MODULE 1: Framework for MCN

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TRANSCRIBED BY: NICOLE ANDREA H. LUNA
LECTURER: F. Saldana

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A. NATIONAL HEALTH SITUATION IN THE PHILIPPINES ON MATERNAL AND CHILD NURSING

The Philippines has made significant investments and advances in health in recent years. Rapid economic growth and strong country capacity have contributed to Filipinos living longer and healthier. However, not all the benefits of this growth have reached the most vulnerable groups, and the health system remains fragmented.

Health insurance now covers 92% of the population. Maternal and child health services have improved, with more children living beyond infancy, a higher number of women delivering at health facilities and more births being attended by professional service providers than ever before.

Access to and provision of preventive, diagnostic and treatment services for communicable diseases have improved, while there are several initiatives to reduce illness and death due to noncommunicable diseases (NCDs). Despite substantial progress in improving the lives and health of people in the Philippines, achievements have not been uniform and challenges remain.

Deep inequities persist between regions, rich and the poor, and different population groups. Many Filipinos continue to die or suffer from illnesses that have well-proven, cost-effective interventions, such tuberculosis, HIV and dengue, or diseases affecting mothers and children. Many people lack sufficient knowledge to make informed decisions about their own health. Rapid economic development, urbanization, escalating climate change, and widening exposure to diseases and pathogens in an increasingly global world increase the risks associated with disasters, environmental threats, and emerging and re-emerging infections.

B. STATISTICS ON MCN

Worldwide

→ Maternal mortality refers to deaths due to complications from pregnancy or childbirth.
→ From 2000 to 2017, the global maternal mortality ratio declined by 38% – from 342 deaths to 211 deaths per 100,000 live births, according to UN inter-agency estimates. This translates into an average annual rate of reduction of 2.9%
→ While substantive, this is less than half the 6.4% annual rate needed to achieve the Sustainable Development global goal of 70 maternal deaths per 100,000 live births.
→ There has been significant progress since 2000.
→ Between 2000 and 2017, South Asia achieved the greatest overall percentage reduction in MMR, with a reduction of 59% (from 395 to 163 maternal deaths per 100,000 live births).
→ Sub-Saharan Africa achieved a substantial reduction of 39 per cent of maternal mortality during this period.

Key Facts

- Every day in 2017, approximately 810 women died from preventable causes related to pregnancy and childbirth.

- Between 2000 and 2017, the maternal mortality ratio (MMR, number of maternal deaths per 100,000 live births) dropped by about 38% worldwide.
- 94% of all maternal deaths occur in low and lower middle-income countries.
- Young adolescents (ages 10-14) face a higher risk of complications and death as a result of pregnancy than other women.
- Skilled care before, during and after childbirth can save the lives of women and newborns.

Causes

Women die as a result of complications during and following pregnancy and childbirth. Most of these complications develop during pregnancy and most are preventable or treatable. Other complications may exist before pregnancy but are worsened during pregnancy, especially if not managed as part of the woman's care.

The major complications that account for nearly 75% of all maternal deaths are (4):

→ Severe bleeding (mostly bleeding after childbirth)
→ Infections (usually after childbirth)
→ High blood pressure during pregnancy (pre-eclampsia and eclampsia)
→ Complications from delivery
→ Unsafe abortion

The remainder are caused by or associated with infections such malaria or related to chronic conditions like cardiac diseases or diabetes

Philippines

Chart and table of the Philippines infant mortality rate from 1950 to 2021. United Nations projections are also included through the year 2100.

- The current infant mortality rate for the Philippines in 2021 is **18.392** deaths per 1, 000 live births, a **2.25% decline** from 2022
- The infant mortality rate for the Philippines in 2020 was **18.815** deaths per 1,000 live births, a **2.2% decline** from 2019
- The infant mortality rate for the Philippines in 2019 was **19.239 deaths** per 1,000 live births, a **2.16% decline** from 2018
- The infant mortality rate for the Philippines was **19.663** deaths per 1,000 live births, a **3.96%** decline from 2017.

Philippines' Maternal mortality rate – Demographics

According to data from the Philippines Department of Health the top 4 causes of maternal mortality are **labor complications, pregnancy-related hypertension, postpartum hemorrhage,** and **unsafe abortion.**

The areas of the Philippines where these causes of **death and morbidity are the highest are rural, isolated regions/communities that exist far from the country's capital and urban centers**, including ARMM, SOCCKSARGEN, MIMAROPA and the Cagayan Valley as can be seen on the adjacent map. Similar trends can be found for child health statistics as well, given that the areas plagued by these health outcomes are characterized by *poor access to health care services, poverty, and geographic isolation*, among other social determinants.

Specifically, **maternal mortality and morbidity are associated with poor birthing practices** including *home births or births without a skilled birth attendant*, and *inadequate perinatal health care by a health provider*- all of which are closely tied to wealth, socioeconomic status and the urban-rural divide. The statistical breakdown of birthplace, perinatal health care and maternal health equity of the Philippines can be referenced below courtesy of the 2014 WHO report on Maternal and Perinatal health by the Department of Maternal, Newborn, Child and Adolescent Health.

According to UNICEF

- 160 women for every 100,000 births die.
- Roughly over 11 women die every day.
- 7 out of 10 deaths occur at childbirth or within a day after delivery.
- 4 out of 10 deaths are due to complications and widespread infections
- For every death, 40 more women get sick.

- 8 out of 10 births in rural areas are delivered outside a health facility

Western Visayas

The Center for Health Development (CHD) in Western Visayas has admitted the need to double their efforts to address the high rate of childhood mortality in the region.

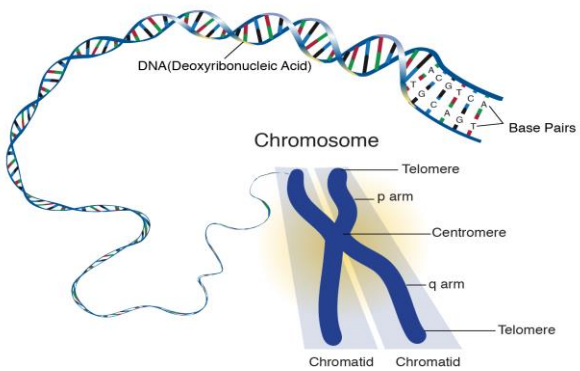
Results of the 2017 National Demographic and Health Survey (NDHS), conducted by the Philippine Statistics Authority (PSA), showed that 31% of 1,000 pregnancies of at least seven months duration are stillbirth; 33% of 1,000 live births do not reach their first month of life; 38% die before their first birthday; and 46% of children die before reaching their fifth birthday. The result is higher than the national childhood mortality rate, which is 22% for under-five mortality, 21% for infant, and 14% for neonatal mortality. Neonatal mortality refers to children who die before their 28th day while infant mortality refers to those who die before they reach one year old.

C. GENETICS AND GENETIC COUNSELING

Nature of Inheritance

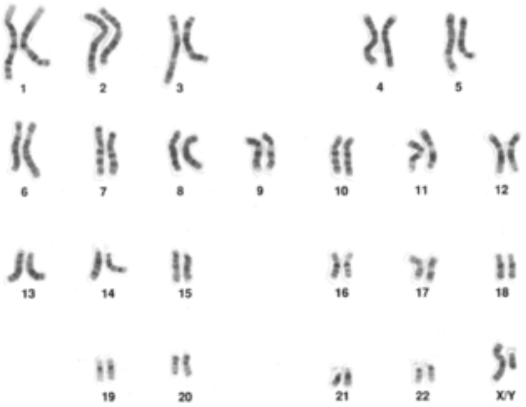
Genes – Basic units of heredity that determine both the physical and cognitive characteristics of people.

- Composed of DNA segments, they are woven into strands in the nucleus of all body cells to form chromosomes.



Chromosomes

- In humans, each cell, with the exception of the sperm and ovum, contains 46 chromosomes (22 pairs of autosomes, and 1 pair of sex chromosomes)
- The *spermatozoa and ova* each carry only half of the chromosome number, or **23 chromosomes**.
- For each chromosome in the sperm cell, there is a like chromosome of similar size, shape, and function (autosome or homologous chromosomes) in the ovum.
- Because genes are always located at fixed positions on chromosomes, two genes (***alleles***) for every trait are represented in the ovum and sperm on autosomes.



- The one chromosome in which this does not occurs is the chromosome for determining gender.
  - XX – Female
  - XY – Male

Phenotype

- Refers to his or her outward appearance or the expression of genes.

Genome

- Complete set of genes present (46 XX or 46 XY)

Genotype

- Actual gene composition

Nature of Inheritance: Dominant and Recessive Pattern

- The principle of inheritance of disease are the same as those that govern genetic inheritance of other physical characteristics, such as eye or hair color.
- These principles are discovered and described by Gregor Mendel, an Austrian naturalist (Mendelian Laws)

Homozygous

- A person who has two like genes for a trait on two like chromosomes

Heterozygous

- If gene differs for a trait on two like chromosomes

Dominant

- Dominant in their action over others, if paired with other genes

Recessive

- Nondominant gene

Mendelian Law

- Permits the prediction of inheritance traits such as eye color or the chance that a child born to parents with a certain genotype will be born with a disorder.
- If the father is homozygous dominant (has 2 dominant genes for brown eye color) and the mother is homozygous recessive (has 2 genes for blue eye color) it can be predicted that their children have a 100% chance of being a heterozygous for a trait (brown eyed – phenotype) that will carry a recessive gene for blue eyes (genotype).
- If the father is homozygous dominant (has 2 dominant genes for brown eye color) and the mother is homozygous recessive (has 2 genes for blue eye color) it can be predicted that their children have a 100% chance of being a heterozygous for a trait (brown eyed – phenotype) that will carry a recessive gene for blue eyes (genotype).
- Suppose the mother is heterozygous, and the father is homozygous dominant, the chances are equal that their child will be homozygous dominant like the father or heterozygous like the mother. All the children's phenotype will be brown eyes
- Suppose both parents are heterogenous.
- There is a 25% chance of their child to being homozygous recessive (appearing blue eyed), 50% chance of being heterozygous (appearing brown eyed) and 25% homozygous dominant (appearing brown eyed)
- This is how 2 brown eyed parents can produce a blue eyed child.

Inheritance of Disease

Autosomal Dominant Disorders (ADD)

- More than 3, 000 ADD are known, only a few are commonly seen
- Most of them cause structural defects
- With ADD, either a person has 2 unhealthy genes (homozygous dominant) or is heterozygous, with the gene causing the disease stronger than the corresponding healthy recessive gene for the same trait.

Rule (ADD)

- A person who is heterozygous for an ADD mates with a person who is free of that trait:
  → 50% chance – the child will have the disorder or would be disease-free and carrier-free
- 2 heterogenous people with a dominantly inherited disorder to choose each other, because if they do, their chances of having children free from the disorder decline.
- There would only be 25% chance of a child's being disease and carrier-free.50% chance that the child would have the disorder, 25% chance that the child would be homozygous dominant (incompatible with life).

When Assessing the Family Genogram

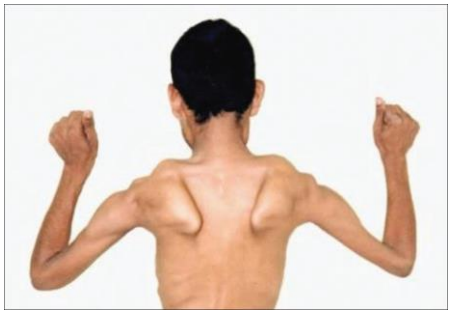
- One of the parents of a child with the disorder also will also have the disorder (vertical transmission).
- The sex of the affected individual is unimportant in terms of inheritance.
- There is usually a history of the disorder in other family members.

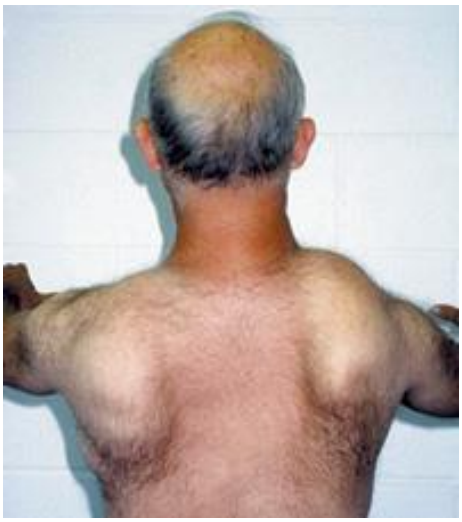
e.g.

- **Huntington Disease**
  - A progressive neurologic disorder that usually manifests symptoms between 35 – 45

years of age and is characterized by loss of motor control and intellectual deterioration.
- Analyzing a specific gene in chromosome 4
- No cure

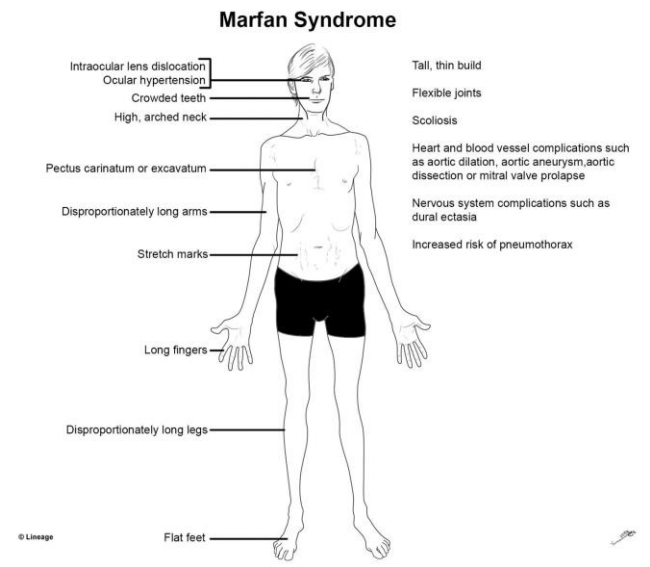
- **Facioscapulohumeral muscular dystrophy**
  - A disorder that results to muscle weakness





- **Osteogenesis imperfecta**
  - A disorder in which bones are exceedingly brittle

- **Marfan Syndrome**
  - A disorder of connective tissue in which the child is thinner and taller than normal and may have associated heart defects.



Autosomal Recessive Disorders

- Biochemical or enzymatic
- Do not occur unless 2 genes for a disease are present (homozygous recessive pattern)
- *e.g. Cystic fibrosis, Adrenogenital syndrome, Albinism, tay-sachs disease, Galactosemia, Phenylketonuria, Limb-girdle muscular dystrophy, and RH factor incompatibility*

Genogram

- Both parents are disease free but both are heterozygous in genotype
- The sex of the affected individual is unimportant
- The family history for the disorder is negative
- A known common ancestor between the parents sometimes exist

Genetic counseling

- Results in making individuals feel "well or free of guilt for the first time in their lives if they discover that the disorder they were worried about was not an inherited one, but was rather a chance occurrence.
- In other instances, counseling results in informing individuals that they are carriers of

a trait that is responsible for a child's condition. Even when people understand that they have no control over this, knowledge about passing a genetic disorder to a child can cause guilt and self-blame. Marriages and relationships can end unless both partners receive adequate support.

- It is essential that information revealed in genetic screening be kept confidential, because such information could be used to damage a person's reputation or harm a future career or relationship.

Nursing Responsibilities

Nurses play important roles in assessing for signs and symptoms of genetic disorders, in offering support to individuals who seek genetic counseling, and in helping with reproductive genetic testing procedures by such actions as:

- Explaining to a couple what procedures they can expect to undergo
- Explaining how different genetic screening tests are done and when they are usually offered
- Supporting a couple that are waiting for test results
- Assisting couples in various clarification, planning, and decision making based on test results

Assessment

History

- Remember to include half brothers and sisters or anyone related in any way as family.
- Document the mother's age because some disorders increase in incidence with age.
- Document also whether the parents are consanguineous or related to each other.
- Documenting the family's ethnic background can reveal risks for certain disorders that occur more commonly in some ethnic groups than others.
- If the couple seeking counseling is unfamiliar with their family history, ask them to talk to senior family members about other relatives (grandparents, aunts, uncles) before they come for an interview. Have them ask specifically for instances of spontaneous miscarriage or children in the family who died at birth.

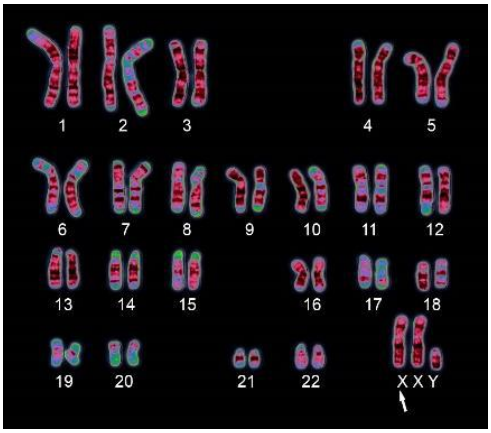
- Attempt to obtain more information by asking the couple to describe the appearance or activities of the affected individual or asking for permission to obtain health records.
- An extensive prenatal history of any affected person should be obtained to determine whether environmental conditions could account for the condition. Based on the above information, draw a family genogram.
- When a child is born dead, parents are advised to have a chromosomal analysis and autopsy performed on the infant.

Physical examination of Family Members

- During inspection, pay particular attention to certain body areas, such as the space between the eyes. The height, contour, shape of the ears, number of fingers and toes, and the presence of webbing.
- Dermatoglyphics (the study of surface markings of the skin) can also be helpful. Note any abnormal fingerprints or palmar creases as these are present with some disorders.
- Abnormal hair whorls or coloring of hair can also be present.

Laboratory Analysis

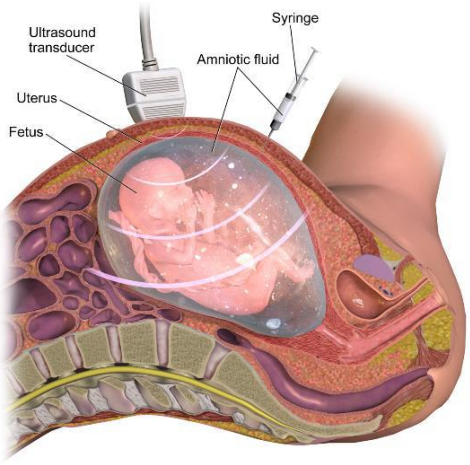
- *Karyotyping or DNA Analysis*
  - Sample of peripheral venous blood or a scraping of cells from the buccal membrane is taken.
  - Cells are allowed to grow until they reach metaphase, the most easily observable phase
  - Cells are then stained, placed under a microscope, and photographed
  - Chromosomes are identified according to size, shape, and stain; cut from the photograph and arranged.
  - DNA analysis or Karyotyping of both parents and an already affected child (provides a picture of the family's genetic pattern and can be used for prediction in future child).



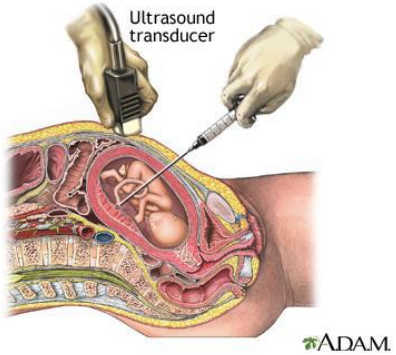
- *Maternal Serum Screening*
  - Alpha-fetoprotein (AFP) is a glycoprotein produced by the fetal liver that reaches a peak in maternal serum between the 13th and 32nd week of pregnancy.
  - This level is elevated with fetal spinal cord disease (more than twice the value of the mean for that gestational age) and is decreased in a fetal chromosomal disorder such as trisomy 21.

- *Chorionic Villi Sampling*
  - A diagnostic technique that involves the retrieval and analysis of chorionic villi from the growing placenta for chromosome or DNA analysis.
  - Highly accurate and yields no more false-positive results than amniocentesis.

- *Amniocentesis*
  - Withdrawal of amniotic fluid through the abdominal wall for analysis at the 14th – 15th week of pregnancy
  - Since amniotic fluid has reached 200 mL at this point, enough fluid can be withdrawn for karyotyping of skin cells found in the fluid as well as an analysis of AFP or acetylcholinesterase.
  - If no AFP, a breakdown product of blood, is found in the specimen it confirms that an elevated AFP level is not a false-positive reading caused by blood in the fluid.

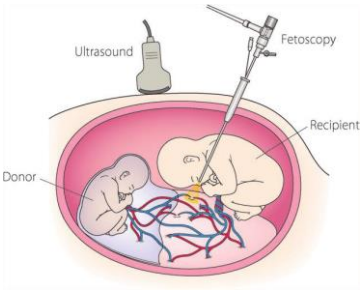


- *Percutaneous Umbilical Blood Sampling (PUBS) or Cordocentesis*
  - Removal of blood from the fetal umbilical cord at about 17 weeks using an amniocentesis technique.
  - This allows analysis of blood components as well as more rapid karyotyping than is possible wen only skin cells are removed



- *Fetal Imaging*
  - Magnetic Resonance Imaging (MRI) and ultrasound are diagnostic tools used to assess a fetus for general size and structural disorders of the internal organs, spine, and limbs
  - Because some genetic disorders are associated with physical appearance, both of these methods may be helpful.
  - Ultrasound is used concurrently with amniocentesis.

- *Fetoscopy*
  - Insertion of a fiberoptic fetoscope through a small incision in the mother's abdomen into the uterus and membranes to visually inspect the fetus for gross abnormalities.
  - It can be used to confirm an ultrasound finding, to remove skin cells for DNA analysis, or to perform surgery for a congenital disorder such as a stenosed urethra.



- *Preimplantation Diagnosis*
  - Possible for in vitro fertilization procedures
  - It may be possible in the future for a naturally fertilized ovum to be removed from the uterus by lavage before implantation and studied for DNA analysis this same way. The ovum would then be reinserted or not, depending on the findings and the parents' wishes. This would provide genetic information extremely early in a pregnancy.

Genetic Disorder Screening and Diagnostic Test

Test Type	Timing	Process	Risk	Results
Nuchal translucency	11-14 weeks	UTZ to assess thickness at fetus' neck, maternal blood draw	Noninvasive	Screening test for Trisomy 21, 18, 13
Chorionic Villi Sampling	10-12 weeks	Biopsy of placenta	Invasive, risk of miscarriage	Diagnostic test for chromosomal disorder
Amniocentesis	15-18 weeks	Collection of amniotic fluid containing fetal skin cells through maternal	Invasive, risk of miscarriage	Diagnostic test for chromosomal disorders
PUBS	>17 weeks	Fetal umbilical blood sampling through maternal	Invasive, risk for miscarriage	Diagnostic for fetal blood disease
Fetal anatomy ultrasound	18 – 22 weeks ideal timing	UTZ of the fetal anatomy	Noninvasive	Screening test for visual fetal anomalies
Fetoscopy	2nd and 3rd trimester	Small camera and instruments passed into the amniotic sac to view and treat anomalies	Risk of miscarriage	Often used to treat disorders like twin-to-twin transfusion
Newborn Screening	Day 2 – several weeks after birth	A blood sample via heel prick or blood draw from newborn	Noninvasive	Screening for genetic disorders

Legal and Ethical Aspects of Genetic Screening and Counseling

Nurses can be instrumental in seeing that couples who seek genetic counseling receive results in a timely manner and with compassion about what their results may mean to future childbearing. Always keep in mind several **legal responsibilities of genetic testing, counseling, and therapy including:**

- Participation by couples or individuals in genetic screening must be elective
- People desiring genetic screening must sign an informed consent for the procedure

- Results must be interpreted correctly yet provided to the individuals as quickly as possible.
- The results must not be withheld from the individuals and must be given only to those persons directly involved
- After genetic counseling, persons must not be coerced to undergo procedures such as abortion or sterilization. Any procedure must be a free and individual decision.

Failure to heed these guidelines could result to charges of invasion of privacy, breech of confidentiality or psychological injury caused by "labeling: someone or imparting unwarranted fear and wear about the significance of a disease or carrier state.

Candidates for Referral for Genetic Testing or Counseling

- A couple who has a child with a congenital disorder or an inborn error of metabolism
- A couple whose close relatives have a child with a genetic disorder such as chromosomal disorder or an inborn error of metabolism
- Any individual who is a known carrier of a chromosomal disorder
- Any individual who has an inborn error of metabolism or chromosomal disorder.
- A consanguineous couple
- Any woman older than 35 years of age and any man older than 55 years
- Couples of ethnic background in which specific illnesses are known to occur.

Reproductive Alternatives

Some couples are reluctant to seek genetic counseling because they are afraid they will be told it would be unwise to have children. Helping them to realize viable alternatives for having a family exist can allow them to seek the help they need.

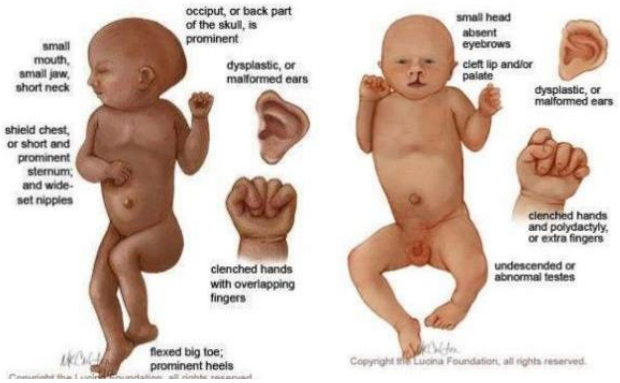
- Alternative Insemination by Donor (AID) – if inherited by male partner or is recessively inherited disorder carried by both partners
- Surrogate embryo transfer – if inherited by a female
- Surrogate mother
- Adoption

Chromosomal Disorders

- *Trisomy 13 Syndrome (47XY13 or 47XX13) or Patau Syndrome*
  - The child has an extra chromosome 13 and is severely cognitively challenged.
  - Incidence of the syndrome is low, approx. 0.45 per 1,000 live births

Manifestations:

- Midline body disorders (cleft lip and palate), heart defects – particularly ventricular septal defects, and abnormal genitalia are present
- Other common findings include: microcephaly with abnormalities of the forebrain and forehead; eyes that are smaller than normal (microphthalmos) or absent; and low-set ears.
- Most children do not survive beyond early childhood.



- *Trisomy 18 Syndrome (47XY18 or 47XX18) or Edwards' Syndrome*
  - Three copies of chromosome 18
  - Incidence is approx. 0.23 per 1,000 live births
  - Children are severely and cognitively challenged and tend to be small for gestational age at birth
  - Have markedly low-set ears, small jaw, congenital heart defects, and usually misshapen fingers and toes (the index finger deviated or crosses over other fingers)
  - The soles of their feet are rounded instead of flat (rocker-bottom feet)
  - Most of these children do not survive beyond early infancy.

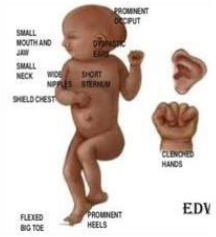
Edwards' syndrome

Edwards' syndrome (trisomy 18):

- It is the second most common autosomal trisomy, occurring in approximately 1 in 7500 live births.
- Greater than 95% of conceptuses with trisomy 18 are spontaneously aborted in the first trimester.
- Trisomy 18 is usually lethal, less than 10% of affected infants survive until their first birthday.
- The diagnosis is confirmed by chromosome analysis.

Clinical Features:

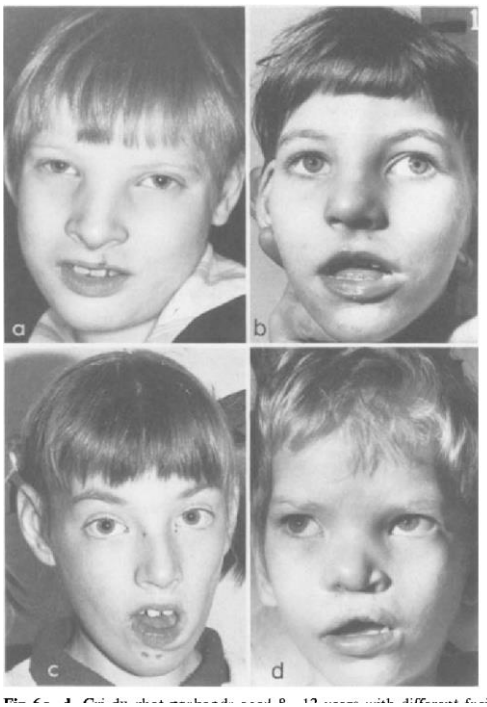
- Low birth weight.
- Prominent occiput.
- Small mouth and chin.
- Cleft lip or palate.
- Low set & malformed ears.
- Short sternum.
- Flexed, overlapping fingers.
- Rocker-bottom feet.
- Cardiac and renal malformations.
- Severe developmental delays.

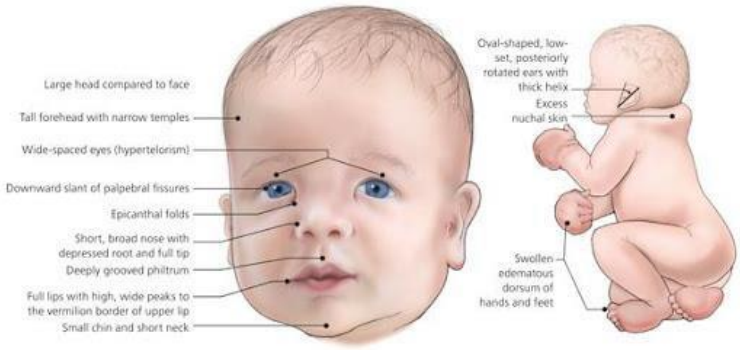


Management:

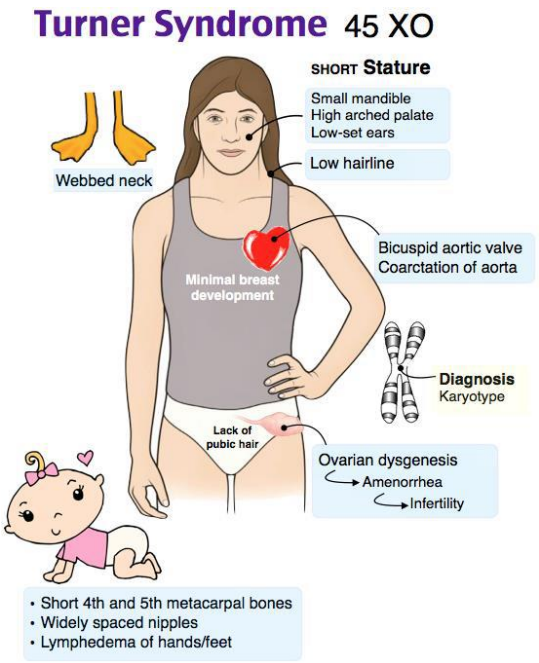
- Neonatal intensive care (NICU) management:
  - Resuscitation, respiratory support, and surgical procedures.
- Medical care:
  - A goal-directed management plan based on careful risk-benefit assessment for the individual patient and developed collaboratively between health professionals and parents is recommended.
  - Treat infections as appropriate (RTI, UTI, Sepsis).
  - Provide nasogastric and gastrostomy supplementation for feeding problems.
  - Orthopedic management of scoliosis may be needed secondary to hemivertebrae.

- *Cri-du-Chat Syndrome (46XX5P or 46XY5P)*
  - Result of a missing portion of chromosome 5
  - Child has an abnormal cry, which sounds much more of a cat's
  - Small head, wide-set eyes, and a downward slant to the palpebral fissure of the eye
  - The children are severely and cognitively challenged.

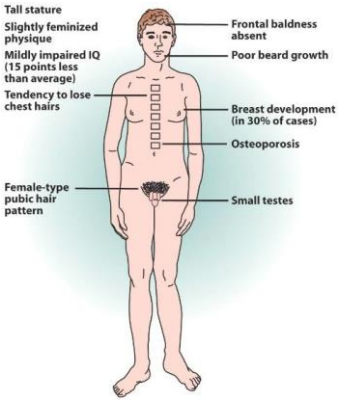


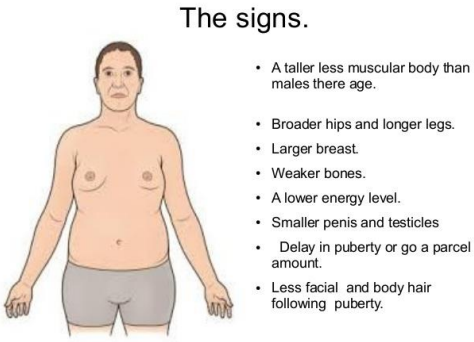


- *Turner Syndrome (45X0) or Gonadal dysgenesis*
  - Child has only one functional X chromosome
  - Short in stature, has only streak (small and nonfunctional) ovaries.
  - She is sterile with the exception of pubic hair; secondary sex characteristics do not develop at puberty.
  - The hairline at the nape of the neck is low set, and the neck may appear to be webbed and short.
  - A newborn may have appreciable edema of the hands and feet and a number of congenital anomalies, most frequently coarctation (stricture) of the aorta and kidney disorders.
  - Incidence is approx. 1 per 10, 000 live births
  - The disorder can be identified with an ultrasound during pregnancy because of the increased neck folds.
  - Although children with Turner syndrome may be severely cognitively challenged, difficulty in this area is more commonly limited to learning disabilities.
  - Socioemotional adjustment problems may accompany the syndrome because of the lack of fertility and if the nuchal folds are prominent.

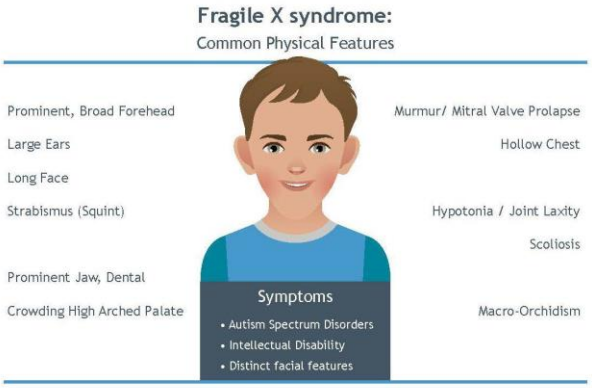


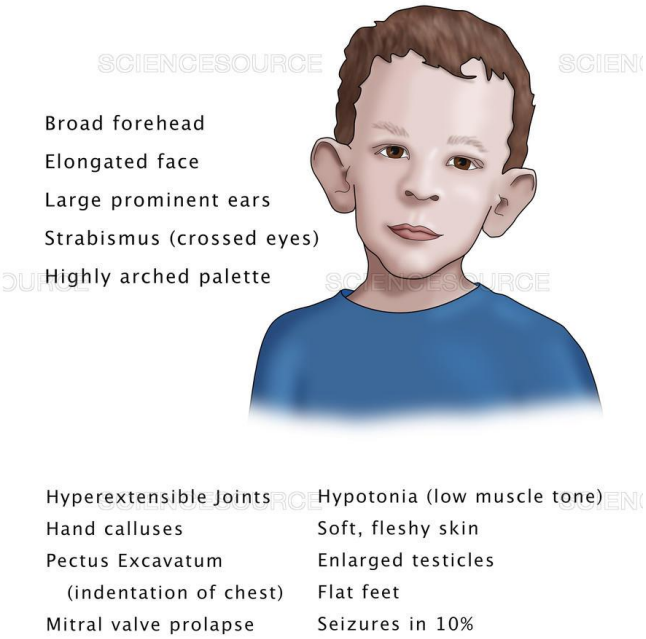
- *Klinefelter Syndrome (47XXY)*
  - Males with an extra X chromosome.
  - Characteristics of the syndrome may not be noticeable at birth.
  - At puberty, secondary sex characteristics do not develop. The child has small testes that produce ineffective sperm (Porche, 2007).
  - Affected individuals tend to develop gynecomastia (increased breast size) and have an increased risk of male breast cancer (Pyeritz, 2009).
  - Incidence is about 1 per 1,000 live births.
  - Karyotyping can be used to reveal the additional X chromosome.



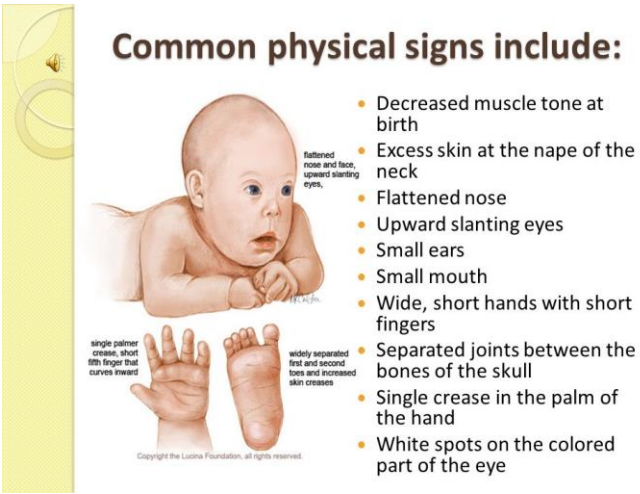


- *Fragile X Syndrome (46XY23q)*
  - The most common cause of cognitive challenge in males
  - X-linked disorder in which one long arm of an X chromosome is defective, which results in inadequate protein synaptic responses.
  - Incidence is about 1 in 1,000 live births.
  - Before puberty, boys with fragile X syndrome typically may demonstrate maladaptive behaviors such as hyperactivity and autism. They may have reduced intellectual functioning, with marked deficits in speech and arithmetic.
  - May be identified by the presence of a large head, a long face with a high forehead, a prominent lower jaw, and large protruding ears. Hyperextensive joints and cardiac disorders may also be present.
  - After puberty, enlarged testicles may become evident. Affected individuals are fertile and can reproduce.





- *Trisomy 21 (47XY21 OR 47XX21) or Down Syndrome*
  - Most frequently occurring chromosomal abnormality
  - Occurs in about 1 in 800 pregnancies.
  - The number of children born with the disorder is considerably less as many women choose to end pregnancies when the diagnosis is made (Witters & Fryns, 2008).
  - The physical features of children with Down syndrome are so marked that fetal diagnosis is possible by ultrasound in utero.
  - The nose is broad and flat. The eyelids have an extra fold of tissue at the inner canthus (an epicanthal fold), and the palpebral fissure (opening between the eyelids) tends to slant laterally upward. The iris of the eye may have white specks, called **Brushfield spots**.
  - The tongue may protrude from the mouth because the oral cavity is smaller than usual. The back of the head is flat, the neck is short, and an extra pad of fat at the base of the head causes the skin to be so loose it can be lifted easily. The ears may be low-set.
  - Muscle tone is poor, giving the baby a rag-doll appearance. This can be so lax that the child's toe can be touched against the nose (not possible in the average mature newborn).
  - The fingers of many children with Down syndrome are short and thick, and the little finger is often curved inward.
  - There may be a wide space between the first and second toes and between the first and second fingers.
  - The palm of the hand shows a peculiar crease (a simian line), which is a single horizontal palm crease rather than the usual three creases in the palm
  - Children with Down syndrome are usually cognitively challenged to some degree. The challenge can range from an intelligence quotient (IQ) of 50 to 70 to a child who is profoundly affected (IQ less than 20). The extent of the cognitive challenge is not evident at birth.
  - The fact that the brain is not developing well is evidenced by a head size that is usually smaller than the 10th or 20th percentile at well-child health care visits.
  - These children also appear to have altered immune function as they are prone to upper respiratory tract infections.
  - Congenital heart disease, especially atrioventricular defects, is common. Stenosis or atresia of the duodenum, strabismus, and cataract disorders are also common.
  - For as yet undetected reasons, acute lymphocytic leukemia occurs approximately 20 times more frequently in children with Down syndrome than in the general population.



Mnemonics

Clinical Features of **Down Syndrome-**

SHIT-DOWN-SYNDROME

S- Short neck
H- Hair thin
I- IQ level (20-75)
T- Transient Leukemoid reaction
D- Duodenal Atresia
O- Occiput Flat
W-Wide Ulnar loop
S- Simian crease
Y- Yellow skin (jaundice)
N- No fertility
D- Delayed fontanelles closure
R- Rolling eye
O- Only male infertility, female fertile
M-Mentally ill
E- Epicanthic folds

egurukul
Inspired Learning