Mental retardation

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Mental retardation

Intellectual disability

Cognitive impairment

Global learning disability

Mental retardation (MR) is one of the more common developmental disabilities.

It can be idiopathic and challenging to recognize in normal-appearing children who have developmental delays.

Conversely, MR can be easily recognized when the child presents with dysmorphic features associated with a known genetic MR disorder.

Mental retardation currently is defined by the American Association on Mental Retardation as

"significantly sub-average general intellectual functioning accompanied by significant limitations in adaptive functioning in a least two of the following skills areas:

communication,
self-care,
social skills,
self-direction,
academic skills,
work,
leisure,
health and/or safety.

These limitations manifest themselves before 18 years of age."

Mental retardation	IQ
Mild	50-69
Moderate	35-49
Severe	20-34
Profound	< 20

BNO-10

Etiology and pathogenesis

1. Organic
2. Polygenic
3. Environmental

Organic etiology

severe mental retardation	55-75 %
mild mental retardation	10-25 %

Etiology

Prenatal:
- genetic defects (> 1000)
- chromosome disorders
- intrauterine infections
- chronic alcohol ingestion, drug

Perinatal:
- prematurity
- extreme low birth weight
- hypoxia
- intracranial hemorrhage
- CNS infections

Postnatal:
- transient hypoxia (drowning, cardiac arrest)
- post-traumatic
- CNS infection
- environmental, psycho-social deprivation
- lead

Multifactorial etiology !

Screening and early diagnosis

History !

PHYSICAL EXAMINATION:

CLUES TO CERTAIN CONDITIONS ASSOCIATED WITH DEVELOPMENTAL DISABILITY

Growth parameters

- Head circumference
  - Microcephaly (< 3rd percentile of age)
  - Macrocephaly (> 97th percentile for age): hydrocephalus, lysosomal storage disorders
- Stature
  - Short stature: Turner sy. (ass. With learning disabilities)
  - Tall stature: Sotos sy.
- Obesity
  - Prader-Willy syndrome
  - Beckwith-Wiedemann sy.

Skin

- Café au lait spots, neurofibromas
  - Neurofibromatosis (learning disability)
- adenoma sebaceum
  - tuberous sclerosis (ment. retard. 50 %)
- facial port wine haemangioma
  - Sturge-Weber sy. (ment. retard.)
- nail hypoplasia or dysplasia : fetal alcohol sy.

Organomegaly

- Neurodegenerative disorders
  - MPS
  - gangliosidoses
  - Gaucher, Niemann-Pick, Zellweger sy.
  - glycogen storage disease
  - galactosaemia

Genitalia

Macroorchidism

Fragile X sy.

Hypogonadism

Prader-Willi sy.

Movement patterns

Hand preference

Dysmorphic features, congenital malformations

CO-MORBIDITIES

Behavior challenges

Psychiatric disorders

Seizures

Sensory impairments

Motor impairments

Sleep disorders

Recurrent vomiting

Autism

Examinations

- Cytogenetic/molecular genetic examinations
- Metabolic work-up
- Imaging (UH, CT, MRI, SPECT, PET)
- Histology
- Evoked potentials
- EEG
- Tests (Bayley, Brunet-Lezine, Budapest-Binet)

Down syndrome

Incidence 1:600-800
IQ 40-60

Characteristic morphologic signs

flat facial profile,
upward slanted palpebral fissures,
abnormal, small ears,
single palmar crease,
generalized hypotonia,
hyperflexibility of joints,
Congenital heart disease (40 %)

Fragile X syndrome

1/4000 males, 1:6000 females
IQ 25-65

FMR1 gene

Characteristics:
Large head, long and narrow face,
large ears,
macroorchidism,
autistic signs, attention deficit,
hyperactivity,
behavioral problems,
delayed speech development

Williams syndrome

Cause:

microdeletion
chromosome 7

Prevalence: 1 : 20 000

Signs and symptoms:

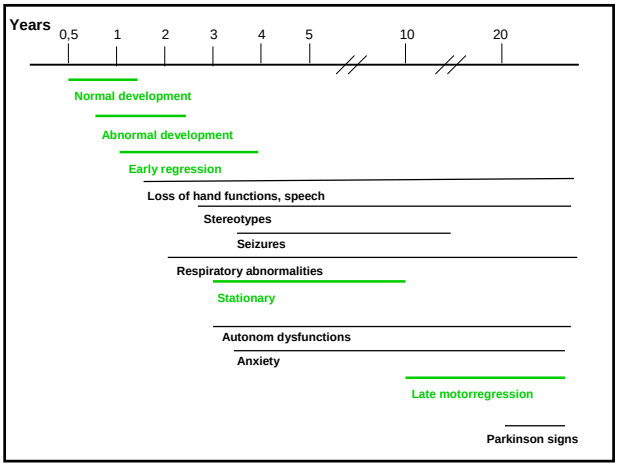
in the first year of life feeding difficulties
crying
constipation
abnormal teeth
congenital heart anomalies
hypercalcaemia
short stature (152,4-167,6 cm)

Angelman syndrome

Deletion on the chromosome 15
Mental retardation, ataxia, stiff puppet-like gait, happy puppet, laughing, dysmorphic face
CT, MRI not characteristic
EEG characteristic

Rett syndrome

Prevalence: 1:15.000



Fetal alcohol syndrome

1-3/1000 children

Family history !

IQ 20-120 (mean 65)

Etiologic diagnosis of mental retardation

50 %

IDENTIFICATION RATES OF DEVELOPMENTAL DISABILITIES

	Prevalence cases/1000	MD is first to make dg	Mean age at identification (mo)
Ment. ret.	25	76	39
Learning disab.	75	12	69
ADHD	150	44	59
CP	2-3	99	10
Visual disab.	0,3-0,6	87	55
Hearing disab.	0,8-2	64	39

Treatment

Prognosis

Early intervention

Rehabilitation

Prevention