AFFORDABLE GENETIC TESTING

COMPiled by HOWIE BAUM
THE MIRACLE OF HOW OUR BODY WORKS

THE FIRST PART OF THIS PRESENTATION IS ABOUT DNA, CHROMOSOMES, GENES, THE DIVISION OF BODY CELLS AND OTHER AMAZING THINGS THAT ARE GOING ON IN OUR BODIES ALL OF THE TIME, TO KEEP US AS HEALTHY AS POSSIBLE.

AS THEY ARE DISCUSSED, PLEASE CONSIDER:

WHAT MIRACULOUS PROCESSES ARE TAKING PLACE, TO MAKE ALL OF THEM HAPPEN?

HOW DO ALL OF THESE MINIATURE BODY CELLS AND PARTS, KNOW HOW TO MAKE THIS HAPPEN?
WHAT IS GENETIC TESTING?

Genetic testing is a type of medical test that identifies changes in a person’s 1) Chromosomes, 2) Genes, or 3) 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 75,000 genetic tests are currently in use, and more are being developed all of the time.

This presentation is about Genetic Testing for adults.

In order to discuss it, we need to first talk about what DNA, Chromosomes, and Genes are.
The human body contains 37.2 trillion cells.

There is a nucleus inside each human cell (except red blood cells).

Each nucleus contains 46 chromosomes, arranged in 23 pairs.

One chromosome of every pair is from each parent.

The chromosomes are filled with tightly coiled strands of DNA.

Genes are segments of DNA that contain instructions to make proteins—the building blocks of life.

A chromosome contains hundreds to thousands of genes.

Note that there are 2 very special things that DNA does in each of our cells–

1) It makes each of the 42 million proteins that are in each of our cells that our body needs

2) The ability to be duplicated accurately, to make new cells that replace old ones that die.
DNA

Often referred to as the molecule of life, DNA (deoxyribonucleic acid) is found in almost all living things.

It acts as a type of chemical code that contains instructions, known as genes, for how the body and all its different parts grow, develop, function, and maintain themselves.

It is tightly packaged into 46 X-shaped elements called chromosomes, which are in every cell’s nucleus!

DNA’s enormous list of instructions takes the form of long, thin molecules, one strand per chromosome, each taking the shape of a double-helix.
GENETIC CODE

The double helix of DNA consists of two corkscrew-like backbones joined by cross-rungs, which are pairs of chemical bases, of four kinds,

Adenine (A)
Thymine (T)
Guanine (G)
Cytosine (C).

The bases always pair in a specific way - A to T and G to C and are called a base pair.

There are almost 3 billion base pairs in each strand of DNA!

https://www.youtube.com/watch?v=zwibgNGe4aY 5.3 minutes
This extra coiling process allows the 3 billion base pairs in each cell to fit into a space just **6 microns** (6 millionths of a meter) across, which is about .00004 (4 hundred thousandths) of an inch.

**This is geometrically equivalent to packing 40 km (24 miles) of extremely fine thread into a tennis ball!**

For size comparison, a human red blood cell is about **5 microns** across. A human hair is about **75 microns** across.

If you stretched the DNA in one cell all the way out, it would be about 2 meters (over 6 feet) long!!
IF ALL OF THE DNA IN ALL OF YOUR CELLS WERE STRETCHED OUT AND PUT TOGETHER, IT WOULD BE ABOUT TWICE THE DIAMETER OF THE SOLAR SYSTEM.
All humans have 99.9% identical genetic makeup.

The remaining 0.1% difference may provide useful information about diseases.
DNA REPLICATION IS PROBABLY ONE OF THE MOST AMAZING TRICKS THAT DNA DOES.

- Each cell contains all the DNA you need to make the other cells.
- We start out from a single cell and we end up with 37.2 trillion cells.
- During that process of cell division, all the information in a cell, has to be copied, and it has to be copied perfectly.
- There are almost three billion base pairs in each strand of DNA to be copied which is done by molecules called polymerases that are specifically dedicated to just copying DNA.
- This takes about 8 hours of copying time.
- At the end of this process, the cell actually has twice the amount of DNA that it needs
- It can then divide and parcel this DNA into the daughter cell, so that the daughter cell and the parental cell are absolutely, genetically identical.

Think of the miracle going on inside each of our body cells. We have our own miniature manufacturing process going on to create a duplicate copy of new DNA, for each cell!

https://www.youtube.com/watch?v=bee6PWUgPo8 1 minute
Most of the time, DNA looks like a tangled ball of yarn or big bowl of noodles in each cell—diffuse, disordered, chaotic.

This is the time that copies of the long DNA strand is copied.

But that messiness poses a problem during mitosis, when the cell has to make a copy of its genetic material and divide in two.

In preparation, it tidies up by packing the DNA into dense, sausage-like rods, called Chromatids, with 2 identical ones together to make up the chromosomes’ most familiar form, in the shape of an X.

They are bonded at the center with a part called the Centromere.

https://www.youtube.com/watch?v=gbSIBhFwQ4s
GO TO 1.12 MINUTES

https://www.youtube.com/watch?v=57Q5V0HcIWU
A genome is the full set of genetic instructions for a living thing, controlling its development from a single cell into a complex, adult body.

The human genome consists of an estimated 20,000–25,000 genes, carried on the double set of 46 chromosomes found in nearly every kind of body cell.

The chromosomes are curved as shown in the diagram but for discussion sake, the next slides show them straight and parallel to each other.

This image of all of the chromosomes is called a Karyotype or Karyogram.

Except for the sex chromosomes X and Y, the rest are called Autosomes.
CHROMOSOMES

There are 46 chromosomes in every human cell, except for red blood cells which don’t have a nucleus.

These consist of 22 equivalent pairs, one of each pair derived from the mother and one from the father and are numbered from 1 (largest) to 22 (smallest).

The 23rd pair is the sex chromosomes, XX signifying female and XY (as here) male.

Men determine the sex of a baby depending on whether their sperm is carrying an X or Y chromosome. An X chromosome combines with the mother’s X chromosome to make a baby girl (XX) and a Y chromosome will combine with the mother’s, to make a boy (XY).

When colored by chemical stains, dark and pale stripes called banding patterns show up on each chromosome.

These allow researchers to “map” the locations of particular genes within the chromosome.
Chromosome seven

One of the first chromosomes to be sequenced, it contains more than 5 per cent of the genome’s total DNA, with about 159 million pairs of bases.

Almost 60 million are in the short arm, 7p, with the rest in the longer arm, 7q.

The conventions of labelling a chromosome make it possible to find the site of the gene if you know its “address”.

The cystic fibrosis gene (CFTR), for example, is located at 7q31.2.
Myotonic Dystrophy
Form of adult muscular dystrophy

Amyloidosis
Accumulation in the tissues of an insoluble fibrillar protein

Neurofibromatosis (NF1)
Benign tumors of nerve tissue below the skin

Breast Cancer
5% of all cases

Polycystic Kidney Disease
Cysts resulting in enlarged kidneys and renal failure

Tay-Sachs Disease
Fatal hereditary disorder involving lipid metabolism often occurring in Ashkenazi Jews

Alzheimer Disease
Degenerative brain disorder marked by premature senility

Retinoblastoma
Childhood tumor of the eye

Human chromosome number

Spinocerebellar Ataxia
Abnormally high absorption of iron from the diet

Cystic Fibrosis
Mucus in lungs, interfering with breathing

Werner Syndrome
Premature aging

Melanoma
Tumors originating in the skin

Multiple Endocrine Neoplasia, Type 2
Tumors in endocrine gland and other tissues

Sickle-Cell Anemia
Chronic inherited anemia, in which red blood cells sickle, clogging arterioles and capillaries

Phenylketonuria (PKU)
An inborn error of metabolism; if untreated, results in mental retardation
96 million cells in our body die every minute but luckily, 96 million new cells are created each minute, to replace them.

To do this, there has to be an efficient process in place to do that.

This is called **Mitosis**, when one cell divides to produce two genetically identical cells, with the same DNA in them.

This happens for all of our cells except for the red blood cells and the egg and sperm cells.

**It takes 8 hours for each cell to create a new one!!**
MEIOSIS

The other type of cell division is called Meiosis which is a process where a single cell divides twice to produce **four cells**, with each containing half the original amount of genetic information.

These cells are our sex cells – sperm in males, eggs in females.
WOMEN’S EGG CELLS

- Virtually all (99.9%) sex cells in a woman's ovaries never develop beyond the primary eggs stage and eventually are reabsorbed by her body.

- By 20 weeks after conception, there are approximately 7,000,000 primary egg cells in the growing female fetus.

- All but about 1,200,000 are lost by birth.

- At puberty, there are only around 400,000 of them remaining. This is when the Meiosis process starts, to make new eggs.

- Throughout life, there is a constant decline in the number of potential eggs.

- Each time one is successfully ovulated, as many as 2000 are reabsorbed into the body.

The woman’s egg cell is the biggest cell in the body and the man’s sperm cell is the smallest!
DOMINANT AND RECESSIVE GENES THAT CREATE OUR PHYSICAL TRAITS

Physical traits are observable characteristics that children inherit from their parents – eye and hair color, height, etc.

Dominant refers to the relationship between two versions of a gene.

Individuals receive two versions of each gene, known as alleles, from each parent.

If the alleles of a gene are different, the allele that will be expressed is called the dominant gene.

The effect of the other allele, called recessive, is masked.
WHAT ARE PHYSICAL TRAITS?
Physical traits are observable characteristics that children inherit from their parents.

Some physical traits, such as freckles, are expressed completely due to dominant or recessive inheritance of a single gene.

Other traits are expressed in varying degrees because they are influenced by multiple genes, such as left or right-handedness.

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<th>Inherited Traits</th>
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<tr>
<td><strong>Dominant Traits</strong></td>
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<tr>
<td>Black or brown hair</td>
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<td>Full lips</td>
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<td>Free earlobes</td>
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<tr>
<td>Dimples in cheeks</td>
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<td>High and narrow nose</td>
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<td>Brown eyes</td>
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This chart shows some of the inherited traits that dominate over recessive traits.
Widow's peak or not?
Can roll the tongue or not?
Which thumb is on the top?
Length of second toe?
Is it longer than your big toe or not?
Little finger straight or crooked?
Ear lobe hangs free
Ear lobe attached
Darwin's point
No Darwin's point
Ear lobe
Front teeth close together
Front teeth with a definite gap
GENETIC TESTING

- **Genetic testing**, also known as **DNA testing**, is used to identify changes in the **DNA sequence** or a chromosomal condition.

- In many cases, genetic testing is used to confirm a diagnosis when a particular condition is suspected based on **physical mutations in the DNA** and symptoms.

- The results of a diagnostic test can influence a person's choices about health care and the management of the disease.

- According to the National Institutes of Health, there are tests available for more than 6,000 genetic disorders.

*Why should I consider genetic testing? [https://www.youtube.com/watch?v=WUQ2-n3h5LM](https://www.youtube.com/watch?v=WUQ2-n3h5LM) 2.2 minutes*
WHAT ARE THE 3 KINDS OF DNA USED TO TRACE ANCESTRY?

Because the chromosomes mothers and fathers pass on to their children are reshuffled versions of the ones they inherited from their own parents, it is hard to use most chromosomes to trace genealogy back very far.

Most of the Y chromosome, however, is handed down from father to son without changes so they use Y-DNA.

Likewise, in humans, the tiny bit of DNA contained in an unusual package of genetic material in each body cell is known as mitochondrial DNA (MtDNA) is passed down from mother to child without any recombination.

The other type is called Autosomal DNA (atDNA) tests, which measure the main 22 pairs of chromosomes called the autosomes.

This is usually a more expensive test.
<table>
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<tr>
<th>TYPES OF GENETIC TESTS</th>
<th>Description</th>
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<tr>
<td>Diagnostic</td>
<td>Used to confirm a diagnosis based on physical signs</td>
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<tr>
<td>Predictive</td>
<td>Used to detect gene mutations associated with disorders that appear later in life</td>
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<td>Carrier Identification</td>
<td>Used by people with a family history of recessive genetic disorders</td>
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<tr>
<td>Prenatal</td>
<td>Used to test a foetus when there is risk of bearing a child with mental or physical disabilities</td>
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<tr>
<td>Newborn Screening</td>
<td>Used as a preventative health measure once the baby is born</td>
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<tr>
<td>Forensic testing</td>
<td>Used to identify an individual for legal purposes</td>
</tr>
<tr>
<td>Research testing</td>
<td>Used for finding unknown genes and identifying the function of a gene</td>
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Interest in getting a genetic test

AMONG ADULTS AGE 50–64

60% for estimating future risk of disease
60% for determining ancestry
54% for guiding diagnosis or treatment of medical condition

5% have gotten a genetic test for medical purposes
10% have gotten a direct-to-consumer genetic test

October 2018 Report: Older Adults’ Views on Genetic Testing
LEVELS OF GENETIC TESTING

**DNA**

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**Protein**

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**Protein Function**

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- **Analysis of whole chromosomes** – for large changes; extra chromosome, very large deletions or insertions

- **Analysis of the sequence of bases in our DNA** – for small changes; mutations in the sequence, small deletions or insertions

- **Analysis of protein shape** – for any change that may affect the folding of the protein

- **Analysis of protein function** – if the functional protein is supposed to make something, then some tests can detect the presence or absence of the product
Abnormal Number of Chromosomes

Trisomies - 3 copies rather than 2 copies of a chromosome

Monosomies – 1 copy rather than the usual 2
CHROMOSOME ABNORMALITIES THAT CAN HAPPEN

- Terminal deletion
- Terminal duplication
- Pericentric inversion
- Reciprocal translocation
- Ring chromosome
Examples of Mutations in the DNA Sequence...

aaaccatctaggctatatattcgagatctatctctactagctactacgactacgccggactactacgacgtctctactactagctactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccggactactacgactacgccg
Examples of Mutations in the DNA Sequence...

aaaccatctaggctatattcggatctatcctcagctactacgatcagggactactacgagctgactactacgagcatc
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Examples of Mutations in the DNA Sequence…

aaaccatctaggctatatctggtatcgtatctggtatctactagactactacgctcagggactactacgacagcatcgactacgaggcttctagaggtatcatatttcgagcataataaaaaaaaacgtagctaggtgtgggtgtgggtgtgtgggtgtgtgtgggtgtgtgtgtgtgggtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtgtg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Examples of Mutations in the DNA Sequence...

dna_sequence

deletion

(Duchenne muscular dystrophy)
Examples of Mutations in the DNA Sequence…

aaaccatctaggctatattcggatatcgtatctatcgtatctactagctactacgatcagggactactacgagcatcgactacgaggcttc
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ing Insertion

(Huntington’s disease)
Examples of Mutations in the DNA Sequence...

Multiple mutations

(Diabetes, susceptibility to breast cancer)
It published recommendations about reporting incidental findings described as ACMG-59 Genes.

In brief, the ACMG recommends that if DNA mutations are found in one of 59 different genes, they are considered as “medically actionable” and should be reported to individuals.

This is because of their potential high medical importance, even though they only apply to a small percentage of the population.

To see a listing of the 59 different genes and genetic disorders that they represent (it is very detailed), go to: https://www.coriell.org/1/NIGMS/Collections/ACMG-59-Genes
ACMG 59 DIFFERENT GENES

It’s currently estimated that approximately 3% of the US population—nearly 10 million people—has a variant in one of these 59 genes.

This list of genes is called the ACMG 59 and covers 3 types of important medical conditions:

1) Cancer predispositions
2) Cardiovascular predispositions
3) Metabolic conditions.

In all three cases, knowing about these conditions ahead of time can give you the power to make informed decisions and potentially avoid health complications.
TYPICAL COSTS:

❖ Detailed Genetic testing typically is covered by health insurance when it is recommended by the patient's doctor.

❖ For patients covered by health insurance, out-of-pocket costs for genetic testing typically would consist of doctor visit(s) and laboratory copays.

❖ Some covered patients choose to pay out of pocket for testing due to concerns about genetic discrimination.

❖ For patients not covered by health insurance, genetic testing cost ranges from about $100-$3,000 or more, depending on the individual, the type of test, and the comprehensiveness of the test.

❖ From the date that a sample is taken, it may take a few weeks to several months to receive the test results.
The basic DNA test kits that cost around $99.00, are taken at home and are not given by a doctor or professional. They also do not require a prescription.

This process is called Consumer testing and use your saliva to do the test.

They are not meant to diagnose any illnesses or provide any guarantees.

They are primarily for entertainment value and to learn some new information about yourself and best guesses of where your Family came from – your Ancestry.

Some of the basic testing can also tell people to which diseases they are susceptible.

Any health reports you receive in your DNA analysis are simply suggestions to improve your health.

A dietician or nutritionist could have made most of these 'actionable' suggestions even without seeing your DNA.

To review the “Top 23” companies who are providing the different types of genetic testing, go to https://medicalfuturist.com/top-companies-genomics/
The basic testing companies are supposed to make it clear that their health test is not meant to be diagnostic. Investigating your genetic health is only intended to provide general information to inform future health decisions.

For specifics and guarantees, you’ll need to talk to a healthcare professional.

**Ancestry.com costs $99.00** – mostly used for ancestry information

**Ancestry.com Health costs $179.00** and does require a doctor’s prescription

**23 and Me costs $199.00** - used for ancestry information and some medical advice

It is the only consumer test to also offer **BRCA1 and BRCA2 screening for breast cancer risk.**

**CRI Genetics costs $199.00** and claim that they provide you with the items below:
The Medical Genomics Laboratory - for common and rare genetic disorders, ranging from $250 for some targeted tests for specific, previously known gene mutations in at-risk family members to $3,350 or more for some types of comprehensive testing.

THE LABORATORY FOR MOLECULAR MEDICINE AT PARTNERS HEALTHCARE CHARGES:

- $400 for a targeted test for a familial known variant
- $500-$1,100 for certain types of genetic alterations linked to various types of cancer
- $400-$3,700 for testing for certain types of genetic alterations related to cardio-myopathy
- $600-$1,300 for tests for certain genetic alterations related to congenital heart disease or defects
- $550-$1,600 for testing for genetic alterations related to connective tissue disorders
- $250-$3,800 for testing for genetic alterations related to hearing loss
- $1,650 for testing for genetic alterations related to cystic fibrosis.
SERVICES OFFERED BY THE VERITAS COMPANY

As an example of one of the screening companies, they offer 3 levels of testing and other services.

1. *myGenome Standard*

   It was built to cover the 59 gene variants listed in ACMG 59 which are associated with highly actionable conditions – that can have immediate, and often dire, health consequences.

   It also includes the other items shown to the right.

Current cost is $599.00!
2) To develop the **Premium** items, they started with all of the conditions included in the ACMG standard, and then traced those back to ALL of the associated genes.

That resulted in a list of more than 400 genes related to over 20 highly actionable conditions, plus 125 genes associated with more than 200 carrier conditions.

This new level of testing and interpretation gives consumers an option to delve deeper into their risks for cancer, heart disease, and conditions that could affect their children.

**Current cost is $1,599.00 !**
**MYGENOME DIAGNOSTIC**

Even if other genetic tests have shown a negative result, with myGenome Diagnostic, individuals may learn about genetic variants that explain signs and symptoms of their condition.

Results can have implications for treatment decisions and outcomes, medical management options, and even for other family members.

This level of screening is available for adults ages 18 and up, as well as for children (with parental or guardian consent).

**Current cost is $2,599.00 to $3,599.00 !!**

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<th>PREMIUM</th>
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<td>ACMG 59</td>
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You Want to be Proactive about Your Health

- Expanded ACMG
- Focus on cancer and cardiovascular

You Have a Complex Medical History

- Insights on the relevant condition
- Clinical expert panel review
- Diagnostic interpretation based on clinical signs and symptoms
ADDITIONAL COSTS:

For more detailed testing, a genetic consultation is recommended to help a patient understand and make decisions about genetic testing. This typically costs about $150 per hour, and the length of time needed depends on the complexity of the situation.

Detailed Genetic testing can take more than a month in some cases. Some laboratories provide rush service for an added fee of $500 or more.

DISCOUNTS:

Many doctors and hospitals give discounts of up to 30% or more to uninsured/cash-paying patients. For example, Washington Hospital Healthcare System in California offers a 35% discount.

SHOPPING FOR GENETIC TESTING:

A primary care physician can order a genetic test or provide a referral to a geneticist. The American Board of Medical Genetics offers a genetic clinic locator by city and state at https://clinics.acmg.net/
GENETIC DISORDERS

1) There are well over 6,000 known genetic disorders, and new genetic disorders are constantly being described in medical literature.

2) Around 1 in 50 people are affected by a known **single-gene disorder**

3) About 1 in 263 are affected by a **chromosomal disorder**.

4) Around 65% of people have some kind of health problem as a result of **congenital genetic mutations**.
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<th>Genetic Disorders</th>
<th>Chromosomal Disorders</th>
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<tr>
<td><strong>DEFINITION</strong></td>
<td>Diseases caused due to the changes occur in the genetic material of an organism.</td>
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<tr>
<td></td>
<td>A type of genetic disorders that occur due to the changes of structure and number of the chromosomes.</td>
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<tr>
<td><strong>TYPES</strong></td>
<td>Single gene mutations, chromosomal disorders and complex disorders.</td>
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<td></td>
<td>Structural abnormalities and numeral abnormalities.</td>
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<tr>
<td><strong>DISEASES</strong></td>
<td>Sickle cell anemia, cystic fibrosis, etc.</td>
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<td>Down syndrome, Klinefelter’s syndrome, Turner’s syndrome, etc.</td>
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How do you determine that you are “related” to someone else?

Once your DNA is analyzed, it is compared to everyone else in the database.

Segments of DNA that match someone else’s DNA are identified and measured.

These segment lengths are measured in centimorgans (cM).

The longer the segment, the more likely that you share an ancestor with the match.

Some programs enable you to graphically visualize your matches.
WHAT ARE THE BENEFITS OF DNA TESTING / GENETIC TESTING?

- Confirm or exclude the suspected diagnosis of a genetic medical condition or illness you may have
- Identify whether you could be a carrier and at high risk of transmitting a genetic medical condition or illness to your future child or children
- Establish whether you are biologically related to other people
- Determine whether someone is a suitable match to you for organ or tissue transplantation
- Make lifestyle and dietary changes for a healthier life
- Obtain better clinical care which could potentially delay or prevent the onset of some genetic medical conditions and could potentially lead to living a healthier and longer life
Genetic Profiling or Sequencing

- Take a sample of cells (blood, hair root)
- Extract the DNA from cells
- Cut up the DNA
- Separate the DNA fragments
- Analyse the DNA fragments

https://www.youtube.com/watch?v=2JUu1WqidC4 2.2 minutes
The output from an automated DNA sequencing machine used by the Human Genome Project to determine the complete human DNA sequence.
Direct DNA studies look directly at the gene in question for an error. This technology is called FISH or PCR.

Fluorescence in situ hybridization (FISH) provides researchers with a way to visualize and map the genetic material in an individual's cells, including specific genes or portions of genes.

This may be used for understanding a variety of chromosomal abnormalities and other genetic mutations.

Different types of errors or mutations are found in different disorders.
PCR STANDS FOR THE POLYMERASE CHAIN REACTION

It is a method widely used in molecular biology to rapidly make millions to billions of copies of a specific DNA sample, allowing scientists to take a very small sample of DNA and amplify it to a large enough amount to study in detail.

It was invented in 1983 by Kary Mullis.

1.5 minute video about the PCR process

https://www.youtube.com/watch?v=2KoLnIwoZKU
KEY ISSUES TO CONSIDER FOR GENETIC TESTING

• Can we claim confidentiality over our genetic information?

• What personal consequences does genetic information have?

• What implications does it have on family members?

• Who should have access to the information?
  • Employers?
  • Insurance companies?
  • Government?

• The physical risks of testing are almost non-existent. But there are emotional and practical risks, including:

  • Feeling a high level of anxiety while awaiting results

  • The possibility of losing life insurance coverage if a result is positive

  • The possibility a test result may impact your job if you work for a company with 14 or fewer employees
• Should over-the-counter genetic tests be available?

• Should there be more regulation?

• Are genes patentable?

• Are we perusing eugenics? (eugenics: ‘well born’)

• Is health strictly a matter of biology?

• Is it a burden or a relief for doctors/parents to learn about genetic traits that do not have any treatment?
• Does genetic testing lead to labelling of people as ‘defective’?

• Can genetic testing lead to discrimination?

• How much do we know about what is and isn’t genetic?

• Behavior genetics: what people do or what people are?

• Scientific discoveries are exciting but they carry with them a responsibility to use the knowledge with wisdom
# Privacy Best Practices for Consumer Genetic Testing Services

**Promotes Transparency**

Consumers are provided:
- A high level overview of key privacy practices and detailed explanation of how Genetic Data is collected, used, and shared
- Educational resources about the basics, benefits, and risks of genetic testing
- Annual transparency reports describing law enforcement requests

**Provides Choices**

Consumers can:
- Give express consent for the collection and use of Genetic Data
- Give informed consent for research
- Access, correct, and delete their Genetic Data
- Request destruction of biological samples

**Enhances Protections**

Companies will:
- NOT share Genetic Data with employers, insurance companies, and educational institutions without consent
- Require valid legal process for disclosing Genetic Data to law enforcement
- Employ strong data security practices and privacy by design

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DRAWBACKS:

Genetic testing generally has a low risk of negatively impacting your physical health.

It can be difficult financially or emotionally to find out your results.

**Emotional:** Learning that you or someone in your family has or is at risk for a disease can be scary. Some people can also feel guilty, angry, anxious, or depressed when they find out their results.

**Financial:** Genetic testing can cost anywhere from less than $100 to more than $2,000. Health insurance companies may cover part or all of the cost of testing.
Once you unfold one of these things, it's never the same.

Map of the Human Genome

Ethical Questions

Medical Dilemmas

Legal Tangles

Privacy Concerns

Moral Issues

Insurance Applications
THE END