PERSONS WITH INTELLECTUAL DISABILITIES 2
https://doi.org/10.33591/sfp.48.7.u1

Unit No. 1

INTELLECTUAL DISABILITY AND AUTISM SPECTRUM DISORDER: DEFINITION, DIAGNOSIS, AND THE CLINICAL APPROACH TO DIFFERENTIATING BETWEEN THEM

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ABSTRACT

Neurodevelopmental disorders are a diverse group of conditions that occur during the developmental period and result in functional impairments. Intellectual Disability (ID) and Autism Spectrum Disorder (ASD) belong in this group and the criteria for diagnosis is described in DSM-5. There is overlap in the clinical symptoms between the conditions and careful assessment is required to establish diagnosis and determine if there is comorbidity and to plan treatment.

Keywords: ID, ASD, Intellectual Disability, Autism Spectrum Disorder

SFP2023; 48(7): 6-10

INTRODUCTION

Neurodevelopmental Disorders (NDD) are defined as a group of conditions with onset in the developmental period which cause functional impairments. These conditions include Intellectual Disability (ID); Communication Disorders; Autism Spectrum Disorder (ASD); Attention-Deficit/ Hyperactivity Disorder (ADHD); Neurodevelopmental Motor Disorders, including Tic Disorders; and Specific Learning Disorders.

This article will discuss the definition, diagnosis, and assessment approach to differentiate between ID and ASD.

INTELLECTUAL DISABILITY (ID)

Definition of ID

In the fifth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-5), the diagnosis of intellectual disability (intellectual developmental disorder) was revised from the DSM-IV diagnosis of mental retardation. It limits a person's ability to learn at an expected level and function in daily life. Intellectual disability involves impairments of general mental abilities that impact adaptive functioning in three domains. These domains determine how well an individual copes with everyday tasks:

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- The conceptual domain includes skills in language, reading, writing, math, reasoning, knowledge, and memory.
- The social domain refers to empathy, social judgement, interpersonal communication skills, the ability to make and retain friendships, and similar capacities.
- The practical domain centres on self-management in areas such as personal care, job responsibilities, money management, recreation, and organising school and work tasks.

While intellectual disability does not have a specific age requirement, an individual's symptoms must begin during the developmental period (usually before age 18) and severity is based on the extent of deficits in adaptive functioning. The disorder is considered chronic and often co-occurs with other developmental and mental conditions.[1]

Diagnosis of ID

The clinical approach to diagnosing ID is to use both clinical assessment and standardised testing of intelligence, with the severity of impairment based on adaptive functioning rather than IQ test scores alone. It is important to not use IQ scores as the defining factor of a person's overall ability without adequately considering their functioning levels. Intellectual disability is identified by problems in both intellectual and adaptive functioning (see **Box 1** for DSM-5 diagnostic criteria for ID).[5]

Intellectual functioning is measured with individually administered and psychometrically valid, comprehensive, culturally appropriate, psychometrically sound tests of intelligence. While a specific full-scale IQ test score is no longer required for diagnosis, standardised testing is often used as part of diagnosing the condition. A full-scale IQ score of around 70 to 75 indicates a significant limitation in intellectual functioning. However, the IQ score must be interpreted in the context of the person's difficulties in general mental abilities. Moreover, scores on subtests can vary considerably so that the full-scale IQ score may not accurately reflect overall intellectual functioning. Therefore, clinical judgement is needed in interpreting the results of IQ tests.[2]

Adaptive functioning in the various domains is assessed through clinical interviews and history-taking with the individual and others, such as family members, teachers, and caregivers, and can be aided by standardised measures. The commonly used standardised tests for assessing adaptive functioning include:

- Vineland Adaptive Behaviour Scales, Third Edition (VABS) (Vineland-3)

cians an opportunity to document factors that may have played a role in the etiology of the disorder, as well as those that might affect the clinical course. Examples include genetic disorders, such as fragile X syndrome, tuberous sclerosis, and Rett syndrome; medical conditions such as epilepsy; and environmental factors, including very low birth weight and fetal alcohol exposure (even in the absence of stigmata of fetal alcohol syndrome).

Intellectual Disabilities

Intellectual Disability (Intellectual Developmental Disorder)

Diagnostic Criteria

Intellectual disability (intellectual developmental disorder) is a disorder with onset during the developmental period that includes both intellectual and adaptive functioning deficits in conceptual, social, and practical domains. The following three criteria must be met:

A. Deficits in intellectual functions, such as reasoning, problem solving, planning, abstract thinking, judgment, academic learning, and learning from experience, confirmed by both clinical assessment and individualized, standardized intelligence testing.
B. Deficits in adaptive functioning that result in failure to meet developmental and sociocultural standards for personal independence and social responsibility. Without ongoing support, the adaptive deficits limit functioning in one or more activities of daily life, such as communication, social participation, and independent living, across multiple environments, such as home, school, work, and community.
C. Onset of intellectual and adaptive deficits during the developmental period.

Note: The diagnostic term *intellectual disability* is the equivalent term for the ICD-11 diagnosis of *intellectual developmental disorders*. Although the term *intellectual disability* is used throughout this manual, both terms are used in the title to clarify relationships with other classification systems. Moreover, a federal statute in the United States (Public Law 111-256, Rosa's Law) replaces the term *mental retardation* with *intellectual disability*, and research journals use the term *intellectual disability*. Thus, *intellectual disability* is the term in common use by medical, educational, and other professions and by the lay public and advocacy groups.

Specify current severity (see Table 1):
317 (F70) Mild
318.0 (F71) Moderate
318.1 (F72) Severe
318.2 (F73) Profound

Specifiers

The various levels of severity are defined on the basis of adaptive functioning, and not IQ scores, because it is adaptive functioning that determines the level of supports required. Moreover, IQ measures are less valid in the lower end of the IQ range.

    - The VABS measures three broad domains of adaptive functioning: communication, daily living skills, and socialisation.

- The Diagnostic Adaptive Behaviour Scale (DABS) (AAIDD, 2013)
    - The DABS was constructed with items across three domains: conceptual, social, and practical adaptive skills. The DABS was developed to focus its assessment around the decision point for determining the presence or absence of significant limitations of adaptive behaviour for the diagnosis of ID.
- Adaptive Behaviour Assessment System 3 (ABAS-3)
    - The ABAS-3 covers three broad domains: conceptual, social, and practical. Tasks focus on everyday activities required to function, meet environmental demands, care for oneself, and interact with others effectively and independently.

Intellectual disability is identified as mild (most people with intellectual disability are in this category), moderate, or severe, and symptoms have to have occurred during the developmental period.

The assessment of intellectual disability across the three domains (conceptual, social, and practical) ensures that clinicians base their diagnosis on the impact of the deficit in general mental abilities on functioning needed for everyday life. This is especially important in the development of a treatment plan.

AUTISM SPECTRUM DISORDER (ASD)

Definition of ASD

Previously under DSM-IV, patients could be diagnosed with four separate disorders: autistic disorder, Asperger's disorder, childhood disintegrative disorder, or the catch-all diagnosis of pervasive developmental disorder not otherwise specified. With DSM-5, there is now a single umbrella disorder of Autism Spectrum Disorder, which is believed to improve the accuracy of the diagnosis of ASD.

People with ASD tend to have communication deficits, such as responding inappropriately in conversations, misreading nonverbal interactions, or having difficulty building friendships appropriate to their age. In addition, people with ASD may be overly dependent on routines, highly sensitive to changes in their environment, or intensely focused on inappropriate items. The symptoms of people with ASD will fall on a continuum, with some individuals showing mild symptoms and others having much more severe symptoms. This spectrum allows clinicians to account for the variations in symptoms and behaviours from person to person. The severity is classified as follows:

- Level 1: deficits cause noticeable social impairments and inflexibility causes significant interference with transitions and hampers independence
- Level 2: marked deficits in nonverbal and verbal social communication, limited or reduced social responses; repetitive behaviour and rigidity is marked and very noticeable
- Level 3: severe deficits in nonverbal and verbal social communication, very limited or no social responses; repetitive behaviour and rigidity severely limits functionality

Under the DSM-5 criteria, individuals with ASD must show symptoms from early childhood, even if those symptoms are not recognised until later. This criterion change encourages earlier diagnosis of ASD but also allows people whose symptoms may not be fully recognised until social demands exceed their capacity to receive the diagnosis.[3]

Diagnosis of ASD

The diagnosis of ASD is made by evaluating the patient's behaviour and development and may be diagnosed from the age of two onwards.

Diagnosis in Young Children

Every child should receive well-child check-ups with a pediatrician, which may include screening for developmental delays at their 9-, 18-, and 24- or 30-month well-child visits,

with specific autism screenings at their 18- and 24-month. A child may receive additional screening if they have a higher likelihood of ASD or developmental problems, e.g., if they have a family member with ASD, show some behaviours that are typical of ASD, have older parents, have certain genetic conditions, or who had a very low birth weight.

The assessment will include obtaining history about the child's behaviours and involve the evaluation of those answers in combination with information from ASD screening tools and clinical observations of the child.[4]

If the screening assessment is positive, then the child can be referred to a service with experience diagnosing ASD for a diagnostic evaluation, which may include medical and neurological examinations, assessment of the child's cognitive abilities and language abilities, observation of the child's behaviour, in-depth conversation with the child's caregivers about the child's behaviour and development, and assessment of age-appropriate skills needed to complete daily activities independently, such as eating, dressing, and toileting.

Diagnosis in Older Children and Adolescents

Caregivers and teachers are often the first to recognise ASD symptoms in older children and adolescents who attend school. The school's special education team may perform an initial evaluation and then refer on for further specialist assessment if required.

At this stage, social difficulties, including problems with social communication, may be the presenting problem. For example, some children may have problems understanding tone of voice, facial expressions, or body language. Older children and adolescents may have trouble understanding figures of speech, humour, or sarcasm. They also may have trouble forming friendships with peers.

Diagnosis in Adults

Diagnosing ASD in adults is often more difficult than diagnosing ASD in children. In adults, some ASD symptoms can overlap with symptoms of other mental health disorders, such as anxiety disorder or attention-deficit/hyperactivity disorder (ADHD). The assessment should enquire about:

- Social interaction and communication challenges
- Sensory issues
- Repetitive behaviours
- Restricted interests

The evaluation should include a conversation with caregivers or other family members to learn about the person's early developmental history, which can help ensure an accurate diagnosis. Receiving a correct diagnosis of ASD as an adult can help a person understand past challenges, identify personal strengths, and find the right kind of help.

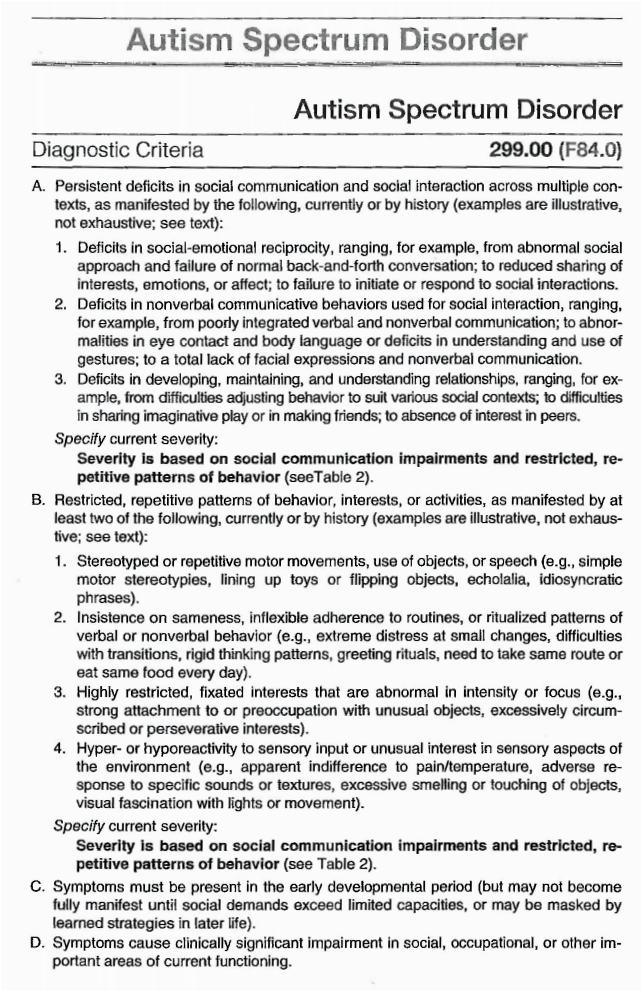
Screening and diagnostic tools for ASD

Examples of screening tools for general development and ASD that may be used include:

- Ages and Stages Questionnaires (ASQ)
    - This is a general developmental screening tool. Parent-completed questionnaire; series of 19 age-specific questionnaires screening communication, gross motor, fine motor, problem-solving, and personal adaptive skills; results in a pass/fail score for domains.
- Communication and Symbolic Behaviour Scales (CSBS)
- Parents' Evaluation of Developmental Status (PEDS)
- Modified Checklist for Autism in Toddlers (MCHAT)
- Screening Tool for Autism in Toddlers and Young Children (STAT)
- Childhood Autism Rating Scale (CARS)
- Gilliam Autism Rating Scale – Second Edition (GARS-2)

Examples of diagnostic tools that may be used include:

- Autism Diagnosis Interview – Revised (ADI-R)
    - A clinical diagnostic instrument for assessing autism in children and adults. The instrument focuses on behaviour in three main areas: reciprocal social interaction; communication and language; and restricted and repetitive, stereotyped interests and behaviours. The ADI-R is appropriate for children and adults with mental ages about 18 months and above.
- Autism Diagnostic Observation Schedule – Generic (ADOS-G)
    - A semi-structured, standardised assessment of social interaction, communication, play, and imaginative use of materials for individuals suspected of having ASD. The observational schedule consists of four 30-minute modules, each designed to be administered to different individuals according to their level of expressive language.
- Ritvo Autism Asperger Diagnostic Scale - Revised (RAADS–R)
- Diagnostic Interview for Social and Communication Disorders (DISCO)
- Royal College of Psychiatrists Interview Guide for the Diagnostic Assessment of Able Adults with Autism Spectrum Disorder (ASD) (Revised edition) 2017



Autism Spectrum Disorder

Autism Spectrum Disorder

Diagnostic Criteria **299.00 (F84.0)**

A. Persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following, currently or by history (examples are illustrative, not exhaustive; see text):

1. Deficits in social-emotional reciprocity, ranging, for example, from abnormal social approach and failure of normal back-and-forth conversation; to reduced sharing of interests, emotions, or affect; to failure to initiate or respond to social interactions.
2. Deficits in nonverbal communicative behaviors used for social interaction, ranging, for example, from poorly integrated verbal and nonverbal communication; to abnormalities in eye contact and body language or deficits in understanding and use of gestures; to a total lack of facial expressions and nonverbal communication.
3. Deficits in developing, maintaining, and understanding relationships, ranging, for example, from difficulties adjusting behavior to suit various social contexts; to difficulties in sharing imaginative play or in making friends; to absence of interest in peers.

Specify current severity:
Severity is based on social communication impairments and restricted, repetitive patterns of behavior (seeTable 2).

B. Restricted, repetitive patterns of behavior, interests, or activities, as manifested by at least two of the following, currently or by history (examples are illustrative, not exhaustive; see text):

1. Stereotyped or repetitive motor movements, use of objects, or speech (e.g., simple motor stereotypies, lining up toys or flipping objects, echolalia, idiosyncratic phrases).
2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior (e.g., extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat same food every day).
3. Highly restricted, fixated interests that are abnormal in intensity or focus (e.g., strong attachment to or preoccupation with unusual objects, excessively circumscribed or perseverative interests).
4. Hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment (e.g., apparent indifference to pain/temperature, adverse response to specific sounds or textures, excessive smelling or touching of objects, visual fascination with lights or movement).

Specify current severity:
Severity is based on social communication impairments and restricted, repetitive patterns of behavior (see Table 2).

C. Symptoms must be present in the early developmental period (but may not become fully manifest until social demands exceed limited capacities, or may be masked by learned strategies in later life).

D. Symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning.

E. These disturbances are not better explained by intellectual disability (intellectual developmental disorder) or global developmental delay. Intellectual disability and autism spectrum disorder frequently co-occur; to make comorbid diagnoses of autism spectrum disorder and intellectual disability, social communication should be below that expected for general developmental level.

Note: Individuals with a well-established DSM-IV diagnosis of autistic disorder, Asperger's disorder, or pervasive developmental disorder not otherwise specified should be given the diagnosis of autism spectrum disorder. Individuals who have marked deficits in social communication, but whose symptoms do not otherwise meet criteria for autism spectrum disorder, should be evaluated for social (pragmatic) communication disorder.

Specify if:
With or without accompanying intellectual impairment
With or without accompanying language impairment
Associated with a known medical or genetic condition or environmental factor (**Coding note:** Use additional code to identify the associated medical or genetic condition.)
Associated with another neurodevelopmental, mental, or behavioral disorder (**Coding note:** Use additional code[s] to identify the associated neurodevelopmental, mental, or behavioral disorder[s].)
With catatonia (refer to the criteria for catatonia associated with another mental disorder, pp. 119–120, for definition) (**Coding note:** Use additional code 293.89 [F06.1] catatonia associated with autism spectrum disorder to indicate the presence of the comorbid catatonia.)

Recording Procedures

For autism spectrum disorder that is associated with a known medical or genetic condition or environmental factor, or with another neurodevelopmental, mental, or behavioral disorder, record autism spectrum disorder associated with (name of condition, disorder, or factor) (e.g., autism spectrum disorder associated with Rett syndrome). Severity should be recorded as level of support needed for each of the two psychopathological domains in Table 2 (e.g., "requiring very substantial support for deficits in social communication and requiring substantial support for restricted, repetitive behaviors"). Specification of "with accompanying intellectual impairment" or "without accompanying intellectual impairment" should be recorded next. Language impairment specification should be recorded thereafter. If there is accompanying language impairment, the current level of verbal functioning should be recorded (e.g., "with accompanying language impairment—no intelligible speech" or "with accompanying language impairment—phrase speech"). If catatonia is present, record separately "catatonia associated with autism spectrum disorder."

Specifiers

The severity specifiers (see Table 2) may be used to describe succinctly the current symptomatology (which might fall below level 1), with the recognition that severity may vary by context and fluctuate over time. Severity of social communication difficulties and restricted, repetitive behaviors should be separately rated. The descriptive severity categories should not be used to determine eligibility for and provision of services; these can only be developed at an individual level and through discussion of personal priorities and targets.

Regarding the specifier "with or without accompanying intellectual impairment," understanding the (often uneven) intellectual profile of a child or adult with autism spectrum disorder is necessary for interpreting diagnostic features. Separate estimates of verbal and nonverbal skill are necessary (e.g., using untimed nonverbal tests to assess potential strengths in individuals with limited language).

In addition to the tools above, the American Psychiatric Association's Diagnostic and Statistical Manual, Fifth Edition (DSM-5) provides standardised criteria to help diagnose ASD (see **Box 2** for DSM-5 diagnostic criteria for ASD).[5]

GENERAL APPROACH TO ASSESSMENT AND DIFFERENTIATING BETWEEN ID AND ASD

The features of ID and ASD have been described above. Both are neurodevelopmental disorders with onset in the developmental period and result in functional impairments.

It is estimated that around 40 percent of people with ASD have ID, compared with just 1 percent of people without ASD. Of the remaining, although technically not having ID, many may nonetheless have some form of a disability that requires support. On the other hand, it has been reported that approximately 10 percent of people with ID also have ASD. Of the rest, some may also show some autistic symptoms not meeting criteria for diagnosis of ASD. Hence, there is a lot of clinical overlap between these two conditions.

In more than 75 percent of the patients with ASD and about half of individuals with ID, the causal diagnosis remains unknown, although it is strongly suspected that underlying genetic causes, as yet undiscovered, play a part.[6]

The general approach to diagnostic assessment of ID and/or ASD in an individual and to determine if there is comorbidity or overlap includes:

- Detailed assessment of intellectual functioning (verbal and non-verbal) and of receptive and expressive language abilities
- A developmental history covering progress from infancy onwards. It may be helpful to use a standardised interview tool.
- Observations of the individual in both structured and non-structured settings. It may be helpful to use a standardised observation tool.
- Routine screening for medical and genetic conditions.
- Consideration of other relevant psychosocial factors.

The comorbidity of ID and ASD poses a challenge for diagnosis because of the difficulties that can arise in distinguishing autism specific symptoms from those associated with very low intellectual functioning. Within the group with the most profound ID, it is often near impossible to determine whether the absence of social and communication skills and the stereotyped and repetitive behaviours are attributable to the severe intellectual impairment or due to an additional comorbid ASD condition. In this group, it may be academic to try to delineate the additional ASD diagnosis as the

overriding factor determining their care and outcome will be the severity of their intellectual impairment.

Diagnosing the comorbidity is much more important for those with a less severe ID. For individuals with a mild ID, the presence of ASD will have substantial implications for prognosis and for the nature and intensity of intervention required. Individuals with ID and ASD are likely to need support that is far more individualised, specialised, and structured than do non-ASD individuals of the same level of intellectual functioning, and even then, their prognosis will often be worse.

To diagnose the comorbidity, following detailed assessment, the clinician must consider whether the findings are appropriate for the individual's chronological age, mental age and language age. If not, then special attention needs to be paid to problems in play, social skills, communication, and behaviours of a stereotyped nature. In cases where another disorder also exists, e.g., Down's syndrome, fragile X, tuberous sclerosis, or severe physical or sensory impairments, clinicians must also consider whether the developmental and behavioural patterns are characteristic of that disorder. ASD can coincide with many developmental or genetic conditions and interventions may be delayed if all the individual's difficulties are incorrectly attributed to the earlier diagnosed disorder (diagnostic overshadowing).

Following the assessment approach above, together with knowledge of both atypical and normal developmental patterns, will allow the determination as to whether the individual meets formal diagnostic criteria for ID, ASD or both.[7]

CONCLUSION

Neurodevelopmental disorders including ID and ASD are complex conditions that exist on a spectrum with varying levels of severity. The symptoms of these two conditions may overlap and both can be present in the same individual as co-morbid conditions. This can lead to diagnostic challenges. Careful assessment to arrive at an accurate diagnosis allows for the planning of interventions and treatments.

REFERENCES

1. https://www.psychiatry.org/Filepercent20Library/Psychiatrists/Practice/DSM/APA_DSM-5-Intellectual-Disability.pdf
2. https://www.psychiatry.org/patients-families/intellectual-disability/what-is-intellectual-disability
3. https://www.nimh.nih.gov/health/topics/autism-spectrum-disorders-asd
4. https://www.cdc.gov/ncbddd/autism/hcp-screening.html
5. American Psychiatric Association. Diagnostic and statistical manual of mental disorders. 5th ed. Arlington, VA: American Psychiatric Association; 2013
6. Schwartz CE, Neri G. 2012. Autism and intellectual disability: Two sides of the same coin. Am J Med Genet Part C Semin Med Genet 160C:89-90. doi: 10.1002/ajmg.c.31329. Epub 2012 Apr 12. PMID: 22499552.
7. Howlin P. Autism and intellectual disability: diagnostic and treatment issues. J R Soc Med. 2000 Jul;93(7):351-5. doi: 10.1177/014107680009300704. PMID: 10928021; PMCID: PMC1298058.

LEARNING POINTS

- **Intellectual Disability (ID) and Autism Spectrum Disorder (ASD) are both neurodevelopmental disorders. They occur during the developmental period and result in functional impairments.**
- **In ID, there is impairment of both intellectual and adaptive functioning. In ASD, there are deficits in social communication and interactions as well as restricted interests and repetitive patterns of behaviours.**
- **There is significant clinical overlap in the presentation of the two conditions especially when they are more severe. Diagnosing the conditions and differentiating between the two conditions allows for appropriate interventions and treatment.**