyline, pregabalin and benzodiazepines, all of which had transient and limited efficacy.

Mr X was diagnosed with ASD by a psychiatrist 5 years ago, a diagnosis that was reached following personal pursuit by Mr X, after reading extensively on the subject on NDDs. This diagnosis was confirmed in the University Mental Health Services following full psychiatric assessment, using the RCPsych Diagnostic Interview Guide for the Assessment of Adults with ASD. Mr X demonstrated a range of enduring impairments across all areas of assessment including reciprocal social interaction, communication difficulties with language delay, the use of formal and pedantic language, limited non-verbal communication and a history of repetitive interests. This history was corroborated by his partner. He scored 42 on the AQ (Autism Spectrum Quotient).

Once diagnosed, Mr X developed a different understanding of his difficulties. During his engagement with the psychiatry clinic, the psychologist and occupational therapist over the following 4 years, it was determined that cognitive inflexibility, sensory hypersensitivity and difficulty navigating interpersonal interactions were common impairments. Relationship dysharmony, and procedures around separation, was a particular source of distress in his early 30s and caused a deterioration in his mental health, often leading to Emergency Department attendances. Impaired expression of his emotional discomfort caused his distress to manifest as apparent mental illness. With the support of counselling services, he learned to interpret and navigate stressful social situations. Occupational therapy provided him with strategies to avoid or cope with high levels of sensory exposure. These interventions facilitated gradual medication reduction over a period of 18 months. At the beginning of this process, he was prescribed pregabalin 300 mg OD, sertraline 100 mg

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OD, nortriptyline 50 mg OD, propranolol 40 mg OD, haloperidol 0.5 mg OD and quetiapine 25 mg OD. At the point of discharge he had been stable for 2 years and was prescribed pregabalin 100 mg OD. Trials off all medication had not yet been successful but the eventual aim was to discontinue all medications.

Discussion

Adults with NDD are known to present with unusual symptoms, such as bizarre or dramatic anxiety responses and unusual fears. Disordered thinking and paranoia in response to situational stress, along with poor social communication, may mimic symptoms of schizophrenia (Takara *et al*. 2015). The case described herein exemplifies the ensuing trajectory of a misdiagnosed NDD, which led to polypharmacy, repeated hospital admissions and frustration for clinicians and patient. The benefit of accurate diagnosis was two-fold. First, it empowered the patient to achieve an understanding of self which he had sought since childhood. Additionally, reframing Mr X's difficulties as primarily neurodevelopmental facilitated targeted multidisciplinary management, with a focus on stress reduction, interpersonal effectiveness and sensory modulation, and a de-emphasis on medication.

One feature of this case that is particularly worthy of discussion was Mr X's over-responsivity to sensory exposure. Abnormal sensory sensitivity was added to criterion B in DSM-5 diagnostic criteria for ASD. In Mr X's case, acute sensory exposure (especially in auditory or visual modalities) led to a sensory crisis, in which he would become acutely agitated and behaviourally disturbed. Both acute sensory exposure and cumulative, low-grade sensory over-responsivity are a common source of distress in adults with autism (Tavossoli *et al*. 2014), but are inherently difficult to identify. The experience of cumulative over-responsivity occurs in response to seemingly innocuous stimuli across the full range of sensory domains, for example, chewing (auditory), light flickering (visual), clothing labels (tactile). This highlights the importance of screening for sensory over-responsivity in community mental health patients who suffer with unexplained and seemingly random emotional distress.

Cases such as this are diagnostically challenging, especially in general adult psychiatry where NDDs are not traditionally managed, however, a number of resources provide guidance in this regard (Murphy *et al*. 2016; NDA, 2017). The *Royal College of Psychiatrists Diagnostic Interview Guide for the Assessment of Adults with Autism Spectrum Disorder* is a comprehensive and straightforward guide for the diagnosis of ASD (Berney *et al*. 2011). Maudsley Guidelines provide direction on medical investigation and prescribing in individuals with autism (the familiar 'start low and go slow' maxim is applicable) (Murphy *et al*. 2016). The evidence base for the role of psychological intervention is very limited. Cognitive behavioural therapy may help individuals with ASD to understand their difficulties and express their needs (Hesselmark *et al*. 2014) and there is evidence that supports the use of cognitive enhancement therapy as a treatment for the core features of ASD (Eack *et al*. 2018). In the case of Mr X, practical support around managing social stress was a beneficial intervention, and did not require expertise particular to NDD. The role of occupational therapy in these types of cases includes sensory modulation and improved occupational functioning, both of which pose practical challenges for individuals with ASD (Ben-Sasson *et al*. 2009; Roux *et al*. 2013). There is some evidence supporting the benefits of mindfulness-based therapy to reduce depression and anxiety in high functioning adults with ASD (Spek *et al*. 2013).

This case report outlines the role of adult mental health services to manage the mental health difficulties associated with NDD. Recognition of ASD and other NDDs can prove to be a valuable therapeutic intervention for seemingly refractory cases of anxiety disorders, depressive disorders, psychotic illness and other clinical presentations. Optimising treatment of such cases will ultimately facilitate recovery, reduce pressure on resources and improve satisfaction for clinicians and patients.

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Conflicts of interest

Neither author has a conflict of interest to disclose.

Ethical Standards

The authors assert that all procedures contributing to this work comply with the ethical standards of the relevant national and institutional committee on human experimentation with the Helsinki Declaration of 1975, as revised in 2008. The authors assert that ethical approval for publication of this case report was not required by their local Ethics Committee. Informed written consent was obtained.

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