MEDICAL ACADMY NAMED AFTER S.I.GEORGIEVSKY OF VERNADSKY CFU

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GROUP LA1 – 202(2)

TOPIC – HEREDITARY BLOOD DISEASES AND HEREDITARY DISEASE OF ENDOCRINE SYSTEM

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Hemolytic disease of newborn

Hemolytic disease of the new born and fetus (HDN) is a destruction of the red blood cells (RBCs) of the fetus and neonate by antibodies produced by the mother

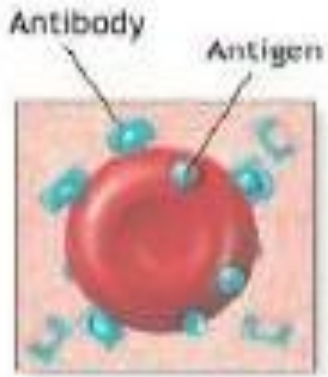
It is a condition in which the life span of the fetal/neonatal red cells is shortened due to maternal allo-antibodies against red cell antigens acquired from the father

Rh incompatibility (Rh–induced Hemolytic Disease of Newborn) :

Rh incompatibility is that develops when a pregnant woman has Rh-Negative blood and the baby in her womb has Rh-Positive blood.

During pregnancy, red blood cells from the fetus can get into the mother's bloodstream as she nourishes her child through the placenta. If the mother is Rh-Negative blood her system cannot tolerate the presence of Rh-Positive red blood cells. In such case the, the mother's *immune system* treats the Rh-Positive fetal cells as if they were a foreign substance and makes *antibodies* against the fetal blood cells. These anti-Rh antibodies (Figure 1) may cross the placenta into the fetus, where they destroy the fetus's circulation red blood cells (Figure 2).

First-born infants are often not affected because it takes time for the mother to develop antibodies against the fetal blood, unless the mother has had previous mis-carriages or abortions, which could have sensitized her system for developing antibodies. Hence second children who are also Rh-Positive may be harmed.



Red blood cell

An antibody is a protein produced by the immune system in response to the presence of an antigen

HEMOGLOBINOPATHY

- It's a blood disorder that is caused by a genetically determined change in the molecular structure of haemoglobin.
- Defects in these genes can produce abnormal haemoglobins and anaemia
- The most important of the hemoglobinopathies are sickle-cell anaemia and thalassemia.

TREATMENT FOR HEMOGLOBINOPATHY

- Treatment for hemoglobinopathy includes:
  - Periodic blood transfusions
  - Iron chelation therapy
- Will use drugs to treat the symptoms of sickle-cell disease include:
  - Analgesics,
  - Antibiotics,
  - ACE inhibitors
  - Hydroxyurea.

PREVENTION FOR HEMOGLOBINOPATHY

- Genetic counselling can be provided to at-risk couples.
- Should be tested to determine if the couple is at risk of having a child with clinically significant disease.
- Chorionic villus sampling and amniocentesis also can be used to diagnosis at-risk fetuses.

- **Endocrine system** – together with the nervous system, acts as the **body´s communication network**

- it is composed of **various endocrine glands and endocrine cells**

- the glands are capable of synthesizing and releasing special chemical messengers –
  - **hormones**

ADRENOGENITAL SYNDROME

Adrenogenital syndrome, also known as congenital adrenal hyperplasia (CAH), is caused by an inherited enzyme deficiency in the adrenal cortex that leads to altered levels of adrenal cortical hormones. Adrenal cortical hormones include mineralocorticoids (ie, aldosterone), glucocorticoids (Ie, cortisol), and sex steroids (ie, testosterone and estrogen). The syndrome occurs when an enzyme deficiency leads to decreased adrenal synthesis of glucocorticoid, which impairs feedback inhibition on the pituitary

RESULT

As a result, the pituitary secretes increased levels of adrenocorticotropic hormone (ACTH), which stimulates the adrenal glands to enlarge and produce more intermediate substrates. These intermediate substrates are shunted toward functioning arms of the hormone synthesis pathways, where increased levels of other hormones are produced (either mineralocorticoids or androgens, depending on the enzyme deficiency). Altered levels of mineralocorticoids and sex hormones lead to electrolyte abnormalities, problems with sexual differentiation, and other signs and symptoms, depending on the deficient enzyme and extent of the deficiency.

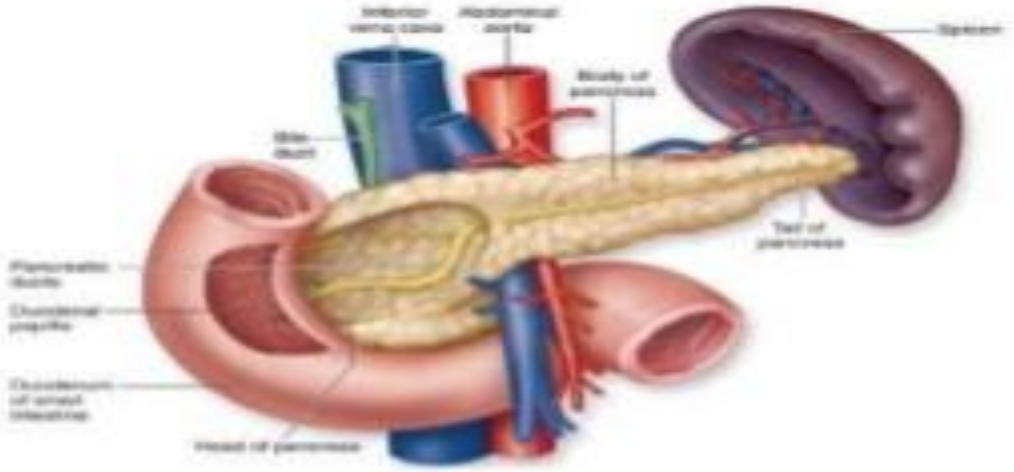
TREATMENT AND DIAGNOSIS

Treatment with relatively small doses of cortisone is effective in suppressing the excessive secretion of adrenal androgen without causing abnormal metabolic or toxic effects. The minimum maintenance dose of intramuscular or oral cortisone must be determined in each case, following the urinary 17-ketosteroids and the rates of somatic growth and development as guides. In individuals of either sex who have reached a level of somatic development comparable to that of puberty (i.e., a bone age of 11 years or greater) suppression of the adrenal hyperactivity with cortisone results promptly in normal adolescent sexual development corresponding to the sex of the patient.

- **Diabetes mellitus** (**DM**), is a group of metabolic diseases in which there are high blood sugar levels over a prolonged period.

- Symptoms of high blood sugar include frequent urination, increased thirst, and increased hunger.

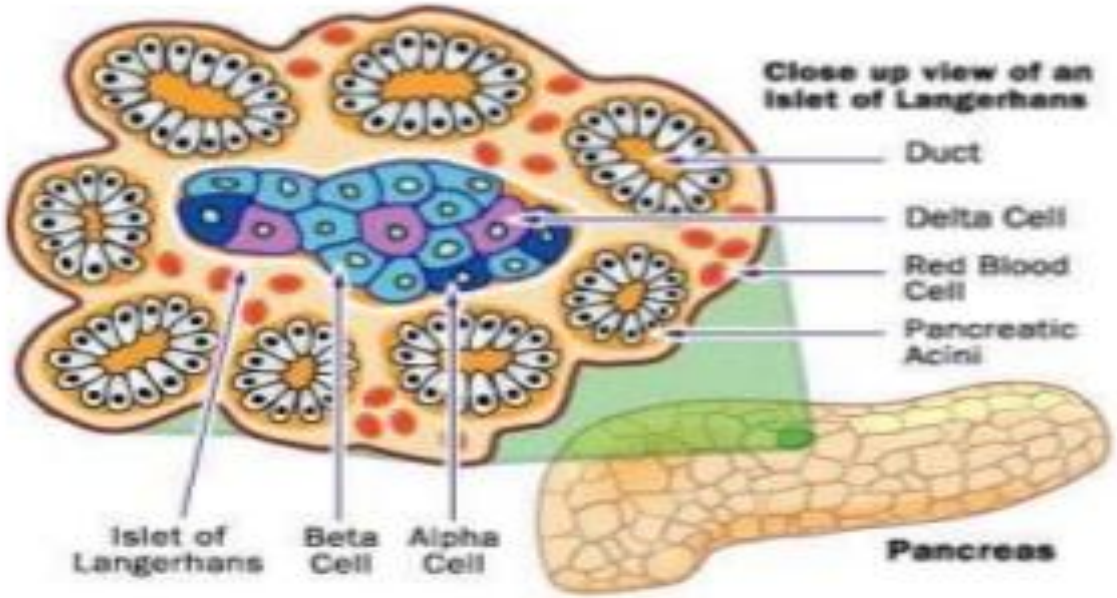
- Diabetes is due to either the pancreas not producing enough insulin or the cells of the body not responding properly to the insulin produced.



- There are three main types of diabetes mellitus:
  - Type 1 DM
  - Type 2 DM
  - Gestational Diabetes

Type 1 DM

- Results from the pancreas's failure to produce enough insulin.



- This form was previously referred to as "insulin-dependent diabetes mellitus" (IDDM) or "juvenile diabetes".

- The cause is unknown.

Type 2 DM

- Begins with insulin resistance, a condition in which cells fail to respond to insulin properly.

- This form was previously referred to as "non insulin-dependent diabetes mellitus" (NIDDM) or "adult-onset diabetes".
- The primary cause is excessive body weight and not enough exercise.

TREATMENT

Treatment of **diabetes** with insulin

Insulin remains the mainstay of **treatment** for patients with type 1 **diabetes**. Insulin is also an important therapy for type 2 **diabetes** when blood glucose levels cannot be controlled by diet, weight loss, exercise, and oral **medications**.

THANK YOU