AUTISM SPECTRUM DISORDER

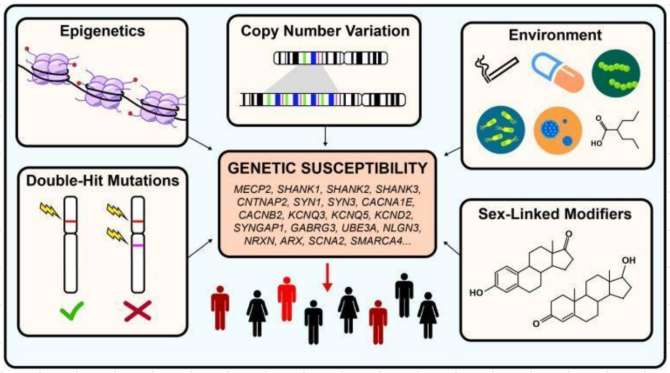
DEFINITION

Autism Spectrum Disorder is a collective term for a group of neurodevelopmental disorder characterised by **deficits** in **2 core domains:**

- Social communication and interaction (verbal and non-verbal)
- Restricted repetitive patterns of behaviour, interests and activities

EPIDEMIOLOGY

- One of the most prevalent neurodevelopmental disorders and increasing in incidence
  - ~1% of population estimated to have ASD
- Boys are 4x more likely than girls to be diagnosed
  - "**Masking**" phenomenon: girls have different presentations, tend to manage challenges differently, possess more capability for social mimicry leading to underdiagnosis



AETIOLOGY

Unknown, however complex interaction of likely genetic and environmental factors.

Risk Factors

- Prenatal exposure to toxins and infections
- Maternal use of sodium valproate in pregnancy
- Parental age
- Gestation age <35 weeks
- Parental psychosis or affective disorder
- Siblings with autism (pts who have a child with ASD have a 25% chance of having a second child who is also affected)
- Birth injuries or defects associated CNS dysfunction (e.g. cerebral palsy)
- Neonatal encephalopathy or epileptic encephalopathy
- Number of genetic syndromes can have an ASD component to their phenotype
  - *e.g. Tuberous sclerosis, Rett syndrome, Fragile X*

Common Misconceptions of What Causes Autism

- Vaccination
- Medical Treatment
- Parenting ("refrigerator mothers")
- Social circumstances

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CLINICAL FEATURES

Presentation may be **greatly impacted by comorbid mental and physical health conditions, as well as by other individual factors such as sex or gender identity, language and cognitive ability**. 31% of individuals with ASD may have some level of intellectual disability. Comorbidity of ADHD, anxiety disorders is high. Children with ASD have a higher incidence of epilepsy, chronic sleep problems and GIT disorders. Comorbid conditions may also overshadow a diagnosis of ASD.

Sensory Issues

- Over or under-sensitivities to touch, sounds, lights, tastes, smells, pain, other stimuli, etc.

Social Communication Challenges

May not understand or appropriately use...

- Spoken language (around 1/3 of people with autism are non-verbal)
- Gestures and body/ other non-verbal language (hand gestures, often don't point at things to share their interest etc.)
- Eye contact
- Facial expressions
- Tone of voice
- Expressions not meant to be taken literally (e.g. jokes, sarcasm)
- May exhibit 'stilted speech'

Social Challenges

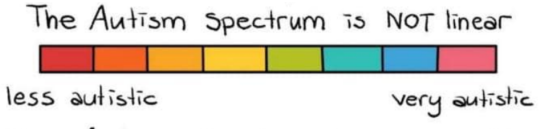
- Difficulty with theory of mind (thinking of/realising that other people have their own thoughts and feelings)
- Recognising emotions and intentions in others
- Recognising one's own emotions (interoception)
- Seeking emotional comfort from others
- Feeling overwhelmed in social situations
    - Expectations, routines in interactions are very important and interactions outside these familiar routines cause huge anxiety, threshold of being overwhelmed is very low
- Taking turns in conversation (reciprocal)
- Gauging personal space (appropriate distance between people)
- Variability as to whether they prefer to be isolated (preference for 1/2 of people)

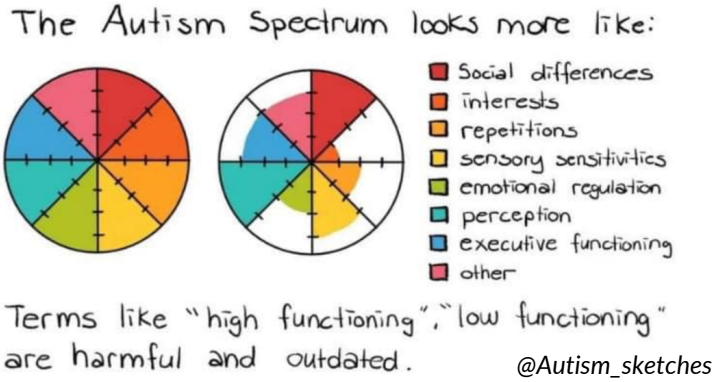
Restricted & Repetitive Behaviours

- Body movements (rocking, flapping, spinning, running back and forth) and repetitive motions with objects (known colloquially as "stimming"
    - Often due to excess stimulus, find it soothing or stimulating
- Staring at lights or spinning objects
- Ritualistic behaviours (lining up objects, repeatedly touching objects in set order)
    - Overlaps in OCD
- Echolalia
- Narrow or extreme interests in specific topic
- Need for unvarying routine/ resistance to change
    - *E.g. Same daily schedule, meal menu, clothes, route to school*
    - Individuals may present with avoidant-restrictive food intake disorder

Other

- Fine or gross motor difficulties





ASSESSMENT

- Antenatal and perinatal history
- Comprehensive developmental history (obtain collateral history to affirm presence or absence of concerns across multiple settings e.g. home, school)
- Past medical history and medical work-up (iron, Vit D and other micronutrient deficiencies)
- Family history
- Evaluate family function and psychosocial history (parental mental health, substance use, parental disability, parental relationship issues, child protection involvement)
- Hearing and vision (especially in a child who has delay in developmental milestones for speech)
- Genetic screening
- For diagnosis refer to specialists - assessment is usually made by a team of professionals: paediatricians, clinical psychologists/ neuropsychologist, speech pathologists and psychiatrists

KEY DSM-V CRITERIA

A. **Persistent deficits** in **social communication** and social **interaction** across multiple contexts

- Deficits in social-emotional reciprocity
- Deficits in nonverbal communicative behaviours
- Deficits in developing, maintaining and understanding relationships

B. **Restricted and repetitive patterns** of behaviour, interests, activities in **≥2 of the following** domains

- Stereotyped or repetitive motor movements
- Insistence on sameness, inflexible adherence to routines or ritualised patterns of verbal or non-verbal behaviour
- Highly restricted, fixated interests that are abnormal in intensity or focus
- Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of the environment

C. Symptoms must be present in early childhood (however may not fully manifest under social demands exceed individual capacity, or may be masked by learned strategies or other compensatory behaviour)

D. Symptoms cause impairment in social, occupation or other areas of functioning

E. Not better explained by intellectual disability, global developmental delay or other disorder.

DIFFERENTIAL DX

- Intellectual disability
- Language or other communication disorders
- Selective mutism
- Stereotypic movement disorder
- ADHD
- Schizophrenia
- Schizoid personality disorder
- Attachment disorders

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MANAGEMENT

PRINCIPLES OF MANAGEMENT

- **Early intervention** (6 and under) affords the best opportunity to support healthy development and deliver benefits across lifespan
- Evaluate function on an individual basis and support the needs of the individual and their caregivers
  - Comprehensive Needs Assessment aims to explore the question: what are the key **strengths, challenges** and **needs** that inform future clinical management and service delivery?
- **Multidisciplinary care** especially where there may be complex support needs
  - Speech pathology
  - Psychology
  - Occupational and behavioural therapy
  - Developmental paediatricians
  - Child psychiatrists
  - Dietician
  - GP
  - Social worker
  - Specialist teachers
- **Consider how to communicate with individuals with ASD**
  - Simplify language
  - Communicate one ideate a time
  - Avoid ambiguous language
  - Consider most appropriate communication style (e.g. written, verbal) or providing a social script or story, using assistive communication devices

BIOLOGICAL

- Prevent, monitor for and treat physical health comorbidities (e.g. commonly bowel disorders, sleep disorders, epilepsy, T1DM)
- Pharmacological interventions such as psychotropic medications may have a role in managing challenging behaviour or comorbid psychiatric conditions

PSYCHOLOGICAL

- Management of challenging behaviours (e.g. aggression, self-injury, PICA) requires
  - understanding behavioural presentations may be triggered by new onset or exacerbations of mental disorders, altered sensory processing, pain, physical health comorbidities or other environmental factors
  - Assessment and management by behavioural support specialists and psychiatrists
- Behavioural and developmental intervention programs provide therapy and specialised support and may help to improve social and adaptive functioning
  - *E.g. ABA, TEACCH, Early Start Denver Model, LEAP*
    - Note: may be appropriate however be aware that some are controversial and therefore not all are always appropriate or readily acceptable to individuals with ASD
- Other therapy-based interventions i.e. art or music therapy, speech and occupational therapy
- Family-based interventions
- Optimise sensory environment
- Provide routines
- Provide augmented or alternative communication aids or assistive technology devices (e.g. Picture Exchange Communication System)

SOCIAL

- Provide psychoeducation to parents, carers and individuals
- Apply for access to NDIS funding and support pathway (Early Child Early Intervention)
- Social worker
- Provide supports in relation employment and training
- Liaise with schools or other educational services to ensure young people with ASD are supported at school
- Provide wellbeing and mental health support for carers
  - Parent and carer support groups
  - Role of support services such as respite care

KEY DSM-V CRITERIA

Note: While autism is most well-known in media for its association with savant-like abilities (e.g. Shaun in The Good Doctor, Raymond from Rain Man), this may be damaging or misleading where this is not an accurate portrayal of a non-neurotypical individual or an individual on the autism spectrum.

Media with positive representation of individuals with ASD:

- Love on the Spectrum
- Leave it to Geege
- Atypical
- People Like Me
- Richard and Jaco: Life with Autism
- The Reason I Jump
- I am Greta
- Sesame Street (Julia)

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