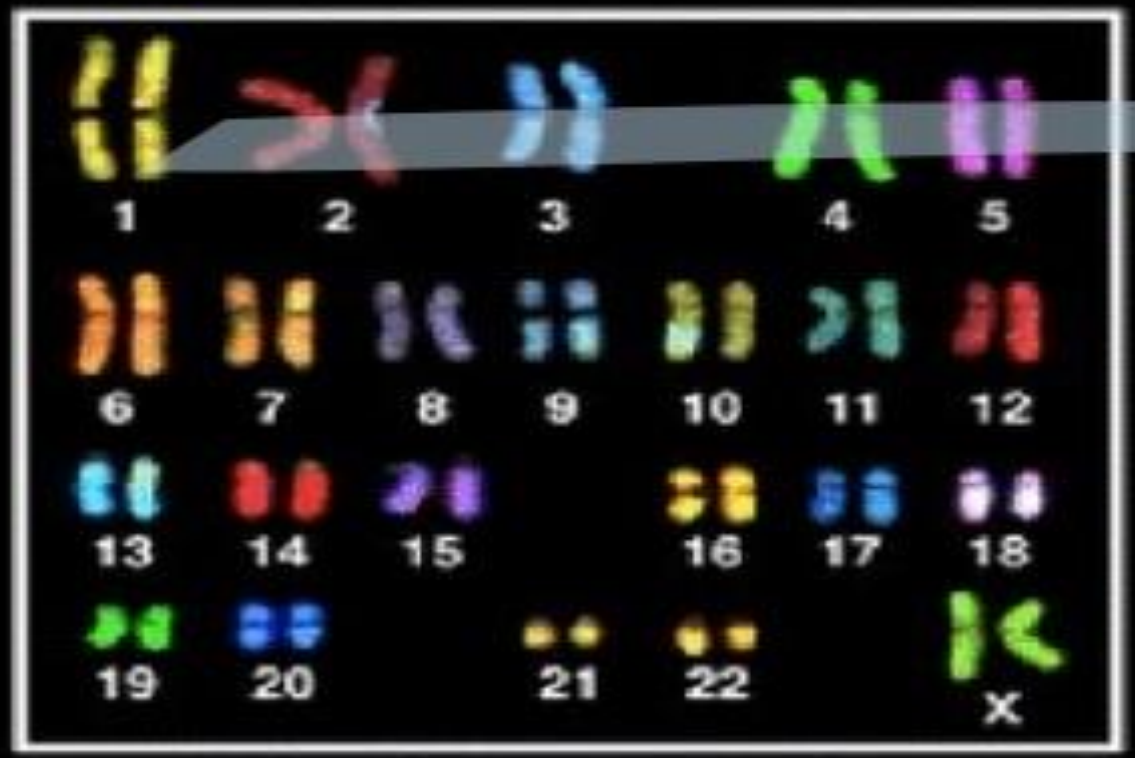
GENETIC DISORDERS

Inheritance of Genetic Traits



SYLLABUS

- Down’s Syndrome,
- Turner’s syndrome,
- Kleinfelter’s syndrome,
- Edwards syndrome,
- Inborn errors of metabolism: phenylketonuria,albinism,galactosemia
- Genetic counseling

DOWN SYNDROME

- Down syndrome (sometimes called Down's syndrome) is a condition in which a child is born with **an extra copy of their 21st chromosome**—hence its other name**, trisomy 21**. This causes physical and mental developmental delays and disabilities

DOWN SYNDROME

Causes

- In all cases of reproduction, both parents pass their genes on to their children. These genes are carried in chromosomes.
- When the baby's cells develop, each cell is supposed to receive 23 pairs of chromosomes, for 46 chromosomes total. Half of the chromosomes are from the mother, and half are from the father.
- In children with Down syndrome, one of the chromosomes doesn't separate properly. The baby ends up with three copies, or an extra partial copy, of chromosome 21, instead of two. This extra chromosome causes problems as the brain and physical features develop.

Types

Trisomy 21

- Trisomy 21 means there's an extra copy of chromosome 21 in every cell.
- This is the most common form of Down syndrome.

Mosaicism

- Mosaicism occurs when a child is born with an extra chromosome in some but not all of their cells.
- People with mosaic Down syndrome tend to have fewer symptoms than those with trisomy 21.

Translocation

- In this type of Down syndrome, children have only an extra part of chromosome 21. There are 46 total chromosomes. However, one of them has an extra piece of chromosome 21 attached.

risk factors

- . According to the Centers for Disease and Prevention, mothers aged 35 and older are more likely to have a baby with Down syndrome than younger mothers. The risk increases the older the mother is.
- people with a family history of Down syndrome
- people who carry the genetic translocation

Symptoms

At birth, babies with Down syndrome usually have certain characteristic signs, including:

- flat facial features
- small head and ears
- short neck
- bulging tongue
- eyes that slant upward
- atypically shaped ears
- poor muscle tone

- An infant with Down syndrome can be born an average size, but will develop more slowly than a child without the condition.
- People with Down syndrome usually have some degree of developmental disability, but it’s often mild to moderate.

Mental and social development delays may mean that the child could have:

✓impulsive behavior

✓poor judgment

✓short attention span

✓slow learning capabilities

Medical complications often accompany Down syndrome. These may include:

✓congenital heart defects

✓hearing loss

✓poor vision

- ✓cataracts (clouded eyes)
- ✓hip problems, such as dislocations
- ✓leukaemia
- ✓chronic constipation
- ✓sleep apnea (interrupted breathing during sleep
- ✓dementia (thought and memory problems)
- ✓hypothyroidism (low thyroid function)
- ✓obesity
- ✓late tooth growth, causing problems with chewing
- ✓Alzheimer’s disease later in life
- ✓People with Down syndrome are also more prone to infection. They may struggle with respiratory infections, urinary tract infections, and skin infections.

HALLS CRITERIA

- Flat facial profile
- Poor Moro reflex
- Excessive skin at the nape of neck
- Slanted palpebral fissures
- Hypotonia ,
- Hyper flexibility of joints
- Dysplasia of pelvis
- Anomalous ears
- Dysplasia of midphalanx of fifth finger
- Transverse palmer crease

Screening for Down syndrome during pregnancy

Screening for Down syndrome is offered as a routine part in some states. If you're a woman over 35, your baby's father is over 40, or there's a family history of Down syndrome, you may want to get an evaluation.

First trimester

- An **ultrasound** evaluation and blood tests can look for Down syndrome in fetus.
- These tests have a higher false-positive rate than tests done at later pregnancy stages.
- If results aren't normal, doctor may follow up with an **amniocentesis** after your 15th week of pregnancy.

Additional prenatal tests

- **Amniocentesis**. Takes a sample of amniotic fluid to examine the number of chromosomes . The test is usually done after 15 weeks.
- **Chorionic villus sampling (CVS)**. doctor will take cells from placenta to analyze fetal chromosomes. This test is done between the 9th and 14th week of pregnancy. It can increase the risk of a miscarriage, but according to the Mayo Clinic, only by less than 1 percent.
- **Percutaneous umbilical blood sampling (PUBS, or cordocentesis).** Take blood from the umbilical cord and examine it for chromosomal defects. It's done after the 18th week of pregnancy. It has a higher risk of miscarriage, so it's performed only if all other tests are uncertain.
- Some women choose not to undergo these tests because of the risk of miscarriage. They'd rather risk having a child with Down syndrome than lose the pregnancy.

Second trimester

- **An ultrasound and quadruple marker screen (QMS) test** can help identify Down syndrome and other defects in the brain and spinal cord. This test is done between 15 and 20 weeks of pregnancy.
- If any of these tests aren’t normal, you’ll be considered at high risk for birth defects.

TRIPLE TEST

- It is an investigation performed in the second trimester for diagnosing the fetus have chromosomal anomalies such as Down syndrome or neural tube defect (15-18weeks)
- Triple screen is a blood test that measures three things called alpha-fetoprotein, human chorionic gonadotropin and unconjugated estriol. The results of the blood test can help your doctor see if your baby may be at higher risk for certain birth defects
- In trisomy 18 all the components are below normal level
- It involves drowning blood from the mother and send to the laboratory

QUADRUPLE TEST

- The test is done to find out if your baby might be at risk for certain birth defects, such as Down syndrome and birth defects of the spinal column and brain (called neural tube defects). ... Certain women are at greater risk of having a baby with these defects, including: Women who are over 35 years old during pregnancy
- The quadruple screen test is a maternal blood screening test that looks for four specific substances**: AFP , hCG , Estriol, and Inhibin-A**.
- The quad screen is a maternal blood screening test which is similar to the Triple Screen Test (also know as AFP Plus and the Multiple Marker Screening).
- The **quad** marker screening **test** (**quad** screen) is a blood **test** administered in pregnancy, typically between the 15th and 20th weeks of gestation.

Tests at birth

- At birth, your doctor will:
- perform a **physical examination** of baby
- order a **blood test** called a karyotype to confirm Down syndrome

Treatment

In these programs, special education teachers and therapists will help your child learn:

- sensory skills
- social skills
- self-help skills
- motor skills
- language and cognitive abilities

- Children with Down syndrome often meet age-related milestones. However, they may learn more slowly than other children.
- School is an important part of the life of a child with Down syndrome, regardless of intellectual ability.
- Public and private schools support people with Down syndrome and their families with integrated classrooms and special education opportunities.
- Schooling allows valuable socialization and helps students with Down syndrome build important life skills.

TURNER SYNDROME

- **Turner syndrome is a genetic condition caused by an abnormality on one of your sex chromosomes. It's also called monosomy X, gonadal dysgenesis, and Bonnevie-Ullrich syndrome.**
- Only the female sex develops this condition

TURNER SYNDROME

- Turner syndrome occurs when part or all of one of your X chromosomes is missing. This condition affects approximately 1 in 2,000 females.

- People with Turner syndrome can lead healthy lives. But they typically require some consistent, ongoing medical supervision to detect and treat complications.

- There is no way to prevent Turner syndrome, and the cause of the genetic abnormality is unknown.

Symptoms

Females with Turner syndrome exhibit certain physical characteristics at birth and in childhood, including:

- swollen hands and feet (in infants)
- short stature
- a high palate
- low-set ears
- obesity
- droopy eyelids
- flat feet

Females with this condition may also have other medical problems associated with Turner syndrome, including:

- heart defects
- infertility
- problems with sexual development
- hearing loss
- high blood pressure
- dry eyes
- frequent ear infections
- scoliosis (spinal curvature)

- These symptoms can appear **early in infancy**. Or, in the case of **sexual development and fertility issues**, they can develop later in **adolescence.**
- Having one or more of these symptoms doesn’t mean that you have Turner syndrome.
- It’s important young females suspected of having this syndrome get a thorough examination from a doctor for an accurate diagnosis.

Diagonosis

- Prenatal genetic testing done before birth can help a doctor diagnose Turner syndrome.
- The condition is identified through karyotyping.
- When performed during prenatal testing, karyotyping can detect if the mother’s chromosomes have any genetic abnormalities.

Tests to look for the physical symptoms of Turner syndrome. These tests may include:

- blood tests to check sex hormone levels
- echocardiogram to examine for heart defects
- pelvic exam
- pelvic and kidney ultrasound
- chest MRI

Treatments

- can still lead a healthy life if you’re diagnosed with Turner syndrome.
- There is no cure, but there are treatments that can ease your symptoms and improve your quality of life.
- Growth hormone injections may help children with Turner syndrome grow taller.
- **Hormone therapy** can also aid in the development of secondary sex characteristics like breasts and pubic hair. It’s usually administered at the start of puberty.
- Women who are infertile because of Turner syndrome can use donor eggs to get pregnant. Finding a support group for women with the condition, or talking to a counselor, can give you **emotional support** and any other challenges you may encounter as a result of your condition.
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Complications

- **Kidney abnormalities** are common.
- Some females with Turner syndrome also have **recurrent urinary tract infections.**
- The **kidneys** may be **malformed** or in the wrong position in the body.
- These abnormalities can increase the risk of **high blood pressure.**
- Hypothyroidism is a condition that the low levels of thyroid hormone. This could be another complication. It's caused by the **inflammation of the thyroid gland.** Supplemental thyroid hormone can treat it.

- People with Turner syndrome also have a **higher-than-average risk of developing celiac disease.**
- Celiac disease causes the body to have an **allergic reaction to the protein gluten, found in foods like wheat and barley.**
- **Heart abnormalities** are common in individuals with Turner syndrome. People with the condition should be monitored for problems with their aorta and high blood pressure.
- **Obesity** may be a complication for some individuals with Turner syndrome. It can increase the risk of developing diabetes.

KLINEFELTER'S SYNDROME

- **Klinefelter syndrome (KS) is a condition that occurs in men who have an extra X chromosome.**
- **The syndrome can affect different stages of physical, language, and social development.**

symptoms

- 1.The most common symptom is infertility.
- 2.Boys may be taller than other boys their age, with more fat around the belly. After puberty, KS boys may have
- 3.Smaller testes and penis
- 4.Breast growth
- 5.Less facial and body hair
- 6.Reduced muscle tone

- 7.Narrower shoulders and wider hips
- 8.Weaker bones
- 9.Decreased sexual interest
- 10.Lower energy
- 11. KS males may have learning or language problems. They may be quiet and shy and have trouble fitting in.

DIAGONOSIS

- A genetic test can diagnose KS.

TREATMENTS

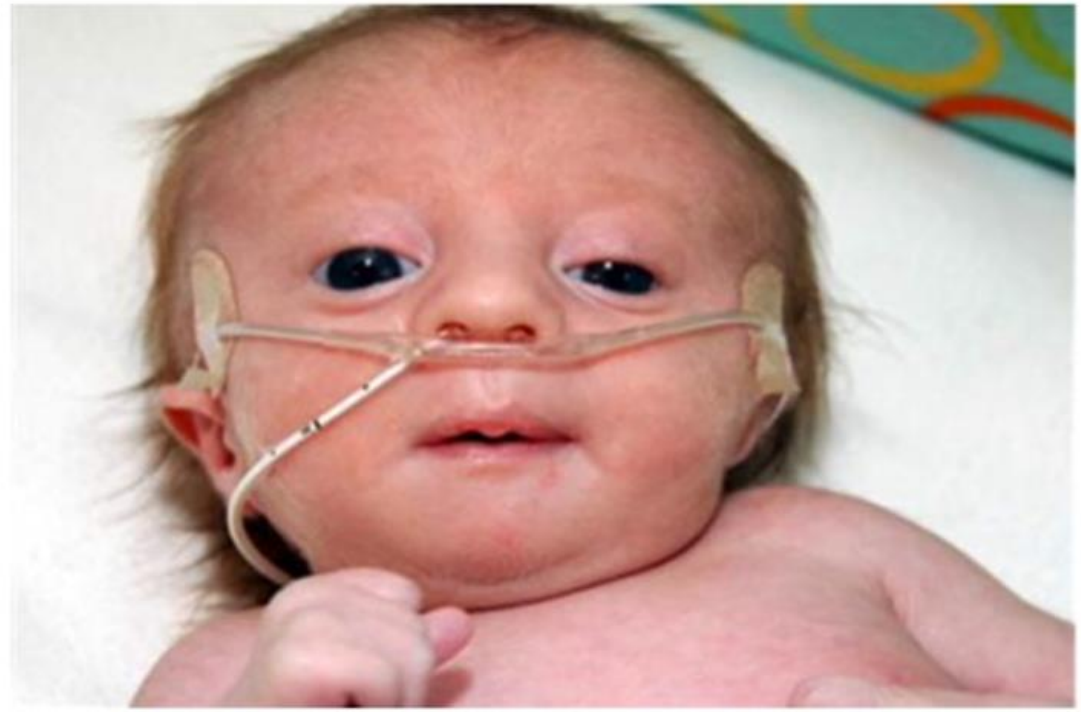
- There is no cure, but treatments are available
- It is important to start treatment as early as possible.
- With treatment, most boys grow up to have normal lives.
- Treatments include testosterone replacement therapy and breast reduction surgery.
- If needed, physical, speech, language, and occupational therapy may also help.

EDWARDS SYNDROME

- Edward syndrome also known **as trisomy 18**, is a genetic disorder caused by a third copy of all or part of chromosome 18. Many parts of the body are affected. Babies are often born small and have **heart defects.** Other features include a **small head, small jaw, clenched fists with overlapping fingers, and severe intellectual disability.**
- Most cases of Edwards syndrome occur due to problems during the formation of the reproductive cells or during early development. The rate of disease increases with the mother's age. Rarely cases may be inherited from a person's parents.

Edwards Syndrome

Also called Trisomy 18



Lexi Ippolito

Causes

- Edwards syndrome is a chromosomal abnormality characterized by the presence of an extra copy of genetic material on the 18th chromosome, either in whole (trisomy 18) or in part (such as due to translocations). The additional chromosome usually occurs before conception. The effects of the extra copy vary greatly, depending on the extent of the extra copy, genetic history, and chance.

- Edwards syndrome occurs in all human populations, but is more prevalent in female offspring.
- Trisomy 18 (47,XX,+18) is caused by a meiotic nondisjunction event. With nondisjunction, a gamete (i.e., a sperm or egg cell) is produced with an extra copy of chromosome 18; the gamete thus has 24 chromosomes. When combined with a normal gamete from the other parent, the embryo has 47 chromosomes, with three copies of chromosome 18.

Signs and symptoms

- Children born with Edwards syndrome may have some or all of these characteristics:
- kidney malformations,
- structural heart defects at birth (i.e., ventricular septal defect, atrial septal defect, patent ductus arteriosus),
- intestines protruding outside the body (omphalocele),
- esophageal atresia,
- intellectual disability, developmental delays,
- growth deficiency, feeding difficulties, breathing difficulties,
- and arthrogryposis (a muscle disorder that causes multiple joint contractures at birth).

Some physical malformations associated with Edwards syndrome include

- small head (microcephaly) accompanied by a prominent back portion of the head (occiput),
- low-set, malformed ears,
- abnormally small jaw (micrognathia),
- cleft lip/cleft palate, upturned nose, narrow eyelid folds (palpebral fissures), widely spaced eyes (ocular hypertelorism), drooping of the upper eyelids (ptosis),
- a short breast bone, clenched hands, choroid plexus cysts, underdeveloped thumbs and/or nails, absent radius, webbing of the second and third toes, clubfoot or rocker bottom feet, and in males, undescended testicles.

In utero,

- the most common characteristic is cardiac anomalies, followed by central nervous system anomalies such as head shape abnormalities. The most common intracranial anomaly is the presence of choroid plexus cysts, which are pockets of fluid on the brain.