

INTELLECTUAL DISABILITY

DEFINITION

Intellectual disability is characterised by significant limitations in both **intellectual functioning** and in **adaptive behaviour**, which covers many everyday social and practical skills. This disability originates **before the age of 18**.

EPIDEMIOLOGY

- 1-3% of the population have an intellectual disability
- Affects males to females 5:1 due to X-linked single gene mutations
- **50% of individuals with intellectual disability will experience major psychopathology**

AETIOLOGY

Genetic

- Chromosomal abnormalities (4-28%)
 - Abnormalities of chromosome number
 - Deletion or duplication
- Single gene disorders (2-9%)
- Inherited causes
 - Autosomal dominant: e.g. tuberous sclerosis
 - Autosomal recessive: e.g. phenylketonuria
 - X-linked recessive inheritance: e.g. Fragile X

Non-Genetic

- Structural CNS (7-17%)
- Complications of prematurity (2-10%)
- Endocrine/ metabolic (1-5%)
- Infections
 - Meningitis and encephalitis in early infancy/childhood
 - Neonatal/congenital infections with CMV, toxoplasmosis, rubella
- Traumatic brain injury
- Chemicals and toxins
 - Radiotherapy, chemotherapy, intrathecal corticosteroids
 - Foetal alcohol spectrum disorder

Unknown (30-50%)

ASSESSMENT

Most patients lack specific findings on history or examination. Suspect a genetic cause in individuals with developmental delay + dysmorphic features, autistic features, epilepsy, congenital anomalies or a family history.

HISTORY

- **Detailed birth & antenatal history**

- Antenatal
 - Previous obstetric history - spontaneous abortions, stillbirths, neonatal deaths
 - Antepartum bleeding - placental abruption, placenta praevia
 - Infections - rubella status, rash and fever during pregnancy, nonspecific illness, known exposures
 - Drug ingestion - medications (valproate), alcohol, nicotine, illicit drugs
 - Toxins - Lead, methylmercury
 - Labour and delivery - Gestation, presentation, mode of delivery, obstetric complications and management
- Neonatal
 - APGAR scores, resuscitation required, anthropometry, congenital anomalies, health in the neonatal period
 - Jaundice - degree of jaundice, Tx used
 - Infection - site of infection, Abx used
 - Feeding difficulties - duration, management
- Infancy
 - Feeding- feeding difficulties, muscle tone
 - Behaviour - irritability, sleep patterns
- Childhood and Adolescence
 - Behaviour - irritability, sleep patterns, eating
 - Play - social interests and skills, make-believe play, range of interests and activities
 - Developmental milestones - smiling, sitting, walking, first words, attending school, puberty
 - Schooling - attendance, type of school, academic achievements and concerns
 - General - parental concerns regarding development, hearing, vision, serious head injury, neurological problems

- **Family history**

- **Parent's education**

- Family history of genetic/ hereditary, psychiatric or developmental conditions

- **Review psychosocial factors**

- **Assess individual ability** in education, self-care, language, social, motor, sensory, behaviour

EXAMINATION

Assess for signs that might suggest an underlying cause of disability, conditions that may contribute to a poorer outcome, conditions that might arise from disability, developmental skills and difficulties.

General Examinations

- Height, weight, BMI, head circumference
- Skin changes - pale or pigmented patches, neurocutaneous stigmata
- Dysmorphic features - face, hands, etc.
- Ears and eyes - visual acuity, hearing
- Heart, lungs, blood pressure
- Abdomen including pubertal development
- Neurological function -posture abnormalities of gait and cranial nerves

ASSESSMENT CONT'D

GENERAL INVESTIGATIONS

- Genetic testing + refer to genetic counselling
 - Chromosome microarray first-line
 - Fragile X DNA testing should be ordered alongside as CMA does not detect gene variants causing Fragile X
- Metabolic screening
 - Thyroid function, CK concentration, urine metabolic screen
- EEG
- Neuroimaging
 - Consider especially in patients with micro or macrocephaly, neurologic signs and abnormal cranial contour

Consider specialist referral for other developmental, adaptive, cognitive or behavioural assessments.

KEY DSM-V CRITERIA

A. **Deficits in intellectual functions**, such as reasoning, problem solving, planning, abstract thinking, judgement, academic learning, and learning from experience, **confirmed by both clinical assessment and individualised, standardised 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 >/1 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.

DIFFERENTIAL DX

- Communication disorders
- Specific learning disorders
- Autism spectrum disorder



MANAGEMENT

PRINCIPLES OF MANAGEMENT

- Understanding behavioural presentations is very important, especially where communication is a barrier. Diagnostic overshadowing significantly contributes to poorer physical and mental health outcomes for individuals with disability.
- Mental health disorders are common and may present in an atypical manner.
- An integrated care model and a multidisciplinary team that ensures continuity of care and support throughout the lifespan.
 - Speech pathology
 - Psychology
 - Occupational and behavioural therapy
 - Developmental paediatricians
 - Psychiatrists
 - Dietician
 - Physiotherapist
 - Exercise physiologist
 - GP
 - Social worker
 - Specialist teachers
- An individualised and strengths-based approach that builds resilience and strength, and minimises the effects of impairments and disabilities on function.
- Provide support that increases protective factors, such as connection and family.
- Provide opportunistic and preventative healthcare.
- Reduce risk of iatrogenic trauma and stress and distress associated with medical care.

BIOLOGICAL

- Annual comprehensive health assessments
 - Oral health
 - Nutrition risk
- Manage common health problems: vision, hearing, constipation, epilepsy, thyroid disease, GORD, H. pylori infection, osteopenia/osteoporosis
- Medication review 3-6 monthly especially in setting of polypharmacy
- Immunisations
- Encourage physical activity
- Mitigate health risks associated with behaviour
 - If unable to make a diagnosis of mental illness (medical, psychiatric and social causes have been excluded) and the behaviour is of concern (places the person or others at risk or negatively impacts on the person's quality of life) targeted treatment of behaviour with psychotropics may be acceptable
 - This is known as chemical restraint and has legal implications regarding authorisation and regular r/v by the DHHS.

PSYCHOSOCIAL

- Behaviour changes
- Look for underlying physical health problems or external factors (e.g. change of support staff, loss of family or friends)
- Refer for assessment with psychologist or psychiatrist
- Financial assistance (Centrelink or NDIS)
- Special Equipment and aids
- Community Access
- Respite care
- Disability support workers
- Social workers
- Consumer and parent support groups
- Improve communication and use alternative communication aids or assistive technology
- Provide support for academic learning and vocational training
- Teach basic interpersonal skills
- Provide routine and supportive physical and sensory environment
- Break down tasks for daily care needs (e.g. dressing, toileting)

MANAGEMENT CONT'D

INPATIENT CARE - preventing/minimising agitation/aggression

(e.g. when admitted for a medical problem)

- Written inpatient care plan
- Maintain a low stimulus environment
- Plan medical or surgical procedures carefully ahead of time
- Avoid unnecessary examinations and interventions (vital signs, blood draws)
- Liberalise diet (allow food from home)
- Explain procedures in a developmentally appropriate way
- Use specific strategies during procedures
- Keep stay as short as possible

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