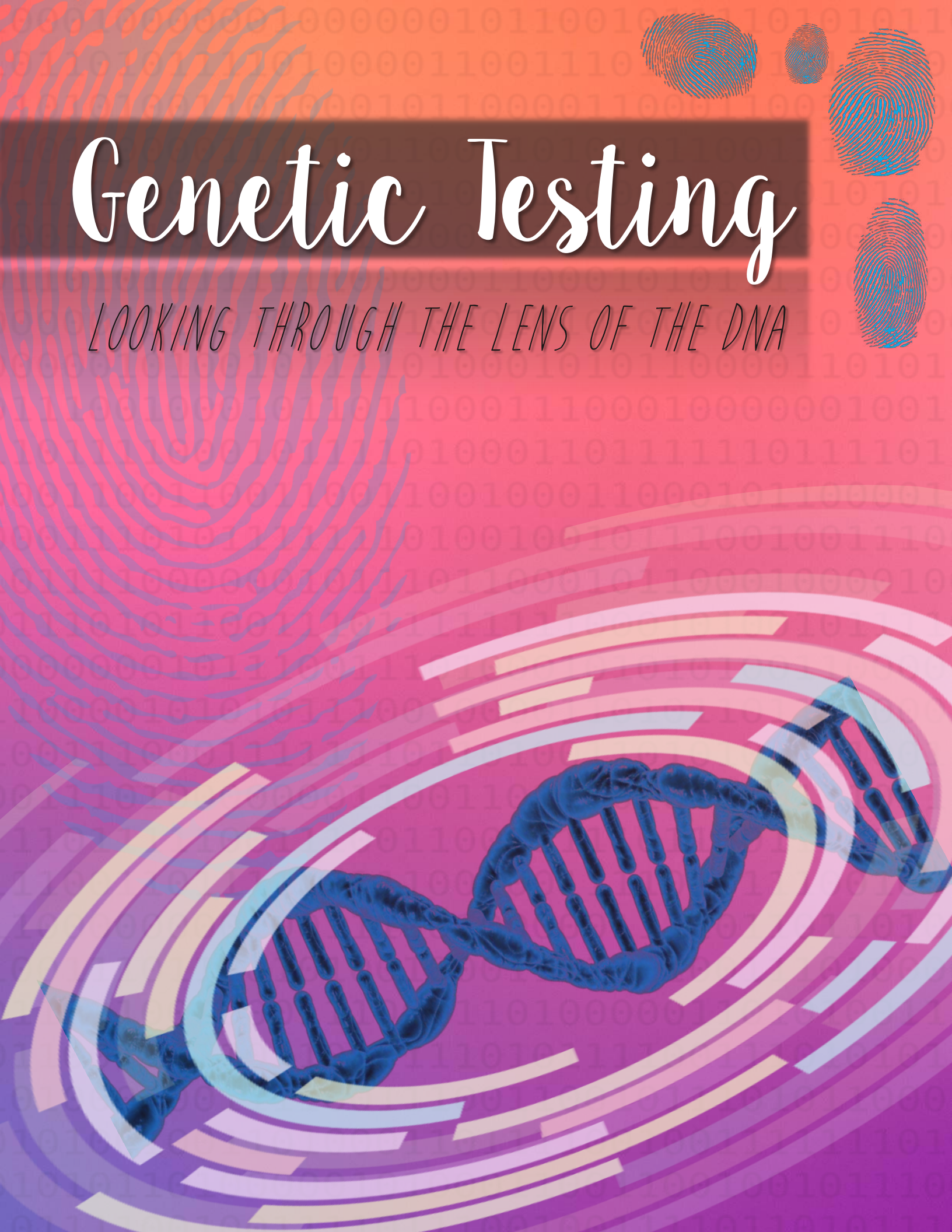


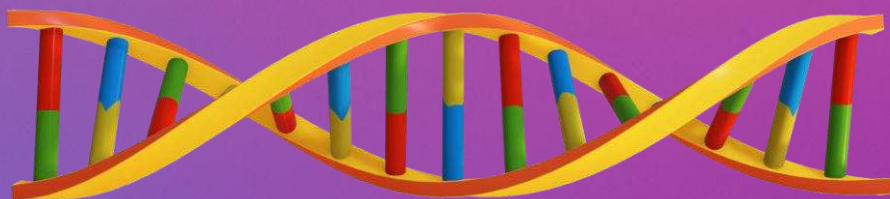
# Genetic Testing

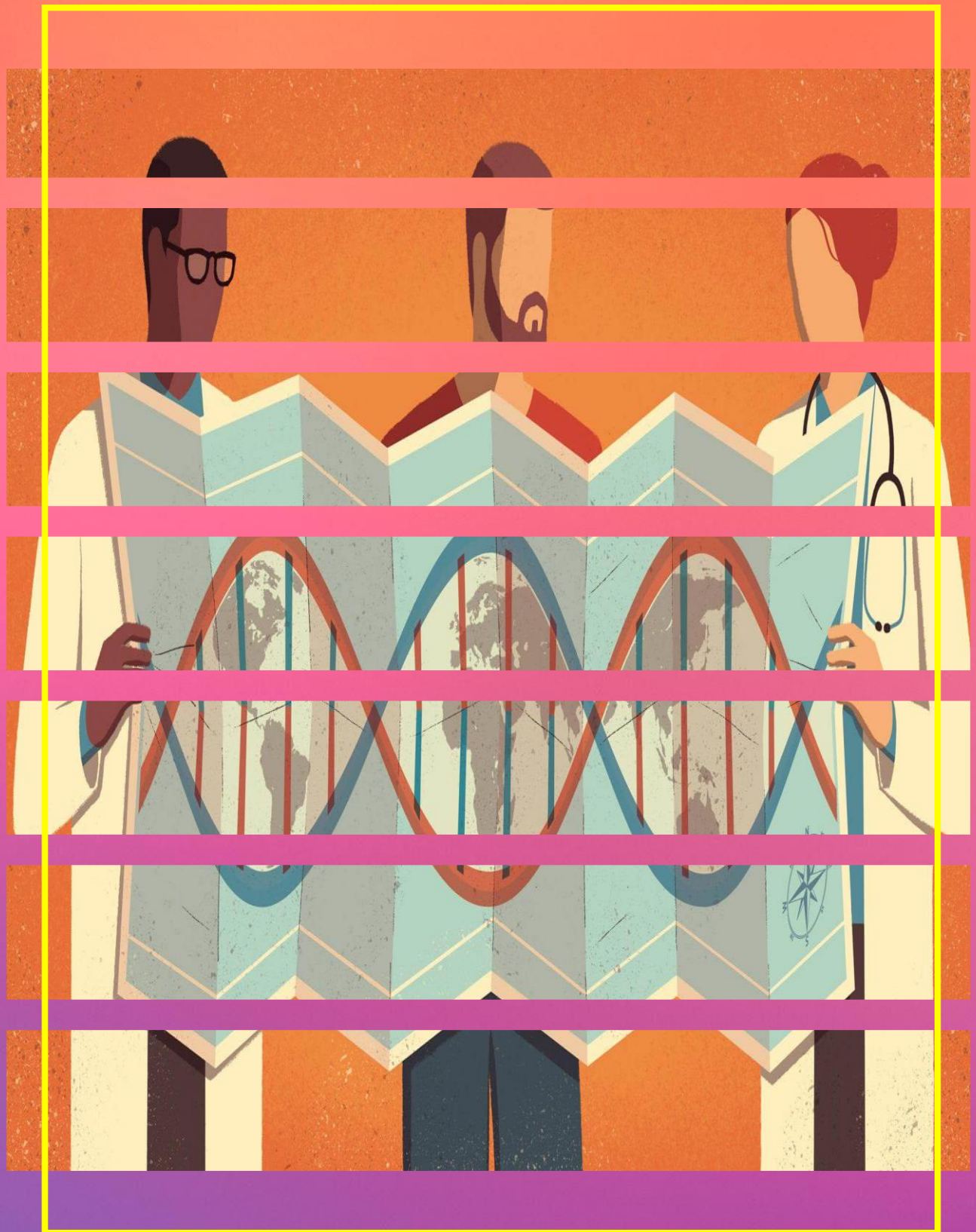
*LOOKING THROUGH THE LENS OF THE DNA*



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# Genetics and Genetic Testing



# What you need to Know

SOME OF THE BASIC CONCEPTS IN GENETICS [12]

## What is a Gene?

a segment of DNA that contains instructions for building a particular protein

contains the information that make you look the way you are

Inheritance – traits that are determined by the genes are passed from parent to child

## Genetic Disorders and Diseases

When there is an abnormal change in the gene, or a **mutation**, it may cause a disease or increase a person's risk of developing a genetic disorder. Genes are passed from parent to child. It is uncontrollable which genes are passed. It is also important to know that not all mutations to our genes are passed from our parents. Sometimes, **people can get genetic disorders if they have copying mistakes or damages in their own DNA.**

## Carriers

are people who have abnormal changes in their genes but do not have the disease

## Inheritance of Genetic Conditions

Genetic conditions caused by a mutation to a single gene can be inherited by one of several patterns. The patterns of inheritance include autosomal dominant, autosomal recessive, X-linked recessive, and X-linked dominant. The pattern of inheritance depends on what gene is mutated and where the mutated gene is located.

Remember, not all mutations to the genes are passed from parents. People can get genetic disorders if there is an error in their own genes. Errors that cause mutations can happen during cell division. Any changes to the chromosome number or structure can cause chromosome abnormality which can lead to different genetic conditions and diseases.

# What is Genetic Testing<sup>[5][9][12]</sup>

- is a type of medical test that identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. More than 1,000 genetic tests are currently in use, and more are being developed.
- The Types Of Genetic Testing are the following:
  1. **Newborn Screening** – is a test done shortly after birth to test for treatable diseases that usually show no symptoms in the newborn period. It helps to identify genetic disorders that can be treated early in life. Using a heel-prick, a small amount of blood is collected shortly after birth.
  2. **Diagnostic Testing** – is used to identify or rule out a specific genetic or chromosomal condition. It can be performed before birth or at any time during a person's life, but is not available for all genes or all genetic conditions.
  3. **Carrier Testing** – is used to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder. This type of testing is offered to individuals who have a family history of a genetic disorder.

# What is Genetic Testing [5][9][12]

4. **Prenatal Testing** – is used to detect changes in a fetus' genes or chromosomes before birth. This type of testing is offered during pregnancy. Amniocentesis and Chronic Villus Sampling (CVS) are two of the most common techniques. Additionally, prenatal testing can lessen a couple's uncertainty or help them make decisions about a pregnancy.
5. **Preimplantation Testing** – this test may be used when one attempts to conceive a child through in vitro fertilization. The embryos are screened for genetic abnormalities. Embryos without abnormalities are implanted in the uterus in hopes of achieving pregnancy
6. **Predictive and Presymptomatic Testing** – is used for people who do not have signs or symptoms but have a family member that has a genetic disorder. This test can find out if you have the gene that causes the disease. It can also find out if the changes in your genes may increase your risk of getting a genetic disorder later in life.
7. **Forensic Testing** - Forensic testing uses DNA sequences to identify an individual for legal purposes. Unlike other tests, forensic testing is not used to detect gene mutations associated with disease. This testing helps identify crime or catastrophe victims, rule out or implicate a crime suspect, or establish biological relationships.

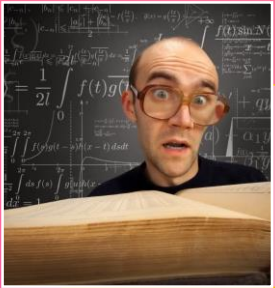


# *Genetic Testing and the Society*

# The Benefits of Genetic Testing <sup>[12][14]</sup>



Sense of relief from uncertainty



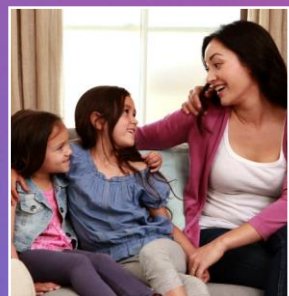
In-depth knowledge about risks for diseases



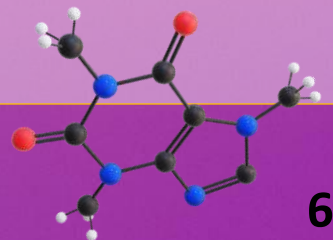
Helps make informed medical and lifestyle decisions



Helps pinpoint genetic factors that caused a certain disease



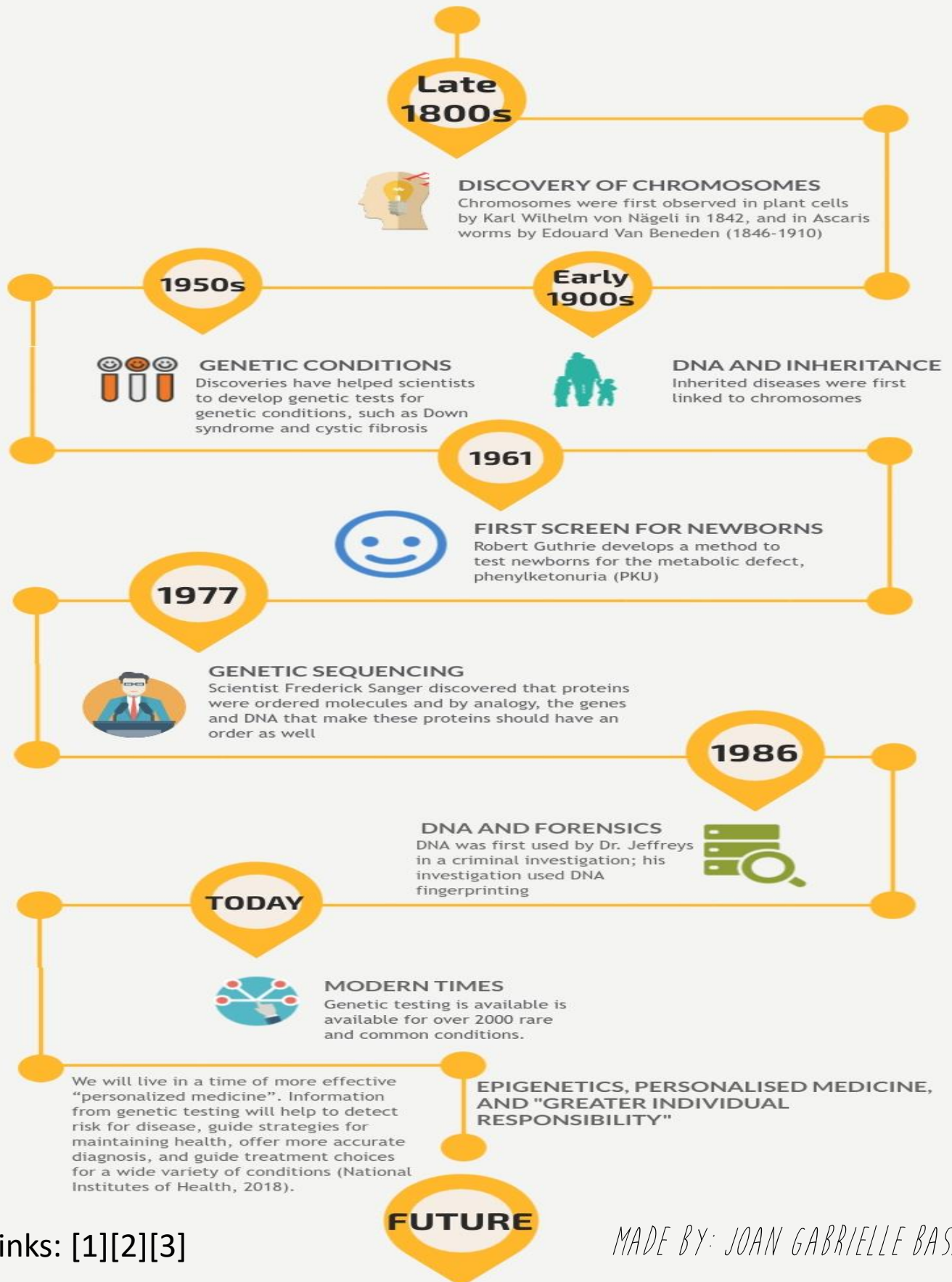
Educate other family members about potential risks



# Genetic Testing

## THROUGH THE AGES

Our world is changing at an extremely rapid rate. As advancements in terms of science and technology continue to emerge through time, questions that have baffled scientists for the past years are now being given answers—most especially in the field of genetics.



# The Future of Genetic Testing <sup>[6][10][11][13]</sup>

## Now

- Genealogical Analysis
- Determine an individual's ancestry

## Future

- Molecular Diagnostics
- Analyses biological markers in the genome

## Future

- DNA Phenotyping Services
- Facial composite can be produced from DNA

## Future

- 3D Facial Modelling
- Possibility of predicting physical appearance

## Future

- Next Generation Sequencing
- Can analyze every location of gene in a traditional DNA barcode

## Future

- Disease Report Cards
- Parents could leave with a cost-effective and accurate genetic profile of their child

Note: Future genetic testing technologies are currently under research and have no projected completion

- **Emotional Burden**: The result of a genetic test can take many months to return from the laboratory. During this time, you may experience emotions of worry, confusion, and uncertainty<sup>[12]</sup>
- **Exposure of Family History**: A positive result may change family relationships as the result may disclose information about the genetic make-up of other relatives, even if they have not been tested. Furthermore, a genetic test may reveal unexpected relationships, such as having a non-biological relative<sup>[12]</sup>
- **Insurance Coverage**: In Canada, primary health insurance is not affected, but there can be problems in getting life insurance, extended or private health insurance, private disability insurance or mortgage insurance if you are diagnosed with a disorder<sup>[12]</sup>
- **Inconclusive or Equivocal Results**: usually means that a gene change has not been previously identified as the cause of a specific genetic disorder<sup>[12]</sup>
- **Genetic Discrimination in Insurance or Employment**: occurs when people are treated differently by their employer or insurance company because they have a gene mutation that causes or increases the risk of an inherited disorder<sup>[7]</sup>



*Controversies and Downsides behind Genetic Testing*

# Genetic Testing Research

## (A PERSONAL OPINION)

Scientific research has come a long way. It has brought numerous contributions in the past and is currently continuing to do so. Specifically, research in the field of genetics is significant to our lives since it has a lot of benefits in terms of health and medicine. For example, when the discovery of mutation in genes has come forth, it opened a wider opportunity for scientists to discover therapies for genetic conditions. In genetic testing, the goals of research are: finding unknown genes, learning how genes work, developing tests for future clinical use, and advancing our understanding of genetic conditions (Genetics Home Reference, 2019). With the aforementioned goals, I deeply believe that research or further development of genetic testing should happen. There is still a vast amount of questions in genetic testing, or genetics in general, that needs to be answered or even unknown questions that are waiting to be discovered. Through undergoing research, we will have a more deeper understanding of genetic concepts and we can develop more efficient and effective technologies and treatments, most especially in medicine, that would benefit all of humanity.



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[3]<https://ghr.nlm.nih.gov/>

[4]<https://www.genome.gov/pages/education/genetictimeline.pdf>

[5]<https://www.mayoclinic.org/tests-procedures/genetic-testing/about/pac-20384827>

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[7]<https://ghr.nlm.nih.gov/primer/testing/discrimination>

[8]<https://report.nih.gov/nihfactsheets/ViewFactSheet.aspx?csid=43&key=G>

[9]<https://www.newbornscreening.on.ca/en/about-screening/whatscreening>

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[15] <https://ghr.nlm.nih.gov/primer/testing/researchtesting>

