Genetics and Cancer

- Genes are made up of deoxyribonucleic acid (DNA) and are found in each cell in the body
- It controls how each cell functions, including how quickly it grows, how often it divides and how long it lives
- Researchers estimate that there are 30,000 different genes in each cell

Genes and Cancer

Genes

- Genes control how the cells work by making proteins that have specific functions and act as messengers for the cell

- All cancers begin when one or more genes in a cell are mutated (changed), creating an abnormal protein or no protein at all

- The information provided by an abnormal protein is different from that of a normal protein, which can cause cells to multiply uncontrollably and become cancerous

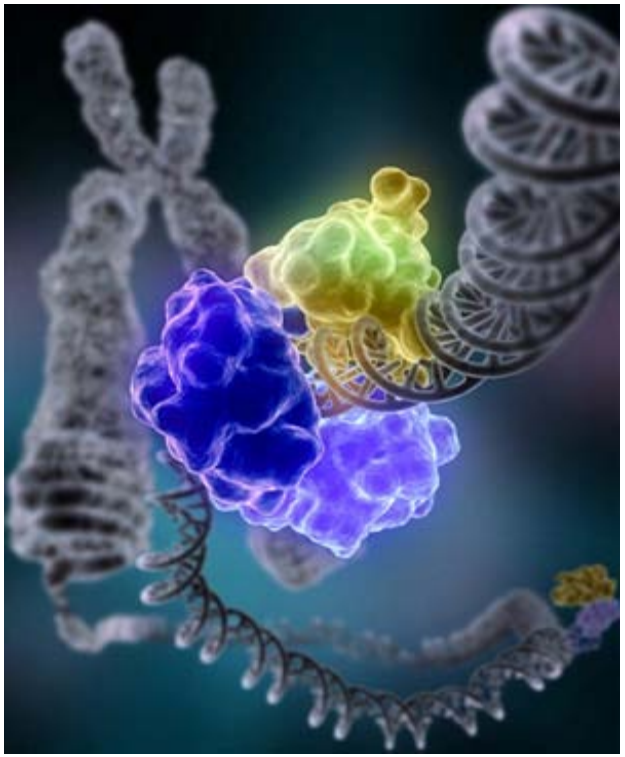
Gene mutations

- Mutations are abnormal changes in the DNA of a gene
- The building blocks of DNA are called bases that determine the gene and its function
- Even a change in just one base among the thousands of bases that make up a gene can have a major effect

- Cells become cancer cells largely because of mutations
- Often many mutations are needed before a cell becomes a cancer cell
- The mutations may affect different genes that control cell growth and division. Some of these genes are called tumor suppressor genes
- Mutations may also cause some normal genes to become cancer causing genes known as oncogenes

Tumor suppressor genes

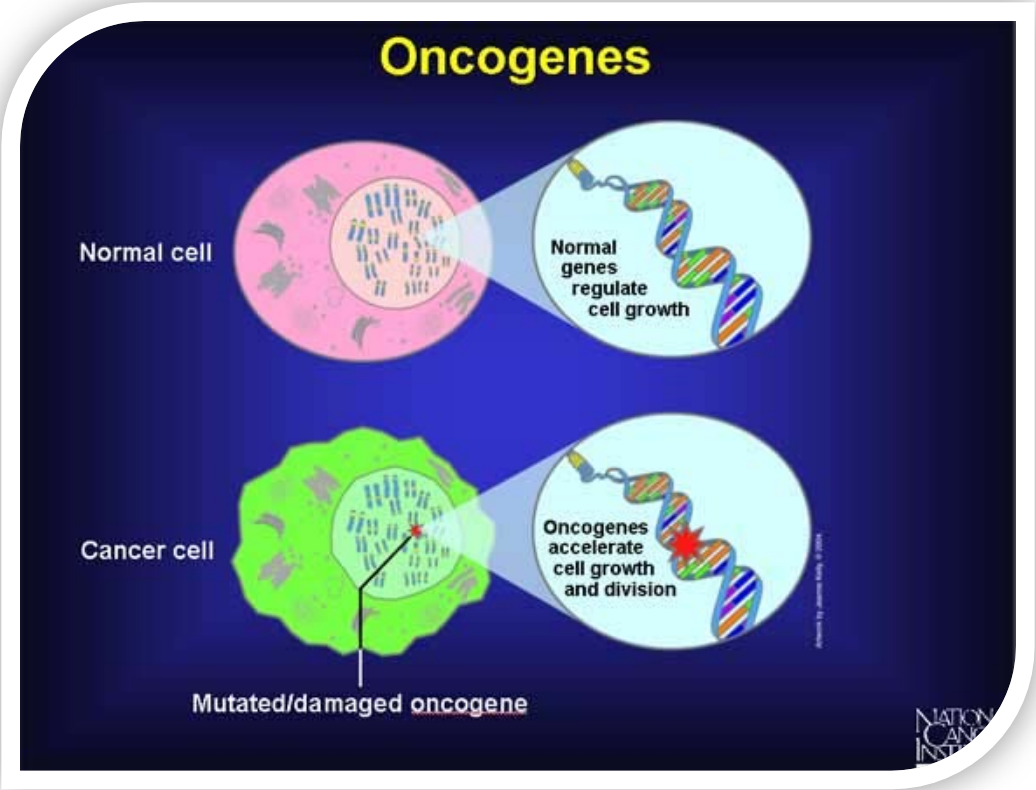
- Tumor suppressor genes are protective genes
- Normally, they suppress (limit) cell growth by monitoring how quickly cells divide into new cells, repairing mismatched DNA and controlling when a cell dies
- When a tumor suppressor gene is mutated, cells grow uncontrollably and may eventually form a mass called a tumor



Oncogenes

- Oncogenes turn a healthy cell into a cancerous cell

- Mutations in these genes are almost always acquired (not inherited)

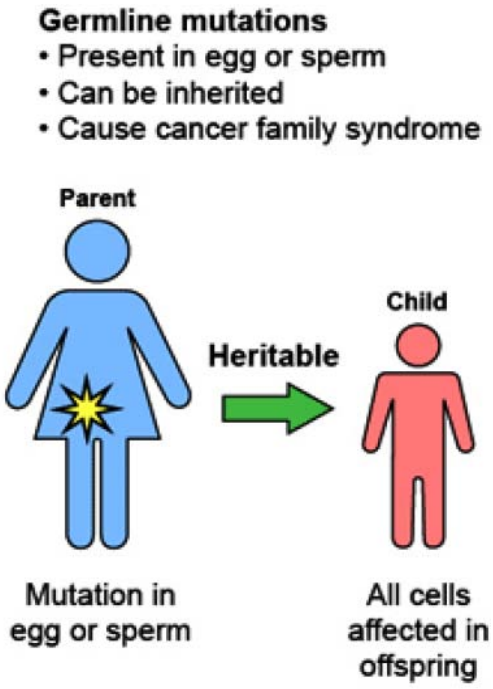


Types of mutations

There are 2 major types of gene mutations:

1. **Inherited**

- An inherited gene mutation is present in the egg or sperm that formed the child
- After the egg is fertilized by the sperm, it created one cell called a zygote that then divided to create a fetus (which became a baby)
- Since all the cells in the body came from this first cell, this kind of mutation is in every cell in the body and so can be passed on to the next generation
- This type of mutation is also called germline (because the cells that develop into eggs and sperm are called germ cells) or hereditary
- Inherited mutations are thought to be a direct cause of only a small fraction of cancers



2. Acquired mutation

- An acquired mutation is not present in the zygote, but is acquired some time later in life
- It occurs in one cell, and then is passed on to any new cells that are the offspring of that cell
- This kind of mutation is not present in the egg or sperm that formed the fetus, so it cannot be passed on to the next generation
- Acquired mutations are much more common than inherited mutations
- Most cancers are caused by acquired mutations. This type of mutation is also called sporadic, or somatic

Somatic mutations
- Occur in ***nongermline*** tissues
- Cannot be inherited

Nonheritable

Mutation in tumor only
(for example, breast)

How genes can help in the diagnosis and treatment of cancer

- Certain mutations are commonly found in the cells of some types of cancers
- Finding certain mutations in cells can confirm the diagnosis of that cancer
- Testing cells for the mutation can also be used after diagnosis to see how the cancer is responding to treatment

Family Cancer Syndromes

Family Cancer Syndromes

- Many families have at least a few members who have had cancer
- Certain types of cancer seem to run in some families, this is because family members have certain risk factors in common, such as smoking, which can cause many types of cancer
- It can also be due in part to other factors, like obesity, that tend to run in families and influence cancer risk

Family cancer syndromes - when should I worry?

Certain things make it more likely cancers in a family are caused by a family cancer syndrome, such as

- Many cases of an uncommon or rare type of cancer like kidney cancer
- Cancers occurring at younger ages than usual like colon cancer in a 20 year old
- More than one type of cancer in a single person like a woman with both breast and ovarian cancer
- Cancers occurring in both of a pair of organs like both eyes, both kidneys, both breasts
- More than one childhood cancer in a set of siblings like sarcoma in both a brother and a sister
- Cancer occurring in the sex not usually affected like breast cancer in a man

Examples of family cancer syndromes

Hereditary Breast and Ovarian Cancer syndrome

- Doctors noticed that in some families, many of the women developed breast and or ovarian cancer
- Often the cancers were found at younger than usual ages, and some of the women had more than one cancer
- Some had breast cancer in both breasts, and some got both breast and ovarian cancer.
- Doctors studying these families coined the term Hereditary Breast and Ovarian Cancer Syndrome (HBOC)
- Scientists studied the genes of these families, and discovered the genes BRCA1 and BRCA2. Because some women (and families) have HBOC based on cancer history, but don't have mutations in these genes
- If someone in a family is found to have a BRCA mutation, it means that their close relatives like parents, siblings and children have a 50% chance of having a mutation, too

Examples of family cancer syndromes

Hereditary non-polyposis colorectal cancer

- The most common inherited syndrome that increases a person's risk for colon cancer is called hereditary non-polyposis colorectal cancer (HNPCC) or Lynch syndrome
- People with this syndrome have a high risk of colorectal cancer
- Most of these cancers occur before age 50
- HNPCC also leads to a high risk of endometrial cancer in women
- Other cancers linked with HNPCC include cancer of the ovary, stomach, small intestine, pancreas, kidney, brain, ureters and bile duct
- If someone in a family is found to have HNPCC, it means that their close relatives like parents, siblings and children have a 50% chance of having a mutation, too

Examples of family cancer syndromes

Li-Fraumeni syndrome

- Li-Fraumeni syndrome is a rare syndrome that can lead to the development of a number of cancers, including sarcoma, leukemia, brain cancers, cancer of the adrenal cortex and breast cancer
- The cancers most often occur in childhood, although the breast cancers occur in young adults
- People with Li-Fraumeni can also be affected by more than one cancer in their lifetime

Genetic testing

Genetic testing

- Genetic testing is the process of using medical tests to look for changes in a person's genes or chromosomes

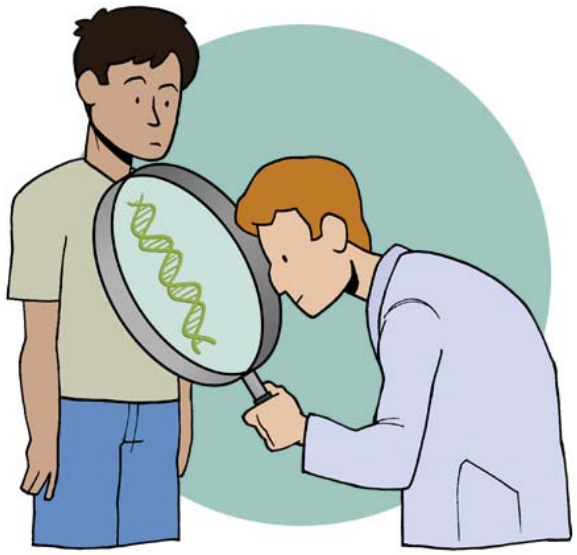
- Hundreds of different genetic tests are used today, and more are being developed

Genetic testing for diseases that can be inherited

- Genetic testing can be used in different situations
- The type of testing most often used to check for cancer risk is called predictive gene testing
- It’s used to look for gene mutations that might put a person at risk of getting a disease
- Genetic testing is also includes Carrier testing , Prenatal screening and Newborn screening

Genetic testing for cancer risk

Cancer-related genetic tests are most commonly done as predictive genetic tests. They may be used:



- To see if a person has a certain gene mutation known to increase the risk for a certain cancer

- To confirm a suspected gene mutation in a person or family

Testing cancer cells for genetic changes

- Sometimes after a person has been diagnosed with cancer, the doctor will order tests to look for gene changes in a sample of the cancer cells

- These tests can give information on a person's prognosis and can sometimes help to tell whether certain types of treatment might be useful

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