MENTAL RETARDATION
LEARNING OBJECTIVES

• To be able to define and classify it on the basis of severity.
• To be able to understand the wide normal variation of intellectual potential in the community and identify developmental delay correctly
• To recognize conditions that can be misdiagnosed as MR.
• To recognize factors that cause mental subnormality and also know common genetic and chromosomal conditions associated with MR.
• To be able to evaluate a child with MR for etiology and severity.
• To enable early recognition of treatable causes.
• Parental counseling about treatment options and prognosis.
• To be able to institute community measures for prevention of mental retardation including genetic counseling.
DEFINITION

What is mental retardation?
U.S special education Law; The Individuals with Disability Education Act defines:

• Significant sub average intellectual function
  IQ < 2SD below the mean/ score of 70 or below.
• With deficits or impairment in adaptive behavior in at least 2 of the following area.
  – Communication, social/interpersonal skill, Self care, use of community resources,
  – Home living, functional academic skill, Self direction, work, leisure, health and safety.
  – Manifests during the development period (<18 years)
### A) American Psychiatric Association (APA) Traditional classification

<table>
<thead>
<tr>
<th>LEVEL</th>
<th>IQ</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mild</td>
<td>51 – 70</td>
</tr>
<tr>
<td>Moderate</td>
<td>36 – 50</td>
</tr>
<tr>
<td>Severe</td>
<td>21 – 35</td>
</tr>
<tr>
<td>Profound</td>
<td>&lt;21</td>
</tr>
<tr>
<td>Severity unspecified</td>
<td>Untestable by standard tests</td>
</tr>
</tbody>
</table>
Now a days most authorities prefer to classify in 2 major groups to identify etiology

<table>
<thead>
<tr>
<th>Mild</th>
<th>IQ 50 – 70</th>
<th>Environmental causes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Severe</td>
<td>IQ &lt; 50</td>
<td>Biological causes</td>
</tr>
</tbody>
</table>
## B) AMERICAN ACADEMY OF MENTAL RETARDATION (AAMR) RECENT CLASSIFICATION

<table>
<thead>
<tr>
<th>Classification</th>
<th>Support system required</th>
<th>Major Deficits</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intermittent</td>
<td>Education</td>
<td>Functional reading &amp; writing skills, lack of judgment</td>
</tr>
<tr>
<td>Limited</td>
<td>Employment</td>
<td>Needs economic support but capable of employment in a sheltered setting</td>
</tr>
<tr>
<td>Extensive</td>
<td>Daily routine activities</td>
<td>Self care skills, communication skills</td>
</tr>
<tr>
<td>Pervasive</td>
<td>Structured environmental &amp; Institutional care</td>
<td>Health / Medical</td>
</tr>
</tbody>
</table>
EPIDEMIOLOGY

• **PREVALENCE**: depends on the definition, the method of ascertainment and the population.

• 2-3% general population,
  – Mild MR 20-30/1000, severe MR 3-4/1000
  – In US, based on APA definition 2.5% out of which 85% mild MR

• The AAMR definition increased the IQ threshold for MR from 70 to 75 to reflect the standard error of IQ measurement. This definition doubles the prevalence of mental retardation.

• Frequently in boys than in girls
  2:1 (mild MR), 1.5:1 (severe MR)
  (mainly because of X linked disorders)
PATHOLOGY

• 10-20% of brains of individuals with severe MR are normal.

• Majority of brains show mild non specific changes:
  ✓ Microcephaly
  ✓ Gray matter heterotopia in the sub cortical white matter
  ✓ Unusually regular columnar arrangement of the cortex
  ✓ Tightly packed neurons

• Minority of brains show specific changes:
  ✓ Dendritic and synaptic organization with digenesis of dendritic spikes
  ✓ Cortical pyramidal neurons or impaired growth of dendritic trees.
## ETIOLOGY – WHY DID IT HAPPEN?

<table>
<thead>
<tr>
<th>Mild (IQ &gt; 50), Idiopathic 50%</th>
<th>Severe (IQ &lt;50) Specific Biological causes in 75%</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Environmental Cause</strong></td>
<td><strong>Genetic</strong></td>
</tr>
<tr>
<td>lead toxin</td>
<td>22% Genetic</td>
</tr>
<tr>
<td>iodine and iron deficiency</td>
<td>21% (Fragile X, Prader-Willi)</td>
</tr>
<tr>
<td></td>
<td><strong>Socioeconomic factor</strong></td>
</tr>
<tr>
<td>(poor stimulation in developing</td>
<td>CNS malformations 9%</td>
</tr>
<tr>
<td>years)</td>
<td>(Hydrocephalus, meningomyelocele, Lissencephaly)</td>
</tr>
<tr>
<td><strong>Low parental intelligence</strong></td>
<td>Congenital infections 4%</td>
</tr>
<tr>
<td></td>
<td>(TORCH)</td>
</tr>
<tr>
<td></td>
<td>Perinatal insult 4%</td>
</tr>
<tr>
<td></td>
<td>(HIE, IVH)</td>
</tr>
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</tbody>
</table>

**Significant overlapping populations due to Multifactorial/ Treatable cause/ Onset and progress of the disease and insult**
DIAGNOSIS OF MR

1. Presence of risk factors
   Family history
   Prematurity
   Maternal substance abuse
   Perinatal insult

2. Clinical manifestation
   a) Dysmorphism
      Identify genetic syndrome - Down syndrome
      Fragile X syndrome
   b) Developmental delay - global/significant
      vision/hearing impairment by 6 months
      gross motor delay by 2 yrs
      language delay by 3 yrs
      fine motor delay by 5 yrs
c) Associated dysfunctions

neurological disorder- cerebral palsy, autism
microcephaly
unusual muscle tone(hypo/hypertonia)
abnormal posture and feeding difficulties
seizure
behavior difficulties(below 5 yrs):
(attention span, anxiety, mood and conduct)
academic underachievement(above 5 yrs)

d) Typical symptoms- hypothyroidism
DIAGNOSIS OF MR

3. Intelligence Quotient Testing (IQ)
   Calculated by IQ = mental age/chronological age

   **Standard Tests**
   • Bayley Scales of Infant Development (BSID II) till 3 yrs
   • Wechsler preschool and primary scale intelligence
   • Wechsler intelligence scale for children, above 6 yrs
   • Malin’s Intelligence Scale for Indian Children 6-15 yrs
   • Vineland Social Maturity Scale (VSMS) - Indian Adaptation, till 15 yrs
   • Stanford-Binet Intelligence Scale
   • Binet Kamat Test of intelligence (BKT) – 3-22 yr (revision of Binet’s scale by Kamat to suit indians)
DIAGNOSIS OF MR Q TESTING – CONT'D

Screening Test

• **Siguin Form Board (SFB):** used in case of speech impairment and to select appropriate IQ test.

• **Goodenough’s Draw- A - man test** for Indian children (DMTC-P), by Pramila Pattak, used from 3-16 yrs of age. The child is asked to draw a man and he/she receives 1 point for each of the items present in the drawing. For each 4 points, 1 year is added to the basal age of 3.

• **Developmental Screening Tests**
DIAGNOSIS OF MR

4) Laboratory Investigations:
Plan and select appropriate tests as per history/clinical findings

Metabolic screening

Urine:
– Ferric chloride test
– DNPH
– Benedict’s test
– Nitroprusside-cyanide spot test
– MPS (Mucopolysaccharides) spot test

Blood:
– Aminoacids
– Organic acids
– Lactate, ammonia
• Neuroimaging - CT, MRI
• EEG
• Genetic evaluation:
  karyotype, cytogenetic, specific gene probes
• Specific tissue/Biochemical Analysis
  skin, liver biopsy
  serum lead, cerryloplasmin
  Thyroid Function Test
  TORCH, HIV
DIFFERENTIAL DIAGNOSIS

Conditions that mimic MR and other conditions with intellectual disability as an associated impairment.

• Sensory deficits-severe vision/hearing loss
• Communication disorders
• Poorly controlled seizures-epileptic syndromes
• Neuromuscular disorders
• Progressive neurological disorders-regression of milestones
• Isolated cerebral palsy-motor skills more affected than cognitive skills
• Autism-social&language skills more affected
• Normal variation till 3 yrs
• Severe PEM/chronic illness
• Emotional deprivation
• Childhood psychosis
EVALUATION OF MR

1) Identification of children at risk for delayed development: plan preventive/follow up programme.

2) Is the child really retarded? check list of milestones

   Identify significant delay

   • Global delay-equivalent deficits in social, motor, adaptive & cognitive skills
   • Consider prematurity, recent illness and compare with siblings and peers
   • Consider condition misdiagnosed as MR
EVALUATION OF MR CONTD...

4) Assess the severity-screen and select the appropriate IQ test

3) Look for associated impairments/anomalies
   - Organ dysfunction/defects
   - Behavioral/psychological abnormalities-ADHD, autism
5) Identify etiology
75% cases of severe MR-biological causes, 50% cases of mild MR-idiopathic causes

**History**
- Iodine deficiency/lead exposure/perinatal insult
- Specific symptoms-hypothyroidism
- Affected family members-AD, AR, X linked Inheritance
- Consanguinity in parents
- Maternal age, educational level, socioeconomic status of parents

**Clinical examination**
- Dysmorphic features-identify syndrome
- Ophthalmological evaluation- ‘cherry red’ spot
- Micro/macrocephaly
- Neurological signs
- Neurocutaneous markers
CLINICAL EXAMINATION

- Dysmorphic features-identify syndrome
- Ophthalmological evaluation- ‘cherry red’ spot
- Micro/macrocephaly
- Neurological signs
- Neurocutaneous markers
Downs syndrome
- oblique eye fissures with epicanthic skin folds on the inner canthi of the eyes
- flat nasal bridge
- a single palmar crease
- protruding tongue (macroglossia)
- excessive space between large toe and second toe
FRAGILE X SYNDROME

- Elongated face
- Large or protruding ears
- Flat feet
- Larger testes
  (macro-orchidism)
TURNERS SYNDROME

- Short stature
- Lymphedema (swelling) of the hands and feet
- Broad chest (*shield chest*) and widely spaced nipples
- Low hairline
- Low-set ears
- Webbed neck
- Wide Carrying Angle
CHERRY RED SPOT

- Mucopolysaccharidosis
- Hurler's disease
- Tay-Sachs disease
- MPS VII
- Farber's disease
- GM1 gangliosidoses
- Niemann Pick's disease
- Lysosomal Storage Diseases
PHENYLKETONURIA

- Fail to attain early developmental milestones
- Microcephaly
- Seizures
- Severe learning disabilities
- A "musty or mousy" odour of skin, hair, sweat and urine
- Hypopigmentation
- Eczema
NEUROCUTANEOUS MARKERS

Café-au-lait spot

Ash leaf spots

Shagreen patches
Lab examination

Specific diagnostic test:—

  *if specific disorder is suspected*

Intensive investigations:—

  *in absence of clinical indicators*

Plan depending on severity of MR/family wish

  *(if prenatal diagnosis is required)*

Emphasis on **simple test** to start with

  *(screening/non invasive/cost effective)*

Look for **treatable conditions**

  *(IDA/PKU/Hypothyroidism)*
EVALUATION OF MR

SPECIFIC DIAGNOSTIC TEST

Ex: Karyotyping for Down Syndrome
MANAGEMENT OF MR

a) Specific treatment
  Early identification & prevention - PKU
  Treatable conditions - IDA, hypothyroidism
  Symptomatic - anticonvulsants, Medical/surgical intervention for associated anomalies eg: CHD

a) Associated impairments
  Behavior management techniques
  Psychopharmacological agents
    Stimulant agents - ADHD
    Neuroleptics - self-injurious behavior
    SSRIs - anxiety & depression
C) Supportive

*normalising and mainstreaming-integrate the children in society and discourage institutionalization.*

- Early stimulation
- Special schooling
- Vocational training
- Primary health care – nutrition, immunization
- Periodic evaluation
- Co-ordinate interdisciplinary services
  - Psychology
  - Speech & language
  - Physiotherapy
  - Audiology
  - Social work
d) **Parental counseling**

- Individualized approach based on severity, etiology, prognosis
- Breaking the news-phased manner

- ✓ Not to be done abruptly/should not use offending terms like madness.
  - ✓ Positive aspects of the disease should be discussed first, followed by the problems but do not hold the truth.

- Ensure full participation of family members with special support to the mother.
- Protect against possible physical or sexual abuse.
- Information regarding parent groups, support organizations and NGO.
- Counseling about risk of recurrence and prevention of disease in future pregnancies.
MANAGEMENT OF MR  CONT'D

E) Prevention - Primary:
   —For all
     • iodine and iron supplementation
     • Prevent exposure against environmental toxin
     • Improve socioeconomic status
     • Avoid consanguinity
   —Pregnant mothers
     • Safe motherhood years 20-35 yrs
     • Periconceptional folate
     • Good antenatal care/perinatal care
   —Children
     • Routine immunization
MANAGEMENT OF MR

—Secondary Prevention
  • Early diagnosis and treatment of curable illnesses - hypothyroidism, galactosemia
  • Prenatal diagnosis -
    • chorionic villus sampling
    • Amniocentesis
    • Cord blood sampling
  – Genetic counseling - exact etiology/empiric risk figures*
—Tertiary prevention
  – Early rehabilitative interventions and support.
### MANAGEMENT OF MR (CONTD)

<table>
<thead>
<tr>
<th>Etiology established</th>
<th>Recurrence risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>A) Down’s syndrome</td>
<td></td>
</tr>
<tr>
<td>- Non dysjunctional&amp;</td>
<td></td>
</tr>
<tr>
<td>denovo translocation</td>
<td>1%</td>
</tr>
<tr>
<td>- Inherited translocation</td>
<td></td>
</tr>
<tr>
<td>• Mother carrier</td>
<td>10%</td>
</tr>
<tr>
<td>• Father carrier</td>
<td>5%</td>
</tr>
<tr>
<td>B) Fragile X syndrome</td>
<td></td>
</tr>
<tr>
<td>- Female carrier</td>
<td>50% (affected males)</td>
</tr>
<tr>
<td>- Affected males</td>
<td>100% (carrier females)</td>
</tr>
<tr>
<td>C) Autosomal Recessive Disorder</td>
<td>25%</td>
</tr>
<tr>
<td>D) Autosomal Dominant Disorder</td>
<td>50%</td>
</tr>
</tbody>
</table>
## Management of MR (Contd)

<table>
<thead>
<tr>
<th>Etiology not established</th>
<th>Recurrence Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Isolated case</td>
<td>1:35</td>
</tr>
<tr>
<td>Isolated case (parents consanguinous)</td>
<td>1:7</td>
</tr>
<tr>
<td>1 affected parent (either sex)</td>
<td>1:10</td>
</tr>
<tr>
<td>1 affected parent &amp; 1 affected child</td>
<td>1:5</td>
</tr>
<tr>
<td>2 siblings (either sex)</td>
<td>1:4</td>
</tr>
<tr>
<td>2 affected parents</td>
<td>1:2</td>
</tr>
<tr>
<td>Affected male (affected maternal uncle)</td>
<td>1:2 (males)</td>
</tr>
</tbody>
</table>
REFERENCES


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• IAP Textbook of Pediatrics, R Parthasarthy, 4th Edition

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www.findmeacure.com
THANK YOU