What is autism and how do we make a diagnosis in the new era of DSM-5 and ICD-11

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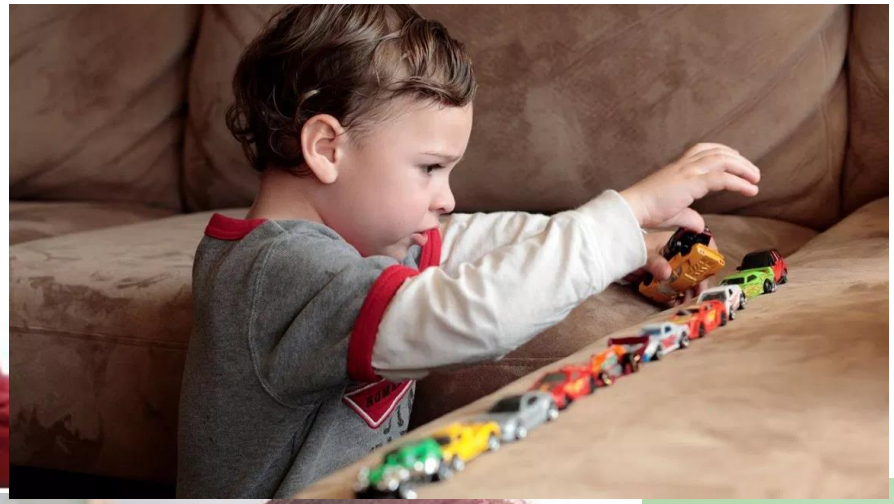

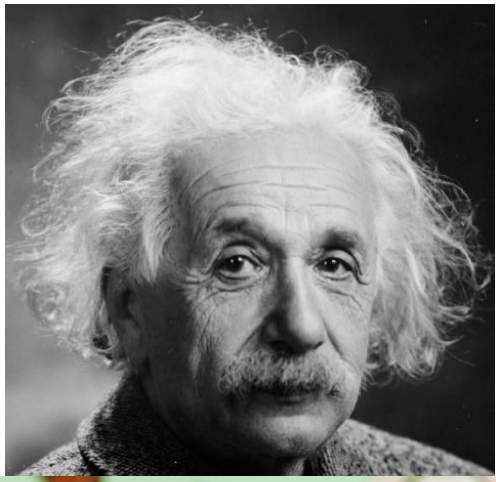


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1799

Dr. Jean Marc Gaspard Itard, Worked with the ferrel boy 'Victor of Aveyron' and described the symptoms of Autism for the first time

1911

Dr. Paul Eugen Bleuler, Swiss psychiatrist, first coined the term autism

1943

Dr. Leo Kanner, Austrian Psychiatrist; published a series of 11 children who displayed 'strong desire to be alone'

1943

Dr. Hans Asperger, Austrian Psychiatrist; published a research paper on autistic psychopathy in childhood

1981

Dr. Lorna Wing, English Psychiatrist; Introduced the term Asperger syndrome

1980 DSM III
1994 DSM IV
1994 ICD-10
2013 DSM-5
2019 ICD-11

- Neurodevelopmental disorder
- Lifetime prevalence 1%
- Core symptoms:
  - Social communication and interaction deficits
  - Restricted and repetitive interests
  - Sensory abnormalities
- Symptoms manifest from early developmental period
- More prevalent in males
- increase in prevalence as intellectual functioning decreases

Complex presentation

- Multifactorial aetiology
- Lifespan presentation
- Co-occurring disorders
- Lifelong difficulties needing long term support
- Heterogeneous presentation

Genetic risk factors

80-90% heritability

10-20% risk of recurrence in a sibling

- 10-15% cases of ASD are single gene syndromes
  - Eg Phelan-McDermid syndrome, Fragile X, Tuberous schlerosis
- De novo single gene loss of function mutations and copy number variants
- Most cases of ASD the genetic risk is polygenic- multiple single nucleotide polymorphisms

Environmental risk factors

Association with increased relative risk:

- Prematurity
- Perinatal hypoxia
- Maternal pre/perinatal infections
- Maternal Vit D deficiency
- Higher paternal age
- Gestational valproate exposure
- Maternal obesity
- Very low birth weight

Brain differences

- Brain structural differences
  - Early brain overgrowth and later growth trajectory flattening
  - ?subset of individuals with ASD
- Serotonin system abnormalities
- Excitatory/inhibitory GABA system imbalance
- Oxytocin system
- Maternal and postnatal immune dysregulation

Diagnostic criteria

- DSM-IV (1994) replaced by DSM-5 (2013)
- ICD-10 (1994) replaced by ICD-11 (2019)
  - Pervasive developmental disorders
  - Triad of impairments
  - Subtypes and partial diagnoses
  - Onset by age 3

Problems with DSM-IV/ICD-10

- Subtypes not distinguished by prognosis, aetiology, neuropsychological profiles, treatment
- Inconsistent diagnosis
- Difficult to distinguish between social communication and interaction
- Age of onset exclusion
- Excluded co-morbid diagnoses

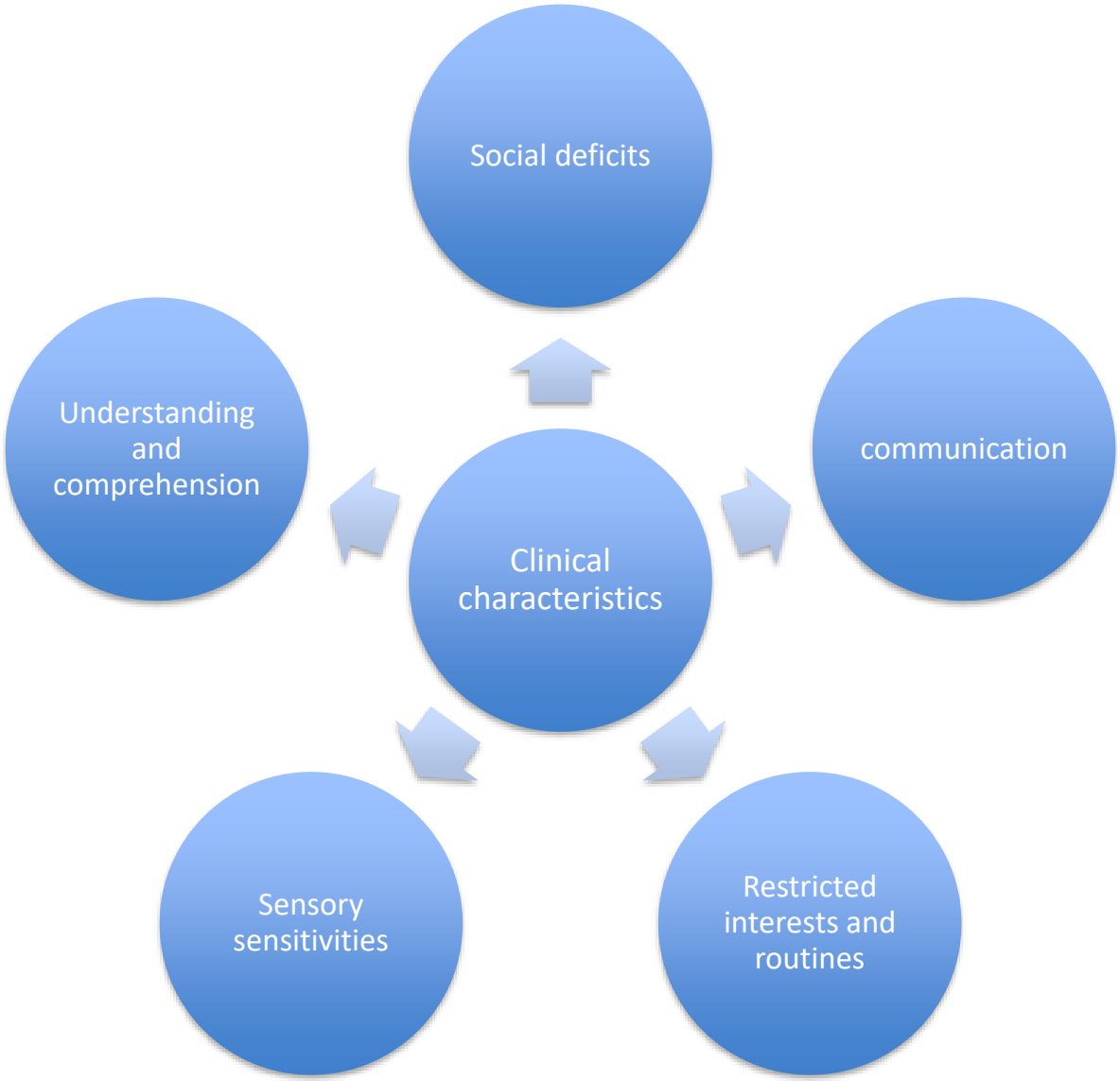
ICD-10	DSM-IV	DSM-5	ICD-11
Pervasive Developmental Disorders: Childhood autism Asperger syndrome Atypical autism PDD-other PDD-unspecified	Pervasive Developmental Disorders: Autistic disorder Asperger's disorder PDD NOS	Autism spectrum disorder	Autism spectrum disorder
3 symptom domains	3 symptom domains	2 symptom domains Including sensory sensitivities	2 symptom domains Including sensory sensitivities
Onset by age 3	Onset by age 3	Removes age of onset	Removes age of onset
Atypical autism	PDD NOS	Social (pragmatic) communication disorder	Developmental language disorder
Comorbidity exclusions	Comorbidity exclusions	Allows co-occurring diagnoses	Allows co-occurring diagnoses

DSM-5

- Spectrum disorder
- Includes sensory sensitivities
- social (pragmatic) communication disorder
  - Classified as a communication disorder
  - Diagnostic reliability, validity, prognosis and common features still to be determined
- Clinical and severity specifiers
- A lack of information about early childhood should not preclude a diagnosis
- Impairment can present later

Comparison of ICD-10R, DSM-IV-TR and DSM-5 in an Adult ASD Diagnostic Clinic (Wilson et al., 2013)

	Above / below ASD threshold		Diagnosis of below ASD threshold participants	
	ASD full-threshold	Below ASD threshold	PDD-unspecified or SCD	No diagnosis
ICD-10R, % (N)	51(76)	50(74)	25(37)	25(37)
DSM-IV-TR, % (N)	53(80)	47(70)	N/A	N/A
DSM-5, % (N)	42(63)	58(87)	14(21)	44(66)


Social deficits
Understanding and comprehension
communication
Clinical characteristics
Sensory sensitivities
Restricted interests and routines

ASD: Impact and impairment

- Educational, occupational and social outcomes can be poor – regardless of intellectual ability
- Often a need for ongoing involvement from health and social care providers (Barnard et al., 2001)
- Carer burden and stress: higher in comparison to non-clinical, and other clinical populations (Cadman et al., 2012)

Recognising autism

- social interaction
- Communication
- Alexithymia
- Unusual or intense interests
- Sensory responses
- Rigidity or inflexibility
- An atypical psychiatric presentation
- Childhood onset

Why is it not recognised

- What are friendships
- Compensation
- Camouflaging
- A lack of developmental history
- Cultures/settings that mask autism
- Being a different cultures/language
- Not being a man
- Diagnostic overshadowing

Assessment (NICE 2012)

- Multi-stage process
- Multidisciplinary approach
- Developmental history from primary caregiver
- Current symptoms
- Exclusion of alternative diagnosis

Diagnostic challenges

- Accuracy of developmental history informant
- Availability of developmental history informant
- Assessment of non-verbal children
- Development of adaptive mechanisms in adults to manage social situations
- Different manifestation in women
  - Different intense interests
  - Fewer stereotyped behaviours
  - More likely to mask social deficits
  - ‘shyness, bossiness, perfectionism’
- Difference between quirk and symptom

Other areas for assessment (NICE, 2012)

- Physical health problems
- Genetic disorders
- Psychiatric co-morbidity
- Medication history (and response)
- Risks
- Vulnerability / exploitation
- Education / occupational status
- Neuropsychological functioning
- Mental capacity

tools

- 20+ tools available
- Mapped onto DSM-IV and ICD-10
- Child v adult tools
- Screening v diagnostic
- Observational v care giver interview

ASD screening questionnaires

- Self-report questionnaires:

  - Autism Quotient: short and full versions

  - Empathy Quotient (AAA, Baron-Cohen et al., 2005)

- Informant-report questionnaires:

  - Social Communication Questionnaire (Berument et al., 1999; Rutter et al., 2003)

  - Social Responsiveness Scale (Constantino et al., 2003)

Diagnostic tools

- Use of a structured diagnostic instrument recommended

But result may depend on

- environment
- Presence of other mental illness
- Experience and training of clinicians using tools
- Time to complete tools
- Are they valid in LD population and non western populations?

Autism Diagnostic Interview (ADI-r)

- Standardised, semi-structured interview, undertaken with an informant
- Assessment of presence/absence of behaviours associated with ASD, and the degree of impairment
- Focuses on two time frames: childhood (aged four-five years) and current behaviour

Autism Diagnostic Interview (ADI-r)

- Five main sections relating to:

  1. Early development
  2. Acquisition and loss of skills
  3. Qualitative impairments in communication
  4. Qualitative abnormalities in reciprocal social interaction
  5. Restricted, repetitive and stereotyped patterns of behaviour

Autism Diagnostic Observation Schedule (ADOS)

- Semi-structured assessment, administrator-rated
- Provides opportunities to elicit spontaneous behaviours in standardised contexts
- Four ADOS modules: choice dependent on language ability
- ADOS (module 4)
  - Low sensitivity in high performing adults
  - Low specificity in low IQ adults
  - Less likely to capture repetitive behaviours or intense preoccupations

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Diagnostic Interview Guide for the Assessment of Adults with Autism Spectrum Disorder (ASD)

ubject's name:	NHS number:	Interviewers name:
ubject's date of birth:	Subject's age:	Date of Interview:

Names of informants (and their relationship with the subject):

Designed and produced by the Royal College of Psychiatrists' Education and Training Centre

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Practical aspects of assessment

- Involvement of MDT
- How to arrange the assessment
- Prepare the individual for the process
- Will they be given a diagnosis on the day
- Keep questions simple
- Avoid leading questions
- Probe ‘yes’ answers
- Examples of behaviour
- Evaluation in different environments
- Is impairment due to ASD or co-occurring disorder?
- Consider cognitive, language and neuropsychological assessment

Equivocal findings ...

- On the balance of probabilities or beyond reasonable doubt?
- ASD = a spectrum of severity
- Sometimes it isn’t possible to reach a definitive conclusion

Differential diagnoses to consider

- Hearing or visual problems
- Intellectual disability
- Other neurodevelopmental disorders

In adults:

- Schizophrenia
- Anankastic PD
- OCD
- Anxiety
- Mood disorder

Points on co-occurring disorders and diagnosis

- ADHD
- Trauma
- Eating disorder
- Depression and suicidality

ASD diagnosis in COVID-19

- The impact on changes to environment and routines
- Reduced social interaction
- Virtual consultation
- mental health impact

In summary

- How we diagnose autism is changing
- Autism is a spectrum disorder and the presentation is highly heterogeneous
- Screening and diagnostic tools are important in diagnosis
- Clinical judgment vital

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