MANAGEMENT OF INTELLECTUAL DISABILITY [ID]

LECTURE

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OBJECTIVES

- Review ID — definition, causes, presentation, comorbidity
- Describe ID clinical assessment and diagnosis
- Outline strategies to treat and prevent intellectual disability [ID]

YOU NEED TO KNOW HOW TO MANAGE INTELLECTUAL DISABILITY BECAUSE:

- Intellectual disability (ID):
    - very common - general population prevalence of ~ 1%
    - preventable
    - pose a huge burden
    - lead to stigmatization
- ID poses risks to those with ID:
    - harmful forms of traditional healing
    - neglect or harsh treatment
- High caregiver stress

DEFINITIONS

- ❑ WHAT IS INTELLIGENCE?
- ❑ WHAT IS INTELLIGENT QUOTIENT [IQ]?
- ❑ WHAT IS ADAPTIVE BEHAVIOR?
- ❑ WHAT IS INTELLECTUAL DISABILITY [ID]?

What is intelligence?

- Intelligence refers to general mental ability such as learning efficiently, learning from experience, understanding complex concepts, ability to solve problems and so on.

What is intelligent quotient [IQ]?

- Intellectual functioning is represented by Intelligence Quotient [IQ] scores, that are obtained from standardized intelligence tests. The intelligence measure IQ is a fraction, which is one's mental age over their chronological age times 100.

$$IQ = \frac{\text{Mental Age (MA)}}{\text{Chronological Age (CA)}} \times 100$$

That is how you get an IQ.

- If MA equals CA, then ratio equals one and IQ is"100" - does things same as other people their age. If mental age is higher than chronological age then of course IQ becomes above "100". If mental age is lower than chronological age then IQ becomes lower than 100 and if mental age is lower that means mentally one is not doing all the things that same chronological aged are doing.
- Intellectual disability [ID] present if an individual scores 70 or below score on IQ tests.

What is adaptive behavior?

Adaptive functioning is one's ability to cope with day-to-day tasks. DSM-5 3 dimensions of adaptive skills are:

1) Conceptual skills – language and literacy, time and number concepts, self-direction.
2) Social skills – Interpersonal skills, social responsibility, self esteem, gullibility, social problem solving and ability to follow rules and to avoid being victimized.
3) Practical skills – personal care/activities of daily living, health care, occupational skills, travel, schedules, safety, use of money, use of the telephone.

What is intellectual disability [ID]?

- D is limited mental abilities or arrested/incomplete development of mental abilities, during developmental period, resulting in impairing subnormal intelligence [as measured by IQ] and adaptive functioning deficits in conceptual, social, and practical domains which commence in early life and persist with development.
- **ID core symptoms:** [1] low intellectual functioning [IQ $\leq$70], [2] impaired adaptive behavior & [3] onset during developmental period.

Sociological definition of Intellectual Disability

•Constructive, as ID defined in terms of the support needs of an individual and the effect of ID seen as something that will vary and can be increased or decreased by external factors.

•This definition does not rely on the capacity of the person being set in stone, but also on the environment and the support that they receive.

•So, adjusting the environment and the support to meet the person's needs can increase the person's capacity and reduce the effect of ID!

•Unlike medically based 'cures', which focuses on the individual & their impairment, social model sees 'cure' to the problem of disability is in restructuring of society. This is an achievable goal and to the benefit of all.

DSM 5 DIAGNOSIS CRITERIA

ID classified under **neurodevelopmental disorders which must meet the below criterion:**

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.

- **Conceptual skills (communication, language, time, money, academic)**
- **Social skills (interpersonal skills, social responsibility, recreation, friendships)**
- **Practical skills (daily living skills, work, travel)**

C. Onset of intellectual and adaptive deficits during the developmental period.

Classification of intellectual disability, stratified by three age groups

	Age	
0 to 5 years *Maturation and development*	*6 to 20 years* *Training and education*	*21 years and older* *Social and vocational adequacy*
Degree: Mild General develop communicative and social skills. May not be distinguishable until beginning school.	Can learn up to 4th/5th primary school grade skills when reaching the ages of 18 or 19 years. Can be integrated into society.	Is capable of acquiring social and work skills for integration into the work force at minimum wage.
Degree: Moderate Can speak or learn to communicate. Some difficulties with motor skills.	Difficulty meeting 2nd primary school grade academic objectives.	May be able to partially maintain oneself economically in manual work under protected conditions
Degree: Severe Marked limitations in motor skills. Minimal language ability.	Can speak or learn to communicate. Can learn elemental self-care and health habits.	Can partially contribute to maintaining oneself economically under total supervision.
Degree: Profound Significant delay, minimal functional ability in sensorimotor areas. Needs basic care.		Some motor and language development. Can learn very limited personal care skills.

CLASSIFICATIONS OF INTELLECTUAL DISABILITY SEVERITY

- Wide range of intellectual functional impairments and difficulties with daily life skills in ID.
- Levels of severity of intellectual impairment & need for support are mild, moderate, severe, profound.
- 5 Dimensions - Intellectual aptitudes; Adaptation level [conceptual, practical, social]; Participation [interaction, social roles]; Health [physical health, mental health, etiology]; and the Social context [environment, culture, opportunities]

Severity Category	% of ID	DSM-IV just IQ	DSM-5 - based on daily skills	American Association Intellectual and Developmental Disabilities (AAIDD) -intensity of support needed
Mild	85%	IQ range 50–69	Can live independently with minimum levels of support.	Intermittent support needduring transitions or periods of uncertainty.
Moderate	10%	IQ range 36–49	Independent living may be achieved with moderate levels of support, such as those available in group homes.	Limited support needed in daily situations.
Severe	3.5%	IQ range 20–35	Requires daily assistance with self-care activities and safety supervision.	Extensive support needed for daily activities.
Profound	1.5%	IQ <20	Requires 24-hour care.	Pervasive support needed for every aspect of daily routines.

MAJOR INFLUENCES ON NEEDED SUPPORTS

DIMENSIONS OF SUPPORT NEEDS

Causes of Intellectual Disability

CAUSES

1. Chromosomal genetic factors
2. Hereditary Genetic conditions
3. Acquired – congenital and developmental
4. Environmental and sociocultural factors
5. Unknown

ETIOLOGICAL FACTORS IN INTELLECTUAL DISABILITY

Causes are heterogeneous. **Mild ID:** 40% no specific cause. **Marked ID**: specific cause > often

1. **Chromosomal genetic factors** - Down, Fragile X, Prader-Will, Rett & Lesch-Nyhan syndromes; Neurofibromatosis, TS
2. **Hereditary genetic factors** – PKU, Galactosemia, Mowat-Wilson syndrome, Tay-Sachs, microcephaly, hydrocephaly
3. **Acquired factors**

i. Congenital

a) Metabolic –neonatal hypothyroidism

b) Toxic – lead poisoning, fetal alcohol syndrome, prenatal exposure to substances

c) Maternal infections - toxoplasmosis, rubella, cytomegalovirus, herpes, syphilis [TORCHES]+ HIV

ii. Developmental

a) Prenatal period -toxemia, intrauterine malnutrition, placenta previa, umbilical cord prolapse

b) Perinatal period – prolonged fetal suffering with neonatal anoxia, asphyxia

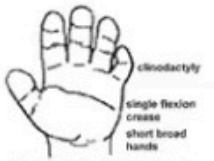
c) Postnatal period – kernicterus, head injury, lead poisoning, carbon monoxide poisoning, infections – encephalitis, meningitis, cerebral malaria

4. **Environmental and sociocultural factors** –poverty, living in adverse economic conditions, poor nutritional and medical care, environment that offers few opportunities for learning

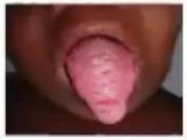
5. None of the above - 2/3 of all ID, the cause is unknown

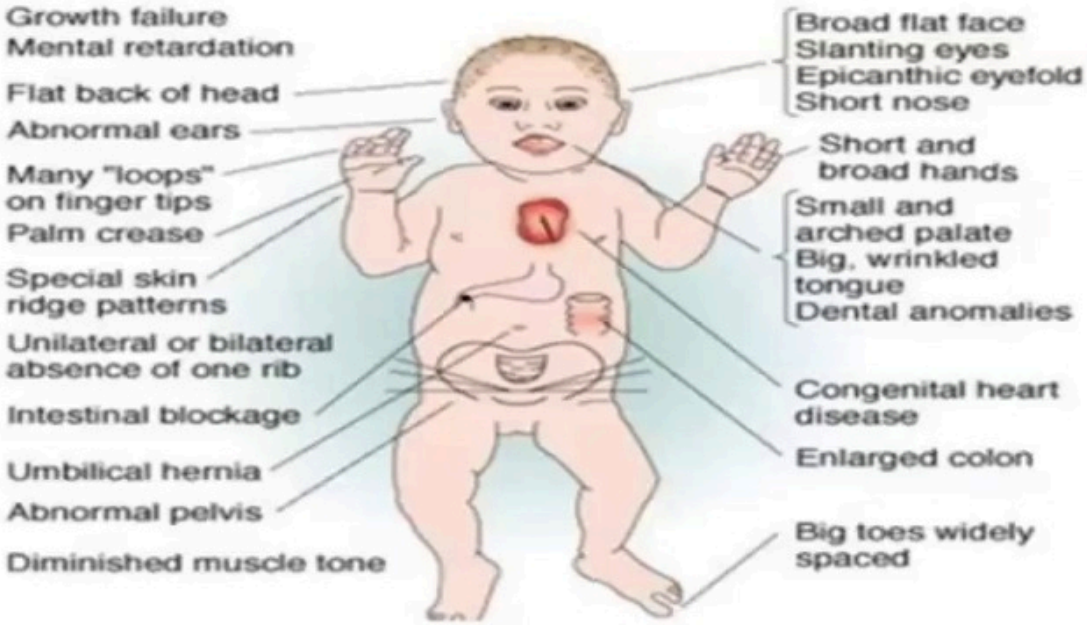
Down syndrome (trisomy21), 1:1000, commonest identifiable cause of ID.

- DS characteristic features - up-slanting eyes, single horizontal palmar crease, flat short nose, short stature, short neck, prominent epicanthic skin folds, small, low-set ears, protruding tongue, small hands and feet, loose joints, weak muscle tone; relative strengths visual processing, musical, social interaction and relative weakness in langauage expression and pronunciation

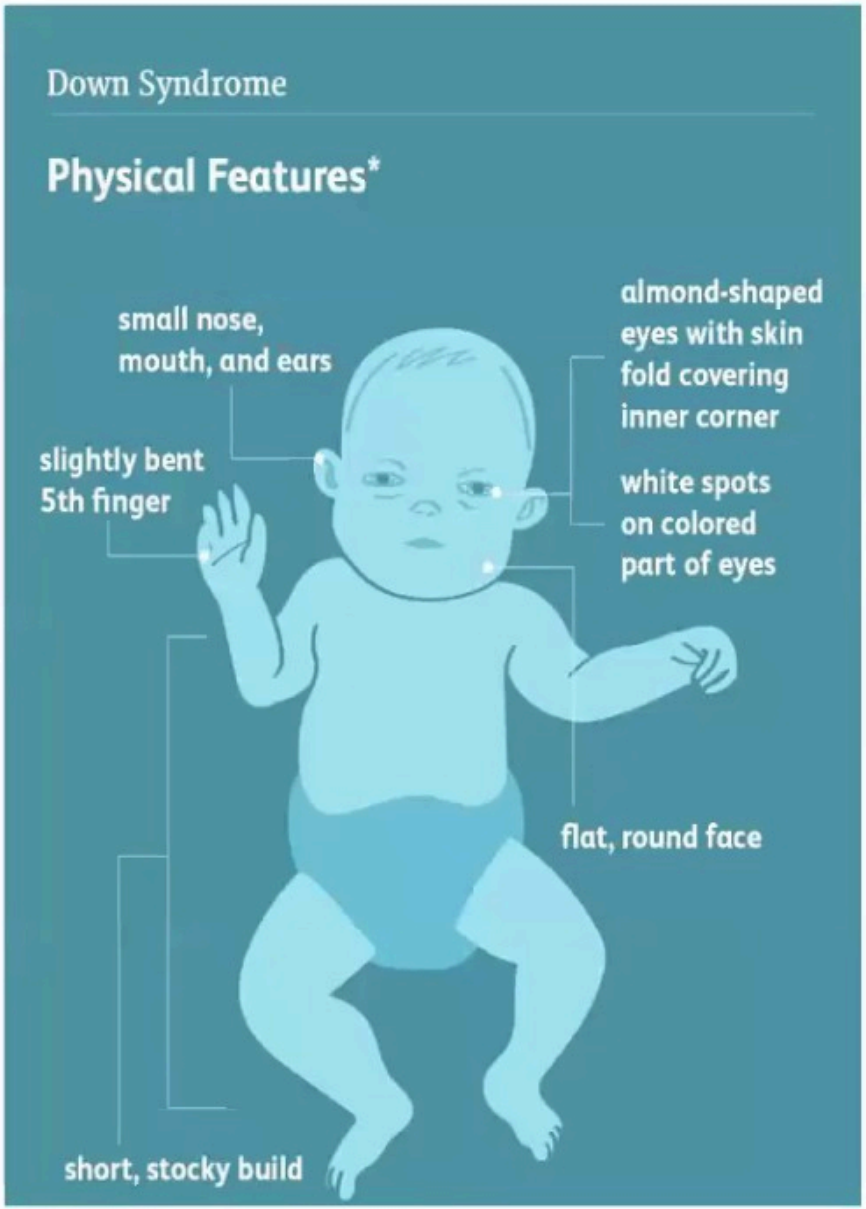


Trisomy 21 - Hand Features





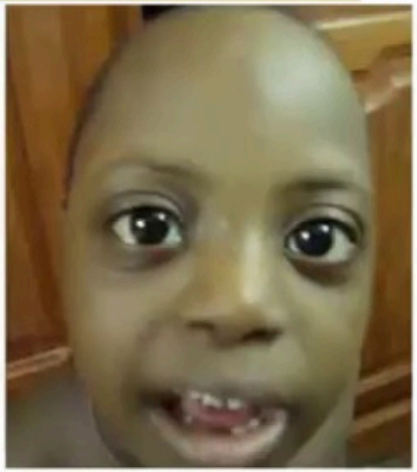
Characteristics (%)		Characteristicss(%)	
Mental retardation	100	Clinodactyly	52
Stunted growth	100	Umbilical hernia	51
Short stature	100	Short neck	50
Flexible ligaments	80	Shortened hands	50
Hypotonia	80	Cong. heart disease	45
Brachycephaly	75		
Short extremities	70	Simian crease	45
Low set/round ears	60	Macroglassia	43
Flattened nose	60	Epicanthic fold	42
Small teeth	60	Strabismus	40
		Brushfield spots	35



Down Syndrome Facial dismorphism with

- ❑coarse face
- ❑hypertelorism
- ❑big nose
- ❑relative midline dimple and
- ❑single transvers palmar crease

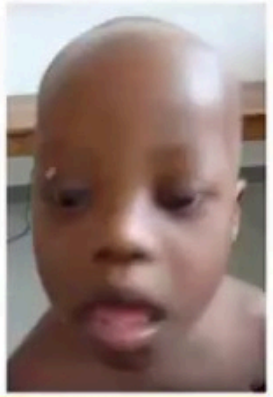
Down Syndrome



- ❑DS facial features in
- ❑6 years old boy with
- ❑strabismus and
- ❑exophthalmos

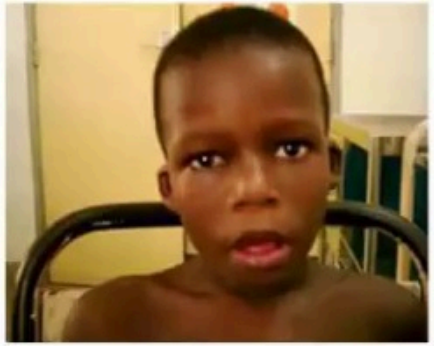
Down Syndrome

- Down Syndrome facial features
- in a 5 years old girl with
- strabismus and
- early teeth renewal

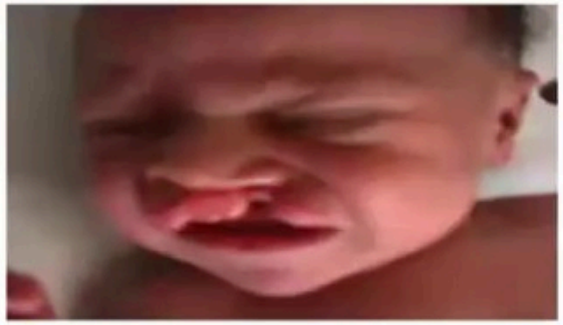


Down syndrome facial features in 2-year-old-boy

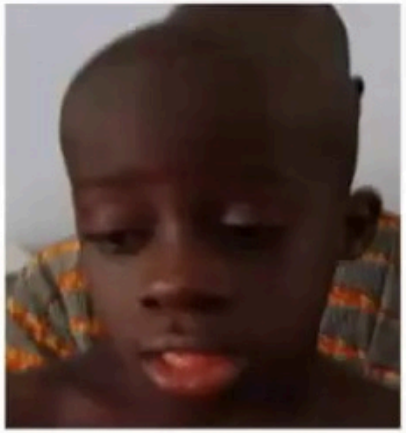
Di George syndrome



- Facial dismorphism with
- bulbing nasal tip,
- long philtrum and
- microstomia

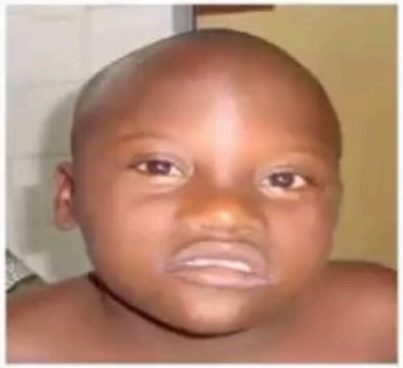


Birth defect with lateral clefting



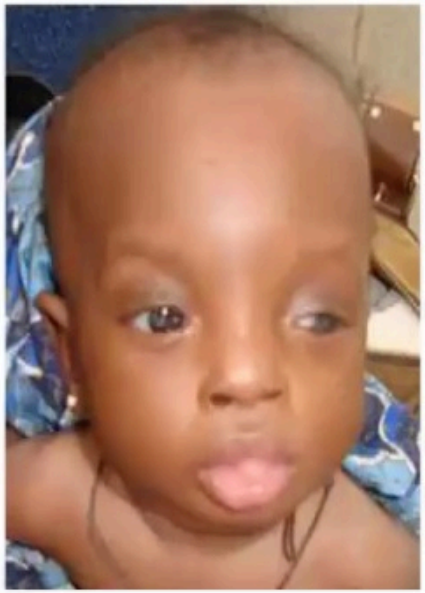
Cranial facial dismorphism with

- ❑Brachycephaly
- ❑Triangular face, large pinae
- ❑Broad nose
- ❑Relative macrostomia and
- ❑Lingual protruding



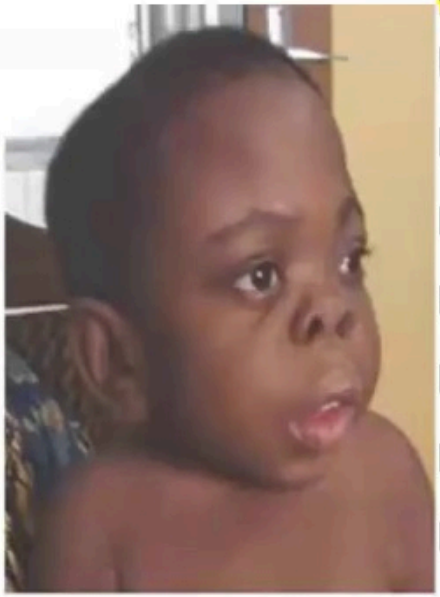
Cranial facial dismorphism with

- ❑microcephaly
- ❑Hypertelorism
- ❑upslanted palpebrate fissures
- ❑large pinnae and
- ❑light facial asymmetry



Facial dismorphism with

- ❑microcephaly,
- ❑strabismus,
- ❑broad nasal bridge,
- ❑anteverted nostril,
- ❑short/large philtrum and
- ❑lingual protruding



Macrocephaly and coarse face with

- ❑Large head
- ❑Bulging frontal bones
- ❑Depressed nasal bridge
- ❑Broad nasal tip, a
- ❑Anteverted nostrils
- ❑Very large ears and
- ❑Enlarged cheeks

Fragile X syndrome (FXS): 1:2000-5000

➡most common form of inherited/genetically acquired ID

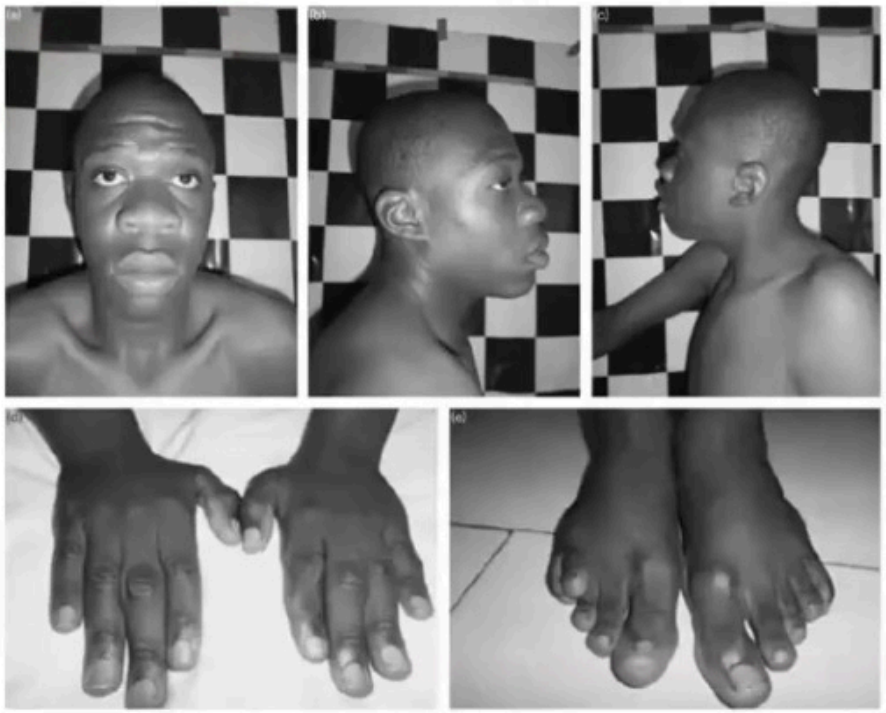
➡Characteristics: broad forehead, elongated face, high arched palate, large ears, hypotonia.

➡ adaptive/intellectual functioning deficits increase with age

➡inceases risk for autism [20-60%], ADHD, social phobia

➡relative strengths in verbal long-term memory

➡relative weakness in short-term memory, math



Congenital hypothyroidism (1:2000-4000)

On the right, a 17-year-old girl with a height of 100 cm,

★severe intellectual disability, marked delayed puberty,

★myxedema, flat, broad nose, hypoplastic mandibule,

★dry, scaly skin, dry, brittle hair, prominent abdomen.

★pseudomuscular hypertrophy, muscular weakness,

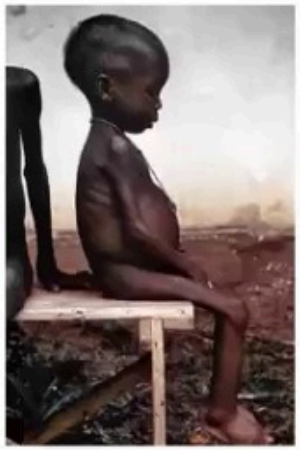
★flat feet, and genu valgum are present;

➡Thyroid gland was not palpable. Her serum level of

➡thyrotropin 288 μU/mL,

➡ thyroxine 0.1 μldl (1.29), and

➡triiodothyronine 10 ngldl (0.154 nM)

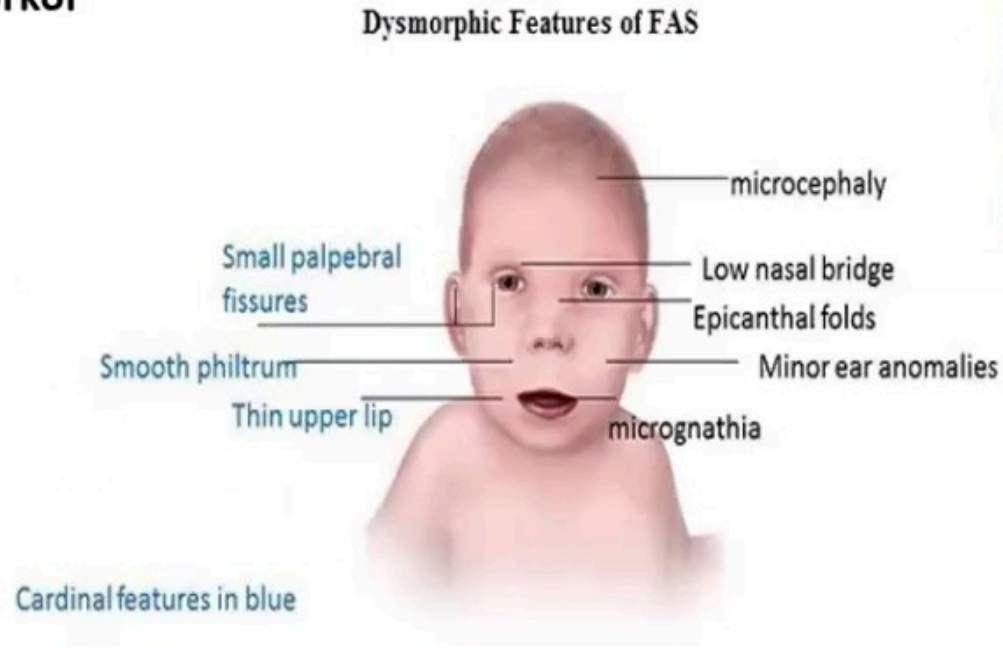


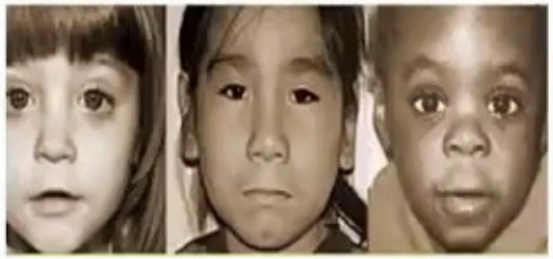
Kwashiorkor

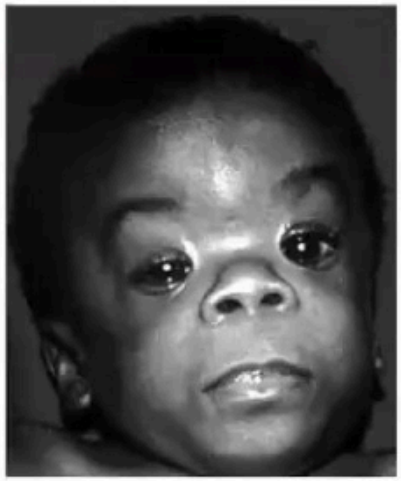
Fetal alcohol syndrome [FAS] preventable ID

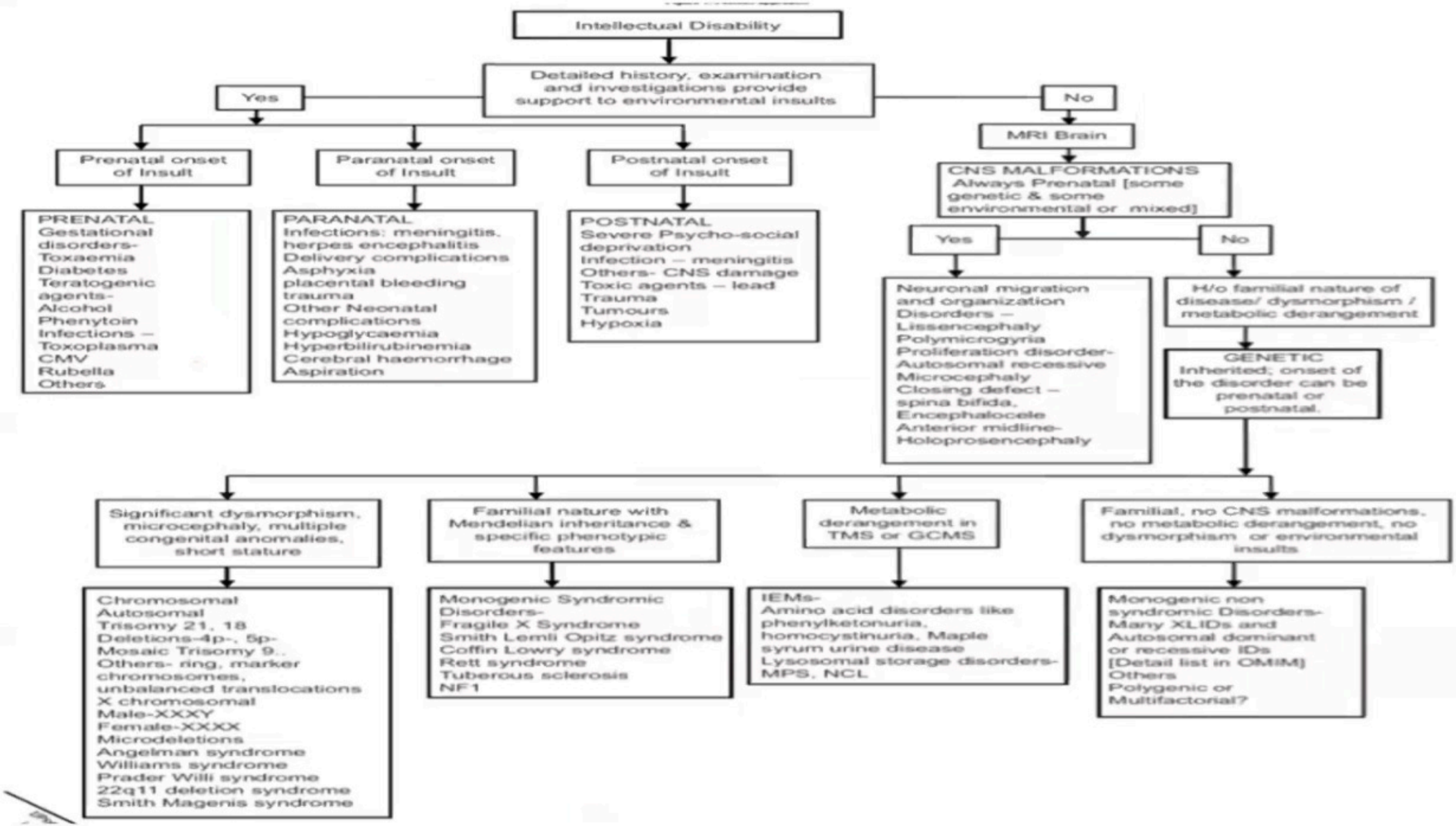
➡FAS is on the most severe end of the FASD spectrum. It describes people with the greatest alcohol effects, causing signs so distinct that the diagnosis is based on special measurements and findings in each of the 3 following area:

1. 3 specific facial abnormalities: smooth philtrum (the area between nose and upper lip), thin upper lip, small palpebral fissures (the horizontal eye openings)
2. Growth deficit (lower than average height, weight or both)
3. Central nervous system (CNS) abnormalities (structural, neurologic, functional, or a combination of these)








Intellectual Disability
Detailed history, examination and investigations provide support to environmental insults
Yes
No
Prenatal onset of Insult
Paranatal onset of Insult
Postnatal onset of Insult
PRENATAL
Gestational disorders-
Toxaemia
Diabetes
Teratogenic agents-
Alcohol
Phenytoin
Infections –
Toxoplasma
CMV
Rubella
Others
PARANATAL
Infections: meningitis, herpes encephalitis
Delivery complications
Asphyxia
placental bleeding
trauma
Other Neonatal complications
Hypoglycaemia
Hyperbilirubinemia
Cerebral haemorrhage
Aspiration
POSTNATAL
Severe Psycho-social deprivation
Infection – meningitis
Others- CNS damage
Toxic agents – lead
Trauma
Tumours
Hypoxia
MRI Brain
CNS MALFORMATIONS
Always Prenatal [some genetic & some environmental or mixed]
Yes
No
Neuronal migration and organization Disorders –
Lissencephaly
Polymicrogyria
Proliferation disorder-
Autosomal recessive
Microcephaly
Closing defect –
spina bifida,
Encephalocele
Anterior midline-
Holoprosencephaly
H/o familial nature of disease/ dysmorphism / metabolic derangement
GENETIC
Inherited; onset of the disorder can be prenatal or postnatal.
Significant dysmorphism, microcephaly, multiple congenital anomalies, short stature
Familial nature with Mendelian inheritance & specific phenotypic features
Metabolic derangement in TMS or GCMS
Familial, no CNS malformations, no metabolic derangement, no dysmorphism or environmental insults
Chromosomal
Autosomal
Trisomy 21, 18
Deletions-4p-, 5p-
Mosaic Trisomy 9..
Others- ring, marker chromosomes, unbalanced translocations
X chromosomal
Male-XXXY
Female-XXXX
Microdeletions
Angelman syndrome
Williams syndrome
Prader Willi syndrome
22q11 deletion syndrome
Smith Magenis syndrome
Monogenic Syndromic Disorders-
Fragile X Syndrome
Smith Lemli Opitz syndrome
Coffin Lowry syndrome
Rett syndrome
Tuberous sclerosis
NF1
IEMs-
Amino acid disorders like phenylketonuria, homocystinuria, Maple syrum urine disease
Lysosomal storage disorders-
MPS, NCL
Monogenic non syndromic Disorders-
Many XLIDs and Autosomal dominant or recessive IDs [Detail list in OMIM]
Others
Polygenic or Multifactorial?

Intellectual Disability Clinical Presentation

ID: Clinical manifestations

- **ID manifestations** mainly developmental delay in intellectual functioning and social adaptive skills deficits arising before age 18. Children with ID progress with normal milestones in similar pattern to normal but the rate of developmental progress is slower and there is a ceiling on ultimate achievement. Abilities of children with ID significantly lower than children of same age and their adaptability in daily life is adversely affected.
- **ID presenting symptoms/signs** typically include delays in cognitive, language and adaptive skills. The developmental delays vary depending on level of ID and etiology, eg, in mild non-syndromic ID, delays may not be notable until preschool years, whereas with severe/profound ID associated with syndromes or extreme prematurity, significant delays in milestones may be noted from birth.
- Language delay: One early sign of ID may be language delays and parents c/o child may be deaf?
- Fine motor/adaptive delay: Significant delays in self-feeding, toileting, dressing typically reported in children with ID. Also prolonged, messy finger feeding & drooling oral-motor incoordination.
- Cognitive delay: Early preacademic difficulties following directions, esp. multipart directions.
- Behavioral disturbances: eg, aggression, self-injury, defiance, hyperactivity and sleep disturbances.
- Neurologic and physical abnormalities: increased rates of seizure disorders, microcephaly, macrocephaly
- ***ID severity levels*** as per severity of deficits in adaptive function, IQ and support need - [1]. mild;[2]. moderate;[3]. severe and [4]. profound ID severity levels.

CLASSIFICATION OF INTELLECTUAL DISABILITY, STRATIFIED BY THREE AGE GROUPS

	Age	
0 to 5 years *Maturation and development*	*6 to 20 years* *Training and education*	*21 years and older* *Social and vocational adequacy*
Degree: Mild General develop communicative and social skills. May not be distinguishable until beginning school.	Can learn up to 4th/5th primary school grade skills when reaching the ages of 18 or 19 years. Can be integrated into society.	Is capable of acquiring social and work skills for integration into the work force at minimum wage.
Degree: Moderate Can speak or learn to communicate. Some difficulties with motor skills.	Difficulty meeting 2nd primary school grade academic objectives.	May be able to partially maintain oneself economically in manual work under protected conditions
Degree: Severe Marked limitations in motor skills. Minimal language ability.	Can speak or learn to communicate. Can learn elemental self-care and health habits.	Can partially contribute to maintaining oneself economically under total supervision.
Degree: Profound Significant delay, minimal functional ability in sensorimotor areas. Needs basic care.		Some motor and language development. Can learn very limited personal care skills.

ID presents differently in the various developmental periods:

❑***Infancy***: Mild and moderate ID may go undetected in infancy, otherwise, for severe ID, most children with ID have physical malformations that identify them at birth as being at high risk for ID and these infants may show significantly low intellectual functioning, characterized by significant limitations in emotional expressiveness, language, purposeful behavior, gross and fine motor skills and apparent cognitive abilities.

❑***Early Childhood:*** Young children with ID may be identified by parents or pediatricians after failure to meet developmental milestones in several functional areas such as significant delays in walking, speech, social skills or self-care skills capacity-self feeding, toilet learning; or on scoring <70 on standardized IQ tests.

❑***Middle Childhood/Adolescence:*** Developmental delays often clear by this age. Evaluation further identify strengths and weaknesses in cognitive and adaptive abilities.

❑**Adults** -ID affects the person's ability to adapt to new or unfamiliar situations and it can involve the person presenting well " "cloak of competence".

❑**Legal rights** - since persons are not criminally responsible when they are chronologically less than 12 yrs, the same ought to be extended to those with MA of less than 12 yrs.

ID: Psychiatric and Medical Co-morbidity

Compared with normal children, children with ID higher risk of other health problems. Co-morbid mental, neurodevelopmental, medical| physical conditions frequent.

•Medical co-morbidity common-3 to 4 times higher than general population; often undetected: epilepsy (22%), cerebral palsy (20%), anxiety disorders (17%), ODD (12%) and autistic disorder (10%).

•Visual and hearing problems present in ~ 5%-10% of persons with ID.

•Challenging behavior: Difficulties accepting criticism, aggression or self-injury common.

•**Diagnosing psychiatric disorders** in ID difficult, yet, ~ 40-70% have psychiatric disorders eg,

➢ ADHD, ODD, depression, bipolar disorders, anxiety disorders, conduct problems, autism, stereotypic movement disorder [with or without self- injurious behavior] and major neurocognitive disorder.

MENTAL ILLNESS IN PERSONS WITH ID

MENTAL ILLNESS IN PERSONS WITH ID: Though ID – not a psychiatric illness, per se, ID associated with 10% to 39% psychiatric morbidity rate. [Deb et al, 2001]. ID/psychiatric morbidity risk factors include:

BIOLOGICAL FACTORS

- Genetic liability
- Brain frontal lobe structural abnormality can cause apathy, social withdrawal and disinhibition.
- Interaction between environment & physical disabilities, may indirectly cause psychopathology.
- Epilepsy occurs in 14–24% people with ID, and could predispose them to psychopathology.
- Thyroid disorder could predispose to PM.
- Prescribed and non-prescribed drugs can cause PM.

PSYCHOLOGICAL FACTORS

- Low intelligence/ Inability to solve problems
- Temporal lobes dysfunction/Impaired memory
- Frontal lobes damage/judgement & apathy issues
- Lower thresholds for stress tolerance
- Poor self-image

PSYCHOLOGICAL FACTORS cont'd

- Immature psychological defense mechanism such as 'regression' when under stress
- Learned dysfunctional coping strategies eg, poor anger Mx under stressful situations

SOCIAL FACTORS

- Under- or over-stimulating environment
- Carer stress/conflicts with family or others
- Issues around the lack of social support
- Difficulties developing fulfilling relationships
- Problems in finding employment
- Physical and psychological abuse
- Lack of appropriate social exposure,
- Stigmatization, and discrimination
- Bereavement and other life events

Challenges in diagnosising psychiatric co morbidities in ID

A. **Diagnostic overshadowing** is tendency to attribute any behavioral/psychological disturbance to ID.

B. **Baseline Exaggeration.** Challenging behavior that exists at a low rate and low intensity may increase dramatically when one has stress or a mental health condition for people with ID, the behavior becomes the focus of the referral, but i**t is only a symptom**

C. **Intellectual Distortion.** Due to challenges in abstract thinking, receptive and expressive language skills questions will be too complex and answers often meaningless. To, "Do you ever hear voices when no one is there? the person with ID responds "Yes" having no concept of a *hallucination*

D. **Cognitive Disintegration.** Due to lack of cognitive reserve, those with ID may dramatically decompensate under stress and e.g., lose skills, become mute or hallucinate. Difficulty communicating feelings of stress may result in anxiety-induced behavior, that can be incorrectly diagnosed as psychosis, bipolar disorder, or dementia

E. **Psychosocial Masking**

- Misunderstanding of what may be developmentally appropriate
- A delusion of being the chieef of police may be mistaken for a harmless fantasy
- An imaginary friend may be mistaken for a delusion

Issues of Dual Diagnosis: ID and mental illness

4 Factors Affecting Presentation Sovner (1986)

- Intellectual Distortion
- Psychosocial masking
- Cognitive disintegration
- Baseline exaggeration

Intellectual distortion

- Emotional symptoms are difficult to elicit because of deficits in abstract thinking and in receptive and expressive language skills
- (Silka & Hauser, 1997)

Cognitive disintegration

- Decreased ability to tolerate stress, leading to anxiety induced decompensation (maybe misinterpreted as psychosis) (Silka & Hauser, 1997)

Psychosocial masking

- Limited social experiences can influence the content of psychiatric symptoms
- example - mania presents as "I can drive a car"
- Silka & Hauser, 1997)

Baseline exaggeration	Increase in challenging behavior frequency and/or intensity during the course of a mental illness. During times of stress, escalating behavior will prompt a mental health evaluation.
Intellectual distortion	The individual cannot accurately understand the questions posed by the evaluator, nor can he or she assemble the correct information to respond.
Psychosocial masking	Because of developmental delay, the individual might present symptomatology that occurs within a developmental framework that would be common in much younger individuals.
Cognitive disintegration	The individual may become grossly disorganized and psychotic because of the lack of "cognitive reserve" available to cope with the illness.

DIFFERRENTIAL DIAGNOSIS

- Borderline intellectual functioning
- Posttraumatic Stress Disorder
- Cognitive Deficits
- Learning Disorders
- Pediatric Depression
- Autism Spectrum Disorder
- Severe communication/language disorders

CLINICAL ASSESSMENT

1. History
2. Phyiscal exam/Dysmorphic features assessment
3. IQ and Adaptive skills assessment
4. MSE - psychiatric comorbidity / challenging behavior assessment

- ID is a permanent condition so it creates special needs for both the individual and family across the life span.
- The needs could be related to independent mobility, physical care, communication needs, modified curricula, aids and appliances, occupational and vocational opportunities and medication if there are treatable, comorbid medical conditions.
- The special needs may necessitate support in varying degrees throughout the life span. So, holistic programs should address the lifelong needs in a step-by-step fashion. For instance, when a child with ID is in preschool years, the needs may center around self-care, sociocommunication skills, and school readiness skills but not so much about independent living or literacy. Similarly, for an adolescent with ID, the needs could be about education, prevocational training, and future independent living.
- Also ID will imply long-term, multidisciplinary approach to intervention for optimum outcome.

CLINICAL ASSESSMENT

The purpose of the assessment is:

1. To identify the condition based on specific criteria.
2. To identify comorbidities, any challenging behaviors and other co —occurenes
3. To identify strentths and weakness as well as the support and needs implicated by the condition.
4. To identify etiological factors and risk factors for ID

HISTORY TAKING

- Family history
  - ■ID, psychiatric illness, epilepsy or others
  - ■Quality of relationships with family members
- Personal and developmental history
  - ■Ask about pregnancy, birth, progress to date
  - ■Developmental years, including milestones
  - ■Family's management of child with ID
  - ■Education, job, relationships at school, work
  - ■Personality/behavior prior psychiatric illness
  - ■Psychosexual history; life events - loss, abuse,
  - ■Highest level of functioning patient reached
- Medical history
  - ■Cause of the ID, eg, genetic cause, if known
  - ■Past/present physical illnesses eg epilepsy,
  - ■Past and present physical disability
  - ■Impairment in vision, hearing, speech
  - ■Recurrent physical illnesses & how person communicates pain or other bodily symptoms
- Psychiatric history
  - ■Previous Health Care services and diagnoses (as previous diagnosis could be wrong, if possible find out exact clinical picture of previous illness)
  - ■Risk assessment (risk to the person and others)
- Drug history
  - ■Past and present medication and dosage
  - ■Drug adverse effects, known allergy
  - ■Substance and alcohol use (if relevant)
- Social history
  - ■Current and previous level of functioning in different areas of adaptive behaviors
  - ■Current and previous social circumstances
  - ■Current and previous living arrangements
  - ■Ask about person's daily/weekly routine to elicit daytime, social, leisure activities and carer support
- Forensic history
  - ■Past and present history of problem with the law both in patients, their friends and relations

Physical examination with ID consists of three parts which are as follows:

1. **Anthropometry**.

- This provides indication of nutritional status and underlying medical or genetic condition. The measures should include the following: height (length in case of neonates and infants), arm span, upper segment and lower segment lengths, sitting height, weight, head circumference, chest circumference, abdominal circumference, intercanthal and interpupillary distances, and palm and foot lengths.

1. **Dysmorphology examination**

2. **Examination of major organ systems**. A systematic examination of all the organ systems to rule out multiorgan involvement and comorbid medical conditions.

- **Interview techniques**

  ■ Asking some general, easy questions at the start of the interview will help put the subject at ease.

  ■ Assess subject's communication ability at the outset of the assessment

  ■ Visual aids eg, drawings or picture books

  ■ As much as possible, speak to subjects

  ■ Avoid use of leading questions, where possible.

  ■ Use appropriate language eg simple phrases, short sentences; avoid words or medical jargon that the subject may not understand).

  ■There may be need to repeat questions.

  ■ Minimise suggestibility

- **Patient observation**

  ■ In some, it is necessary to ask the carer/family, about any changes in the subject's mental state.

  ■ It may also be necessary to ask carer or family to monitor for a period of time, certain variables such as sleep, appetite, weight, activity level, particular behaviors and so on, to aid in the diagnosis.

  ■MSE. Direct observation of patient - a must.

  ■ Physical examination

  ■ Certain investigations such as thyroid function test, EEG, brain scan etc (if relevant).

  ■ Multi-professional assessment (nurse, general practitioner, psychiatrist, clinical psychologist),

PHYSICAL EXAMINATION

- Head circumference: Microcephaly may correlates with cognitive deficits. Macrocephaly with hydrocephalus.
- Height: Short may suggest a genetic disorder, hypothyroidism. Tall may suggest Fragile X
- Sensory: visual impairment and hearing deficits
- Neurologic: assess for hypotonia or spasticity, strength and coordination, deep tendon reflexes, persistent primitive reflexes, ataxia, other abnormal movements-dystonia or athetosis.
- Skin exam: hyperpigmented/hypopigmented macules café-au-lait macules > tuberous sclerosis, fibromas,
- Extremities: face and digits anomalies
- Kilifi Developmental Inventory- culturally appropriate to describe development of at-risk children in resource-limited settings in Kenya

DYSMORPHIC FEATURES ASSESSMENT.

- Observations and also use of photography helpful.
- With appropriate parental consent a full face view, right and left lateral views, a full body view and close up views of any specific dysmorphic features are a vital part of the medical record. Also of skull, face, neck, hands, feet, chest, mouth.

COGNITIVE AND ADAPTIVE ASSESSMENTS

- IQ and adaptive skills evaluation :

➢ IQ Tests such as WISC, Stanford-Binet

➢ Adaptive functioning assessment include interviews with carers/family/ teacher/others, who have observed their adaptive functioning — i.e., their conceptual, social & practical functioning — whether or not someone has the skills necessary to live independently

➢ Vineland Adaptive Behavior Scales

MENTAL STATE EXAMINATION

- For mild, moderate ID the standard MSE format can usually be followed, though some observations may differ in significance from similar findings in patients who don't have ID and some phenomena may take a different form.
- Severe, profound ID may - not able to describe mental phenomena. MSE reliant on patient observation. Ask carers-behavior; sleep, activity changes. Use visual aids.
- Psychiatric morbidity substantial - ADHD, ASD, Mood Disorders, Stereotypic Movement Disorder but intellectual distortion, psychosocial masking, cognitive disintegration, baseline exaggeration aspects of ID may obscure usual indicators of psychiatric illness

MSE among those with ID – key points

- Appearance/behavior- restlessness, spasticity, gait, abnormal movements; social response at interview-language, gesture, eye contact, social style eg reserved, friendly, disinhibited
- Language – comprehension, speech, gesture use

MSE finding differences in ID:

- For mood symptoms - focus on biologic symptoms and behavioral equivalents to subjective states.
- Delusions –may include new avoidance or fears; glaring with intense anger at strangers or previously liked others; sudden medication refusal
- Hallucinations-more likely to observe interaction with hallucinations; they may be agitation or SBI in response to hallucinations; may be seen covering eyes or ears to 'block out ' hallucinations and may also include sniffing the air, as if smelling something not smelt by others
- Symptoms not indicative of psychosis –some symptoms, though dramatic in presentation almost never indications of psychosis:

➢ Volitional self talk and self answering; shouts and screeches almost always vocal tics; symptoms that very clearly being copied from others;

➢ gestures that person can start and stop at will or when asked to start or stop; gestures/vocalizations which have been explicitly taught to the person

MANAGEMENT OF ID

1. **Biopsychosocial Investigations**
2. **Biopsychosocial Treatment Interventions**

BIOPSYCHOSOCIAL INVESTIGATIONS

Biological investigations - none specific good hx and physical/MSE guide to appropriate ones -

- blood hemogram, thyroid profile; urinalysis, lead levels etc as needed
- EEG/echo,
- EEG; Xray skull, CSF exam; CT/MRI scan - hydrocephalus, tuberous sclerosis, cortical atrophy,
- chromosomal study for down/turner's/fragile X syndromes;
- biopsy [bone marrow, liver, rectum, brain, skin] storage disorders;
- hearing, speech and vision tests, occupational and physical therapy evaluations

Psychological tests and psychoeducational assessments— special education tests

Social investigations – Assessment of family needs and functioning—collaborative history, home/ school/job visits to elicit more about the family background, family interactions, history of similar disorder etc.

TREATMENT

- There is no "cure" for ID. ID not a single entity, it is myriad of different disorders, some with known etiology.
- The aims of treatment are:

[1]. To identify and treat reversible causes of ID,

[2]. to alleviate suffering for individual and family and

[3]. to minimize consequences, maximize functioning and promote healthy development so as to develop the person's potential to the fullest in terms of: education, social and life skills.

- Treatment interventions and support plans for ID focus on person's: strengths, weakness and additional conditions as well as support needed to function as determined by the severity of ID and supports available as determined by input from multiple disciplines. Early, ongoing, appropriate interventions/support, do improve functioning. The treatment/support interventions can be grouped into:

1. Parental counselling
2. Specific treatment
3. Associated impairments treatment/minimizing complicating co-morbidities
4. Supportive treatment/Environmental optimization

No human has ever been conducted. Issues relating to sexuality/family planning/custodial care/estate planning/guardianship as well as brain— or— head transplant important but beyond scope of this lecture.

1. Parental counselling

- Breaking the news-phased manner.

Management of ID begins with breaking the news, compassionately, culturally appropriately and in an individualized approach as per the severity, cause and prognosis. Positive aspects of the ID should be discussed first, followed by the problems but do not withhold the truth. Development of correct attitudes towards their child by giving correct information on nature, causes & treatment ID . In order to deal with false guilt, parents should be explained that ID is generally due to causes over which they have no direct control.

- Psychoeducation:

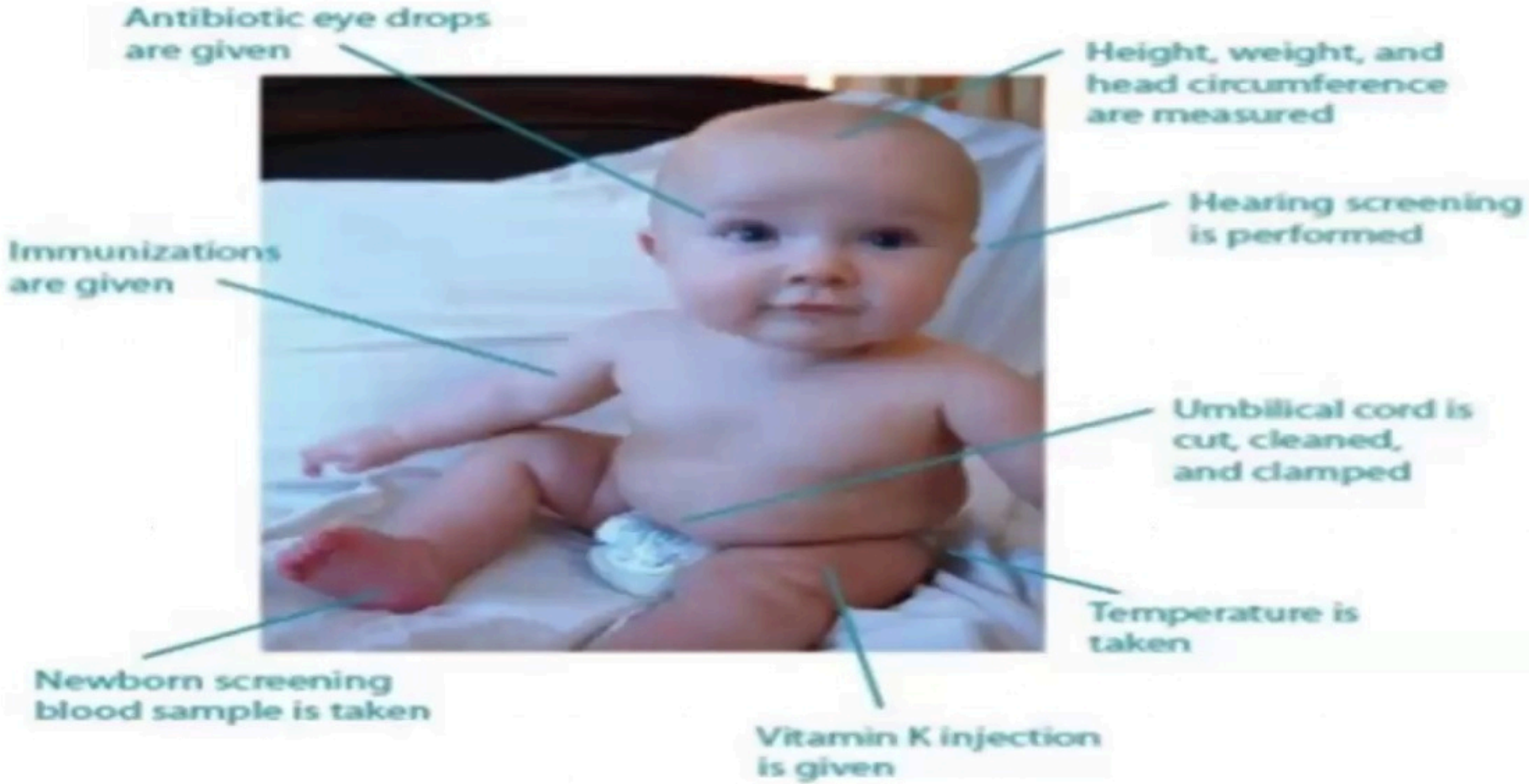
Provide information regarding child's ID, management of associated medical problem and other disabilities must be made available to the parents. Ensure full participation of family members with special support to the mother. Facilitate protection against physical or sexual abuse

- Parent skills training. Educate parents about their role in training child.
- Parent support: Information regarding parent group, support organizations and counseling about the risk of recurrence prevention of disease in future pregnancies.

2. Specific treatment

- Early identification and etiological treatment if cause is known and treatable, for example: PKU - dietary restriction for newborns identified with PKU, who if left untreated develop an IQ of less than 30. Successful treatment of PKU has produced a generation of healthy adults with PKU.
- Symptomatic –anticonvulsants, medical/surgery and interventions for associated anomalies eg, CHD
- Preventative treatment: prenatal treatments examples include education, avoidance of neurotoxic compounds such as alcohol, treatment of maternal hypothyroidism or enzyme replacement therapy (ERT) and metabolic amelioration for some inborn errors of metabolism. There are promising treatment directions for targeted therapeutics for Down/Fragile X/Rett/syndromes and Tuberous sclerosis.
- Physiologic therapies, including deep brain stimulation and transcranial magnetic stimulation, offer yet another direction to enhance cognitive functioning.
- However, there are still areas, that remain challenging for development of treatments: congenital structural brain lesions e.g. hydrocephalus; ID of unknown etiology and untreated consequences of known disorders, such as neurodegenerative conditions, inborn errors of metabolism, kernicterus etc.

NEWBORN SCREENING AT BIRTH



DOWN SYNDROME

- ❑ Incidence (0.01%) 1: 700 -1000 (live births). Ideal maternal age is 20s, over 35 increased risk of DS. Ages - <20yr 1: 4000; 35yr 1: 400; 40yr 1: 110 & 45yr 1:35. Paternal age - mild effect/abnormal sperms after 42 yr.
- ❑ Examination and investigations at birth:

- Red reflex r/o congenital cataract, eye exam for strabismus, hearing test, exam for imperforate anus, TFTs to r/o hypothyroidism, echo to r/o CHD, CBC to r/o congenital/transient leukemia, USG
- Antenatal Screening [non- Invasive tests] -maternal blood triple test - comprises alpha fetoproteins[AFP], b human chorionic gonadotropin (B-hCG) & unconjugated Oestriol (uE3). Done at 15-18 wks. Low AFP & uE3 with high B-hCG+ maternal age, diagnose 69% DS
- Invasive tests: Detects up to 99.8% with rare false +ve result: (i) chorionic Villous sampling: At 10-12/52. Fetal cells are obtained, cultured & karyotyping is done. (ii)Amniocentesis: At 14-16/52

- ❑ Management:

- TOP. UK/Europe 92% - abortion after antenatal Dx
- Parents counselling: Nature/problem, future antenatal screening, family planning, baby & parents karyotyping, Mx of menstruation & contraception-adolescent
- Specific treatment for medical conditions-eye/ear #; underactive thyroid; CHD, GIT abnormalities, hip/joint #, Respiratory Tract Infections [RTI]
- Glasses for refractive errors/hearing aids. Avoid risky athletics/games; gluten diet if coeliac disease
- For hypothyroidism, D/M, hearing loss and eye#- annual checkup and investigations; early dx, Mx, prophylactic medications and special additional vaccines to avoid RTI
- Plastic surgery-partial glossectomy > 2/3 speech & 1/3 oral competence improve. Surgery-GIT/heart/other repairable anomalies
- Speech delay Mx – speech therapy, speech augmentative & alternative communication eg, pointing, body language
- Early interventions from birth, to coordinate/plan effective strategies for learning development
- Special schooling – main stream or special abilities within class sports, outing, breaks, meals & art
- Prevention – avoiding late child bearing esp. maternal age > 35 yrs

CONGENITAL HYPOTHYROIDISM [CH]

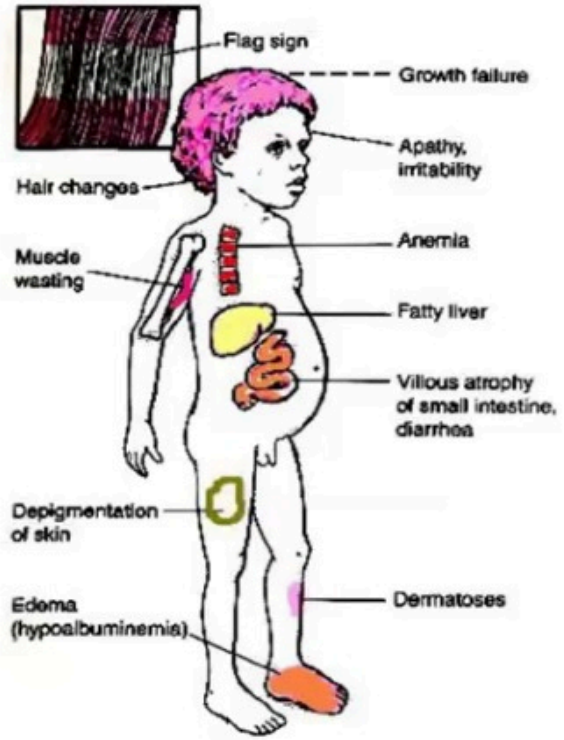
- **Definition:** thyroid hormone deficiency present at birth. 1: 4,000
- **Screening** –universal newborn at 3-4 days of age -T4/TSH using cord blood or from heel prick; otherwise, newborns screened if: 1. clinical features or family history of CH. 2. Maternal thyroid# 3. Presence of Down's, neural tube defects, CHD, metabolic disorders, etc, associated with higher prevalence of CH.
- **Treatment.** Recommended dietary iodine -30ug/kg/OD for infants, 90-120ug/OD for children and 150ug/OD for adolescent and adults. Infants with low T4/elevated TSH - start L-Thyroxine at dose of 10-15µg/Kg/day. If severe -very low T4/very high TSH & radiograph of knee# - start at highest dose 15µg/Kg/day.
- **Monitoring:** In less than a week T4 rises and in 4-5/52 TSH normalizes. Overtreatment-craniosynotosis & temperament problem. T4/TSH monitoring: 0 to 6/12 - every month; 6 months to 2 years-every 2- 3/12; >2 years - every 6/12/ 6-8/52 after dose change.
- **Prognosis:** Intellectual impairment preventable if treatment is initiated early, otherwise prognosis related to the - nature & severity of CH, age at diagnosis & onset of treatment, adequacy & regularity of treatment.

PHENYLKETONURIA (PKU):

- **Definition:** genetic disorder - inability to utilize essential amino acid, phenylalanine.
- **Treatment** - tyrosine supplementation [Tetrahydrobioptrin -cofactor of phenylalanine oxidation] & phenylalanine dietary restriction. Diet goal is to maintain phenylalanine blood level between 2 and 10mg/dl as some phenylalanine needed for normal growth. With good dietary control, [with phenylalanine but in lower amounts than usual], potential effects on development minimized.
- Foods that can be given: cereals/other starches, dried beans/peas, fruits, vegetables along with a milk substitute and nutritional supplements-fish oil, selenium, vitamin B12/K [if deficient]
- Periodic check phenylalanine blood levels.
- Fever and illness can cause normal body proteins to break down, raising phenylalanine blood levels, so physician and nutritionist can suggest dietary changes to help maintain levels in the desired range during illness.

KWASHIORKOR

- Kwashiorkor, severe malnutrition, caused by a deficiency in dietary protein. Children with kwashiorkor tend to have low blood sugar levels, as well as low levels of protein, sodium, zinc, magnesium.
- If kwashiorkor suspected -check for enlarged liver; next, blood and urine tests include: blood urea nitrogen, blood creatinine/potassium levels, complete blood count, urinalysis
- How is kwashiorkor treated? Kwashiorkor can be corrected by eating more protein and more calories overall, especially if treatment is started early.



RH INCOMPATIBILITY

- **Difference** in Rh blood type between mother (Rh -) & fetus (Rh+)
- **Screening**- blood group/Rh status
- **Treatment:**1. Antenatal-intra–uterine/peritoneal /vascular blood transfusion. 2. Postnatal-if mild neonatal jaundice-phototherapy-if moderate/ severe exchange transfusion before1/52
- **Prevention:**–RhoGAM immune globulins at 28/52 -If baby Rh positive, another dose is administered within 72 hours after delivery. This prevents mother creating any future antibodies that could cause harm during a pregnancy.

3. Associated impairments/minimizing complicating co-morbidities treatment interventions:

- Behavior management techniques used to control behavior problems. But, aversive (punishment) behavior management techniques should be completely eliminated and reliance be on positive behavioral techniques.
- Behaviour intervention for challenging behaviour
  - Psychotherapy. People with mild ID can benefit from psychotherapy. Cognitive Behavior Therapy [CBT] teaches people with mild ID to recognize the situations in which they get into trouble and to develop alternative behavior and solutions to their problems. Although widely used with the general population, CBT only recently adapted for use with people with ID.
  - Social Skills Training - gradually teaches effective social interactions and appropriate social behavior. It is a cost-effective, time-limited approach with noticeable quality of life/interpersonal behavior improvements.
  - Activity/Music/Art therapy help build positive experiences and self-confidence.
  - Occupational and physical therapy can be helpful for some individuals.
  - Psychopharmacology. Medications are only part of an effective total management program. However, don't over-medicate people with ID and carefully monitor the behavioral effects of medications.
- **Manage risk/contributing factors** - ear /eye#, nutrition, immunization, maternal depression, stimulation.
- **Medical /psychiatric Care** to manage co-occurring epilepsy, behavior problems and psychiatric morbidity. Medications-stimulant agents-ADHD/ Neuroleptics-self injurious behavior/ SSRIs- anxiety, depression

Medical Care

➡**Physical activity and obesity are major contributors to disease in ID.**

★Programs targeting healthy lifestyles (nutrition, exercise, self-care, stress reduction) needed.

★Annual counseling on these issues is recommended.

★Medications (eg, antipsychotics) should be titrated to reduce the risk of obesity and metabolic issues.

➡**Pain:** Manifestations of pain in people with severe to profound ID include crying, screaming, grimacing, protective postures (eg, arching, fetal position), rocking, and aggression. Parent/caregiver input is key to interpretation of these behaviors, though validated tools used as adjuncts eg, Pediatric Pain Profile. Common causes of acute pain include dental caries/abscesses, GERD, constipation, UTI, spasticity (when ID is associated with cerebral palsy), pressure sores, and fractures. In addition, neuropathic pain due to dysautonomia or motor spasms may create chronic disturbances. Treat pain promptly including NSAIDs for mild pain, tramadol or equivalent for moderate pain and opioids for severe pain as indicated and management of sources of pain. Written, verbal and pictoral communication; gestures and demonstrations are helpful for those with ID to ensure mutual understanding and improve treatment adherence.

➡**Sedation/anesthesia**: Patients with ID may have paradoxical reactions eg, to benzodiazepines, so use the lowest dose and titrate slowly.

➡**Sexuality/abuse:** Some with ID experienced abuse, which contributes to mental health issues. This should be addressed at each medical visit, esp. in the setting of changes in behaviors, eg increased aggression.

➡**Cognitive enhancement:** No treatments available specifically for cognitive deficiency. Pharmacologic enhancement of cognition (eg, donepezil) posed, but research on this compounds is limited.

Psychiatric Care

➡ **Comorbid psychiatric disorders:**

★40-70% individuals with ID, have diagnosable psychiatric disorders, such as ADHD, schizophrenia, mood disorders, anxiety disorders, austism spectrum disorder and psychoses.

★Psychopharmacology may have a crucial role in a multidisciplinary/multimodal treatment of comorbid psychiatric disorders. Antidepressants, mood stabilizers, anxiolytics, stimulants & antipsychotics are effective. SSRIs, newer anticonvulsants and atypical neuroleptics preferred in treatment of those with ID.

➡**Challenging behaviors**:

★Also like anyone else, a person who is intellectually disabled may exhibit emotional, behavioral, interpersonal or adjustment problems that do not constitute major psychiatric disorders but that, nonetheless, may benefit from psychiatric input. 25% exhibit stereotypies, physical aggression or SIBs challenging behaviors. Challenging behaviors can be harmful to the child or others, strain social relationships, limit children's access to educational or social opportunities and place a financial burden on families.

★Behavior modification used to address challenging behavior. Functional analysis used to identify and then alter the antecedents or consequences of the identified challenging behavior.

★**Behavior modification** can used to develop a new behavior, strengthen a behavior, maintain an established behavior and to stop inappropriate behavior. Behavior modification core components:

1. Positive punishment, Adding a consequence to decrease problem behaviors is referred to as a positive punishment, eg, assigning an extra chore when child tells a lie; spanking is a form of positive punishment, but most experts discourage any use of corporal punishment when utilizing behavior modification as research suggests spanking may actually increase negative behaviors and negative long-term outcomes.
2. Negative punishment means to remove or take away, such as taking away a privilege or withholding positive attention eg time out, actively ignoring a child's temper tantrum.
3. Positive reinforcement - a child is given something they like to reinforce good behavior, examples are - praise, reward system or a token economy.
4. Negative reinforcement - something unpleasant is removed due to child's positive behavioral choice.

★Play therapy and opportunities for social group interaction often help them express their inner conflicts.

★Behavior therapy, especially positive reinforcement, effective in modifying some maladaptive behaviors.

★Psychotropic medication occasionally used to help remove or modify some target behavioral symptoms, eg., hyperactive and impulsive behavior. Medications, when prescribed, are targeted to specific co morbid psychiatric disease or behavioral disturbances. Atypical antipsychotics eg, risperidone, lithium or propranolol useful to manage aggressive behaviors. Naltrexone has reduced self-injurious behaviors in some ID persons.

4. Environmental optimization/Supportive treatment:

- ✓ AIM – normalizing/mainstreaming – integrate in society & discourage institutionalization.
- ✓ Key–Early intervention and input from multidisciplinary team - special educators, clinicians, nurses, social workers, psychologists, language/speech/behavior/OT/physical therapists etc.,
- ✓ How- multidisciplinary team work- to identify ID, evaluation of strengths, weakness, additional conditions, supports needed as per person's ID degree [mild, moderate, severe, profound] in 5 dimensions

1. Intellectual
2. Adaptation [conceptual, practical, social]
3. Participation [interaction, social roles]
4. Health [physical|mental]
5. Social context [culture, environment, opportunities]

to set the stage for individualized early interventions that specifically tailored to each person's needs

✓ Treatment plan also guided by developmental stage of the individual with ID:

(a) Infancy/early childhood: sensory/motor/communication/self help skills & socialization development.

(b) During childhood and adolescence: basic academic skills, activities of daily life skills, application of reasoning and judgment in the mastery of environment in their use of social skills

(c) During late adolescence and adulthood: vocational performances and social responsibilities

✓ What - interventions?

- early stimulation, education plan [special schooling, school support, vocational],
- transition services to adulthood after high school, day programs, vocational programs, as well as ensuring
- the individual receives proper care, psychological or psychiatric services,
- speech and language pathology or audiology services, therapeutic recreation,
- community rehabilitation, adapted equipment or assistive technology.

PREVENTION

Primary - eliminating the cause/preventing ID

- For all
  - ★Iodine and iron supplementation
  - ★Prevent exposure to environment toxin
  - ★Improve socioeconomic status
  - ★Avoid consanguinity
- Pregnant mothers
  - ★Safe motherhood years 20-35 yrs
  - ★Peri-conceptional folate
  - ★Good antenatal/perinatal care
- Children
  - ★Routine immunization
  - ★Provide stimulation for optimal development in safe enriching environment

Secondary – halting disease progression

★Early diagnosis|treatment of curable illness - hypothyroidism, treating Rh blood incompatibility between mother and fetus, PKU identification & maternal dietary restrictions.

★Prenatal diagnosis –chromosomal analysis, chorionic villus|cord blood sampling, amniocentesis for Down syndrome or other genetic abnormalities > [TOP]

★Genetic counselling –exact etiology/empiric risk figures

Tertiary - minimizing consequences/maximizing functioning

★Support for families

★Early rehabilitative intervention [stimulation, training]

★Vocational opportunities and support.

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