# Congenital Disorders

Set BY Dereje B.

# Trachea-esophageal fistula

- TE fistula is **a birth defect, which occurs in 1 in 5,000 births, and occurs as a fetus is forming in its mother's uterus**.
- When a baby with a TE fistula swallows, the liquid can pass through the abnormal connection between the esophagus and the trachea.
- When this happens, liquid gets into the baby's lungs

# TEF..

- Tracheoesophageal fistula is a connection between the esophagus and the trachea.
- The esophagus is the tube that connects the throat to the stomach.
- The trachea is the tube that connects the throat to the windpipe and lungs.
- Normally, the esophagus and trachea are two tubes that are not connected. This problem is also called TE fistula or TEF. It can happen in one or more places.

# TEF…

- TE fistula is a birth defect. This means it’s a problem you are born with. It happened when your baby was forming during pregnancy.
- When a baby with a TE fistula swallows, liquid can pass through the connection between the esophagus and trachea.
- When this happens, liquid gets into your baby's lungs which can cause pneumonia and other problems. (Aspiration Pneumonia).

# Esophageal atresia

- TE fistula often happens with another birth defect called esophageal atresia.
- This means your baby’s esophagus doesn’t form well during pregnancy. It forms in 2 parts instead of 1. One part connects to the throat.
- The other part connects to the stomach. But the 2 parts don’t connect to each other.
- Since the esophagus is in 2 parts, liquid that your baby swallows doesn’t pass as it should through the esophagus and reach the stomach. This means your baby can't digest milk or other fluids.

## What causes TE fistula and esophageal atresia?

- As a baby is growing during pregnancy, organ systems are forming.
- The trachea and the esophagus start forming as 1 tube.
- At 4 to 8 weeks of pregnancy, a wall forms between the esophagus and trachea.
- This separates them into 2 tubes.
- TE fistula and esophageal atresia happen when this wall doesn’t form as it should.

# Who is risk for TE fistula or esophageal atresia?

- Having the following issues can raise your baby's risk for these conditions:
  - Trisomy 13, 18, or 21
  - Other digestive tract problems, such as diaphragmatic hernia, duodenal atresia, or imperforate anus
  - Heart problems, such as ventricular septal defect, tetralogy of Fallot, or patent ductus arteriosus
  - Kidney and urinary tract problems, such as a horseshoe or polycystic kidney, absent kidney, or hypospadias
  - Muscular or skeletal problems
  - VACTERL syndrome, which involves spinal, anal, heart, TE fistula, kidney, and limb issues
  - up to one half of babies with TE fistula or esophageal atresia also have another birth defect.

## What are the symptoms of TE fistula or esophageal atresia?

- Your child’s healthcare provider will often spot symptoms soon after your baby is born. Symptoms can happen a bit differently in each child. They can include:
  - Frothy, white bubbles in the mouth
  - Coughing or choking when feeding
  - Vomiting
  - Blue color of the skin, especially when the baby is feeding
  - Trouble breathing
  - Very round, full stomach

# Diagnosis of TEF/Atresia

- Your child may need an X-ray of their chest and belly.
- For this test, the healthcare provider will put a small tube into your child's mouth or nose.
- Endoscopy

- It will be guided into the esophagus. With esophageal atresia, the tube often can't go very far into the esophagus.
- The tube's position can be seen with the X-ray.
- This can help your child’s healthcare provider diagnose the condition.

# Treatment of TEF/Atresia

- Surgery based on age/severity of disease
- Child might have more than surgery (other birth defect may occur)…close gab.
- **<u>Complication</u>**
  - Aspiration
  - With esophageal atresia, the esophagus is in 2 parts. Liquid that your baby swallows doesn’t pass normally through the esophagus and reach the stomach. Your baby can't digest milk or other fluids.

# Living with a TE fistula or esophageal atresia

- **TE fistula**
  - Children who had surgery for a TE fistula may develop scar tissue.
  - This can make swallowing tricky.
  - Your child may need another surgery to open the scar tissue.
- **Esophageal atresia**
  - Some children with esophageal atresia have long-term problems. It may be hard for them to swallow foods and liquids. This may be because of the following:
    - Trouble with the movement of foods and liquids down the esophagus (peristalsis).
    - Scarring that can happen after surgery as the wounds heal. This can partially block the passage of foods.

# Nursing interventions.

(1) Monitor respiratory status.

(b) Perform pulmonary physiotherapy.

(c) Suction as necessary.

(2) Administer antibiotics and parenteral fluids as ordered.

(3) Accurate I&O.

(4) Observe for signs of complications (that is, pneumothorax).

(5) Maintain gastrostomy tube feedings.

# Nursing interventions...

(6) Give the baby a pacifier to satisfy his sucking needs but only when he can safely handle secretions.

(7) Offer the parents support and guidance and encourage bonding.

(8) Positioning before and after surgery varies with the doctor’s philosophy and the child’s anatomy.

(a) Supine with his head low to facilitate drainage.

(b) Head elevated to prevent aspiration.

# Sample vedio



Tracheoesophageal Fistula (TEF, TOF) I Nucleus Health.mp4

# CHROMOSOMAL ANOMALIES AND BIRTH DEFECTS

# Cytogenetics

- The study of chromosomes:-
  - their structure
  - inheritance and
  - abnormalities
- Chromosomes are made up of DNA and other protein complexes and contain most of the genetic information that is passed from one generation to the next.

- Chromosome studies are important because abnormal chromosome number
  - e.g., trisomy 13 and
- abnormal chromosomal arrangements

  e.g., micro-deletion may lead to multiple congenital anomalies.

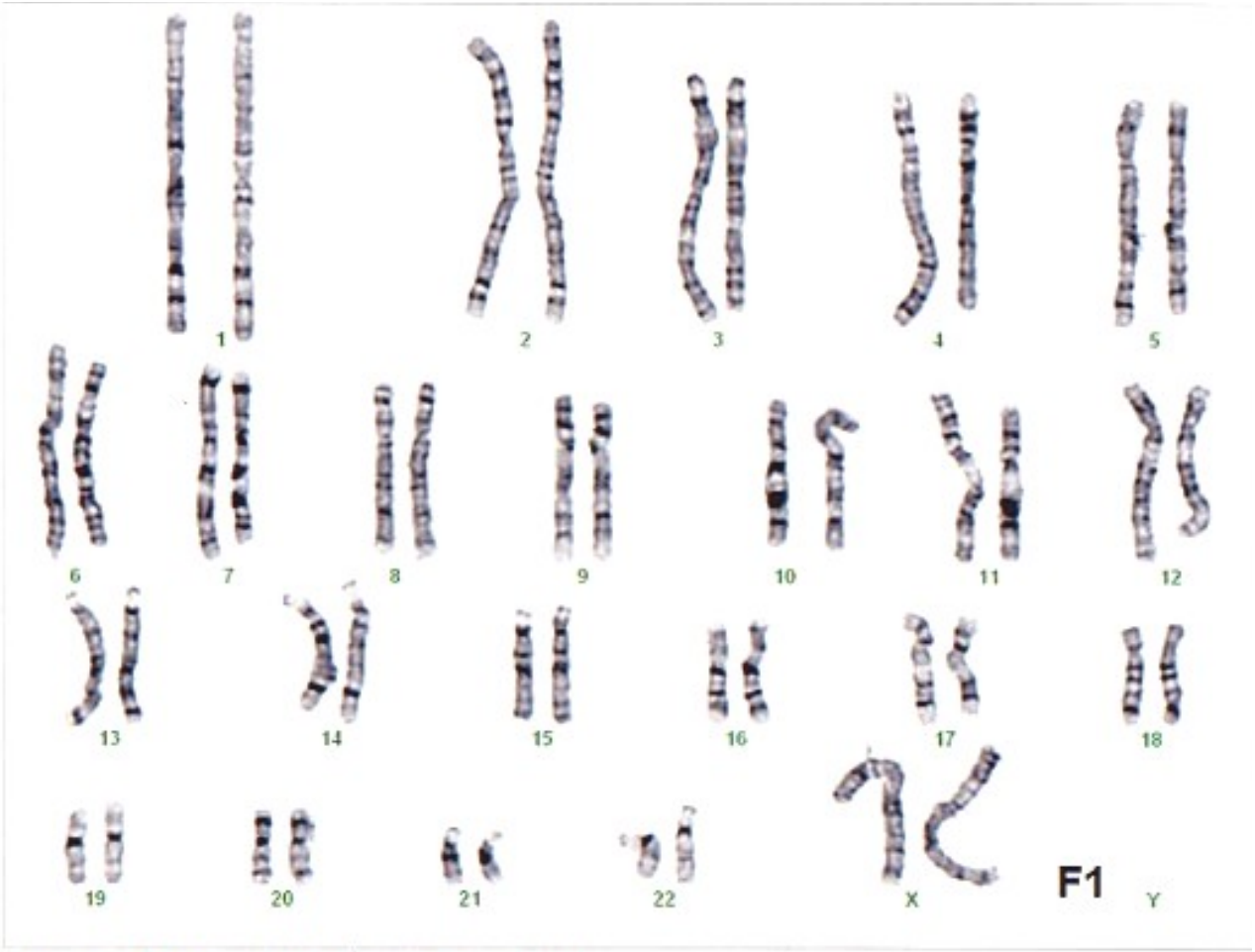
- Chromosome abnormalities:
  - occur in approximately 1% of live births and
  - are responsible for a large proportion of:-
    - early fetal losses
    - multiple congenital malformations, and
    - cases of mental retardation
    - have a significant role in the development of neoplasias
- Chromosome analysis is indicated in the child with multiple congenital anomalies and/or dysmorphic

# Chromosomes

- Human cells contain a multiple of 23 chromosomes ($n$ = 23)
- A haploid cell (n) has 23 chromosomes (ova or sperm)
- Two errors of cell division commonly occur during meiosis or mitosis, and both can result in an abnormal number of chromosomes
  - Nondisjunction
  - Anaphase lag

- **Nondisjunction -** two chromosomes fail to separate and thus migrate together into one of the new cells, producing one cell with two copies of the chromosome and another with no copy.
- ***Anaphase lag* -** a chromatid or chromosome is lost because it fails to move quickly enough during anaphase to become incorporated into one of the new daughter cells.

# Normal Female: 46,XX




Case: STANDARD Slide: alfa Cell: 1 Patient:

# Normal Male: 46,XY



# Chromosomal Anomalies

- Include abnormalities of number and structure
- Are the result of *errors in cell division.*

## Chromosome Errors Results in

- Spontaneous abortuses
- Sexually ambiguous organisms
- Infertile males or females
- Newborns with multiple congenital anomalies
- Mentally retarded
- Mentally ill
- Behaviorally disordered
- Cancers

# ABNORMALITIES OF CHROMOSOME NUMBER

- Trisomy: the presence of an extra chromosome
- Monosomy: the absence of a **whole** chromosome
- Deletion: the absence of a **part of** a chromosome
- Inversion: the **180° rotation** of a part of a chromosome
- Translocation: the **breakage and rejoining** of parts of two, **non-homologous** chromosomes.

- Trisomy, characterized by the presence of 3 instead of the normal 2 of any particular chromosome, is the most common form of aneuploidy
- Trisomy can be complete and present in all cells or it may be in mosaic form.

# ABNORMALITIES OF CHROMOSOME STRUCTURE

- **Translocations**
- **Inversions**
- **Deletions**
- **Duplications**

# chromosomal Anomalies

- General features:
  - Mental retardation (MR)
  - Growth retardation (GR)
  - Multiple congenital anomalies
  - Prenatally diagnosable
  - Associated with spontaneous abortion (Sab)

# chromosomal Anomalies

- Trisomy 13 - Patau Syndrome
- Trisomy 18 - Edward Syndrome
- Trisomy 21 - Down Syndrome
- $5p^-$ deletion - Cri-du-chat Syndrome

# Sex Chromosome Anomalies

- General features:
  - Some growth retardation (GR)
  - Reproductive anomalies/problems
  - Prenatally diagnosable
  - Associated with spontaneous abortion (Sab)

# Sex Chromosome Anomalies

- Monosomy X: Turner's Syndrome (45, X)
- Trisomy X: Triplo-X Syndrome (47, XXX)
- Trisomy (47, XXY): Klinefelter's Syndrome
- Trisomy (47, XYY): XYY Syndrome

# Multifactorial Inheritance

- anencephaly
- atopic allergies
- cleft lip/palate
- club foot
- congenital heart disease
- congential hip dysplasia
- congenital scoliosis
- diabetes mellitus
- epilepsy
- hydrocephalus
- non-specific MR
- NTD
- pyloric stenosis
- urinary tract malformations

# Well recognized Syndromes, sequences, associations

- Down syndrome
- Pierre Robin sequence
- Trisomy 13
- Trisomy 18
- Cri du chat syndrome
- Turner syndrome
- Klinefelter syndrome

# Down Syndrome

- Described by John Landon Down in 1866
- Etiology: nondisjuction mutation resulting in Trisomy 21, translocation
- Prevalence 1:700
  - Most common chromosomal anomaly
- Associated with Maternal age > 35

**Down Syndrome**

# Genetics

- Three types of cytogenetic abnormalities result in the Down syndrome phenotype
  - Trisomy 21 (47,+21), in which an extra copy of chromosome 21 is present — 95 %
  - Robertsonian translocation involving chromosome 21— 4 %
  - Trisomy 21 mosaicism (47,+21/46), which occurs in 1 % of cases.
    - In this form, there are two populations of cell types: a normal cell line with 46 chromosomes and a second line with trisomy 21

# Down Syndrome

- Actual risk for Down Syndrome:
  - 1/10 if female is translocation carrier
  - 1/20 if male is translocation carrier
- Translocations account for 9% of the children with Down syndrome born to mothers younger than 30 yr of age.
- Half the translocations arise de novo in the affected individual, whereas half are inherited from a translocation carrier parent

# Down Syndrome

## DYSMORPHIC FEATURES

- Each characteristic dysmorphic feature is present in 47 to 82 percent of cases
- These features predominantly affect the head and neck and the extremities

# Down Syndrome

- Characteristic dysmorphic features of DS affecting the head and neck include:
  - Brachycephaly /flat occiput
  - Upslanting  palpebral fissures
  - Epicanthic folds
  - Brushfield spots
  - Flat nasal bridge
  - Folded or dysplastic ears
  - Small ears
  - Protruding tongue
  - Narrow palate
  - Short neck
  - Excessive skin at nape of the neck

# Down Syndrome

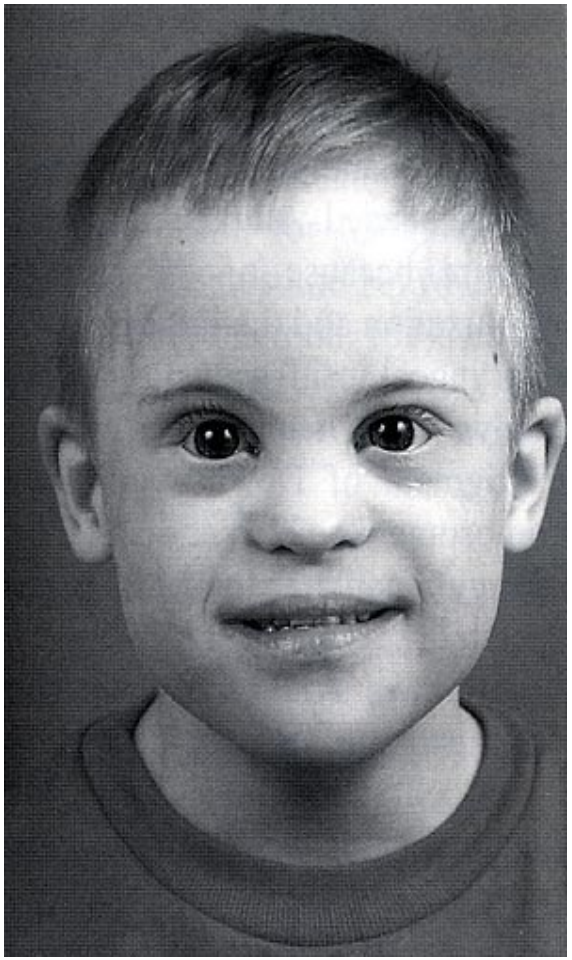
- Characteristic dysmorphic features of DS affecting the extremities include:
  - Short broad hands
  - Hypoplastic mid phalanx of fifth finger
  - Incurved fifth finger
  - Transverse palmar crease
  - Space between the first and second toes (sandal gap deformity)
  - Hyperflexibility of joints

# Down Syndrome



# Down Syndrome

photograph



# Down Syndrome

- Airway Concerns
  - Due to midface hypoplasia, the nasopharynx and oropharynx dimensions are smaller
    - Slight adenoid hypertrophy can cause upper airway obstruction
  - Congenital mild-moderate subglottic narrowing

# Down Syndrome

## Growth and development

- Mental retardation- All individuals with DS have cognitive impairment, although the range is wide
- Developmental impairment becomes apparent in the first year of life
- the average age of sitting (11 months), creeping (17 months), and walking (26 months) is twice the typical age
- Birth weight, length, and head circumference are less in DS compared to typical infants
- Newborns with DS weigh approximately 0.18 to 0.37 kg less than their siblings

# Down Syndrome

- Obstructive Sleep Apnea
  - Prevalence 54-100% in DS patients
  - Combination of anatomic and functional mechanisms
    - Midface hypoplasia, macroglossia, etc
    - Hypotonia of pharyngeal muscles

# Down Syndrome

## Hearing loss

- Hearing impairment affects 38 to 78 percent of individuals with DS
- Congenital and Acquired, overall high incidence
- Conductive hearing loss
  - more common
- Sensorineural hearing loss
  - less common
- Otitis media is a frequent problem ( 50 to 70 %)
  - Monitoring for this condition is important to preserve hearing

# Down Syndrome

- Cardiovascular anomalies (40%)
  - Atrioventricular septal defect (ECD) with or without other lesions — 45 percent
  - VSD- 35%
  - Others -ASD, ,Tetralogy of Fallot,PDA
- GI anomalies (5%)
  - Pyloric stenosis, duodenal atresia, TE fistula
- Malignancy
  - 20 fold higher incidence of leukemia
  - Gonadal tumors

# Down Syndrome

- Reproduction:
  - Women with DS are fertile and may become pregnant
  - Nearly all males with DS are infertile. The mechanism is impairment of spermatogenesis

# Nursing care for DS

- Provide adequate nutrition. Assess the child' ability to swallow, provide information on the proper way of giving food, and provide good nutrition counseling.
- Frequent consultations are a must. ...
- Assess understanding of Down syndrome. ...
- Provide emotional support and motivation.

# Nursing Diagnoses

- Based on the assessment data, the major nursing diagnoses are:
- **Delayed growth and development** related to impaired ability to achieve developmental tasks.
- **Self-care deficit: bathing and hygiene, dressing, feeding, toileting** related to cognitive impairment.
- **Impaired verbal communication** related to impaired receptive or expressive skills.
- **Risk for infection** related to decreased muscle tone and poor drainage of mucus.

# REST and Digest

- **1. Which of the following assessment findings would lead the nurse to suspect Down syndrome in an infant?**

A. Transverse palmar crease
B. Small tongue
C. Large nose
D. Restricted joint movement

# R/D

**2. When planning care for an 8-year-old boy with Down syndrome, the nurse should:**

A. Plan interventions according to the developmental levels of a 5-year-old because the child will have developmental delays.

B. Plan interventions according to the developmental level of a 7-year-old child because that’s the child’s age.

C. Direct all teaching to the parents because the child can’t understand.

D. Assess the child’s current developmental level and plan care accordingly.

# R/D

**3. A nurse is monitoring the intake and output of a toddler with Down syndrome receiving furosemide (Lasix) IV. Which of the following method is the most appropriate for measuring the urine output?**

- A. Weighing the diaper.
  B. Insert Foley catheter.
  C. Ask the mother regarding the number of diaper changes.
  D. Compare intake with output.

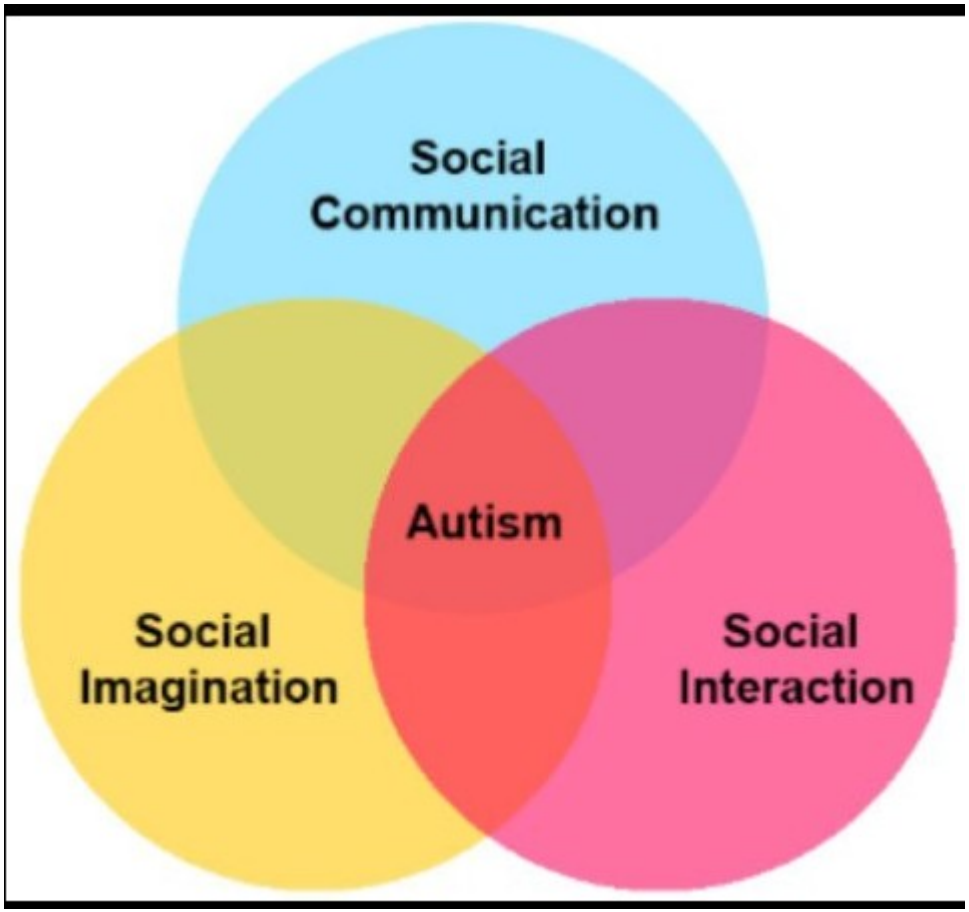
...

# Autism

# Definition

- In the milestone development if there is impairment of socialization and language & communication is result in isolation
- This social interaction and communication isolation called Autism.
- Autism is spectrum disorder b/c  it is Varity and diversity of symptom.
- It cause problem with functioning skill and society

# Cont....


Social
Communication
Autism
Social
Imagination
Social
Interaction

# Causes

- The cause of autism spectrum disorder are unknown.
- Obstetric complication:-intrauterine exposure

  -infection
- Genetic disease
- Toxic exposure
- Extreme preterm babies

# Symptom of ASD



# Symptom of ASD

- Difficulty in social interaction:-
  - Eye contact
  - Faecal expression
  -Body posture
  - Emotional signal
  - Enjoyment with others
  - Difficulty of understanding others feeling

# Con….

- Difficulty in communication:-
  - lack of talk
  - Taking turns
  - Stereotyed
  - Listen respectively
  - Focus on self need
- Difficulty in behaviour:-
  - hypersensitivity
  - Rocking
  - flapping
  - sleep problem

# Types of Autism

## 1 KANNERS AUTISM

- Sever type of autism
- Low functioning autism
- Very low IQ
- Loss of cognition/speech
- Anti social behaviour
- Mental retardation

# Cont…….

## 2 **ASPERGERS SYNDROM**

- High function autism
- Has basic symptom with low severity
- High level of IQ
- Smart on arts, maths, etc
- Difficulty of social interaction and delay of  language or cognitive problem

# Cont.....

**3 PREVASIVE DEVELOPMENTAL**

**DISORDER(PDD)**

- Mild type of autism that present range of symptom
- The child may experience delay in language development, walking and other motor skills
- The most common symptom are

# Cont.....

**4 CHILDHOOD DISINTEGRATIVE DISORDER**

- Defined by delayed onset of developmental problem in motor skill, social and language function.
- More common in boys
- The child may loss toileting skill if they had already been established.
- Acquired language or vocabulary
- Adaptive behaviour and some motor

# Diagnosis

- History

  -careful hx from the parents

- Physical examination

  - neurologic abnormality(macrocephaly and hypotonia)

  - dysmorphic feature

  -skin e.g ash leaf

- Developmental

  -speech and language, motor, social and play skill

# Cont….

- In general:- audiology test
  -vision test

Genetic testing:-

- if the child has intellectual disability
- if there is a family history of mental

retardation.

-dysmorphic features.

- EEG

-to rule out seizure disorder

# MANAGEMENT OF AUTISM

- Speech and language therapy:-

  - To improve verbal and non-verbal communication

- Occupational therapy:-

  - to improve fine motor deficit and improve academic and self care skill

# Cont…

- Applied Behavioural Analysis(ABA):-

- To increase socially appropriate behaviour

- Decrease the severity of and challenge of behaviour .

- Result in improvement in:-

- communication

-social relatedness

- decrease repetitive behaviour

# Cont….

- Obsessive- compulsive disorder

  - second most common co-occurring condition with ASD

  -Unwillingness to change routine

  - may limit their ability to interact socially

  - Ask parents about these symptom

  -**SSRI** use in children with autism found no evidence of effect for either anxiety or obsessive compulsive disorder

# Cont…

- For Disruptive behaviour:-
  - Behavioural therapy
  - Risperidone or aripiprazole
- Complementary and alternative medicine:-
  - Gluten-free and casein-free diet
  -vitamin B6
  - Magnesium
  - vitamin C
  -Fatty acid
  - combined hyper vitamine therapies

# Nursing intervention

- What are 5 strategies used to work with a child with autism?
  - Give advance notice before a transition is going to occur.
  - Use visual supports.
  - Use structure and consistency.
  - Use reduced language.
  - Provide light praise for good transitions.

# Nursing intervention…

- Perform nursing assessment progressively and slowly.
- Talk with the patient about their interests
- Evaluate communication abilities and develop rapport and trust.
- Provide a calm and inviting atmosphere
  - Avoid loud noises, radios, talking
  - Turn off the TV during exam and evaluations
  - Limit number of people in the room to those whom the patient is most comfortable with
  - Provide for safety. Place infants or toddlers in the crib, raise rails on the bed(remove harmful objects)

# Imperforated Anus

Infant to be kept nil by mouth

Requires a gastric tube insertion for decompression

IV and IV fluids to be started



# Imperforated anus ...

- **inborn abnormality characterized by the absence or abnormal localization of the anus**. The rectum or the colon may be connected to the vagina or the bladder by a tunnel (fistula).

# Causes

- Imperforate anus may occur in several forms:
  - The rectum may end in a pouch that does not connect with the colon.
  - The rectum may have openings to other structures. These may include the urethra, bladder, base of the penis or scrotum in boys, or vagina in girls.
  - There may be narrowing (stenosis) of the anus or no anus.
  - It is caused by abnormal development of the fetus.

# C/M

- No passage of first stool within 24 to 48 hours after birth
- Stool passes out of the vagina, base of penis, scrotum or urethra
- Swollen belly

# Dx and RX

- Dx
  - Imaging
  - PE
- Rx
  - Surgical correction
  - The infant should be checked for other problems, especially those affecting the genitals, urinary tract and spine.
  - Surgical reconstruction of the anus is often needed. If the rectum connects with other organs, repair of these organs will also be necessary through abdominal surgery.

# Phimosis, paraphimosis, criptiorchidism)

- Phimosis
  - It is a condition in which the foreskin of the penis cannot be pulled back past the glans.
  - A balloon-like swelling under the foreskin may occur with urination.
  - In teenagers and adults, it may result in pain during an erection, but is otherwise not painful.
  - Those affected are at greater risk of inflammation of the glans.
  - In young children, it is normal to not be able to pull back the foreskin.
- RX: circumcision





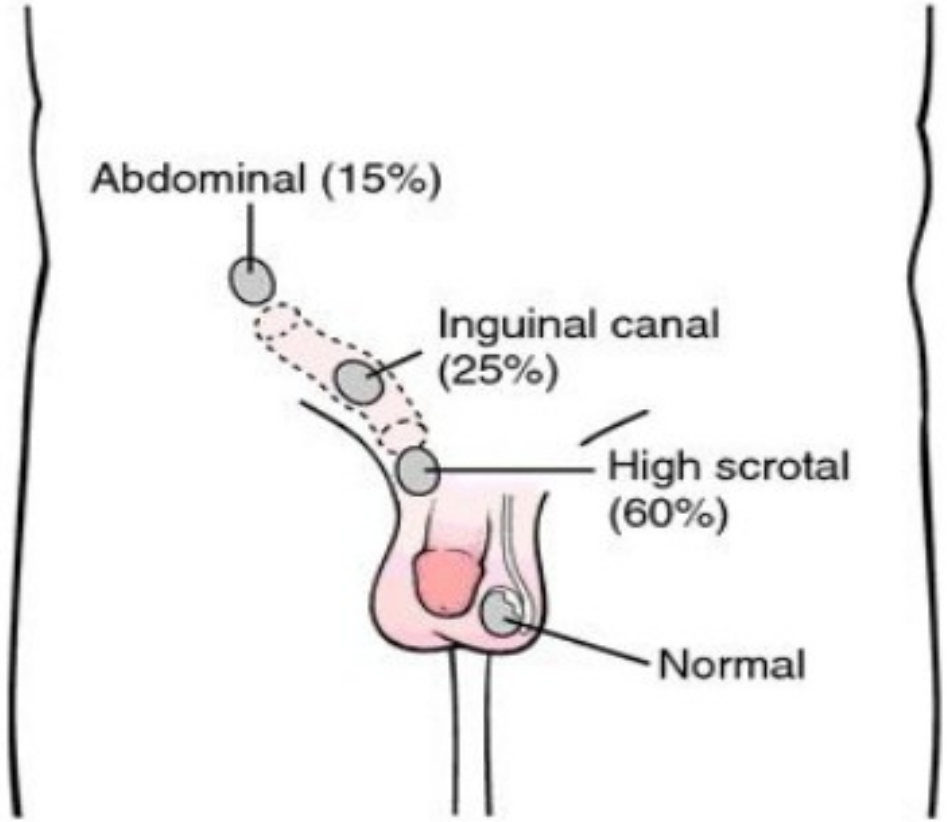
# Paraphimosis

- Paraphimosis can often be effectively treated by manual manipulation of the swollen foreskin tissue.
- This involves compressing the glans and moving the foreskin back to its normal position, perhaps with the aid of a lubricant, cold compression, and local anesthesia as necessary.
- If this fails, the tight edematous band of tissue can be relieved surgically with a dorsal slit or circumcision.



# Cryptorchidism

- is the absence of one or both testes from the scrotum.
- It is the most common birth defect of the male genitals.
- About 3% of full-term and 30% of premature infant boys are born with at least one undescended testis


Abdominal (15%)
Inguinal canal
(25%)
High scrotal
(60%)
Normal

# Cryptorchidism...

- A testis absent from the normal scrotal position may be:

  • anywhere along the "path of descent" from high in the posterior (retroperitoneal) abdomen, just below the kidney, to the inguinal ring;

  • in the inguinal canal;

  • ectopic, having "wandered" from the path of descent, usually outside the inguinal canal and sometimes even under the skin of the thigh, the perineum, the opposite scrotum, or the femoral canal

  • undeveloped (hypoplastic) or severely abnormal (dysgenetic);