Diagnostic Approach for Mental Retardation

Nam, Sang-Ook M.D.
Department of Pediatrics
School of Medicine
Pusan National University

Mental Retardation

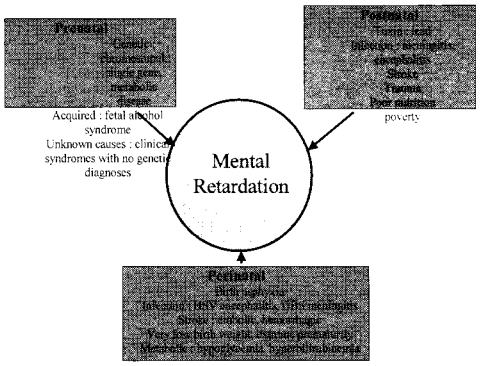
- **Definition**
- **2002 American Association on Mental Retardation (AAMR)**
  - A disability characterized by a significant limitation both in intellectual functioning and in adaptive behavior as expressed in conceptual, social, practical, and adaptive skills
  - Originates before the age of 18

Mental Retardation

- Heterogeneous in etiology
  - Maternal vs birth factor
    - e.g. maternal age, low birth weight
  - Prenatal, perinatal, postnatal
- IQ < 2 SD below the man (IQ < 70)
  - Prevalence 2.2%
  - Sex : Male to female - 1.4:1.0
  - Race : Black > white

Qualitative description of IQ

SCORE	CLASSIFICATION	PERCENT INCLUDED IN THEORETICAL NORMAL CURVE
≥130	Very superior	2.2
120-129	Superior	6.7
110-119	High average	16.1
90-109	Average	50.0
80-89	Low average	16.1
70-79	Borderline	6.7
≤ 69	Extremely low	2.2



Evaluation of the patients

- **History**
  - Family history
  - Birth history
  - Developmental milestone
  - Coexisting medical problem
  - Past medical history
- **Physical examination**
  - Careful observation
  - Facial and gross morphology
  - Body measurement
  - Focal neurologic deficit
  - Developmental assessment
- **Laboratory testing**

History

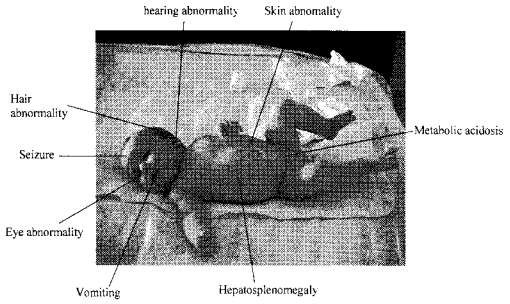
- **Family history**
  - Similar patient : Metabolic, genetic, CNS disease
  - Early postnatal death
  - Consanguinity
- **Birth history**
  - Maternal history : gestation, delivery
  - Mode of delivery
  - Gestational age
  - Body measurement at birth
  - Apgar scores
  - Perinatal or postnatal illness
  - Duration of postnatal hospital stay

History

- **Developmental milestone**
  - Motor skill/language skill
  - Timing of initial parental concern
  - Regression
  - Current functional level
- **Coexisting medical problems**
  - Seizure, feeding difficulty
  - Recurrent or progressive illness
- **Past medical history**
  - Chronic medical conditions
  - hospital admissions
  - Surgery
  - Medication
- **Previous evaluation or treatment**

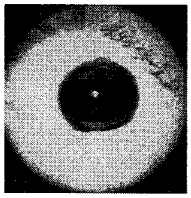
Physical examination

- **Careful observation**
  - During history taking
  - Skill, dexterity, inquisitiveness, behavior, interaction
  - Autistic behavior : Screening for autism or language disorder
- **Body measurement**
  - Weight, height, head circumference, fontanelles
  - Patients and family members
- **Dysmorphic features** : face, trunk, extremities
- **Organomegaly : hepatosplenomegaly**
- **Skin abnormality**
- **Spine** : dermal sinus, hair tuft in sacral region
- **Neurologic examination**
  - Hearing
  - Vision
  - Focal neurologic deficits
  - Developmental screening test



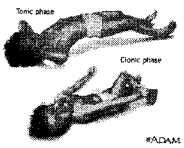
Eye abnormality

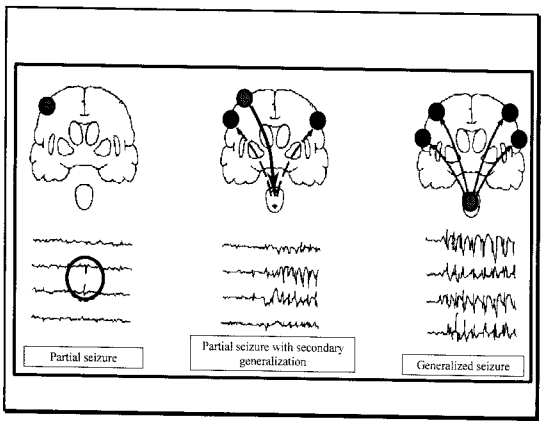
- Cataract
- Cherry-red spot in macular area
- Chorioretinitis
- Corneal ulcers
- Dislocated lenses
- Glaucoma
- Nystagmus
- Photophobia
- Retinitis pigmentosa
- Vertical supranuclear gaze palsy



Seizures

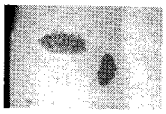
- Seizure type
  - Partial seizure
  - Generalized seizure
  - Unclassified seizure
    - Neonatal seizure
- Acute vs chronic seizure
- Idiopathic vs cryptogenic or symptomatic





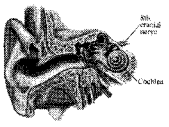
Skin abnormalities

- Café au lait spots
- Depigmented nevi
- Eczema
- Linear nevus
- Malar flush
- Photosensitivity
- Rash
- Synophrys



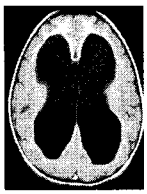
Hearing abnormalities

- Deafness
  - Conduction deafness
  - Sensorineural deafness
- Hyperacusis



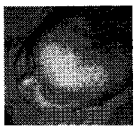
Vomiting

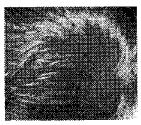
- Metabolic disease
  - Hyperammonemia
  - Hyperglycinemia
  - Hyperlysinemia
  - Hypervalinemia
  - Maple syrup urine disease
- Mitochondrial diseases
  - Lactic acidosis
  - MELAS syndrome
- Increased intracranial pressure



Hair abnormalities

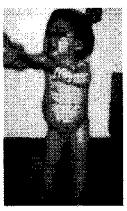
- Fine hair
- Friable and tufted hair
- Loss of scalp hair
- Premature gray hair
- White hair





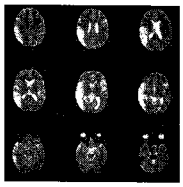
Hepatosplenomegaly

- Argininosuccinic aciduria
- Gaucher's disease
- GM1 gangliosidosis
- Glycogen storage disease type I, III
- Hydroxykynureninuria
- Hyperpipecolatemia
- Mucopolysaccharidoses
- Neuronal ceroid lipofuscinosis
- Niemann-pick disease



Metabolic acidosis

- Ketotic hypoglycemia
- Lactic acidosis
- Mitochondrial encephalopathy
- Maple syrup urine diseaes
- Methylmalonic acidemia
- Propionic acidemia
- Methionine malabsorption syndrome
- 5-Oxoprolinuria



Other abnormalities

- Movement disorders
  - Ataxia
  - Choreoathetosis
  - Dystonia
  - Tremors
- Odors
- Short statures
- Fat pad distribution
- Peripheral neuropathy

Laboratory tests

- **Cytogenetic test**
- **Neuroimage**
- **Metabolic study**
- **EEG**
- **Visual or auditory Evoked potential**

Laboratory tests

- **Cytogenetic study**
  - High resolution banding karyotype
    - FISH (Fluorescence in situ hybridization)
      - routine or specific syndrome
    - Subtelomeric probes
  - Globally delayed or retarded patients
  - Without an apparent cause after history and P/E
  - With or without dysmorphic features

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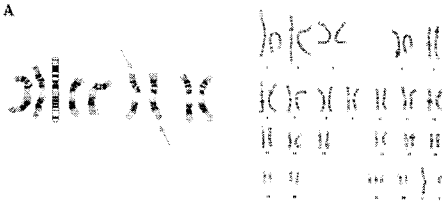
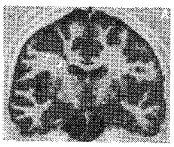


High resolution Low resolution

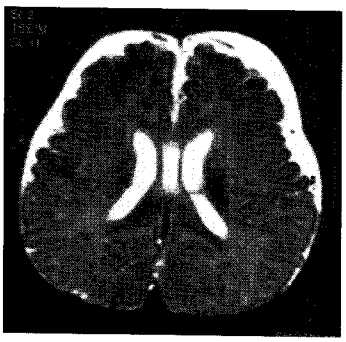
Subtelomeric probes

Laboratory tests

- **Neuroimage**



  - MRI > CT
  - Cerebral dysgenesis
  - Acquired CNS injury of various causes
  - Disturbances in white matter maturation



Laboratory tests

- **Metabolic study**
  - Family history
  - Parental consanguinity
  - Developmental regression
  - Dysmorphology involving nonectodermal origin
  - Possible white matter involvement

Laboratory tests

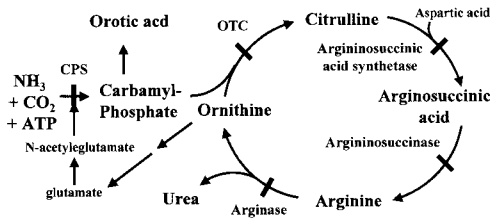
Metabolic study

CBC, LFT, BUN/Cr, ABGA, Na/K/Cl
Glucose, serum ammonia
Serum &/or CSF lactate/pyruvate
Ca/Mg, uric acid, PBS, U/A
Serum & Urine amino acid, Urine organic acid
lipid profile, carnitine, ketone body

Enzyme function study

DNA study

Urea cycle : Pathway of ammonia disposal and ornithine metabolism



CPS : carbamylphosphate synthetase
OTC : ornithine transcarbamylse

Laboratory tests

- **EEG**
  - Suspected coexisting paroxysmal disorder
  - Evidence of language regression or behavioral abnormalities
- **Visual or auditory Evoked potential**
  - Assessing the integrity of the visual and auditory system
  - In the young uncooperative child

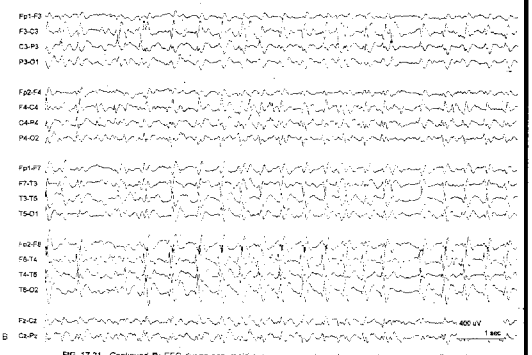
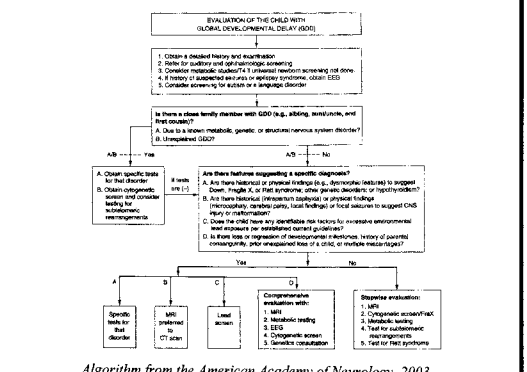


FIG. 17.21 *Continued.* **B:** EEG during non–rapid-eye-movement sleep shows nearly continuous spike-and-wave discharges. Although broadly distributed bilaterally, they are maximal over the right temporal region. TC, 0.1 second; HFF, 70 Hz.



Algorithm from the American Academy of Neurology, 2003