Down Syndrome (Mongol Baby)

It is the most common chromosomal defect wherein there is an extra chromosome 21, so individuals with down syndrome have 47 chromosomes in total instead of the usual 46.

Causes

- It is caused by an error in cell division called nondisjunction. During meiosis one pair doesn't divide & the whole pair goes to one daughter cell. In trisomy 21, one cell has two 21st chromosomes instead of one, so the resulting fertilized egg has three 21st chromosomes.
- Elderly women (Above 35 years)
- Mothers who smoke, use oral contraceptives

Complications

- Growth retardation
- Congenital heart disease
- Pneumonia

- Cataract
- Hearing loss

Signs & Symptoms

- Moonface
- Epicanthic folds
- Flat nasal bridge
- Muscular hypotenia
- Mental retardation
- Behavioral problems
- Hypothyroidism
- Mouth is small & usually open with protrusion of tongue
- Ears are small & located at lower level
- Simian crease
- Little finger is small & incurved
- Sandal toe

Diagnosis

- History collection
- Prenatal screening
  - Chorionic Villus Sampling
  - Amniocentesis

- Percutaneous Umbilical Blood Sampling
- Karyotype

Management.

Down syndrome is not a condition that can be cured. Treatment is directed at addressing the individual concerns of a particular individual e.g certain heart defects may require surgery. Timely surgeries for cardiac & GI anomalies are necessary to prevent serious complications.

Nursing management

- Nurses should obtain history of mother's pregnancy, birth history & genetic testing.
- Observe physical characteristics of down syndrome.
- Assess the following:
- Respiratory functioning due to poor muscle tone.

- Heart sounds for presence of a murmur
- Infant's ability to eat due to protruding tongue & mouth breathing
- Bowel functioning
- In an older child, assess height & weight & compare to appropriate growth chart.
- Determine family knowledge, coping & support
- Observe interaction & bonding between mother & infant.

TURNER SYNDROME
(Ullrich- Turner Syndrome)

Turner syndrome (TS) is a chromosomal condition that describes girls & women with common features that are caused by complete or partial absence of the second sex chromosome.

Causes

Turner syndrome is typically caused by what is called

nondisjunction. This happens when a pair of sex chromosomes fails to separate during the formation of a sperm or egg. When sperm with no X chromosome unites with a normal egg to form an embryo, that embryo will have just one X chromosome. (X rather than XX).

Signs & Symptoms

- Short stature
- Infertility
- Non functioning ovaries.
- Not fully develop breast.
- Webbed neck (extra folds of skin on the neck).
- A broad chest & widely spaced nipples.
- Swelling of the hands & feet.
- Small & narrow fingernails, toenails that turn up.
- Arms that turn out slightly at the elbow.

Dicognosis.

- History collection
- Physical examination
- Prenatal screening
  - Chronionic villus sampling
  - Amniocentesis
- Karyotype

Management.

The best way to treat this disorder is hormone replacement therapy. This therapy is usually started at the time of normal puberty, around 13 years. Teenagers are treated with growth hormone to help them reach a normal height. They may be given low doses of androgens to increase height & encourage hair & muscle growth.

Babies born with a heart murmur or narrowing of the aorta may need surgery

to correct the problem.

ORTHOPEDIC ABNORMALITIES

(1) Congenital talipusequinovarus.

The defect may be on one side or both sides & occurs due to crooked position of the baby in the womb because of less quantity of amniotic fluid.

Causes
- Oligohydramnios
- Abnormal fetal positioning
- Unstretched uterus
- Placental insufficiency.
- Toxins
- Infective pathogens (enterovirus)
- Drugs (abortifacients)
- Electromagnetic radiation

Clinical features
- Foot shorter & wider than normal
- Transverse plantar creases & clefts at the mid foot

- Atrophy of the calf
- The foot is turned downward (plantar flexed) & turned inward (adducted).
- The upper surface of the foot cannot be made to touch the front of the leg (shin) by dorsiflexion.

Diagnosis

History collection
Physical examination
Imaging
Plain radiography

Management

Management should be started as early as possible with the standard foot wear, may be in the first week of life
Early physiotherapy (foot is pulled outward & then dorsiflexed to touch the shin) & applications of plaster of

Paris cast can correct the deformity. This initial management may be continued for 2 to 6 months of age.

Dennis Browne bar/ splint may be used along with corrective shoes after initial period of management. Recently, Wheaton brace or Bebax shoe is used.

Surgical management is usually performed at 4 to 9 months of age with postoperative immobilization before the child begins to walk. Tenotomy is indicated in some cases.

Postoperative immobilization is done by cast for 12 weeks followed by use of brace (ankle-foot-orthosis) or corrective shoes for a period of 2 to 4 years. Parents need adequate explanations & instructions about

correct use of shoes, brace, care of cast, skin care & regular follow-up till the completion of the total schedule of management.

Complications

- Awkward gait
- Callosities
- Bursae

(2) Congenital Dislocation of Hip.

It is one of the most common congenital malformation. It occurs due to faulty development of the head of the femur, the acetabulum & the surrounding capsule & soft tissues. The deformed acetabulum causes dislocation, with the head of the femur becoming displaced upward & backward.

Etiology

Unknown reason

- Hereditary factors
- There is a high risk of hip deformity among females with male to females ratio of 1:4
- Breech position

Because in this position the fetal femur can be easily forced out of the acetabulum.

- Neurological disorders

Clinical Features

- Asymmetry of the gluteal skin folds
- Limited abduction of the affected hip
- Apparent shortening of the femur on the affected side
- Limited range of movements of hip joint on the affected side
- After the child has started

walking the signs including lordosis, protruding abdomen, shortened extremity & a waddling gait.

- Uncorrected dislocation causes limping, easy fatigue, hip & low back discomfort, and postural deformities of spine.

Diagnosis

- History collection
- Physical examination
- X-Ray
- Multiplanar USG
- MRI

Management

Early management is essential for optimal functional recovery. The legs are maintained in the position of abduction & external rotation by the use of VON Rosen Splint.

The Pavlik harness has been

effectively used to treat classical cases of Congenital dislocation of Hip. The recovery is generally complete in a period 3-4 months.

Osteotomies of the upper part of femur may be needed to change the neck-shaft angle. Tenotomy of contracted muscles is often required.

Nursing management

- Maintaining the correct position of the hip.
- Protection of the skin from irritation.
- Encouraging normal physical, emotional & social development.
- Education of parents.