Genetic Diseases and Disorders

What Is a Gene?

- Most living organisms are made up of **cells** that contain a substance called **deoxyribonucleic acid** (DNA).
- DNA is wrapped together to form structures called **chromosomes.** Most cells in the human body have 23 pairs of chromosomes, making a total of 46.
- **Genes** are sections or segments of DNA that are carried on the chromosomes and determine specific human characteristics.

- cell [sel] – клетка
- deoxyribonucleic acid - дезоксирибонуклеиновая кислота, ДНК

What Are Genetic Disorders?

- **Cells** can sometimes contain changes or variants in the information in their genes.
- This is called **gene mutation**, and it often occurs when cells are aging or have been exposed to certain **chemicals** or **radiation**.
- Fortunately, cells usually recognize these mutations and repair them by themselves.
- Other times, however, they can cause **illnesses**.
- And if the gene mutation exists in **egg** or **sperm cells**, c hildren can **inherit** the mutated gene from their parents.

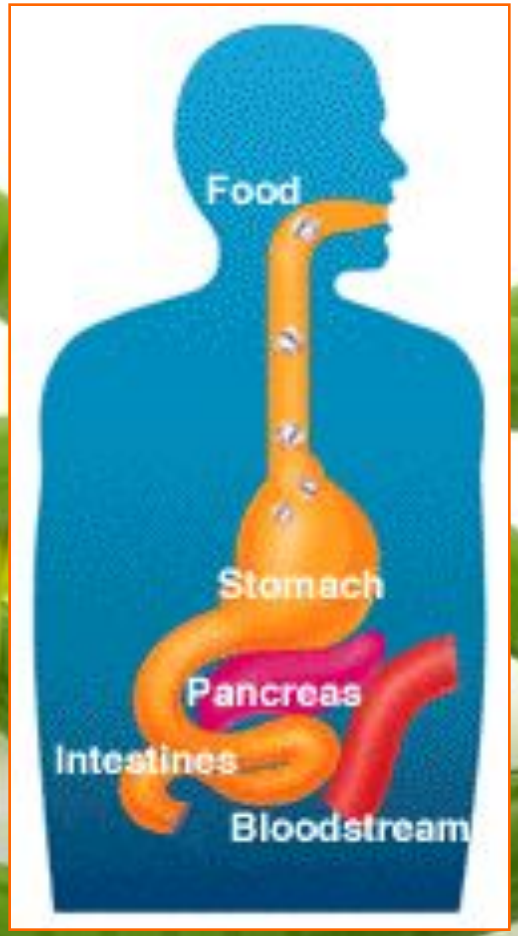
- inherit [ɪn'herɪt] - унаследовать, перенять

- Researchers have identified **more than 4,000 diseases** that are caused by genetic variants.
- On average, people probably carry **from 5 to 10** variant or disease genes in their cells.
- Problems arise when the disease gene is **dominant** or when the same **recessive** disease gene is present on both chromosomes in a pair.
- Problems can also occur when several variant genes interact with each other - or with the environment - to increase **susceptibility** to diseases.

- susceptibility [sə¸septə'bɪlətɪ] - восприимчивость

Diabetes

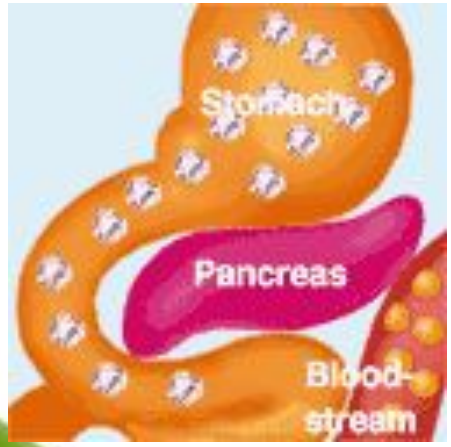
What is diabetes?

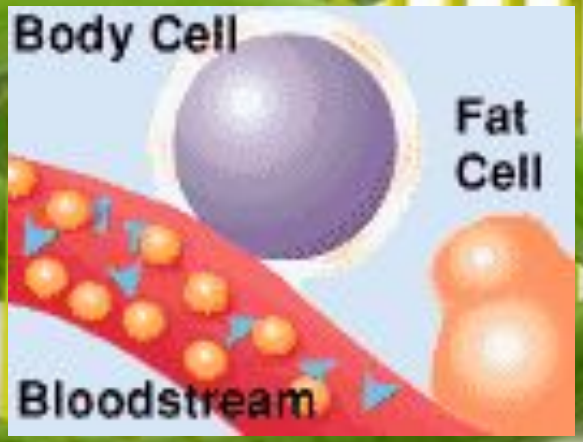


- **Diabetes** is classed as a **metabolism** disorder. **Metabolism** refers to the way our bodies use digested food for energy and growth. Most of what we eat is broken down into **glucose**. **Glucose** is a form of sugar in the blood - it is the principal source of **fuel** for our bodies.

  When our food is digested the **glucose** makes its way into our **bloodstream**. However, **glucose** cannot enter our **cells** without **insulin** being present - **insulin** makes it possible for our **cells** to take in the **glucose**.

- metabolism [mə'tæb(ə)lɪz(ə)m] - метаболизм, обмен веществ
- digest [daɪ'ʤest] - переваривать (пищу)
- break down - распадаться (на части)
- fuel [fju:əl], ['fjuəl] - топливо, горючее
- bloodstream ['blʌdstri:m] – кровоток
- intestine [ɪn'testɪn] - кишечник, кишки





- **Insulin** is a **hormone** that is produced by the **pancreas**. After eating, the pancreas automatically releases an adequate quantity of **insulin** to move the **glucose** present in our blood into the cells, and lowers the **blood sugar level**.
- pancreas ['pæŋkrɪəs] - поджелудочная железа
- release [rɪ'liːs] - избавлять, освобождать

- A person with diabetes has a condition in which the quantity of glucose in the blood is **too elevated**. This is because the body *does not produce enough insulin*, *produces no insulin*, or *has cells that do not respond properly to the insulin the pancreas produces*. This results in too much glucose building up in the blood. This excess blood glucose eventually passes out of the body in **urine**. So, even though the blood has plenty of glucose, the cells are not getting it for their essential energy and growth requirements.
- urine ['juərɪn] моча

Types of diabetes

- There are three main types of diabetes:
- **Type 1** - You produce no insulin at all.
- **Type 2** - You don't produce enough insulin, or your insulin is not working properly.
- **Gestational Diabetes** - You develop diabetes just during your pregnancy.
- **Diabetes Types 1 and 2** are **chronic** medical conditions - this means that they are persistent and perpetual. **Gestational Diabetes** usually resolves itself after the birth of the child.

Symptoms of Diabetes

- Frequent urination
- Disproportionate thirst (жажда)
- Intense hunger
- Weight gain
- Unusual weight loss
- Increased fatigue
- Irritability (раздражительность)
- Blurred (неясное,туманное) vision
- Cuts and bruises (синяк, кровоподтёк; ушиб) don't heal (заживать) properly or quickly
- More skin and/or yeast (грибковые) infections
- Itchy (зудящая) skin
- Gums (десны) are red and/or swollen (опухшие)
- Frequent gum disease/infection
- Sexual dysfunction among men
- Numbness (нечувствительность) or tingling (покалывание, пощипывание), especially in your feet and hands

Treatment

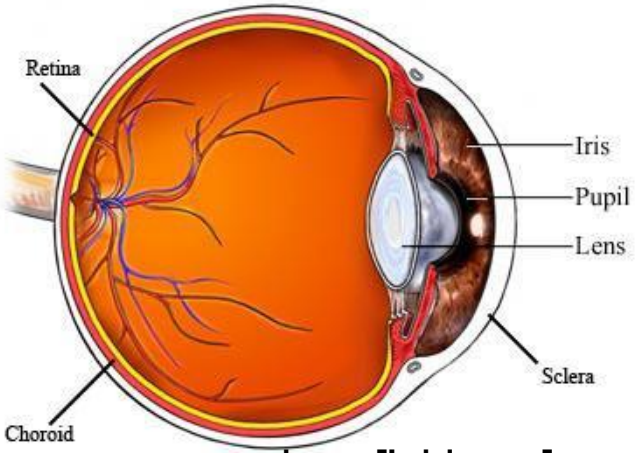
- Type 1 and Type 2 diabetes last a lifetime; there is no known cure. The patient receives regular insulin, which became medically available in 1921.
- The treatment for a patient with Type 1 is mainly **injected insulin**, plus some **dietary** and **exercise adherence**.
  adherence [əd'hɪər(ə)n(t)s] - строгое соблюдение
- Patients with Type 2 diabetes are usually treated with **tablets, exercise** and a **special diet**, but sometimes **insulin injections** are also required.

Colour blindness

What is colour blindness?

- The words 'colour blindness' are misleading. People who cannot see all colours are not 'blind' - they can see things as clearly as people who are not 'colour blind'.
- Colour blindness means that a person cannot 'see' some colours, or sees them differently to other people.
- Very few people who are colour blind are 'blind' to all colours. The usual colours which they see differently are greens, yellows, oranges and reds.

retina ['retɪnə]
сетчатка



iris ['aɪərɪs]
радужка

pupil ['pju:p(ə)l] зрачок

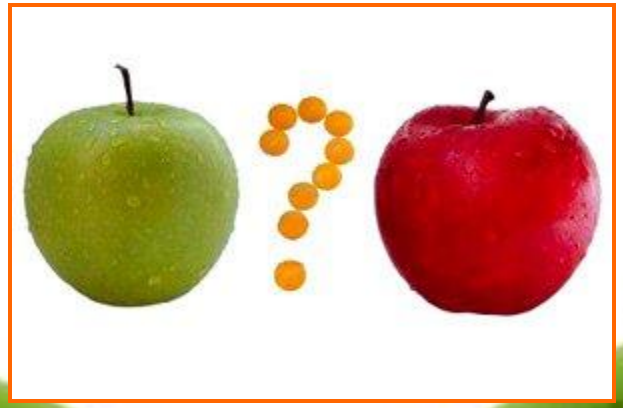
lens [lenz] линза

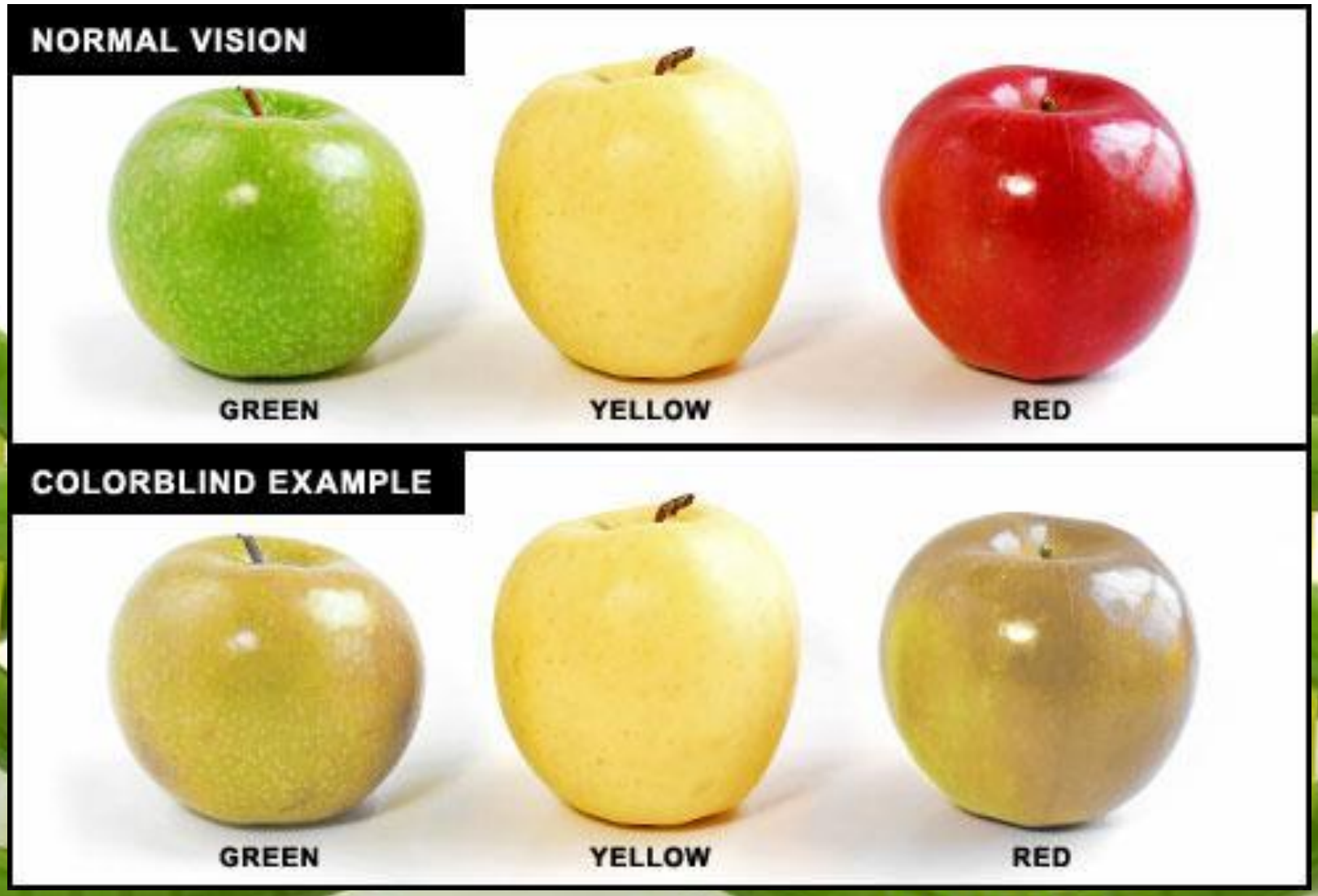
choroid - сосудистая оболочка глаза

sclera ['sklɪərə] склера, белочная оболочка глаза

- Colour blindness arises from the structure of the eye.
- In the **retina** at the back of the eye (the part of the eye that picks up light coming in), there are two types of light-sensitive cells called **'rod cells'** (палочки) and **'cone cells'** (колбочки) , and these react differently to light.
- **Rod cells** are very sensitive to light, and they can react to even very faint (слабый) light, but they do not 'see' different colours. Rod cells allow us to see things around us at night, but only in shades of black, grey and white.
- **Cone cells** react to brighter light, and they help us to see the detail in objects. They also pick up colours.

- There are three types of cone cells; ones that pick up red light, others green and others blue.
- By combining the messages from each set of cone cells, we get the wide range of colours that we can normally see.
- Someone who is colour blind lacks (не хватать, недоставать) one or more of these types of cone cells.

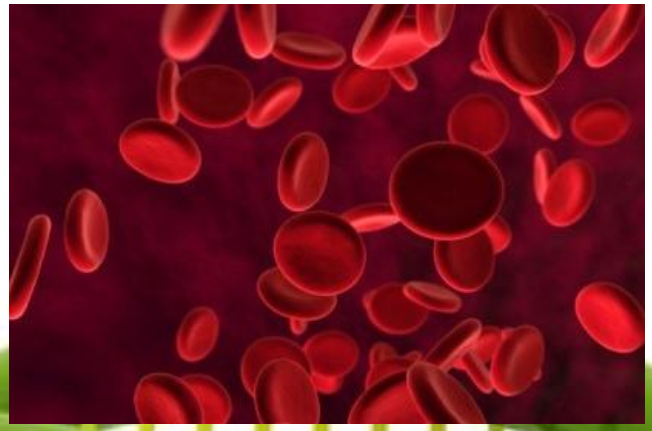



NORMAL VISION
GREEN
YELLOW
RED
COLORBLIND EXAMPLE
GREEN
YELLOW
RED

- Most of the people with colour blindness are **male** – about **1 boy in 12** will be colour blind, while only about **1 girl in 400** will be colour blind.
- The gene for colour vision is on the X chromosome. Females have two X chromosomes and will only be colour blind if both those chromosomes are affected.
- Males have only one X chromosome, so they only need that chromosome to be affected – that's why colour blindness is more common in males.

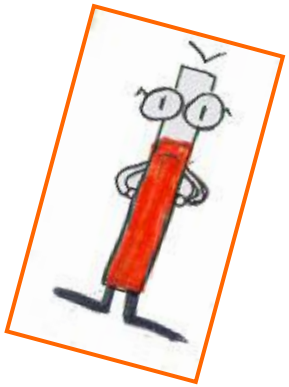
Hemophilia

What Is Hemophilia?



- **Hemophilia** is a genetic disorder in which a person's blood does not clot properly.
- A person who has **hemophilia** has a tendency to **bleed** excessively.
- clot [klɔt] -свёртываться, запекаться (о крови)

Types of Hemophilia



- A person can have one of two types of hemophilia, **A or B**. It depends on which clotting factor they are low on.
- If someone produces **1% or less** of the affected factor, the case of hemophilia is called **severe**. Someone who produces **2% to 5%** has a **moderate** case, and someone who produces **6% to 50%** of the affected factor level is considered to have a **mild** case of hemophilia.

Why Do Kids Get Hemophilia?

- **Hemophilia** almost always affects boys. Why? Because the disease is an **X-linked genetic disorder**, passed **from mother to son**.
- If the mother carries the gene for hemophilia on one of her X chromosomes, each of her sons will have a 50% chance of having hemophilia.
- A mother who is a carrier also has a 50% chance of giving the faulty X chromosome to her daughter. That does not give the daughter the hemophilia disease, but it does result in the daughter becoming **a hemophilia carrier**.

- Even kids with **severe hemophilia** often live long and healthy lives. A **cut**, a **scrape** or minor **wound** is usually no big deal for a person with hemophilia, but **internal bleeding** can be serious. When bleeding occurs in the **joints**, **muscles**, or **internal body organs**, treatment is necessary.
- wound [wuːnd] рана; ранение
- joint [ʤɔɪnt] сочленение, сустав