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Genetic Disorders

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Abstract

A genetic disease is any disease caused by an abnormality in the genetic makeup of an individual. Genes play an important role in shaping how we look and act and even whether we get sick. Genes are particulate factors that passes unchanged from parent to progeny determines a biological character of an organism. The billions of people on earth, no two look exactly alike. Facial characteristics, hair, eye and skin color, hair texture and body build make us distinct from one another. In addition to the many visible differentiating characteristics, there are many more that are invisible like blood chemistry, mental and physical aptitudes, susceptibility to certain diseases, and other traits, which contribute to our individuality. But there is also sameness among human beings, which makes us distinct from other species. What is mechanism responsible for this similarity with differences and how is it transmitted from one generation to another? The science that deals with the study of heredity and variations is known as genetics. There are four major categories under which genetic disorders are listed that is Single gene inheritance, Multi factorial inheritance, Chromosome abnormalities, Mitochondrial inheritance. There are different risk factors, causes, complications for every disorder. There is no definitive treatment for the babies with genetic disorders. It is very challenging and difficult for the parents to take care of a child with genetic disease. So it is important for parents to get support from health care providers to provide the best quality of life for the child.

Keywords: Genetic disease, Genetic disorders, Genes

GENETIC DISORDERS

Introduction

You have ever listen , "It's in my genes"? We mostly talk about a physical characteristic, personality trait, or talent that is shared with other members of our family. Genes play an important role in shaping, IN person's look and act and even whether we get sick. Now scientists are trying to use that knowledge in exciting new ways, such as treating health problems.

Definition of Gene

Most living organisms are made up of cells that contain a substance called Deoxyribonucleic acid. Genes are particular factors that passes unchanged from parent to progeny determines a biological character of an organism. Chromosome is a thread like structure that is visible during cell division.

Genetics

There are billions of people on earth, no two look exactly alike. Their Facial characteristics, hair, eye and skin color, hair texture and body build make distinct from one another.Even many visible differentiating characteristics, there are many more that are invisible like blood chemistry, mental and phyysical aptitudes, susceptibility to certain diseases, and other traits, which contribute to one's individuality. But there are also similarities among human beings, which makes them distinct from other species. The mechanism responsible for this similarity

with differences and how is it transmitted from one generation to another,need to understand. The science that deals with the study of heredity and variations is known as genetics.

Nursing's unique contribution to genomic medicine is its philosophy of holism. Nurses are ideally positioned to implement genetics into their assessment, planning and interventions for patient at different ages and stages across the lifespan and in all settings. The holistic view that characterizes nursing takes into account each person's intellectual, physical, spiritual, social, cultural, biopsychologic, ethical and esthetic experiences while addressing genetics information, gene based testing, diagnosis and treatments. Thus knowledge about genetics is basic and essential to nursing practice.

Gene and hereditary

Genes hold DNA or it's a segment of DNA that is responsible for giving instructions in the production of proteins. It directs particular function of body.When mutations occurs in genes may it cause failure in the working of proteins leading to a condition called genetic disorder. These disorders may be inherited from parents or may occur at any point of lifetime. Genetic disorder may result in the addition or reduction in the number of chromosomes. A genetic disorder is a disease that is caused by an abnormality in an individual's DNA system. Abnormalities can be as small as a single-base mutation in just one gene, or they can involve the addition or subtraction of entire chromosomes. Some people inherit genetic disorders from the parents, while acquired changes or mutations in a preexisting gene or group of genes cause other genetic diseases. Genetic mutations can occur either randomly or because of some environmental exposure.

The four types of genetic disorders

1. Single gene inheritance
2. Multi factorial inheritance
3. Chromosome abnormalities
4. Mitochondrial inheritance

1. Single gene inheritance disorders

Single gene inheritance is also called Mendelian or monogenetic inheritance. It is caused when changes or mutations occur in the DNA sequence of a single gene. 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

- Autosomal dominant inheritance.
- Autosomal recessive inheritance
- X-linked inheritance

Examples

1. Cystic fibrosis
2. Alpha- and beta-thalassemias
3. Sickle cell anemia (sickle cell disease)
4. Marfan syndrome
5. fragile X syndrome
6. Huntington's disease
7. Hemochromatosis

2. Common multi factorial genetic inheritance disorders

Multi factorial inheritance is also called complex or polygenic inheritance. It is cause by multi factorial 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.

Examples

1. Heart disease
2. High blood pressure
3. Alzheimer's disease
4. Arthritis
5. Diabetes
6. Cancer
7. Obesity

3. 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.

Example

1. Down syndrome or trisomy 21
2. Turner syndrome (45,X0)
3. Klinefelter syndrome (47, XXY)
4. Cri du chat syndrome, or the "cry of the cat" syndrome (46, XX or XY,)

4. Mitochondrial genetic inheritance disorders

This type of genetic disorder 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 their mitochondria during fertilization, mitochondrial DNA is always inherited from the female parent.

Examples

1. Leber's hereditary optic atrophy ,xanveye disease
2. myoclonic epilepsy with ragged red fibers
3. mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes

There are well over 6,000 known genetic disorders, and new genetic disorders are constantly being described in medical literature. More than 600 of these disorders are treatable. Around 1 in 50 people are affected by a known single-gene disorder, while around 1 in 263 are affected by a chromosomal disorder.

So with the view of heavy global burden of genetic disorders author has selected this topic and out of 4 major types of genetic disorders, four examples of genetic diseases are explained that are like Huntington disease, Edward syndrome, Turner syndrome and Down syndrome.

1. **HUNTINGTON DISEASE**

Introduction : Huntington disease is a brain disorder in which brain cells, or neurons, in certain areas of your brain start to break down. As the neurons degenerate, the disease can lead to emotional disturbances, loss of intellectual abilities, and uncontrolled movements.

Huntington disease has 2 subtypes:

- Adult-onset Huntington disease.
- Early-onset Huntington disease

Symptoms

- Irritability
- Depression
- Mood swings
- Trouble driving
- Trouble learning new things
- Forgetting facts
- Trouble making decisions

As the disease progresses, the following symptoms become more common:

- Trouble feeding oneself
- Difficulty swallowing
- Strange and uncontrolled movements that are either slow or wild and jerking (chorea)
- Loss of memory and judgment
- Changes in speech
- Personality changes
- Disorientation and confusion
- Hallucinations, paranoia, and psychosis

In children

- Slow movements
- Rigidity
- Tremor

Diagnosis

- Blood tests
- Computed tomography
- Magnetic resonance imaging
- Positron emission tomography

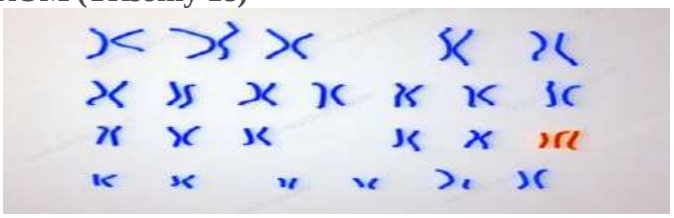
Risk factor

Most people develop it in their 30s or 40s. Huntington disease is a rare disorder. More than 15,000 Americans currently have the disease, but many more are at risk of developing it.

Treatment

It can't cure or slow the progression of Huntington disease, but health care providers can offer medications to help with certain symptoms. Drugs like haloperidol, tetrabenazine, and amantadine are especially helpful for controlling the unusual movements caused by Huntington disease.

3.EDWARD SYNDROM (Trisomy 18)



Introduction: Trisomy 18 is a chromosomal abnormality also called Edwards syndrome. Chromosomes are the thread like structures in cells that hold genes. Sometimes the mother's egg or the father's sperm contains the wrong number of chromosomes. As the egg and sperm combine, this mistake is passed on to the baby. A "trisomy" means that the baby has an extra chromosome in some or all of the body's cells. In the case of trisomy 18, the baby has three copies of chromosome 18. This causes many of the baby's organs to develop in an abnormal way.

Types

- Full trisomy 18
- Partial trisomy 18
- Mosaic trisomy 18

Symptoms

- Intrauterine growth retardation
- Craniofacial features such as abnormalities of the jaw, skull, ears, and neck
- Clenched fists with overriding fingers
- Small fingernails
- Short sternum
- Club feet
- Heart defects
- Kidney defects
- Neuro developmental delays11

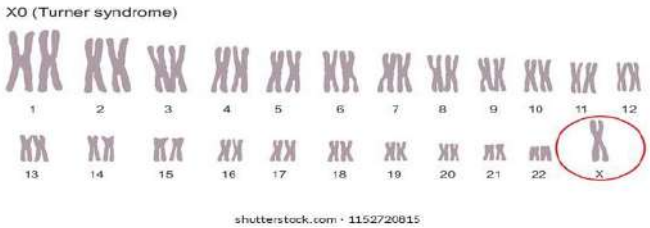
Diagnosis

- Pregnancy ultrasound
- Amniocentesis
- Chorionic villus sampling

Treatment

There is no cure for trisomy 18. Treatment for trisomy 18 consists of supportive medical care to provide the child with the best quality of life possible.

3. **TURNER SYNDROM**



Introduction : Turner syndrome, a condition that affects only females, results when one of the X chromosomes is missing or partially missing. Turner syndrome can cause a variety of

medical and developmental problems, including short height, failure of the ovaries to develop and heart defects.

Symptoms : Before birth

Prenatal ultrasound : Large fluid collection on the back of the neck or other abnormal fluid collections (edema)

- Heart abnormalities
- Abnormal kidneys

At birth or during infancy

- Wide or weblike neck
- Low-set ears
- Broad chest with widely spaced nipples
- High, narrow roof of the mouth (palate)
- Arms that turn outward at the elbows
- Fingernails and toenails that are narrow and turned upward
- Swelling of the hands and feet, especially at birth
- Slightly smaller than average height at birth
- Slowed growth
- Cardiac defects
- Low hairline at the back of the head
- Receding or small lower jaw
- Short fingers and toes

In childhood, teens and adulthood

- Slowed growth
- No growth spurts at expected times in childhood
- Adult height significantly less than might be expected for a female member of the family
- Failure to begin sexual changes expected during puberty
- Sexual development that "stalls" during teenage years
- Early end to menstrual cycles not due to pregnancy

Diagnosis

- Webbed neck
- Broad chest
- Widely spaced nipples
- Ultrasound test
- Chorionic villous sampling
- Amniocentesis
- Karyotype

Causes

Most people are born with two sex chromosomes. Boys inherit the X chromosome from their mothers and the Y chromosome from their fathers. Girls inherit one X chromosome from each parent. In girls who have Turner syndrome, one copy of the X chromosome is missing, partially missing or altered.

The genetic alterations of Turner syndrome may be one of the following:

- Monosomy
- Mosaicism
- X chromosome abnormalities
- Y chromosome material

Effects

The missing or altered X chromosome of Turner syndrome causes errors during fetal development and other developmental problems after birth — for example, short stature, ovarian insufficiency and heart defects. Physical characteristics and health complications that arise from the chromosomal error vary greatly.

Treatment

Turner syndrome is a genetic condition with no cure, but treatment may help resolve issues with short stature, sexual development, and learning difficulties. Estrogen and progesterone replacement therapy will enable sexual development and reduce the risk of osteoporosis.

4.**DOWN'S SYNDROM**

Introduction: Down syndrome is a condition in which a child is born with an extra copy of their 21st chromosome hence it's other name, trisomy 21. This causes physical and mental developmental delays and disabilities. Many of the disabilities are life long, and they can also shorten life expectancy.

Causes

In all cases of reproduction, both parents pass their genes on to their children. These genes are carried in chromosomes. When the baby's cells develop, each cell is supposed to receive 23 pairs of chromosomes, for 46 chromosomes total. In children with Down syndrome, one of the chromosomes doesn't separate properly. The baby ends up with three copies, or an extra partial copy, of chromosome 21, instead of two. This extra chromosome causes problems as the brain and physical features develop.

Types

1. Trisomy 21
2. Mosaicism
3. Translocation

Other parents who are more likely to have a child with Down syndrome include:

- people with a family history of Down syndrome
- people who carry the genetic translocation

Symptoms : At birth

- Flat facial features
- Small head and ears
- Short neck
- Bulging tongue
- Eyes that slant upward
- Atypically shaped ears
- Poor muscle tone
- Impulsive behavior
- Poor judgment
- Short attention span
- Slow learning capabilities

Diagnosis

Screening for Down syndrome is offered as a routine part of prenatal care in the United States. If you're a woman over 35, your baby's father is over 40, or there's a family history of Down syndrome, you may want to get an evaluation.

Treatment

There's no cure for Down syndrome, but there's a wide variety of support and educational programs that can help both people with the condition and their families.

School is an important part of the life of a child with Down syndrome, regardless of intellectual ability. Public and private schools support people with Down syndrome and their families with integrated classrooms and special education opportunities. Schooling allows valuable socialization and helps students with Down syndrome build important life skills.

Conclusion

Five main disorders have been explained in this article with their introduction, sign and symptoms, diagnostic technique, treatment and complications. As the author reviewed that there is NO cure for genetic disorders. So there is a scope for nurses to come forward in the field of genetic studies to provide preconception counseling sessions and psychological support to the parents.

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