SAWA UNIVERSITY

جامعة ساوة الاهلية
كلية التقنيات الصحية والطبية
قسم التخدير - اللجنة العلمية

ADVANCED MEDICINE

LECTURE THREE : GENETICS AND DISEASES

Sawa University — جامعـــــة ســـــاوة الاهلية

College of health and medical techniques — كليـــــة التقنيـــات الصحيـة والطبيـة

Department of Anesthesia — قسم تقنيـــات التخديـــــر

Fourth Stage — المرحلـــــــــة الرابعة

Lecture two

Theoretical

Dr. shaker kadhium

M.Sc. medicine

A genetic disease is any disease caused by an abnormality in the genetic makeup of an individual. The genetic abnormality can range from minuscule to major -- from a discrete mutation in a single base in the DNA of a single gene to a gross chromosomal abnormality involving the addition or subtraction of an entire chromosome or set of chromosomes. Some people inherit genetic disorders from their parents, while other changes are acquired changes or mutations in a preexisting gene or group of genes cause other genetic diseases. Genetic mutations can occur either randomly or due to some environmental exposure.

The environmental influence on genetics is currently one of the rapidly evolving areas of research (epigenetics).

There are four different types of genetic disorders (inherited diseases and disorders) and include:

1. **Single gene inheritance**
2. **Multifactorial inheritance**
3. **Chromosome abnormalities**
4. **Mitochondrial inheritance diseases**

Single gene inheritance disorders

Single gene inheritance is also called Mendelian or monogenetic inheritance. Changes or mutations that occur in the DNA sequence of a single gene cause this type of inheritance. There are thousands of known single-gene disorders. These disorders are known as monogenetic disorders (disorders of a single gene). Single-gene disorders have different patterns of genetic inheritance, including

1. **autosomal dominant inheritance**
2. **autosomal recessive inheritance**
3. **X-linked inheritance**

Some examples of single-gene disorders include

- **cystic fibrosis,**
- **alpha- and beta-thalassemias,**
- **sickle cell anemia (sickle cell disease),**
- **Marfan syndrome,**
- **fragile X syndrome,**
- **Huntington's disease, and**
- **hemochromatosis**

Common multifactorial genetic inheritance disorders

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 example, different genes that influence breast cancer susceptibility have been found on chromosomes 6, 11, 13, 14, 15, 17, and 22. Some common chronic diseases are multifactorial disorders. Examples of multifactorial inheritance include

- **heart disease,**
- **Alzheimer's disease,**
- **diabetes,**
- **Obesity .**
- **. high blood pressure,**
- **. arthritis,**
- **. cancer**

Multifactorial inheritance also is associated with heritable traits such as fingerprint patterns, height, eye color, and skin color.

Chromosomal abnormalities

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

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

For example, Down syndrome (sometimes referred to as "Down's syndrome") or trisomy 21 is a common genetic disorder that occurs when a person has three copies of chromosome 21. There are many other chromosomal disorders including:

- **Turner syndrome (45, X0),**
- **Klinefelter syndrome (47, XXY), and**
- **Cri du chat syndrome, or the "cry of the cat" syndrome (46, XX or XY, 5p-).**

Diseases may also occur because of chromosomal translocation, a type of genetic variation in which portions of two chromosomes are exchanged.

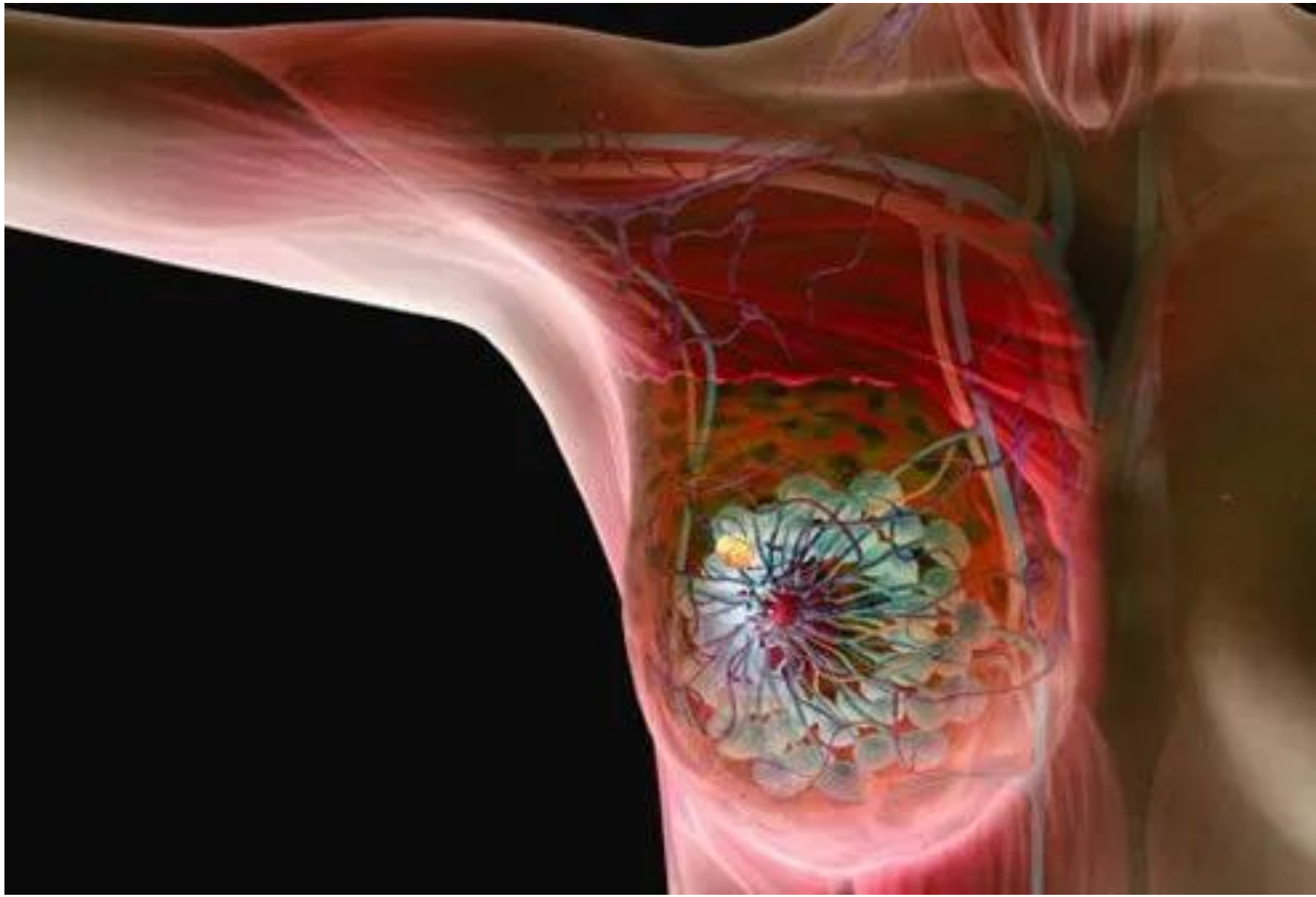
Types of mutations

There are various types of genetic mutations based on where they develop. These include germline mutations and somatic mutations.

Germline mutations

They occur in reproductive cells, such as sperm and eggs. These mutations are significant because they are heritable. This means they can be passed on to offspring, and every cell in the offspring will carry the mutation.

- Germline mutations are inherited from parents and can be passed to future generations.
- These mutations can lead to genetic disorders or contribute to the risk of developing certain diseases, such as hereditary cancers.
- Examples of germline mutations include mutations in the BRCA1 and BRCA2 genes, which increase the risk of breast and ovarian cancers.



Somatic mutations

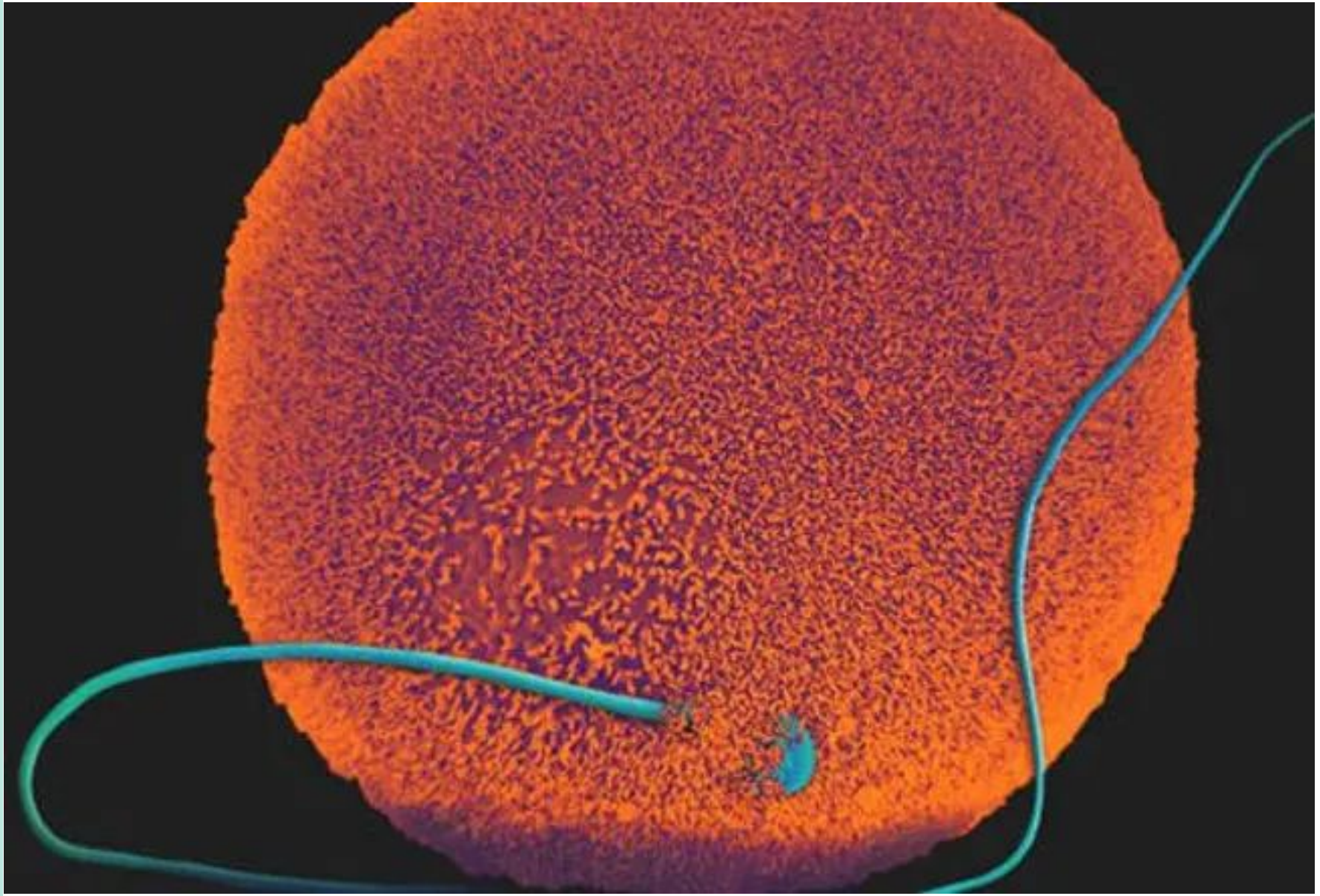
Somatic mutations occur in nonreproductive cells and are not inherited. These mutations happen after conception and can occur in any cell of the body except sperm and egg cells.

Somatic mutations are not passed on to offspring.

These mutations can lead to diseases such as cancer. For example, if a somatic mutation occurs in a gene that controls cell growth, it can lead to uncontrolled cell division and tumor formation.

Examples of somatic mutation include mutations in the TP53 gene, which is involved in many cancers.

Both types of mutations can have important implications for human health and disease.



the human genome

The human genome is the entire "treasury of human inheritance." The sequence of the human genome obtained by the Human Genome Project, completed in April 2003, provides the first holistic view of our genetic heritage. The 46 human chromosomes (22 pairs of autosomal chromosomes and 2 sex chromosomes) between them house almost 3 billion base pairs of DNA that contain about 20,500 protein-coding genes. The coding regions make up less than 5% of the genome (the function of all the remaining DNA is not clear) and some chromosomes have a higher density of genes than others.

- **Most genetic diseases are the direct result of a mutation in one gene.**
- **However, one of the most difficult problems ahead is to further elucidate how genes contribute to diseases that have a complex pattern of inheritance, such as in the cases of diabetes, asthma, cancer, and mental illness.**
- **In all these cases, no one gene has the yes/no power to say whether a person will develop the disease or not.**
- **It is likely that more than one mutation is required before the disease is manifest, and a number of genes may each make a subtle contribution to a person's susceptibility to disease;**
- **Genes may also affect how a person reacts to environmental factors.**

QUIZ

What is the types of genetics diseases ?

Any questions?