MEDICAL ACADEMY NAMED After S.I GEORGIEVSKY OF VERNADSKY CFU

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TOPIC= GENDER BOUND/SEX BOUND INHERITANCE

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GENDER BOUND/SEX BOUND INHERITANCE

- The **inheritance** of a trait (phenotype) that is determined by a gene located on one of the **Sex** chromosomes is called Sex Linked Inheritance.

TYPES OF GENDER/SEX BOUND INHERITANCE

- HEMOPHILIA-A
- HEMOPHILIA-B
- COLOUR BLINDNESS
- PERIODIC PARALYSIS
- PIGMENTOSA RETINITIS

HEMOPHILIA-A

- **Hemophilia** A is a genetic deficiency in clotting factor VIII, which causes increased bleeding and usually affects males.
- In the majority of cases it is inherited as an X-linked recessive trait
- Causes= Factor VIII Deficiency
- SIGNS AND SYMPTOMS:

1. Mild hemophilia A{ episode of heavy bleeding }

a] After you have surgery or another procedure, like having a tooth pulled.

b] After childbirth

c] After a serious injury

d] During your period

2.Moderate hemophilia A

a] Bleed a lot when injured.

b] Bleed without an injury (spontaneous bleeding).

c] Bruise easily.

d] Bleed a lot after a vaccination.

3. Severe hemophilia A {frequent episodes of bleeding often into the joints and muscles}

a]Prolonged headache

b]Throwing up

c] Sudden weakness or problems walking

d] Double vision

e] Convulsions or seizures

- DIAGNOSIS: Partial prothrombin test {ppt}
- TREATMENT: Regular supplementation with intravenous recombinant or plasma concentrate Factor VIII.

HEMOPHILIA-B

- **Haemophilia B** is a blood clotting disorder causing easy bruising and bleeding due to an inherited mutation of the gene for factor IX and resulting in a deficiency of factor IX.
- It is less common than factor VIII deficiency.
- CAUSES= Factor IX deficiency
- SIGNS AND SYMPTOMS:

  a] easy bruising

  b] urinary tract bleeding {haematuria}

  c] nosebleeds [epistaxis]

  d] bleeding into joints (haemarthrosis)

- DIAGNOSIS: a] Coagulation screening test
  b] Bleeding scores
  c] Coagulation factor assays
- TREATMENT : a] intravenous infusion of factor IX
  b] blood transfusions.

COLOUR BLINDNESS

- **Color blindness** is the decreased ability to see color or differences in color.
- It can impair such tasks as selecting ripe fruit, choosing clothing, and reading traffic lights.
- CAUSES=Color vision deficiencies can be classified as acquired or inherited.

  a] Acquired: Diseases, drugs (hydroxychloroquine), chemicals such as styrene or organic solvents may cause color blindness.

  b] Inherited: 1. monochromacy [total color blindness]=is the lack of ability to distinguish colors caused by cone defect or absence.

  2. dichromacy= Protanopia and deuteranopia are hereditary and sex-linked, affecting predominantly males

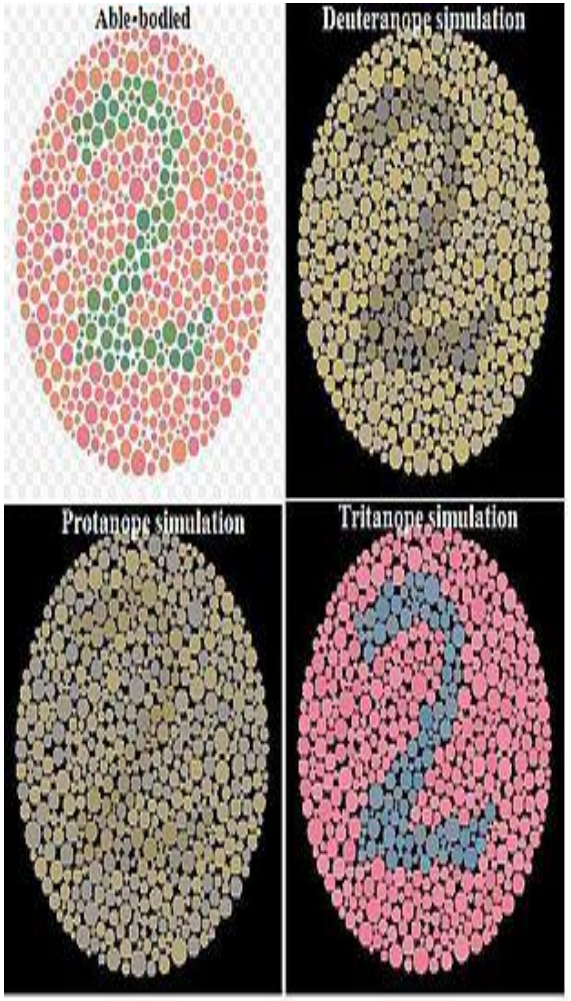
  3. anomalous trichromacy=occurring when one of the three cone pigments is altered in its spectral sensitivity

SIGN AND SYMPTOMS: 1.color blind people retain blue–yellow discrimination

2. Dichromats often confuse red and green

- DIAGNOSIS:

  Ishihara color test = a series of pictures of colored spots, is the test most often used to diagnose red–green color deficiencies .



- TREATMENT: There is no cure for color deficiencies. It can only managed by a contact lens on one eye which increase the ability to differentiate between colors.

PERIODIC PARALYSIS

- **Periodic paralysis** is a group of rare genetic diseases that lead to weakness or paralysis .
- The underlying mechanism of these diseases are malfunctions in the ion channels in skeletal muscle cell membranes that allow electrically charged ions to leak in or out of the muscle cell causing the cell to depolarize and become unable to move.
- CAUSES= episodic attacks of muscle weakness which are commonly associated with serum potassium levels.
- SIGNS AND SYMPTOMS= 1. Weakness in the face
  2. Muscle pain and stiffness
  3. An irregular heartbeat
  4. Trouble breathing or swallowing

- DIAGNOSIS:

  This disease is difficult to diagnose. Patients often report years of wrong diagnosis and treatments that made them worse instead of better. But there are some tests which gives an idea of this disease like:

1. Blood tests to check potassium, thyroid, and other levels
2. Electromyography (EMG) and nerve conduction studies to see how well muscles and nerves are working
3. Electrocardiogram (EKG) to check your heart
4. Muscle biopsy to check for abnormal muscle cells

- TREATMENT:

1. It include carbonic anhydrase inhibitors (such as acetazolamide, methazolamide or dichlorphenamide)
2. supplemental oral potassium chloride and a potassium-sparing diuretic (for hypos) or avoiding potassium (for hyper)
3. thiazide diuretics to increase the amount of potassium excreted by the kidneys (for hyper)
4. lifestyle changes including tightly controlled levels of exercise or activity.

PIGMENTOSA RETINITIS

- **Retinitis pigmentosa** (**RP**) is a genetic disorder of the eyes that causes loss of vision and the back wall of the eye (retina) is damaged.
- Retinitis pigmentosa is a rare, inherited degenerative eye disease that causes severe vision impairment.
- CAUSES=(1) Non-syndromic, that is, it occurs alone, without any other clinical findings
  (2) Syndromic, with other neurosensory disorders
  (3) Secondary to other systemic diseases
- SIGN AND SYMPTOMS: 1. Night blindness
  2.Tunnel vision (due to loss of peripheral vision)
  3.Latticework vision
  4.loss of depth perception
  5.Photopsia (blinking/swirling/shimmering lights)
  6.Photophobia (aversion to bright lights)

7. Blurring of vision
8. Poor color separation
9. Loss of central vision
10. Eventual blindness

- DIAGNOSIS: 1. visual field test
  2. visual acuity test
  3. electroretinography [ERG]
  4. fundus and optical coherence imagery
- TREATMENT:

There is currently no cure for retinitis pigmentosa, but the efficacy and safety of various prospective treatments are currently being evaluated. The efficiency of various supplements, such as vitamin A, DHA, and lutein, in delaying disease progression.

THANK YOU