APPROACH TO DEVELOPMENTAL DELAY

By Dr. Nazia Mukhtar

Definition:

- Delay in the acquisition of developmental skills beyond limit age.
- Types:

  Isolated

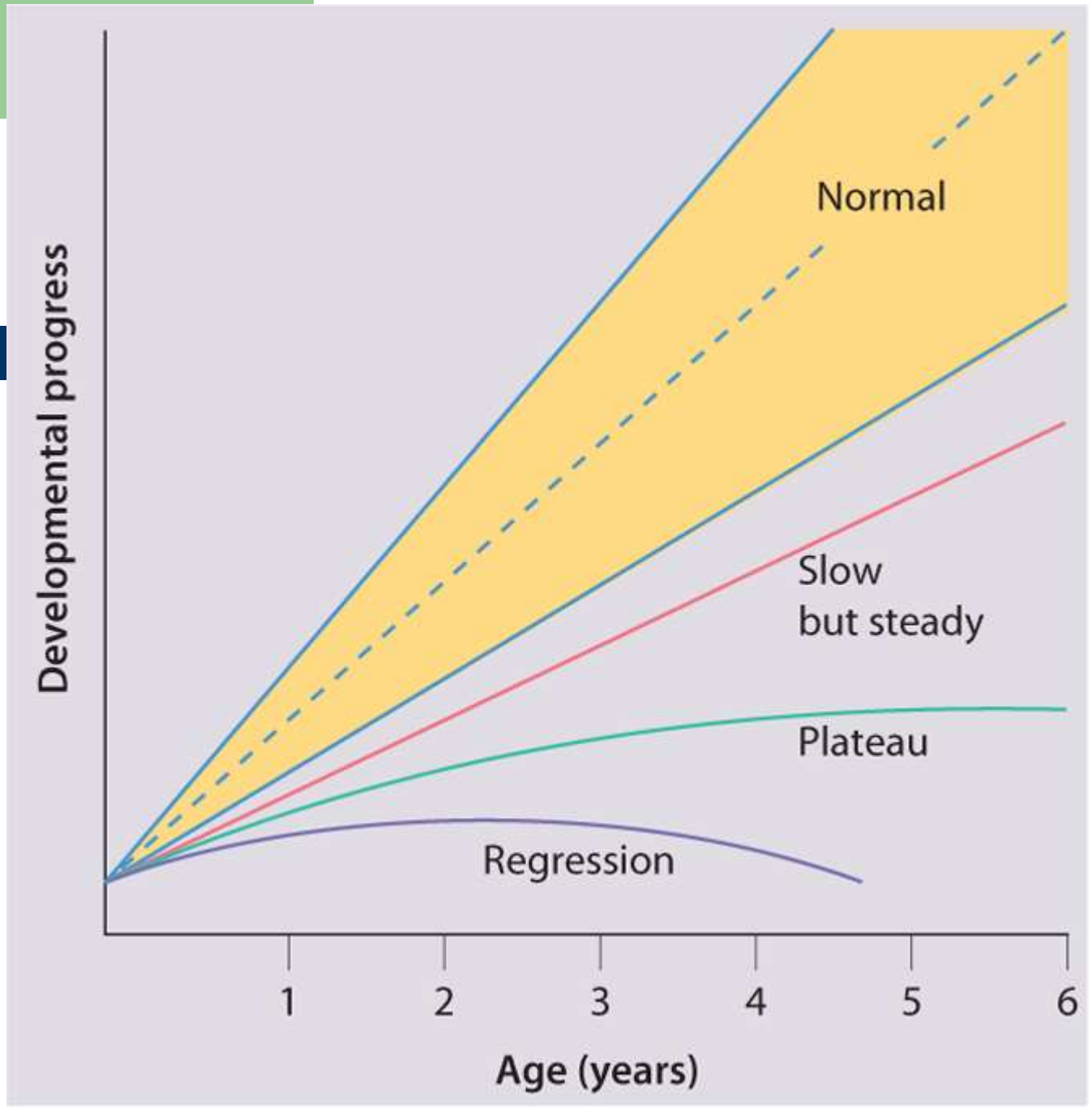
  Global

ISOLATED DELAY

- Gross motor delay
- Impaired speech and language
- Impaired hearing
- Visual impairment
- Impaired cognition/learning difficulties
- Impaired social skills / Autism

GLOBAL DELAY

- Delay in acquisition of two or more skill fields.
- Presents within first two years of life



CAUSES:

- Gross motor delay:
  - Cerebral palsy
  - Neuromuscular disorders

Static Global delay:

- Chromosomal disturbances
- Maternal alcohol or drug abuse
- Cerebral malformations
- Hypoxic ischemic encephalopathy
- Intracranial hemorrhage
- Infection
- Brain tumor

Progressive global delay:

1. NEUROCUTANEOUS DISORDERS
   - Tuberous sclerosis
   - Neurofibromatosis

2. METABOLIC BRAIN DISORDERS

A. Gray matter disorders(without visceral storage)

- Tay Sachs disease
- Lipofuscinosis
- Alpers disease
- Menkes disease
- Rett syndrome

B. Gray matter diseases (with visceral storage)

- GM-1 gangliosidosis
- GM-2 gangliosidosis(sandhoff)
- Niemann-Pick disease
- Gaucher disease

C. Diseases affecting white matter:

- Galactosemia
- Phenylketonurea
- Maple syrup urine disease
- Homocystinuria
- Urea cycle defects

D. Diseases affecting white&Gray matter

MPS I,II,III,VII

Mitochondrial disorders

Peroxisomal disorders

3. Congenital hypothyroidism
4. Malnutrition
5. Chronic illness

CEREBRAL PALSY

- Disorder of tone and posture due to nonprogressive lesion of the developing brain
- Often associated with impaired speech,hearing and cognition.

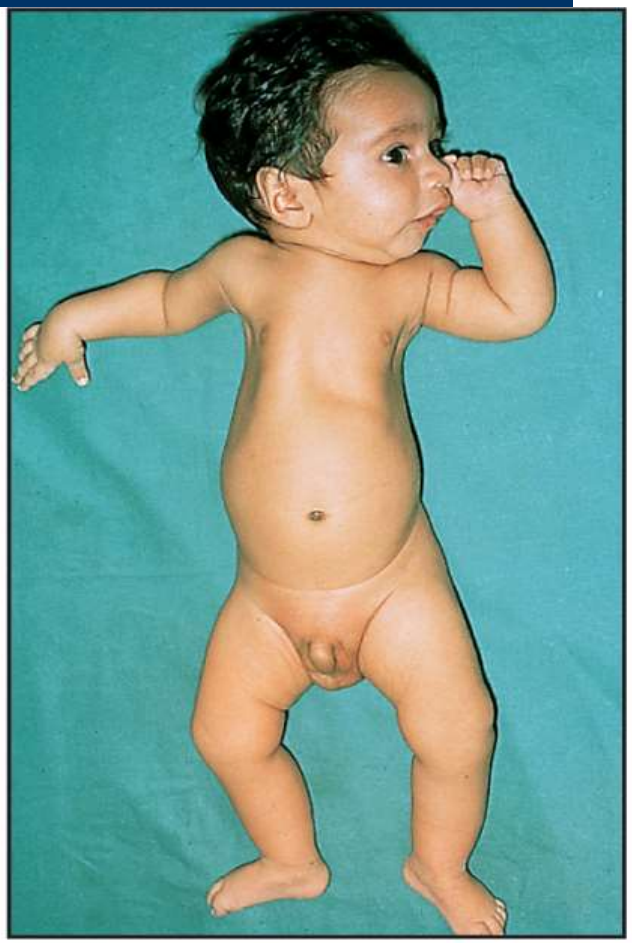
- Birth history: asphyxia,prematurity
- Developmental delay
- Seizures
- Microcephaly

- Quadriplegia
- Paraplegia
- Hemiplegia
- Diplegia

- Spasticity
- Hypotonia & ataxia
- Dyskinetic

Neuromuscular disorders:

- Anterior horn,nerves,NMJ & muscle
- SMA
- Myotonias
- Myopathies
- Mysthenia gravis

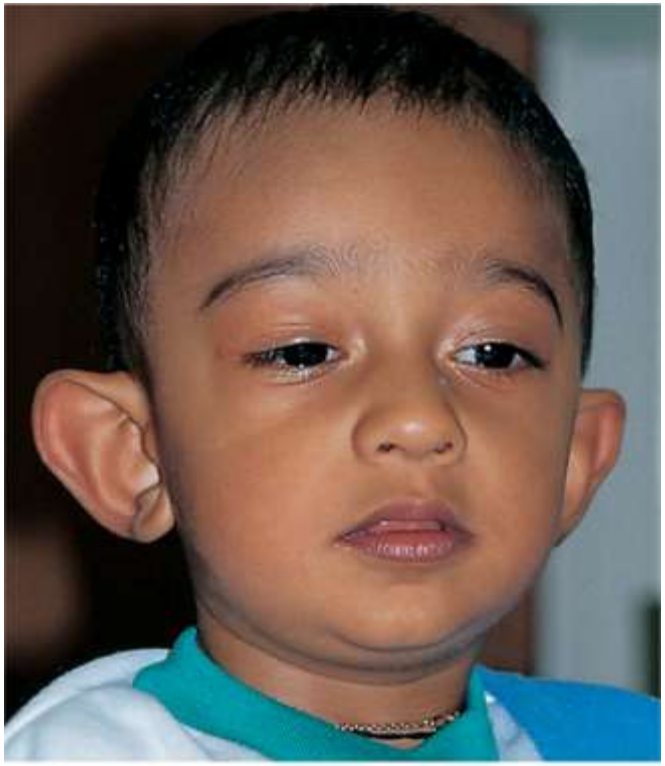


Chromosomal disturbances

Most common---
trisomy 21
Typical facies
Hypotonia
Congenital heart
disease
Karyotyping

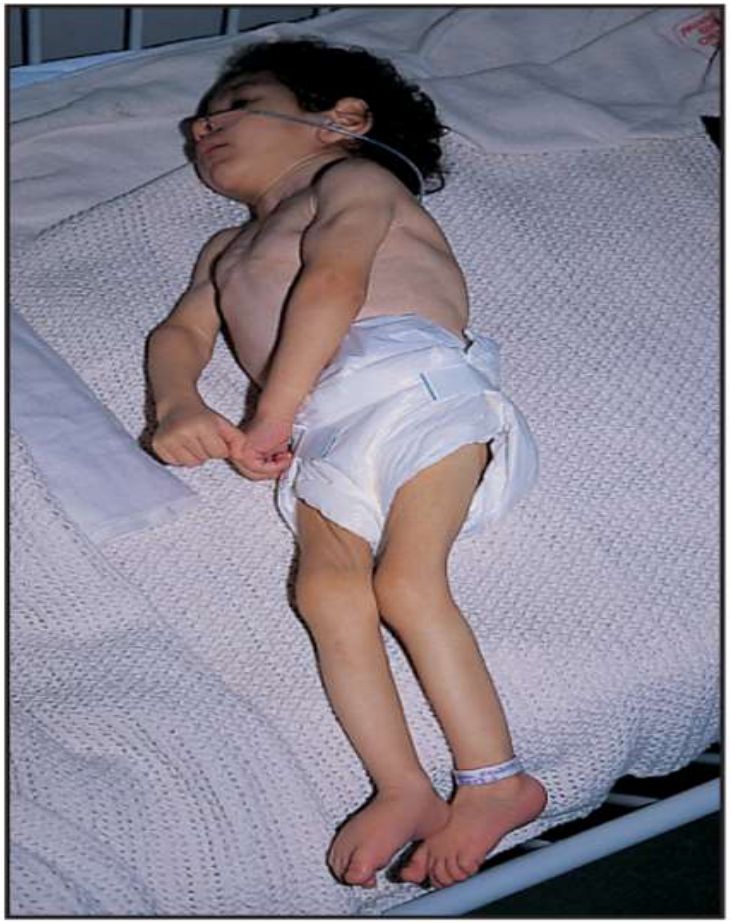
- Edward syndrome(trisomy 18)
- Developmental delay
- Rocker bottom feet
- Finger overlap

- Fragile X syndrome
- Prominent forehead
- Large ears
- Macrocephaly
- Large testes
- Karyotyping



Hypoxic ischemic encephalopathy

- Motor ,cognition ,seizures
- Perinatal asphyxia
- Cardiopulmonary disease
- Meningitis
- Head trauma
- Status epilepticus
- CT/MRI



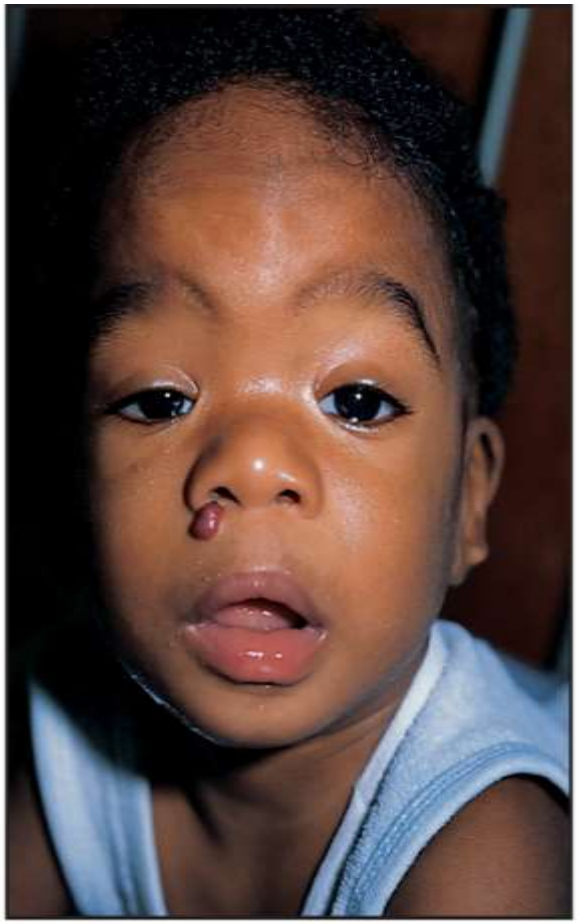
Maternal drugs/alcohol

- Alcohol ---- fetal alcohol syndrome
- Cocaine ---cerebral hemorrhage & infarction
- Anticonvulsants ---- cerebral malformations& dev delay

- FETAL ALCOHOL SYNDROME:

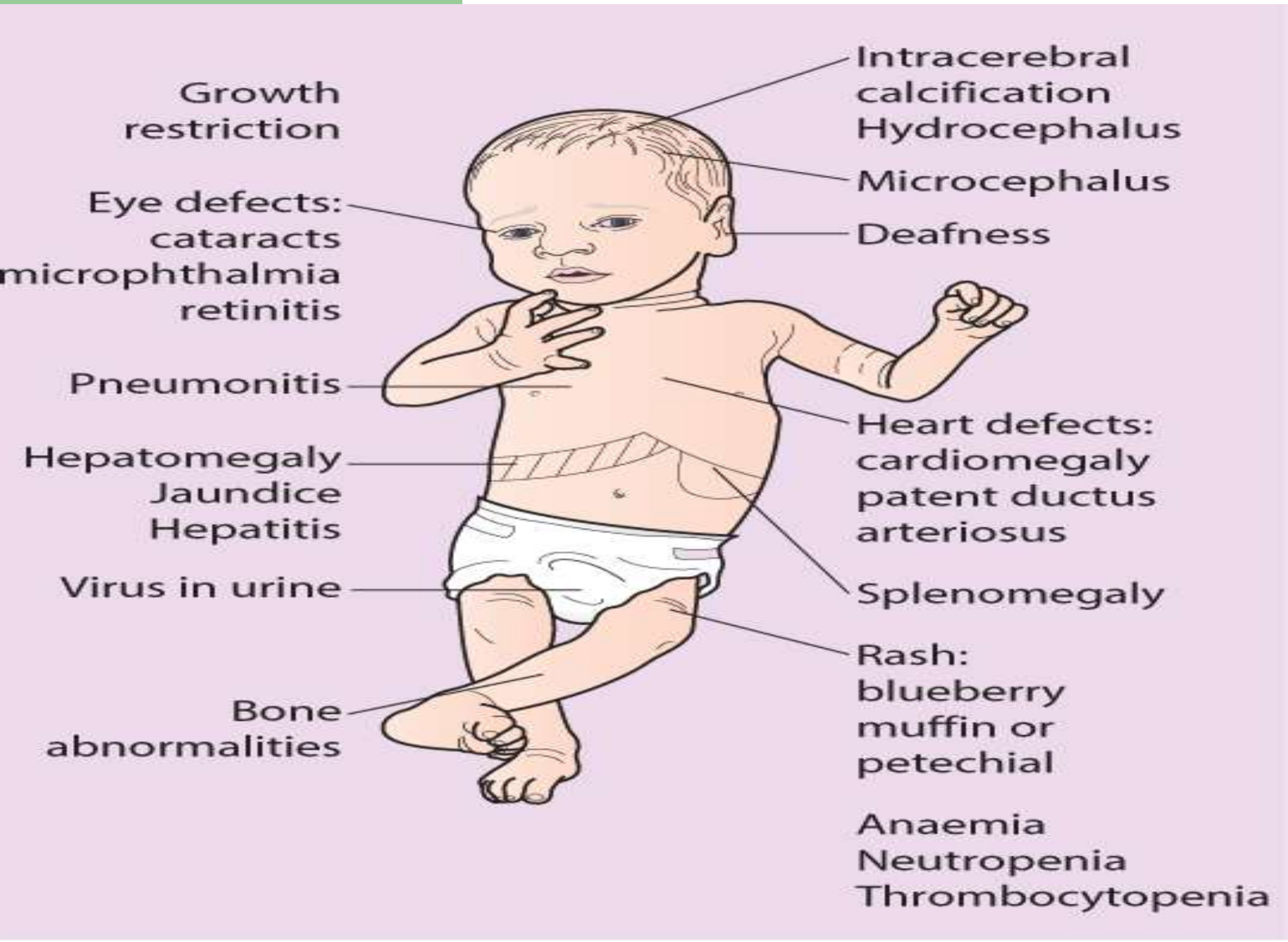
  microcephaly,thin upper lip,flattened midface.

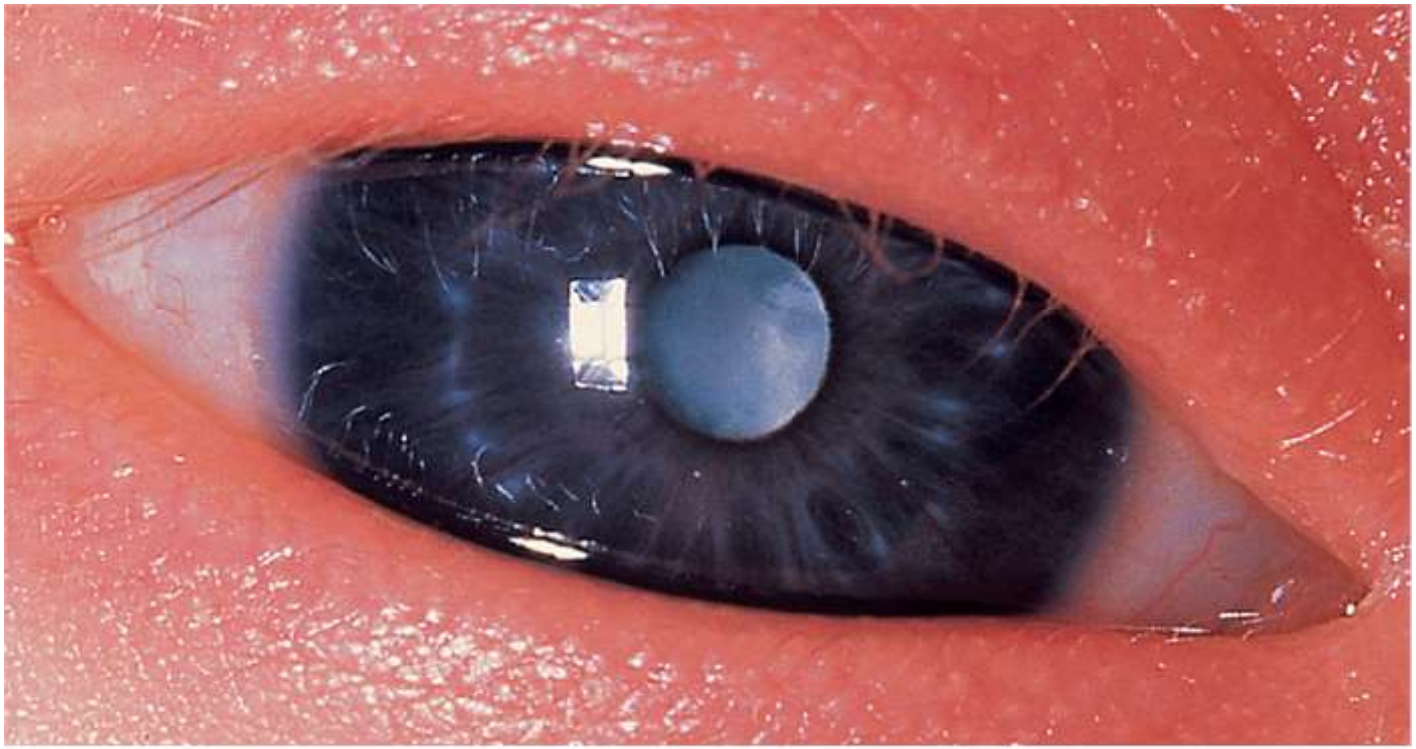
  brain dysfunction,dev delay,poor memory



Congenital infections

- Toxoplasmosis
- Rubella
- CMV
- Herpes simplex
- Syphilis





Brain infections

- Bacterial /tuberculous
- Herpes encephalitis
- Seizures
- Motor&cognitive impairment
- language dysfunction
- Deafness& blindness.

NEUROCUTANEOUS SYNDROMES

- Neurofibromatosis

  Autosomal dominant…1 in 3000

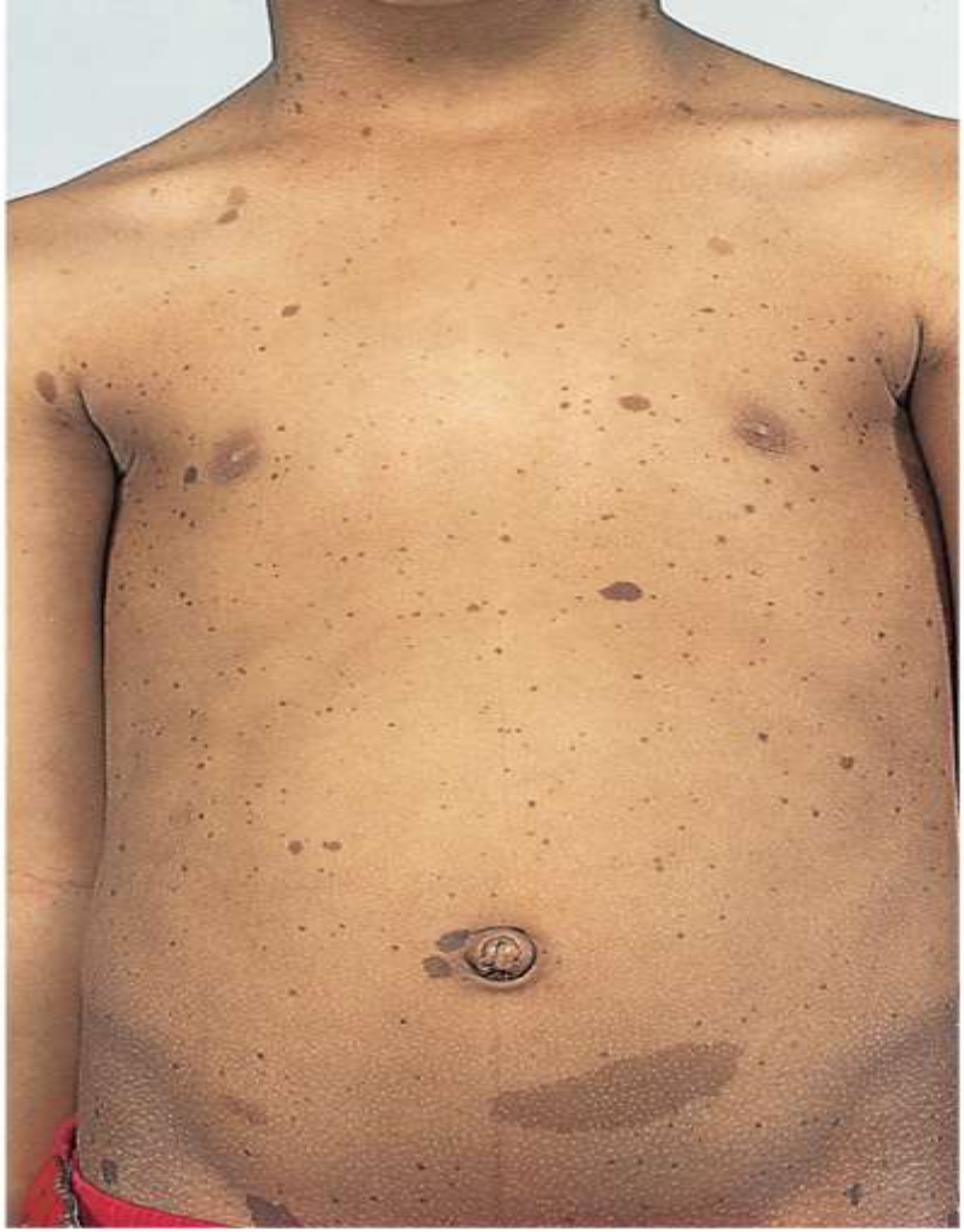
  Diagnostic criteria(2 or more)

  6 or more café-au-lait spots >5mm

  >1 neurofibroma

  axillary freckles

- Optic glioma
- Lisch nodule of iris
- Sphenoid dysplasia
- First degree relative with NF1.



Tuberous sclerosis

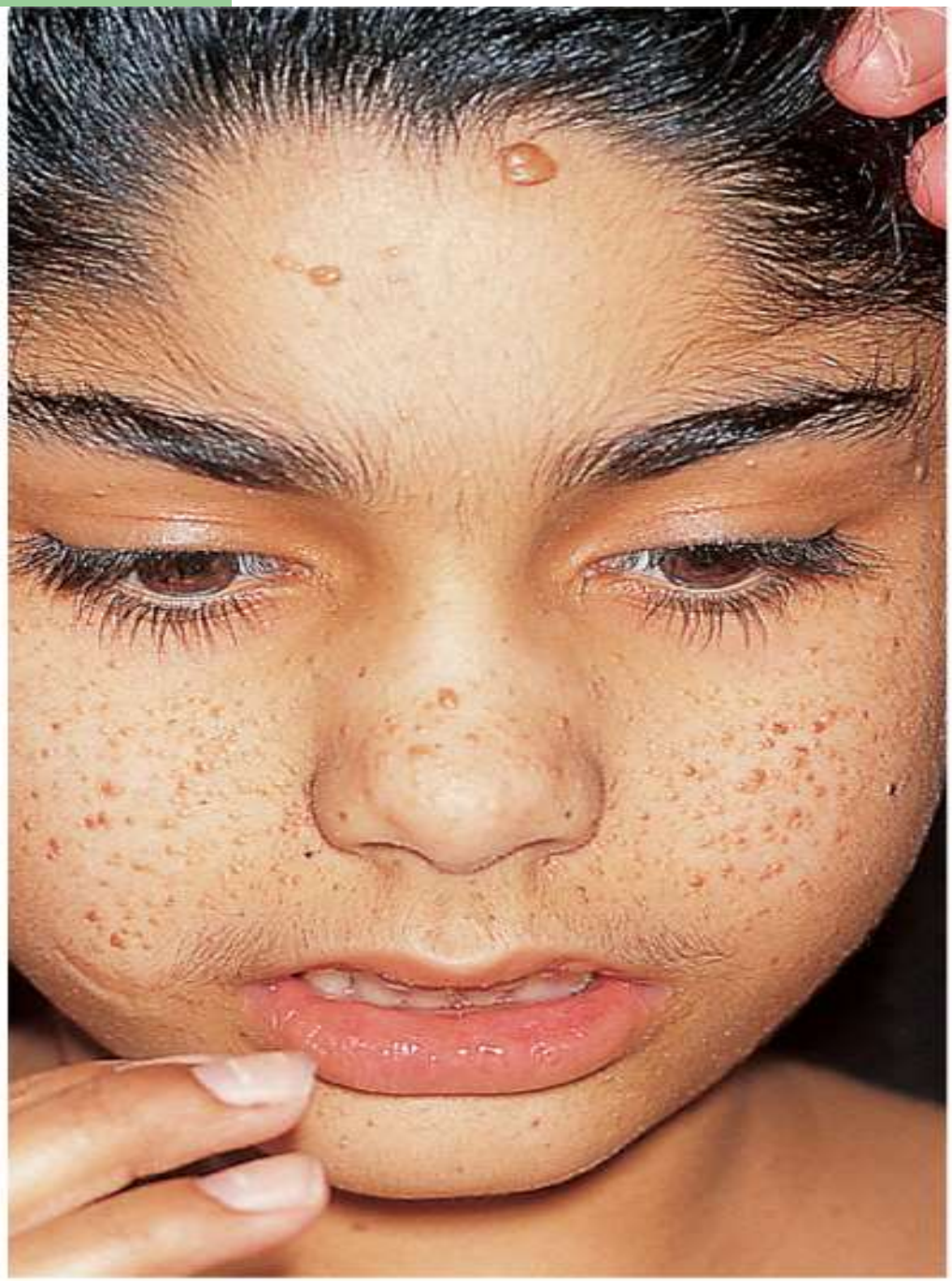
- Autosomal dominant…….1 in 9000
- Cutaneous features:

  ash leaf patches(depigmented)

  shagreen patches(roughened skin)

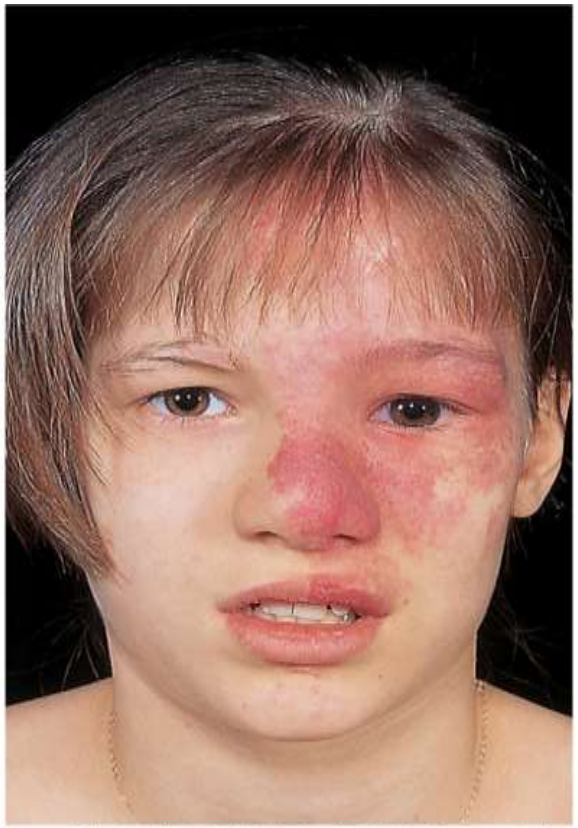
  adenoma sebaceum on face

  (angiofibromata)



Sturge webber syndrome

- Port wine stain
- Seizures
- Learning disability
- Hemiplegia



GM2 Gangliosidosis(tay sach disease)

- Autosomal recessive
- Deficient Hexosaminidase
- Onset 3 – 6 mo
- Abnormal startle reaction
- Developmental delay
- Cherry red spot
- Enzyme analysis in cultured leukocytes

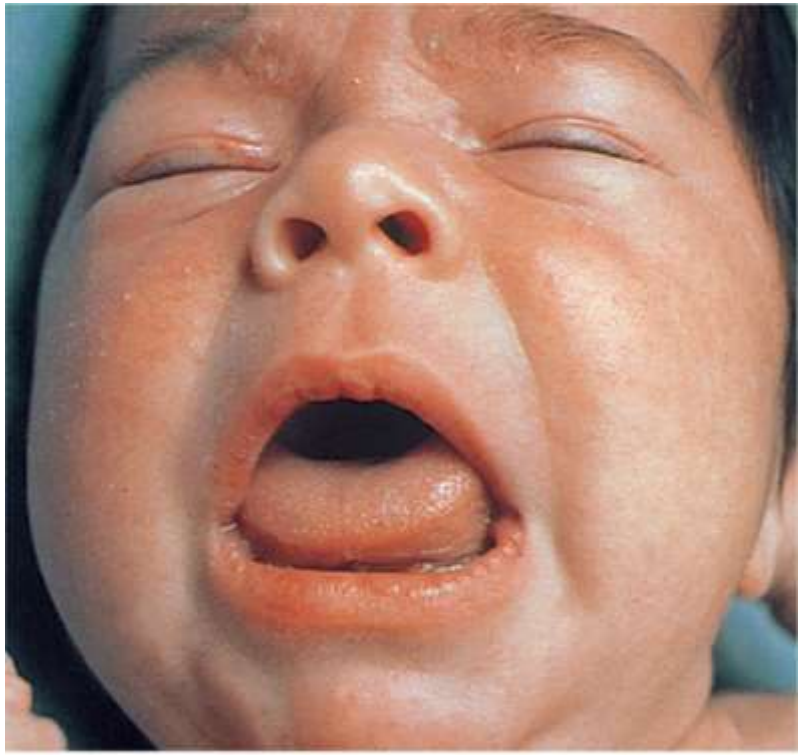
Mucopolysaccharidosis(I,II,III,VII)

- Lysosomal enzymes deficiency
- AR (except II…X linked)
- Coarse facial features,
- Hepatosplenomegaly
- Dysostosis multiplex
- Corneal clouding

(except II&III)

Congenital hypothyroidism

- Developmental delay
- Mental retardation
- Coarse features
- Neonatal jaundice
- Constipation
- Umbilical hernia



Diagnostic approach

HISTORY

- Antenatal history (maternal infections, cardiovascular diseases,vaccination)
- Birth history
- Prematurity
- Fever,jaundice
- Feeding

- FAMILY HISTORY:
  - consanguinity
  - other siblings
  - developmental delay
  - death of siblings

- Developmental history:

  milestones achieved&

  at which ages.

  unAchieved milestones

- Chronic complaints
  - fever
  - cough
  - vomiting
  - diarrhea
  - recurrent ARI
  - cyanosis& fatigability
- Seizures

EXAMINATION

- Anthropometry
- Developmental assessment

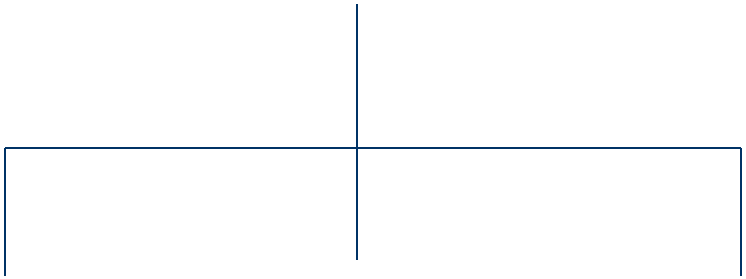
  (Denver dev. Screening test)

  hearing,vision,fine&gross motor,primitive reflexes.

- Neurological examination
- Abdominal examination
- Eye examination

  shape,opacities,cataracts,cherry red spot.
- Neurocutaneous stigmata

- Developmental Delay



speech motor global

- Delayed Speech
  - hearing loss
  - autism
  - pseudobulbar palsy

GLOBAL DELAY

DYSMORPHISM

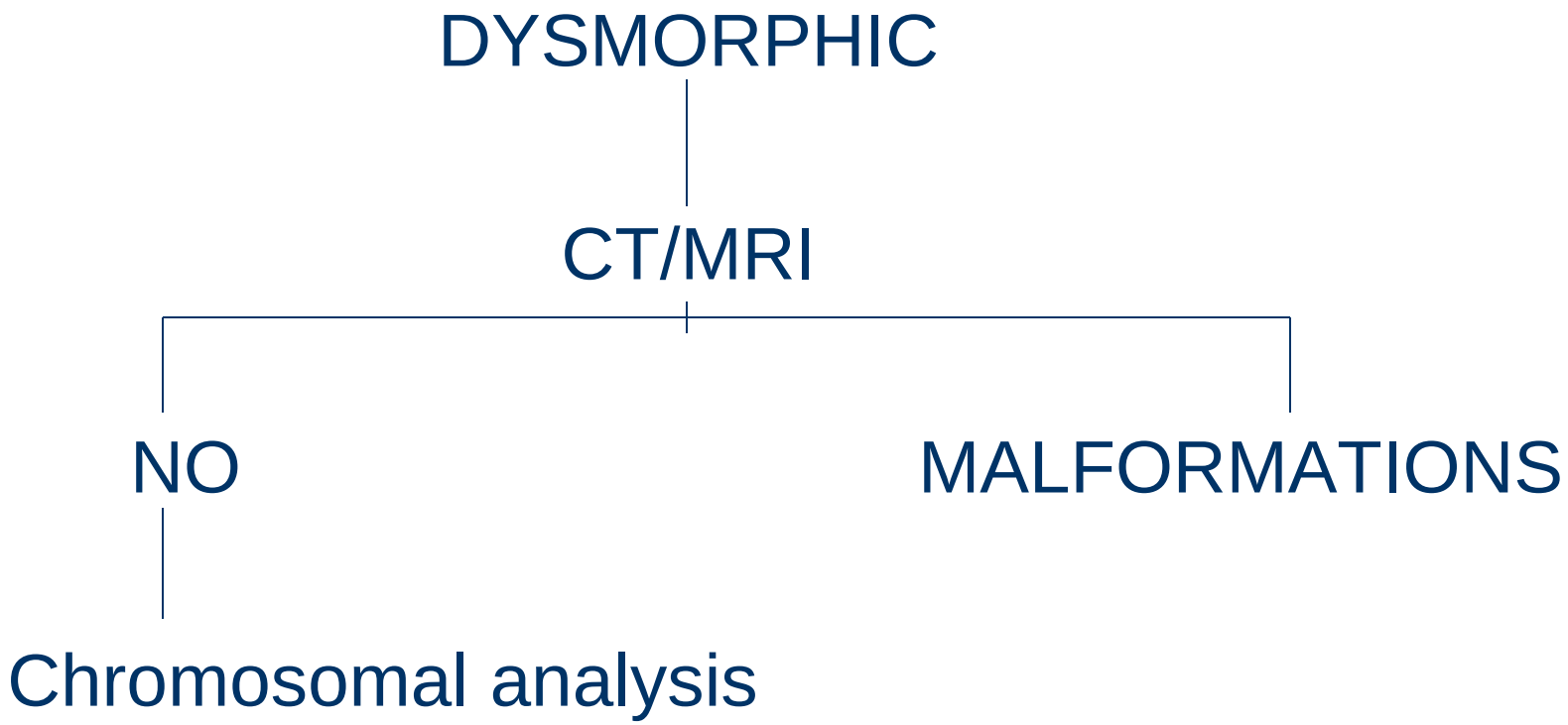
NO

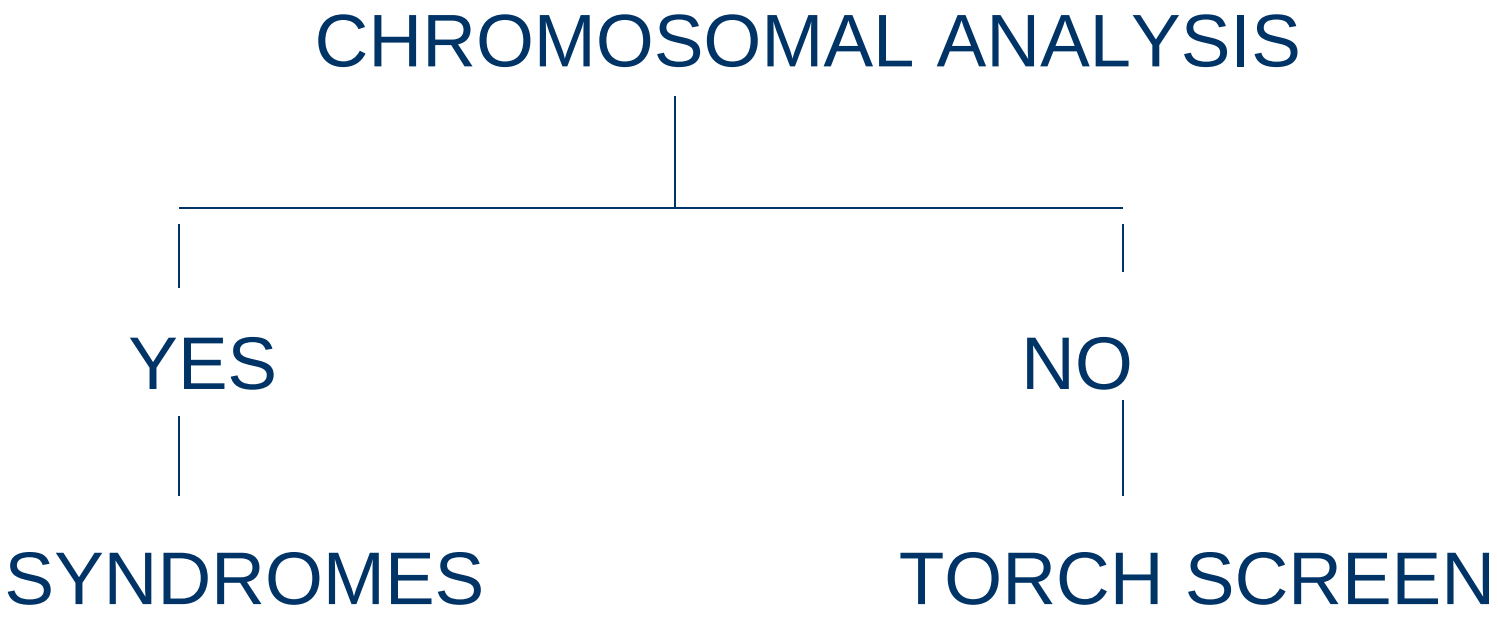
H/O birth asphyxia,infection,trauma,drugs

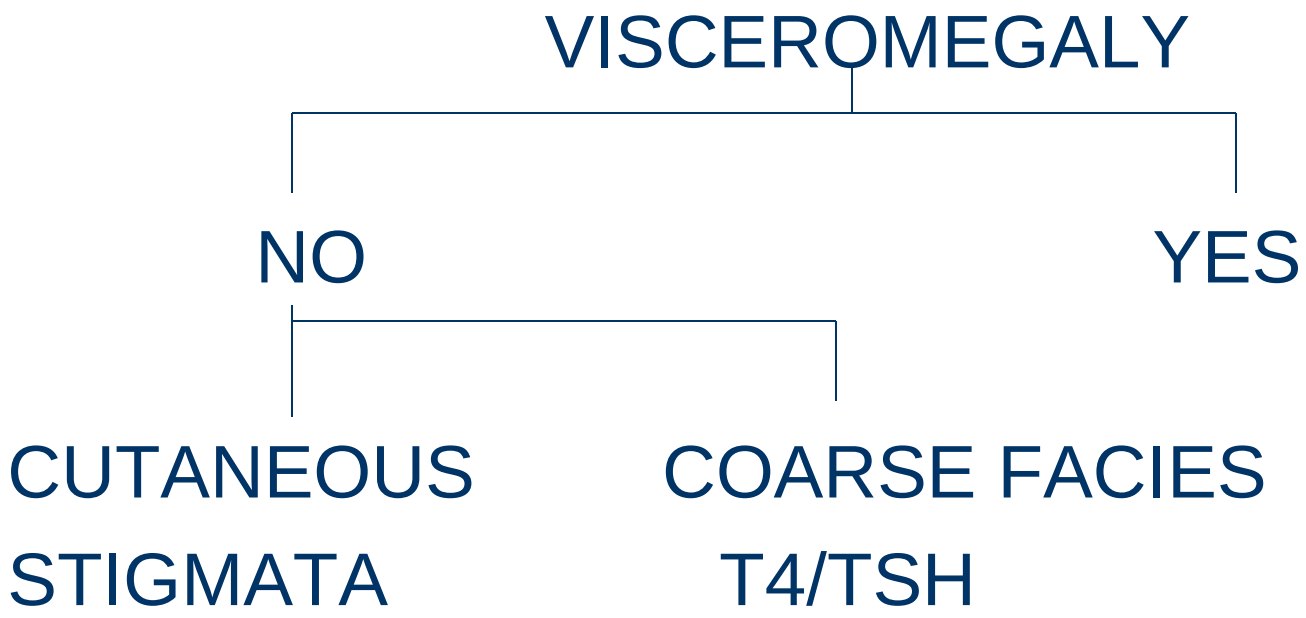
CP

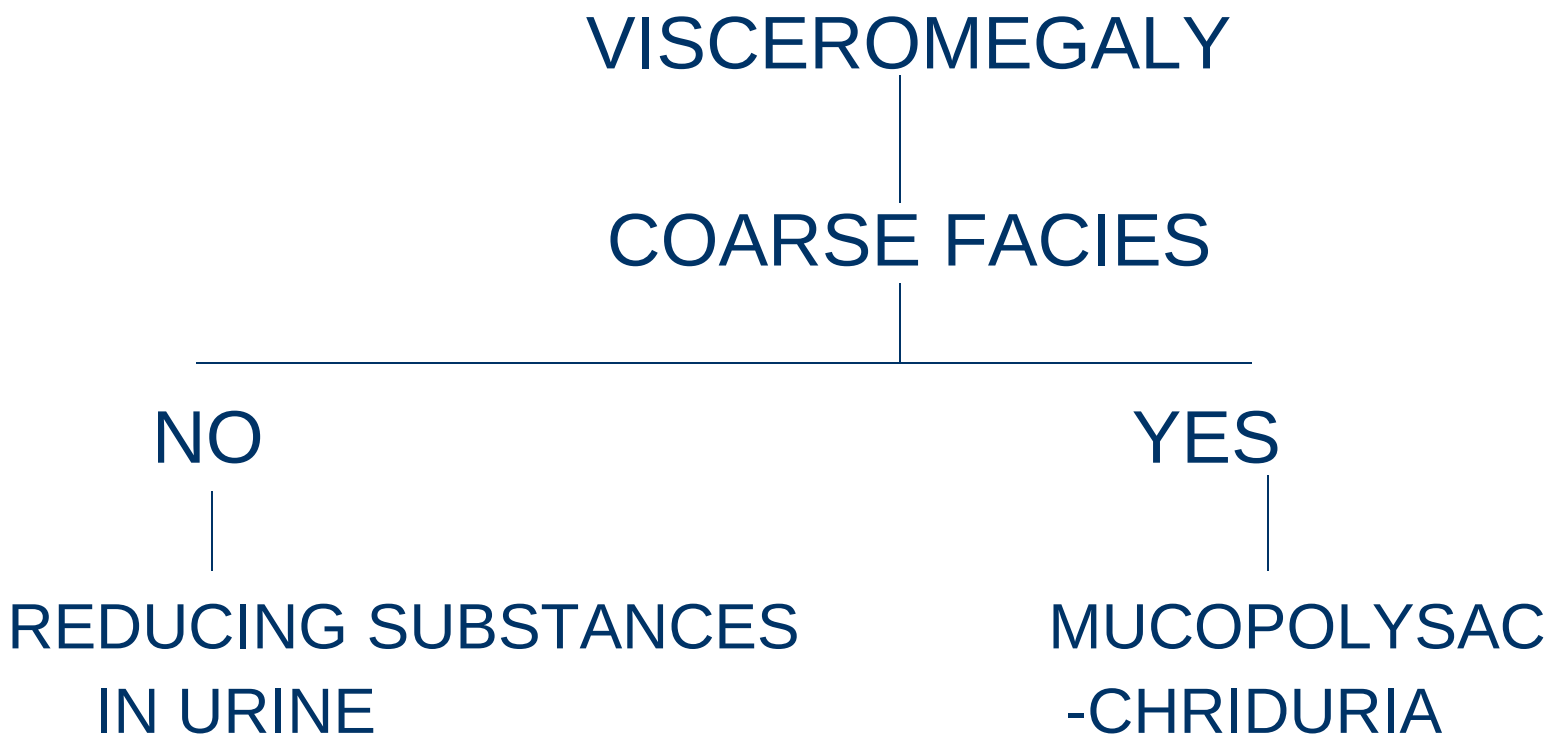
YES

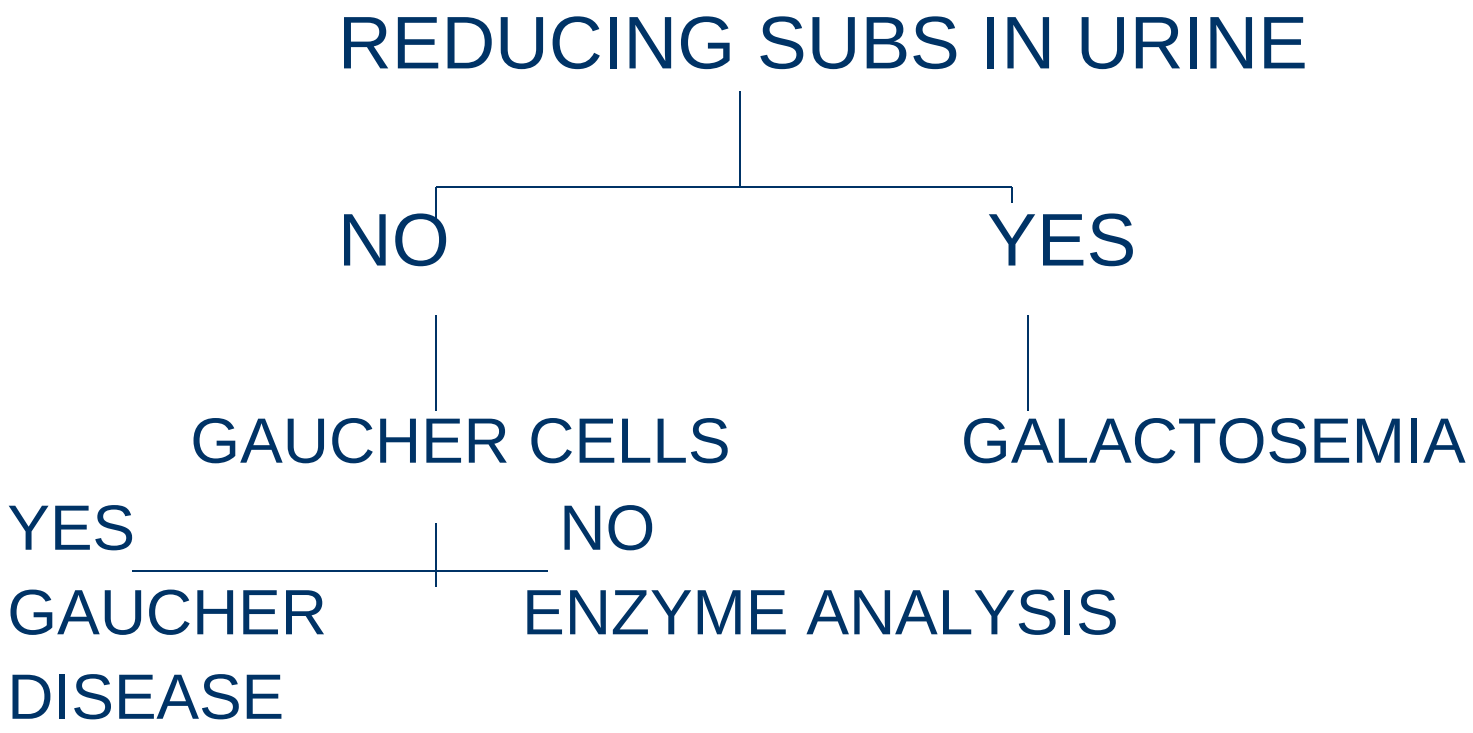
CT/MRI


DYSMORPHIC
CT/MRI
NO
MALFORMATIONS
Chromosomal analysis


CHROMOSOMAL ANALYSIS
YES
NO
SYNDROMES
TORCH SCREEN


VISCEROMEGALY
NO
YES
CUTANEOUS STIGMATA
COARSE FACIES
T4/TSH


VISCEROMEGALY
COARSE FACIES
NO
YES
REDUCING SUBSTANCES IN URINE
MUCOPOLYSAC -CHRIDURIA


REDUCING SUBS IN URINE
NO
YES
GAUCHER CELLS
GALACTOSEMIA
YES
NO
GAUCHER DISEASE
ENZYME ANALYSIS

Other tests

- Audiologic testing
- Opthalmologic evaluation
- EEG
- EMG ,Nerve conduction studies,muscle biopsy

THANK YOU.