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कौमार भृत्य की निरूक्ति– कौमार भृत्य दो शब्दों से मिलकर बना है– कुमार और भृत्य।

1. **कुमार–** कुमार शब्द का व्यवहार बालक, युवराज, राजकुमार, पाँच वर्ष तक के बालक या अविवाहित पुरूष के लिए किया जाता है परंतु कौमार भृत्य के अंर्तगत इन शब्द का प्रयोग बालक के अर्थ में ही किया जाता है।

   कु+मा+र से मिलकर कुमार बना है।

कुमार की निरूक्ति के अंर्तगत–

–कार्तिकेय को कुमार कार्तिकेय कहने से।

–बच्चा जन्म के बाद, उसके मृत्यु की संभावना अधिक होती है।

–कुमारो मारोडस्यति–बच्चे की बुद्वि निर्गम रहती है।

कुत्सितो मारोडस्येति कुमार वा

–कुमारयति क्रीडति, क्रीडायाम्।

बच्चो का ज्यादातर समय खेलने मे जाता है।

2. **भृत्य–** भृ धातु मे त्य प्रत्यय लगाकर भृत्य शब्द बना है, जिसको अर्थ भरण–पोषण करने वाले के लिए किया जाता है।

कौमारभृत्य की व्याख्या–

" कुमारस्य भरणमाधिकृत्य कृर्त कौमारभृत्यम।"(चरक)

अर्थात कुमारो के भरण–पोषण के लिए जो तंत्र है उसे कुमार भृत्य कहते है।

"कौमारभृत्यनाम् कुमारभरण धात्री क्षीरदोष संसोधनार्थ दृष्टस्तन्य ग्रहसमुत्धानां च त्याधीनामुपशमनार्थ।"

(सु.सू.1/7)

अर्थात कौमारभृत्य नामक तंत्र कुमार के भरण–पोषण, धात्री, के दुग्धसंशोधनार्थ तथा विकृत दुग्ध से उत्पन्न होने वाली व्याधियों एवं ग्रहो से उत्पन्न होने वाली व्याधियों एवं अन्य व्याधियों के शमनार्थ हैं।

"गर्भोपक्रमविज्ञानं सूतिकोपक्रमस्तथा। बालानां रोगशमनी क्रिया बालचिकित्सतम्।"

(हा.सॅ.प्रथमस्थान.अ.2/17)

गर्भ के लिए उपक्रम अथवा गर्भकालीन उपक्रम, सूतिकाकालीन व्यवस्था एवं बालकों के रोगों का संशमन इन सबका अन्तभवि (कौमारभृत्य) बाल चिकित्सा में किया जाता है।

आधुनिक मतानुसार– आधुनिक बालचिकित्सा (paediatrics) के विशेषज्ञ भी यही कहते है कि जिस विज्ञान में बच्चे का अध्ययन गर्भाधान से प्रारंभ होता है पूरे शैशवकाल में चलता है और युवा होने तक किया जाता है, वह paediatrics है।

It is a study of the child from very conuption through childhood, up to adoleseence.

Paediatric शब्द ग्रीफ शब्दों के योग से बना है–

Paediatric-paedia + iatrike + ics

Pedia:meaning a child or pertaining to a child: शिशु या शिशु विषयक

Iatrike:meaning treatment: चिकित्सा

ics : meaning a branch of science : विज्ञान की एक शाखा

अर्थात विज्ञान की वह शाखा जिसमें शिशु या शिशु विषयक चिकित्सा का अध्ययन होता है Paediatrecs कही जाती है।

कौमारभृत्य की उपयोगिता एवं महत्व–

1. बलकों की दी जाने वाली औषधि दुध होनी चाहिए। उनकों दी जाने वाली औषधि की मात्रा भिन्न होती है। उनके रोगों के निदान एवं चिकित्सा में प्रयुक्त होने वाले उपक्रम भिन्न होते है इसी कौमारभृत्य के द्वारा पोषित वृद्वि को प्राप्त हुए अन्य लोग भी चिकित्सा करते है।
2. बाल्यावस्था जीवन का निर्माण काल है। गर्भावस्था से लेकर 16 साल की उम्र तक उसमें निरंतर वृद्वि और विकास की प्रक्रिया चलती रहती है। शारिरिक और मानसिक विकार प्रायः समान्तर गति से चलते रहते है। बालक को अपने माता–पिता से जो बीजरूप में मिल जाता है। वही अनुकुल अवसर, पोषण और परिवेश पाकर पनपता/विकसित होता है। गर्भ की अवस्था से प्र्याप्त पोषण मिलता है, प्रसव–काल से ठीक से देख–रेख की जाती है, प्रसव सामान्य होता है, नवजात की देख–रेख के प्रति आवश्यक सावधानी बरती जाती है, उसकी परिवेश के प्रतिकूल प्रभावो से हर संभव प्रकार के रक्षा की जाती, उसकी रोगप्रतिरोधक क्षमता को बनाये रखने के लिए हर आवश्यक कदम उठाया जाता है, खेलने का उचित प्रबंध किया जाता है, प्रज्ञापराध से बचाया जाता है, सदवृत्त का पालन आदि संरूकार किया जाता है।
3. कौमारभृत्य बालक को शारीरिक, बौद्विक,मनोवैज्ञानिक विकास की ओर ध्यान देता है। यदि बालको का लालन–पालन, बतलाये गये नियमों के अनुरू की जाए तो निश्चित हो बालक की आयु सुखायु होगी और वह एक स्वरूप और सुखी प्रौढ़ के रू में विकसित होगा।
4. बाल्यावस्था में कफ की प्रधानता होती है। अतः इस अवस्था में उन्हे कफज या कफ की प्रधानता वाले रोगों से पीड़ित होने की अधिक संभावना रहती है। दुर्घटनाँए भी इस अवस्था में अधिक होती है।

कौमारभृत्य का क्षेत्र– उत्तर तंत्र के विषय का वर्णन करने के बाद आचार्य सुश्रुत कहते हैं कि शुक्र एवं आर्तव आदि के दोष, स्तन के रोग, क्लैत्य, मूढ़गर्भ, गर्भिणी के रोग या गर्भिणी की भासानुमासिक चिकित्सा, दौहद्य व्यवस्था, योनित्यापद यंधयत्व चिकित्सा एवं उत्तरस्थान में वर्णित संबंधित अंश अथवा प्रसवोपरांत होने वाली व्याधियों का संशमन कौमारभृत्य का क्षेत्र है।

काश्यप संहिता की विशिष्टा–

1. वृद्व जीवकीय ही काश्यप संहिता है।
2. कौमारभृत्य का सबसे प्रधान ग्रंथ काश्यप संहिता को मानस जाता है। क्योकि इसमें जो मुख्य विषय है वह बालक है, बालक को केंद्र में रखकर इसकी रचना की गई है।
3. इसमें बालकों की उत्पत्ति, वृद्वि,विकास, परिचर्या,र रोगनिदान, व बालग्रह तथा उनकी चिकित्सा सभी का विस्तार से वर्णन किया गया है।
4. गर्भिणी परिचर्या का विस्तृत वर्णन है।
5. दुष्टप्रजाता, दोषों के निराकरण/उपचार गर्भावस्थाके दौरान या प्रसव के बाद मृत्यु हो जाती है।
6. धात्री का वर्णन किया गया है।
7. दन्तजनिक अध्याय में दंतोद्भव, दंतगत वेदना की चिकित्सा की जाती है। दंतोद्भेद जन्य व्याधि एवं उनकी चिकित्सा एवं दंत सम्पत की चिकित्सा की जाती है।
8. लक्षणाध्याय में सामुदिक लक्षणें का वर्णन किया गया है।
9. स्वेदाध्याय में बच्चों में हस्त स्वेद करने को कहा गया है।
10. प्रसव काल में कुछ आचार्यों के मूसल चलाने के मतका काश्यप ने खण्ड़न किया है।
11. 6 मास में फलरस प्रासन कराने का मत का काश्यप संहिता में मिलता है।
12. कुमारागाह, क्रीडाभूमि, क्रीडानक का विस्तृत वर्णन काश्यप संहिता में मिलता है।
13. वय विभाजन का वर्णन किया गया है।
14. काश्यप ने 20 बालग्रह में रेवतीग्रह को श्रंष्ठ माना है।
15. काश्यप संहिता में वेदनाध्याय का वर्णन है। बच्चे अपनी तकलीफ स्वयं नहीं बता पाते इसलिए उनके लक्षणों द्वारा व्याधि का ज्ञान करना इसमे वर्णित है।
16. ल्हन अध्याय का वर्णन– बच्चो को दवाई चटाना, ऐसी व्याधियाँ जिसमे बच्चों को औषधि अवलेहरूप में (चटाई जाती है) दी जाती है, उनका वर्णन है। किन बालकों में लेहन का निषेध है यह भी वर्जित है।
17. कुमारागार, क्रीडाभूमि, क्रीडानक का विस्तृत वर्णन मिलता है तथा कुपोषण जन्य व्याधियों का एवं उनकी चिकित्सा हेतु भिन्न–भिन्न अध्यायों की रचना की गई है जैसे फक्क रोग के लिए फक्कचिकित्सा अध्याय की रचना की गई है जिसमें उसकी चिकित्सा में फक्क रध का वर्णन मिलता है जिसे फक्क रोगी शिशु को चलाने हेतु प्रयोग कराया जाता है।

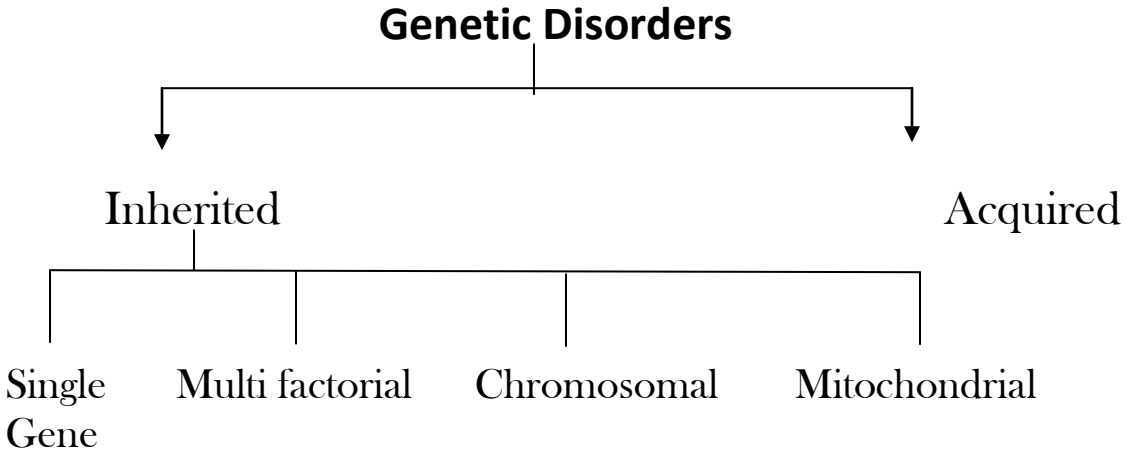
विषय प्रवेश

GENETIC DISORDERSyu

Definition- A genetic problem caused by one as more abuormalities formed in the genome. They can be hereditary or non hereditary.

A genetic disease is any disease caused by an abuormalitaty in the genetic make up of an individual. The genetic abuormality can range from minor to major. It can be inherited from parents or can be required mutations in a preexisting gene/group of genes causing other genetic diseases. Genetic mutations can occur either randomly or due to some environmental exposure.

Types of genetic disorders-



1. **Single gene inheritance-** This is also called mendelian or monogenetic inheritance.

Changes / mutations that occur in the DNA sequence of a single gene cause this type of inheritance. The disorders are called monogenetic disorders. Single gene disorders have different patterns of genetic inheritance, including

- Autosomal dominant inheritance- only one copy of a defective gene is necessary to cause the condition.

- Autosomal recessive inheritance- two copies of a defective gene are necessary to cause the condition.
- X-linked inheritance, in which the defective gene is present on the female. (x-chromo) if may be dominant or recessive. Ex. α&β- thalassemia, sickle cell anemia, fragile x-syndrome, hemo chromatosis.

2. **Multifactorial genetic inheritance-**

Multifactorial inheritance is also called complex or polygenic inheritance. Multifactorial inheritance disorders are caused by a combination of environmental factors and mutations in multiple genes. For eg. heart disease, Alzheimer`s disease, diabetes, cancer, etc. Multifactorial inheritance also is associated with heritable traits much as fingerprint patterns, height, eye color, and skin color.

3. **Chromosomal abnormalities-**

Chromosomes, made up of DNA and protein are located in the nucleus of each cell. Chromosomes are carriers of genetic material. Abnormalities in chromosome number or structure can result in disease.

Chromosomal abnormalities typically occurs due to a problem with cell division.

For ex- down syndrome/21 trisomy is common genetic disorder that occurs when a person has three copies of chromosome 21. There are many other chromosomal adnormalities including:

- Turner's syndrome (45,xo)
- Klinefelter syndrome (47,xxy)

4. **Mitochondrial genetic inheritance-**

This is caused by mutations in the non-nuclear DNA of mitochondria. Mitochondria are small round or rod-like organelles that are involved in cellular respiration and found in the cytoplasm of plant and animal cells. Each mitochondrion may contain 5 to 10 circular pieces of DNA since egg cells, but not sperm cells, keep mitochondrial during fertilization, mitochondrial DNA is always inherited from the female parent.

Ex- mitochondrial encephalopathy, myclonic epilepsy, leber`s hereditary optic atrophy(an eye disease).

Some of the comman genetic disorders are-

- Sickle cell anemia
- Down`s syndrome
- Turner`s syndrome
- Thalasemia
- Muscular dystrophy
- Juvenile diabetes

1. **Sickle cell anemia-**

An inherited from of anemia in which there aren`t enough healthy red blood cells to carry adequate oxygen throughout our body.

This type of anemia affects hemoglobin in RBCs and makes it atypical hemoglobin molecule called hemoglobin S, which can distort red blood cells into a sickle or crescent shape.

It is a single gene inheritance disorder.

Sickle cell RGB	Normal RGB
- stiff sticky	- smooth, round and flexible
- sickle shaped	- can move through the vessels in our body vessels easily
- oxygen less	- live up to 120 days
- cluster together & cannot move through blood	
- live only for 10-20 days	

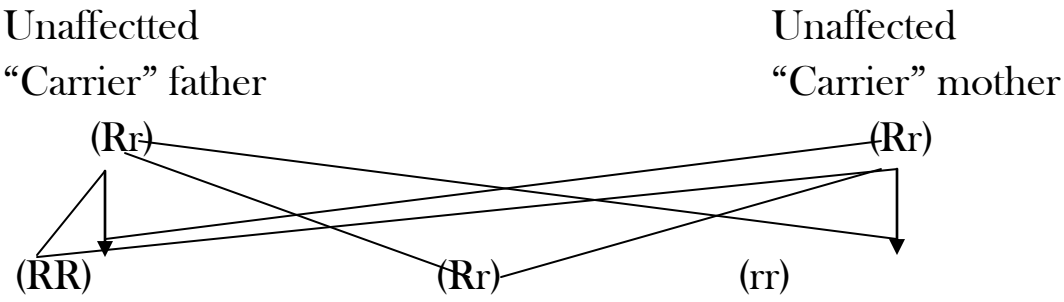
This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene but they typically do not show signs and symptoms of the condition.

- **Symptoms-**

- Fatigue and anemia jaundice

- Swelling and inflammation of hands & feet and arthritis.
- Bacterial infections.
- Sudden pooling of blood in the spleen and liver congestion.
- Lung and heart injury.
- Leg ulcers.
- Aseptic and bone infarcts [death of portions of bones].
- Eye damage.
- In younger children can suffer fever abdominal pain dactylitis.
- Affected infants do not develop symptoms in the first few months of life because the hemoglobin produced by the developing fetus protects the RBC from sickling. By 5 months of age, the sickling of RBC is prominent and symptoms begin.

- **Causes**- Genetic-inherited in an autosomal recessive pattern



Unaffected	unaffected	Affected
1 in 4 chance	2 in 4 chance	1 in 4 chance.

- **<u>Complications</u>**-

  Stroke, acute chest syndrome, pulmonary hypertension, Blindness, organ damage, infections, splenic sequestration (swelling with pain of spleen).

- **Line of treatment**-

Bone marrow transplant (stem cell transplant) offers the only potential use for sickle cell anemia. Its usually reserved for people younger than age 16 because the risk increase for people older than 16.

Medications:-

1. Antibiotics – children with sickle cell anemia may begin taking antibiotics penicillin when they are about 2 months old and continue taking it white 5 years old prevents infection.
2. Pain-relieving medication .
3. Hydroxyurea (droxia hydra)- reduces the frequency of painful cries and might reduce the need for blood transplant.
4. Vaccination (to prevent infection)- such the pneumococcal vaccine and the annual flu shot.
5. Blood transfusion.
6. Experimental treatment-gene therapy.
7. Diet &supplementation- daily intake of folic acid supplement, vitamin supplement.

2. **Down's Syndrome**- Down's Syndrome is the most common chromosomal disorder occurring with a frequency of 1:800 to 1:000 newborns. Chromosome number 21 is present in triplicate, the origin of the extra chromosome 21 being either maternal or paternal (mostly from mother). Often in offspring of mothers conceiving at older age.

Risk ratio	Age of mother
1:1500	15-29yr.
1:1800	30-34yr
1:270	35-39yr
1:100	40-44 yr
1:50	>45yr

(It is a type of chromosomal abnormality.)

- **Clinical features & Diagnosis-**

- Mental & physical retardation
- Flat facial profile
- An upward slant of eyes and epicanthic folds.
- Oblique palpebral fissure is obvious only when eyes are open.
- Nose- small with flat nasal bridge.
- Month- a narrow short palate with small teeth and furrowed protruding tongue.
- Significant hypotonia.
- The skill appears small and brachycephalic with flat occiput
- Ears are small and dysplastic.
- Characteristic facial grimace on crying.
- Hands are short and brood.
- Simian crease are usual.
- Wide gap between the first and second toe (Sandle gap).

▪ Cause of Down Syndrome-

Chromosomal abnormality. Normally, human cells contains 23 pairs of chromosomes. One chromosome in each pair comes from father and the other comes from mother. Down syndrome results when abnormal cell division involving chromosome 21 occurs. This result in on extra partial / full chromosome 21 this extra genetic material is responsible for the genetic characteristic features and developmental problems of Down's syndrome instead of 46 chromosomes plus two sex. Chromosomes there are 47.

▪ Complicatins.

- Heart defects
- Gastrointestinal defects- the baby may face
- Intestinal defect- severe vomiting.

- oesophagus defect- frequent choking (improperly connected with wind pipe)

- Anus defect- A condition called hirschsprung disease
  –intestinal nerve makup it difficult to stools (2-15% of infants with down`s syndrome)
- gastro esophageal reflex (heart burn)
- immune disorders- high risk of auto immune disorders.
- Eye problems- blockage in tear ducts, cataract.
- Hearing loss- ear infections due to narrow ear cands.
- Sleep apnea- due to incomplete developmental of child's soft tissues and skeletons that can obstruct their airways-harder to breathe due to lack of oxygen, sleep is affected-sleep apnea (hard to breathe during sleep)
- Obesity.
- Spinal problems- child can have a misalignment of the top 2 vertebrae in the neck(overextension of the neck).
- Dementia.
- Malfunctioning thyroid.
-

Line of treatment-

There is no standard treatment for downs syndrome but a variety of therapies can be used in early intervention programs and throughout a person's life to promote the greatest possible development, independence and productivity.

Physical therapy : includes activities and exercise that help build motor skills increase muscles strength and improve posture and balance.

Speech- language therapy: can help in improving their communication skills and use language more effectively.

Occupational therapy: helps find ways to adjust everyday tasks and conditions to match a person's need and abilities.

Emotional and behavioral therapy: Works to find useful response to both desirable and undesirable behaviors.

3. **Turner's syndrome-** Turner syndrome having 45X chromosomal constitution, has an incidence of about 1:3000 newborns. It

is also a chromosomal abnormality and is also called gonadal dysgenesis.

Turner syndrome, a condition that only affects females, results when one of the x chromosome (sex chromosome) is missing or partially missing [45,*or 45,*0]

This can cause a variety of medical`s developmental problems, including short height, failure of the ovaries to develop, heart defects. This may be diagnosed before birth (prenatally), during infancy or in early childhood.

- **Symptoms-**

1. **Before birth-** suspected prenatally based on prenatal cell-free DNA screening (through blood sample from the mother or prenatal ultrasound)
   - Heart abnormalities.
   - Abnormal kidneys.

2. **At birth / during pregnancy-**
   - Wide or web- like neck.
   - Low-set ears.
   - Broad chest with widely spaced nipples.
   - High, narrow root of the mouth (palate)
   - Arms that turn outward at the elbows.
   - Fingernails and toenails that are narrow and turned upward.
   - Swelling of the hands & feet.
   - Slowed growth.
   - Cardiac defects.
   - Low hair line at the back of the head.
   - Smaller lower jaw.
3. **In childhood, teens and adulthood-**
   - Slowed growth.
   - Adult height significantly less.
   - Failure to begin sexual changes expected during puberty.
   - Early end to menstrual cycle not due to pregnancy.
   - Inability to conceive child without fertility treatment.

- **Cause of Turners syndrome-** Normally, people are born with two sex chromosomes males, inherit the X chromosome from their mothers.

And the Y chromosome from their fathers. Females inherit one x-chromosome from each parent-when a female has TS, one x chromosome copy either absent or significantly altered.

- **Complications-**
  - Cardiovascular problems.
    - defects in aorta-aortic dissection-a tear in the inner layer of the aorta.
  - Hypertension.
  - Diabetes
  - Hearing problems-hearing loss.
  - Kidney problems-urinary tract infections.
  - Hypothyrodism.
  - Tooth loss- poor / abnormal tooth development.
  - Vision- farsightedness is common.
  - Bones- osteoporosis, forward rounding of the upper back. Scoliosis (sideways curvature of spine)
  - Psychology- having problems with self-esteem, anxiety, depression.

- **Line of Teatment**-

Although there is no cure for turner syndrome some treatment can help minimize its symptoms . these include;

Human growth hormone: If given in early childhood, hormone injection can often increase adult height by a few inches.

Estrogen replacement therapy (ERT): ERT can help start the secondary sexual development that normally begins at puberty (around age 12) this includes breast development and the development of wider hips . ERT also provide protection against bone loss.

4. **Thalasemia-** Thalassemia is an inherited blood disorder in which the body makes an abnormal from of hemoglobin. The disorder results in excessive destruction of RGB which leads to anemia.

Thalasemia is inherited which means at least one of the parent must be a carrier of the disorder it is caused by either a genetic mutation or a deletion of certain key gene fragments. Around 100000 newborns are affected each year and is most common in Mediterranean, South Asia, Africa.

- **Symptons-**
  - Fatigue.
  - Weakness.
  - Pale or yellowish skin.
  - Facial bone deformities.
  - Slow growth.
  - Abnormal swelling.
  - Dark urine.
  - Rapid heart beat.
  - Poor feeding.
  - Cramps.
  - Greater susceptibility to infections.
- **Causes-**

The protein hemoglobin transports oxygen around the body in blood cells. Bone marrow uses the iron we get from food to make hemoglobin in people with thalasemia, the bone marrow does not produce enough healthy RBCS. Sometimes they lack oxygen resulting in anemia and fatigue. It is inherited from parents and is a kind of single gene inheritance

- **Complications-**
  - Iron overload- due to disease or frequent blood transfusion. Too much iron can bad to damage to the heart, liver and endocrine system.
  - Infection- high risk of various infections.
  - Severe thalassemia can cause bone deformities- expended bone marrow- bones get widen, thins brittle- broken bones.
  - Splenomegaly- due to destruction of a large number of RBCS-enlarged spleen-can lead to splenectomy (removal of spleen)
  - Slowed growth rates. Heart problem- congestive heart failure, arrhythmias

5. Muscular dystrophy-

Muscular dystrophy refers to a group of disorders that involve a progressive loss of muscle mass and consequent loss of strength muscular dystrophy may affect up to 1 in every 5,000 males.

Muscular dystrophy is caused by genetic mutations that interfere with the production of muscle protein that are needed to build and maintain healthy muscles.

Duchene muscular dystrophy is the most common type. There is currently no cure for muscular dystrophy

- **Symptoms-**

Duchene muscular dystrophy- most common among children majority of individuals affected are boys. Symptoms are:-

- Trouble walking
- Loss of reflexes
- Difficulty standing up
- Poor posture
- Bone thinking
- Scoliosis
- Mild intellectual impairment
- Breathing difficulties
- Swallowing problems
- Lung and heart weakness

- **Cause-**

Muscular dystrophy is caused by lack of a protein called dystrophin, which is necessary for normal muscle function. Absence of this protein can cause problems with walking, swallowing and muscle coordination.

The causes are genetic. A family history of muscular dystrophy will increase the chance of it affecting an individual.

- **Complications-**

- In shortening of muscles or tendons around joints (contractures)
- Breathing problems.
- Swallowing problems- nutritional problems & aspiration pneumonia may develop.
- Walking problems.

- **Line of treatment-**

There is no cure for any form of muscular dystrophy. But treatment can help prevent or reduce problems in the joint and spine to allow people to remain mobile as long as possible.

Medications-

1) Etepirsen (Exondys51) –may increase muscle strength.
2) Corticosteroids(prednisone)-can help muscle strength and delay progression of MD. But prolonged use of these types of drugs can cause weight gain and weakened bones increasing fracture risk.
3) Heart medications angiotensin converting enzyme (ACE) Inhibitors or beta blockers, if muscular dystrophy damages the heart.

Therapy-

- Range of motion and stretching exercise can help to keep joints as flexible as possible.
- Exercise- strengthening exercise also might be helpful.
- Braces- can help keep muscle and tendons stretched and flexible, slowing the progression and contractures, can also aid mobility and function.
- Mobility aids-cause, walkers and wheelchains.
- Breathing assistance- a sleep apnea device may improve oxygen delivery during the night.

Surgery- May correct a spinal curvature.

Preventing respiration infection- Through vaccination.

6. **Juvenile diabetes-** Type 1 diabetes, once known as juvenile diabetes or insulin dependent diabetes, is a chronic condition in which the pancreases produces little or no insulin.Different factors, including genetics and some viruses may contribute to type 1 diabetes although type 1 diabetes usually appears during childhood or adolescence , it can develop in adults.

- **Symptoms-** type 1 diabetes symptoms can appear relatively suddenly and may include:
    - Increased thirst
    - Frequent urination.
    - Bed wetting in children who previously didn't wet the bad during night.
    - Unintended night loss.
    - Irritability and other mood changes.
    - Fatigue and weakness.
    - Blurred vision.
    - Polyuria.
- **Causes-**

  Exact cause is unkuown. Usually, the body`s own immune system-normally fights harmful bacteria and viruses-mistakenly destroys the insulin producing langerhans cells in the pancrease. Other possible causes include:
    - Genetics
    - Exposure to viruses and other environmental factors.There`s no diabetes gene that gets turned on/off to give you type 1 instead, a punch of then play a role, including they make proteins your immune system uses to keep you healthy. Since type 1 diabetes the cells that make insulin.
- **Complications-**
    - Coronary artery disease with chest pain,heart attack,stroke, hypertension.
    - Neuropathy (nerve damage)- excess sugar can injure the walls of blood capillaries that nourish nerves (especially in legs) – cause tingling, numbness, burning or pain.
    - Nephropathy (kidney damage)
    - Eye damage – blindness, cataracts and glaucoma.
    - Skin & mouth conditions – infections, gum disease, dry mouth.

- **Line of Treatment-**

  Include:
  - Taking insulin.
  - Carbohydrate, fat and protein counting.
  - Frequent blood sugar monitoring.
  - Eating healthy foods fruits, vegetables,wholegrains.
  - Exercising regularly and maintaining a healthy weight.

- **Taking insulin-** Needs lifelong insulin therapy.
  -Short acting insulin – Eg. Humulin R, Novolin R
  -Rapid acting insulin- Eg. Insulin glulisine (Apidra), novolog
  -Intermediate acting insulin-Eg. NPH / novolin N, humulin N
  -Long-acting insulin- Eg. Glargine, insulin detemir.

Insulin administration by injections or by insulin pump.

- **Frequent blood sugar monitoring-** By artificial pancreas or by blood sugar test. Artificial pancreas (closed loop insulin delivery) checks blood sugar level every five minutes. And delivers correct amount of insulin when its needed.

Other medications-

- High lop medications- angiotensin converting enzyme (ACE) inhibitors to help keep kidneys healthy.
- Aspirin- to protect heart.
- Cholestrol – lowering durgs.

People born in or with ancestry Asia (especially southeast Asia, India and china), the middle east. Northern Africa or Mediterranean regions are more likely to have thalasemia but anyone of any race can have of these disorders.

THALASSEMIA

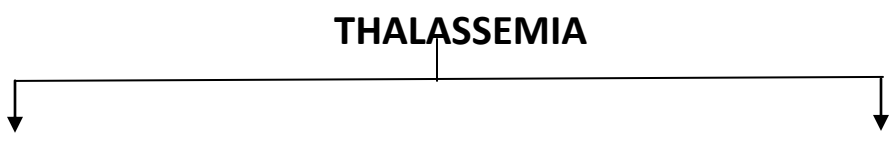
According to WHO, annually there are over 3,32,000 affected conceptions or births from blood related disorders about 56,000 have a major thalasemia, including at least 30,000 who need regular transfusions to survive and 5500 who die prenatally due to α thalassemia major.

1. What is Thalassemia?

Thalassemia is a single gene inherited blood disorder that affects the body's ability to produce hemoglobin and RBCs.

All forms of hemoglobin are made up of two molecules. Heme & globin.
Globin part is made up of four body peptide chains. In normal adult, hemoglobin (HbA), the pre dominant type of hemoglobin after the first year of life two of the global chains are identical to each other and are called alpha chains. The other two are also identical but are different from alpha chains, are called beta chains.
In thalassemia, there is a mutation in one or both of the alpha or beta globin chains. The carriers of heterozygous forms of alpha and beta thalassemia have red cell anomalies that range from very mild to server.

2. Types of thalassemia-



Alpha thalassemia

(hemoglobin does not produce enough Alpha protein)

beta thalassemia

❖ **Alpha thalassemia**-

Alpha globin protein chains = 4 genes -> 2 on each chromosome 16.
(2 from each parent).

It one or more of these genes are missing = alpha thalassemia results.
Severity of thalassemia depends on how many genes are faulty or mutated.

- **One faultygene**- patient has no symptoms. (alpha thalassemia minima).
- **Two faulty genes**- patient has mild anemia. (alpha thalassemia minor).
- **Three faulty genes**- patient has hemoglobin H disease (chronic anemia), needs regular blood transfusions throughout life.
- **Four faulty genes**- alpha thalassemia major is the most server form of alpha thalassemia it can cause hydrops fetalis (serious condition in which fluid accumulates in parts of the fetus body).

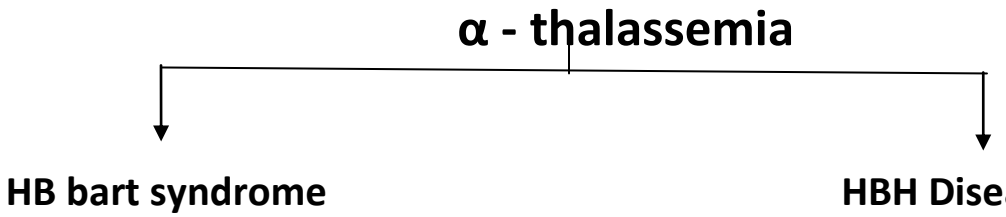
A fetus with four mutated genes cannot produce normal hemoglobin and is unlikely to survive, even with blood transfusions

❖ **Beta thalassemia**-

Beta globin chains = 2 globin genes (me from each, each parent)

It one or both genes are faulty, beta thalassemia will occur. Severity depends no of genes mutated.

- **One faulty gene-** beta thalassemia minor.
- **Two faulty gene-** thalassemia major, used to be called colley`s anemia.



- Characteresed by hydrops fetalis
- Severe anemia.
- Heptosplenomegaly.
- Heart defects, etc.

- causes mild to moderate anemia.
- jaundice.
- usually appear.
- in early childhood.

❖ **Symptoms of α - THALASSEMIA-**

- Pale or yellow skin
- Feeling tired / lathargyness
- Low appetite
- Spleenomegaly
- Bone deformities
- Dark urine

β - thalassemia

Thalassemia major(cooley`s anemia)

- More severe.
- Symptoms appear within first 2 yrs of life .
- Develops life threatening anemia.

Thalassemia intermidia

- milder than thalassemia major.
- symptoms appear in early child-hood or also later.
- bone deformities.

❖Symptoms of β – thalassemia

β – thalassemia major symptoms in an infant (between 6 to 24 months).

- Poor growth and development.
- Feeding problems.
- Diarhhiea.
- Irritability, fussiness.
- Fevers.

β – thalassemia intermedia happen at a later age & includes.

- Pale skin, all stones, leg ulcers, soft bones.

3. **Cause of thalassemia-** Thalassemia is caused by mutations in the DNA of cells that make hemoglobin. It disrupts the normal production of hemoglobin & healthy RBCS. This causes anemia leaving the porson fatigued.

4. Diagnosis of thalassemia-

(I) CBC (complete blood count) – checks RGB,WBCS, platelets. It includes hemoglobin and hematocrit and more details about the RBCS.

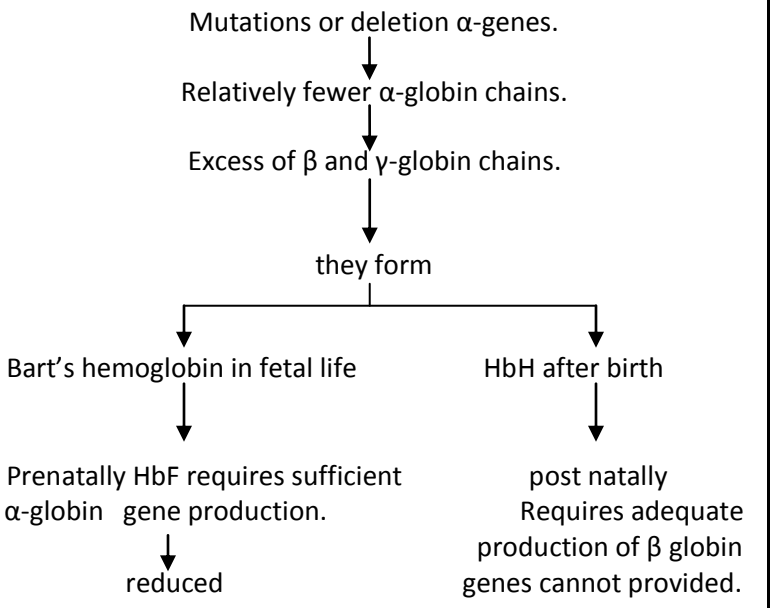
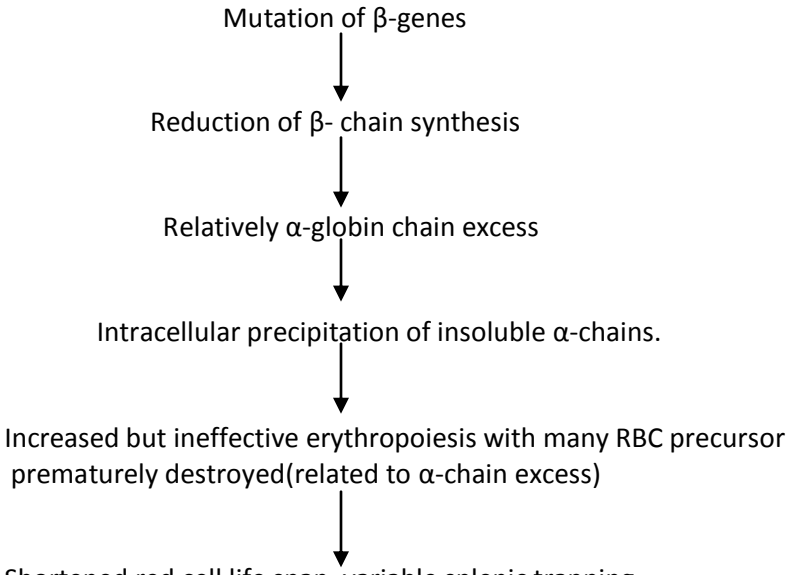
(II) Peripheral smear- A small blood simple is checked under microscope.

(III) Hemoglobin electrophoresis- test that measures types & amount of hemoglobin.

(IV) DNA testing- tests look for gene defects and can find carriers.

(V) Prenatal testing-CVS / chorionic villus sampling- a piece of placenta is removed for testing around 11^{th} week of pregnancy.

5.Pathogenesis of Thalassemia.

Pathogenesis of α- thalassemia	Pathogenesis of β-thalassemia
	

5. **Complications-**

I. Iron overload – due to frequent blood transfusion also causes hepatitis (swollen liver), congestive heart failure.
II. Alloimmunization – a blood transfusion can trigger a reaction where patient`s immune system reacts to the new blood and tries to destroy it is important to have the have the exact blood type match.
III. Enlarged spleen- spleen recycles RBCs in thalassemia RBC, may have an abnormal shape making it harder for the spleen to recycle them the cells accumulate in the spleen, making it grow. Enlarged spleen can become over reactive, it can destroy healthy RBG.
IV. Infection- removal of spleen & regular transfusions pancreases high risk of contracting a blood borne disease.
V. Bone deformities- in some cases, bone marrow expands, deforming the bone around it.

6. **Line of treatment-**

Some of the treatments include:-

- Blood transfusions.
- Bone marrow transplant.
- Medications and supplement.
- Possible surgery to remove spleen or gallbladder.

❖ **Blood transfusion-** more- sever form of thalassemia often require frequent blood transfusion.

❖ **Bone marrow transplant-** A stem cell transplant may be an option in select causes it can dominate the need for lifelong blood transfusion of stem cells from a compatible donor, usually a sibling.

❖ **Medications and supplement-** chelating agents, antimetabolites, antipyretics, antibacterial combinations. Diet for thalassemia- a low fat, plant based diet thalassemia can cause folic acid (folate) deficiencies naturally found in foods such as dark leafy greens and legumes. Doctor may recommend 1 mg supplement taken daily.
To avoid iron excess, fish, meat, fortified. Cereals, breads and juices are avoided.

CONCLUSION

Genetic disorder`s are problems caused by one or more abnormalities formed in the genome. They can be hereditary or non hereditary. According to WHO, around 10,000 of the cases of diseases reported are of genetic abnormality world wide in a year. There are basically two types of genetic disorders : inherited and acquired. Inherited genetic disorder includes single gene, multifactorial, chromosomal and mitochondrial genetic abnormality. Some of the common genetic diseases prevailing world wide are : sickle cell anemia, downs syndrome turner`s syndrome, muscular dystrophy and juvenile diabetes.

All of these diseases, do not have a definite cure but become manageable through medications and councelling,
Among the mentioned diseases, every disease has it`s own mile stone of cases reported per year, starting with sickle cell anemia , which is a single gene inherited abnormality has a remark of 3,00,000 babies born with this disorders. Line of treatment mainly includes bone marrow transplant.
Following comes thalassemia which is also a single gene defect and affects around 280 million people of south east Asia, Africa, greek, and Italy. Line of treatment goes with blood transfusions, bone marrow transplant & medications.

A X-linked chromosomal disorder, is turner`s syndrome, with which 25 to 210 per 1,00,000 of female babies are born. This abnormality affects only female child and its line of treatment includes hormone therapy.
Next comes Down`s syndrome with 1:3000 new born per year . It`s line of treatment goes with many therapies such as physical, speech, occupational and emotional therapy.
Muscular dystrophy and Juvenile diabetes are seen less and the former affects only males. Line of treatment for muscular dystrophy depends completely on symptoms and there is no cure. Juvenile diabetes can be managed by doses of insulin and diet maintanence and keeping a check on blood sugar levels .
Genetic disorder can be diagnosed earlier through family history and by checking on carriers.

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