

## Editorial

# Autism spectrum disorders: current issues and future directions

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### Abstract

This edition of Irish Journal of Psychological Medicine is a Special Themed Issue on Autism Spectrum Disorders (ASD). Mental health services are not currently meeting the needs of autistic people across the lifespan. We have limited evidence based treatments for core symptoms and comorbidities and there is lack of awareness and under-recognition of ASD, particularly in adults and certain groups of individuals. The key themes in this edition focus on challenges with recognition and diagnosis and address these from both clinical and research perspectives. Co-occurring conditions also feature, which are also under-recognised and can contribute to less optimal outcomes. New and existing research developments in stratification for clinical trials and neuroimaging are also discussed. We hope this Issue highlights relevant current issues in ASD, and provides insights which can help address the challenges in providing evidence based pathways to better meet the needs of autistic people into the future.

**Keywords:** Autism spectrum disorders; clinical services; mental health; research.

### Introduction

The current edition of Irish Journal of Psychological Medicine focuses on Autism Spectrum Disorders (ASD), also referred to here as autism. Autism is a neurodevelopmental condition that affects around 1% of the population and is frequently associated with co-occurring mental health disorders (Lai *et al.* 2019). Autism is highly heterogeneous affecting individuals across the age and IQ spectrum. Although increasingly recognised and diagnosed, autism is still under-recognised in some groups of individuals, who often experience a diagnostic odyssey of inaccurate mental health diagnoses. Diagnostic overshadowing, where mental health symptoms are attributed to autism leads to under-recognition and under treatment of co-occurring mental and physical disorders. This may be a factor in the earlier mortality observed in autism, especially where suicide-related (Hirvikoski *et al.* 2016). The papers featuring here reflect the clinical heterogeneity of autism and the challenges these pose to clinical recognition of diagnosis and treatment in both research and clinical care.

### Under-recognition and co-occurring mental health disorders complicate diagnosis

Under-recognition of autism and co-occurring mental health disorders were the subject of five clinically focused papers in this issue. Challenges with recognition and service provision for adults is a global concern (Murphy *et al.* 2016). Crowley *et al.*, (pp 312–318)

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reviewed the unmet need of adults with ASD in mental health services and the need for evidence based approaches to identification and diagnosis. They propose a model of care for autistic individuals within adult mental health and recommend autism awareness training among mental health professionals. Alexander and Farrelly (pp 301–304) provide a nice illustration of underdiagnosis in their description of an autistic individual who received multiple psychiatric diagnosis before autism was correctly identified. They note that stressed autistic adults may present with bizarre symptoms. Access to appropriate autism supports in this case ultimately resulted in discharge from mental health services. Most importantly, they emphasised that recognition of autism was itself a beneficial intervention, allowing for improved understanding of personal difficulties. Both papers call for specialist care pathways and the need for guidelines for diagnosis, prescribing and therapeutic approaches.

Focusing on children, an original research study by Leader *et al.*, (pp 240–250), investigated the relationship between co-occurring ADHD in the context of autism with other aspects of behavioural and physical health. They reported an association between aggression and co-occurring ADHD. This is important as it highlights that ADHD may be a diagnostic consideration and treatment target in the context of aggression. A second paper by Leader *et al.*, (pp 261–271) reported on a pilot study of sleep using actigraphy in families with an autistic child. Sleep difficulties are common in children with ASD based on informant report, and the use of wearable sensors may provide more objective sleep measurement. In their pilot study they noted poor agreement between informant report and actigraphy. Poor compliance with the wearable sensors reduced the data available for children, indicating the sensors may not be a generalisable solution for accurate sleep measurement in this context. The study, although small, highlights an important

and often neglected area of health and wellbeing that contributes to reduced quality of life for families of autistic children with sleep difficulties.

Turning to the impact of the pandemic, Byrne and Longphuir (pp 319–320) noted a paradoxical reduction in anxiety and behavioural challenges among a group of children with autism without intellectual disability (ID) that corresponded with school closures in the early days of the pandemic. This they attributed to reduced demands on children from school and the community. In fact these observations were not entirely borne out over time, as later studies indicated that while some autistic young people may have benefitted from reduced demands, the mental health of young autistic people may have been disproportionately affected during the pandemic (Asbury and Toseeb 2022). Nonetheless, the observations by Byrne and Longphuir certainly raise questions concerning environmental impacts on anxiety in autism that require further study.

### Lack of data concerning autism in special populations

Considering special populations, three articles in this issue address the recognition of autism among older adults with ID and prisoners. Little is known about the prevalence of autism in older adults with ID and the relationship to outcomes. In their review, Maguire *et al.*, (pp 287–300) highlight that the older adult group with both ASD and ID had reduced functional independence and higher rates of psychiatric comorbidity when compared with an older adult group generally or those with ID only. Accordingly, they propose that longitudinal studies are required to support the provision of resources for future care needs in this population. Indeed, this has been highly effective in advocating for the needs of older people with ID in Ireland (McCarron *et al.* 2013) and has led to important developments in models of care such as the memory clinic in Tallaght University Hospital.

Moloney and Gulati (pp 321–323) discuss the overrepresentation of autistic people in prison populations internationally. They suggest that under-detection of autism among prisoners may be related to lack of awareness, training and reliable diagnostic approaches. The importance of recognition is underscored by the disadvantages that autistic people may face in relation to processes in the criminal justice system and in their relationships with other prisoners. In a reply, O'Sullivan stated that knowledge gaps also exist in the UK prison population due to similar factors. Both authors call for more research regarding ASD in the prison population, particularly prevalence studies to inform policy and rehabilitation.

### Research in diagnostic tools, brain mechanisms and biomarker discovery

This edition also contains articles with a research focus on diagnosis, brain mechanisms and biomarker discovery. Anglim *et al.*, (pp 251–260) report on their analysis of the factor substructure of the diagnostic interview for social and communication disorders (DISCO-11) and the relationship to DSM-5 ASD criteria. A two-factor substructure of social communication and restricted and repetitive behaviours has been reported in relation to the Autism Diagnostic Interview-Revised (ADI-R) and partly informed the realignment of symptoms in DSM-5 (Snow *et al.* 2009). This is the first examination of the underlying factors in the DISCO-11. Their findings suggest that the DISCO is a valid measure in the context of DSM-5.

There are numerous neuroimaging studies of autism investigating brain structure and function, focused largely on functional and

structural Magnetic Resonance Imaging (MRI). The imaging research has demonstrated abnormalities in brain structure and function in autism, from which a number of theories of altered cortical connectivity have emerged (Kana *et al.* 2011). PET and SPECT are two nuclear imaging techniques that can detect cellular changes in organs and tissues earlier than MRI scans, but these techniques are used considerably less frequently than MRI. Kowalewska *et al.*, (pp 272–286) provide a review of PET and SPECT studies in autism. They describe inconsistency in study findings and suggest that heterogeneity is a particular difficulty within the autism cohorts that have been studied. It seems unlikely that PET and SPECT will become widespread modalities in ASD neuroscience research, but PET may have utility for studies at the molecular level, for example in researching synaptic density (Serrano *et al.* 2022)

Molloy and Gallagher (pp 305–311) discuss new directions for biomarker research to address diagnostic heterogeneity in ASD. They present the case for biologically valid stratification biomarkers to identify subgroups of autistic individuals and the necessity for these in the context of research, clinical trials and clinical care. Ultimately the vision in the field is to provide biomarkers leading to more personalised care and therapeutics for those requiring it the most.

Considering these papers, it seems that longstanding challenges prevail in autism research and clinical care. Clinical heterogeneity is characteristic of autism and varies across different domains of development, cognition, and behaviour. This renders diagnosis more difficult, particularly in adults and certain vulnerable populations. The developmental nature of autism means that diagnosis frequently focuses on the early years, requiring reliable collateral history that is not always available and comorbidities may further cloud the picture. Consequently, it remains a challenge to provide evidence based mental health services and we lack models of care, an issue which is highly relevant in Ireland. The autism research community are trying to address these challenges, and advances in neuroscience may go some way to provide more appropriate care where it is needed.

### Future directions for research and clinical care

Clearly, to provide better mental health services to autistic people we must increase awareness and training for mental health professionals and develop models of care and service innovations. These seem unlikely without health services research and investment. The disability services in Ireland and elsewhere are currently struggling to meet the needs of autistic children and young people. A whole health strategy is more appropriate for autistic people, which in Ireland will hopefully be addressed in the newly established Oireachtas special committee for autism.

The era of personalised medicine is upon us, and large-scale international studies are pursuing the goal of improved therapeutics and biomarkers. This collides with the growing view of autism as a spectrum of neurodiversity and is associated with heated debate between diverse parts of the autism community (for example among adult self-advocates and parents of autistic children). This calls for serious engagement between researchers, clinicians, and the autism community to co-create research questions that are relevant and that address outcomes which are important to improved quality of life. Examples of excellence in relation to engagement and participant involvement include efforts within AIMS-2-TRIALS. In Ireland, the HRB funded PPI Ignite network exists to support researchers to build participation into their

research. Finally, it is notable that there were no papers in this issue focused on autistic females who have an increased suicide risk in comparison to typically developing peers, possibly moderated by co-occurring ADHD (Hirvikoski *et al.* 2016). Research in autistic females aligns to the policies of funding agencies internationally, and will be essential to understand and appropriately support all autistic individuals (Lai and Szatmari 2020).

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