

# WHAT IS GENETIC COUNSELLING AND WHY IS IT IMPORTANT?

MICHELLE STEWART  
GENETIC COUNSELLOR  
MURRUMBIDGEE LOCAL HEALTH DISTRICT

WITH SPECIAL THANKS TO SALLY BRESCIANI, THE GENE MACHINE AND JUDITH TSIPIS FOR  
SHARING THEIR SLIDES FOR THIS TALK

SPECIAL REPORT

# TIME

## Genetics THE FUTURE IS NOW

New breakthroughs can cure diseases and save lives,  
but how much should nature be engineered?



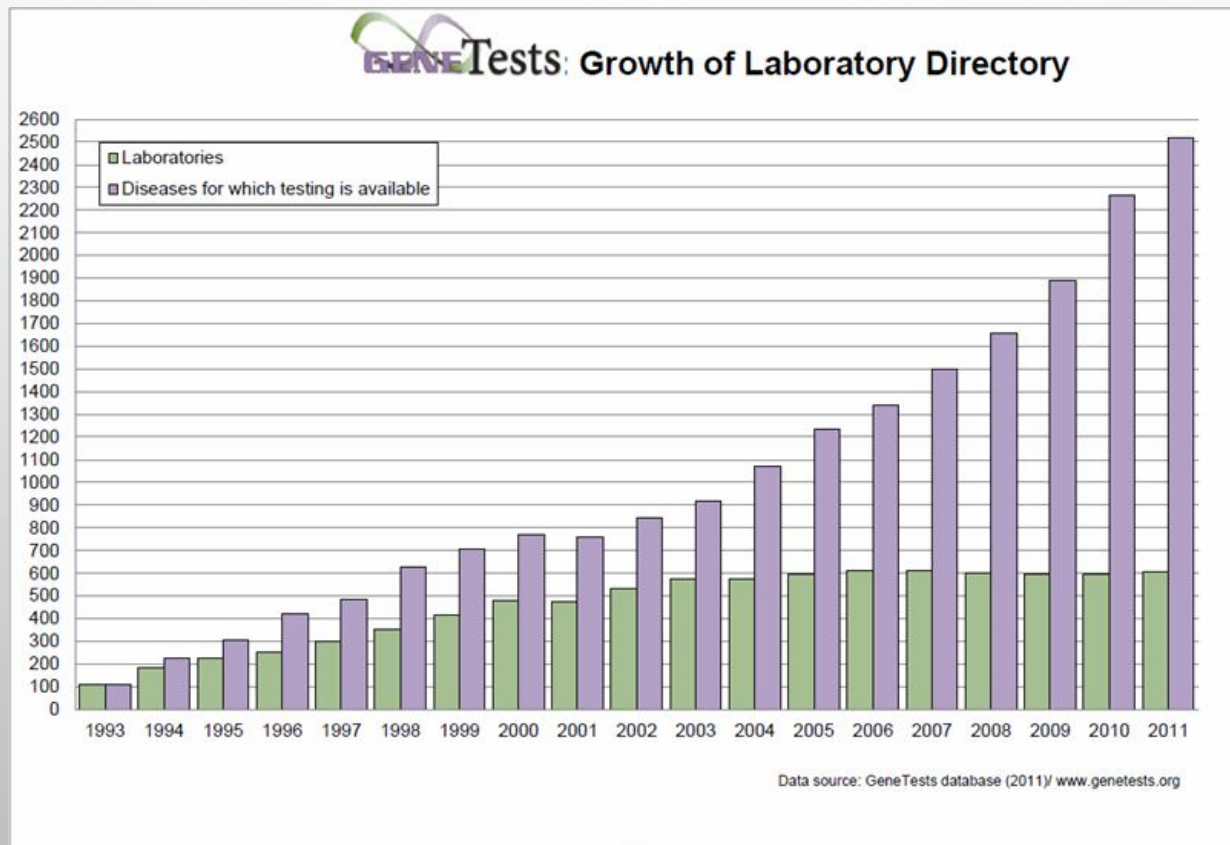
# HOW MANY GENES TO WE HAVE IN OUR GENOME?

- 20,000-25,000 GENES

## HOW MANY GENETIC DISORDERS ARISE FROM CHANGES IN ONE OR MORE OF THESE GENES?

- AS OF APRIL 4, 2018, OMIM REPORTED 3,890 GENES  
KNOWN TO CAUSE A GENETIC DISORDER

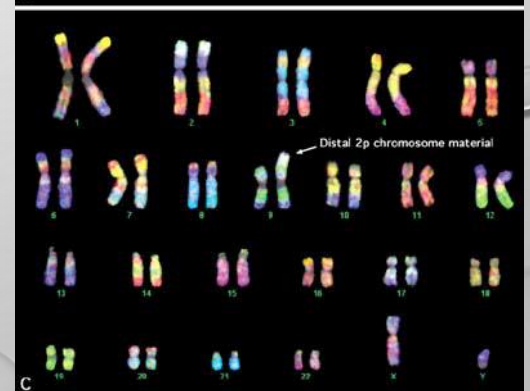
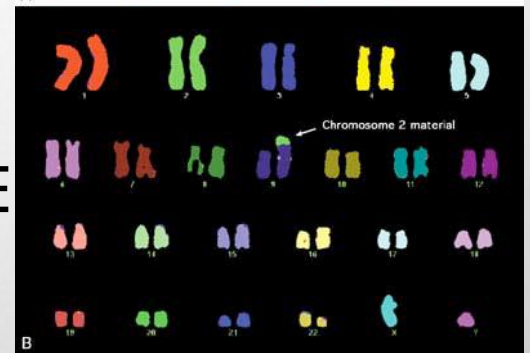
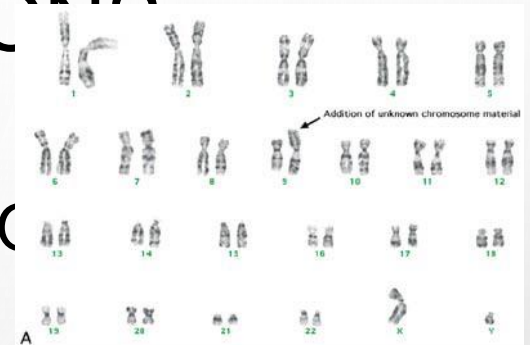
# HOW MANY GENETIC DISEASES OR DISORDERS CAN BE TESTED FOR?





# INCIDENCE/PREVALENCE OF SOME GENETIC CONDITIONS

- 0.3% OF LIVEBORNS ARE ANEUPLOID
- DOWN SYNDROME = TRISOMY 21
  - (1/600-700 LIVE BIRTHS)
- SEX CHROMOSOME ANEUPLOIDIE
  - FEMALE 47XXX 1/1000
  - MALE 47XXY 1/500-1000
  - FEMALE 45X0 1/2500



# INCIDENCE/PREVALENCE OF SOME GENETIC CONDITIONS (CONT.)

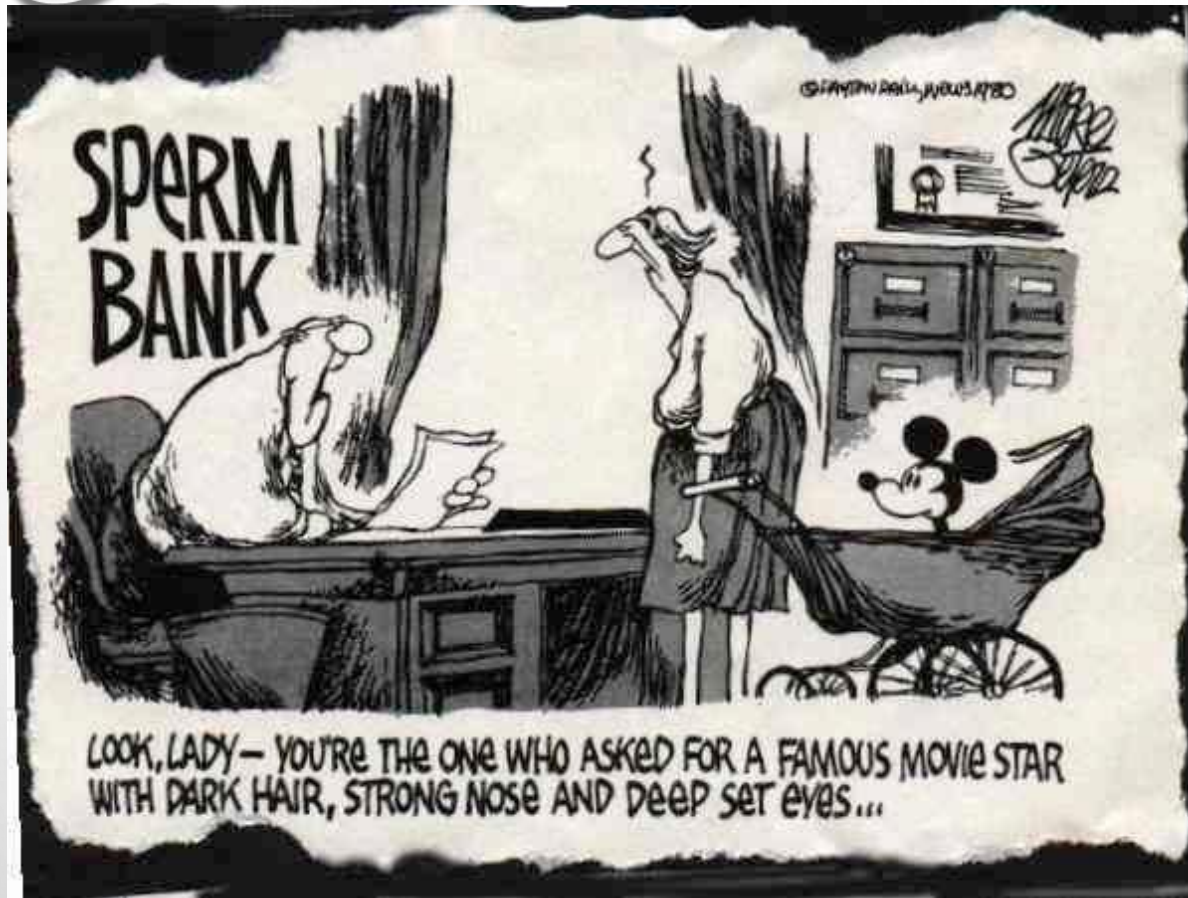
- CYSTIC FIBROSIS (1/2500 CAUCASIAN AUSTRALIANS; 1 IN 25 ARE CARRIERS)
- FRAGILE X SYNDROME (1/1,000 MALES HAVE FXS, AND 1/260 FEMALE ARE “CARRIERS” WITH 30% HAVING ID)
- SPINAL MUSCULAR ATROPHY (4/1000 BIRTHS; 1 IN 90 AUSTRALIANS OF ANGLOSAXON ANCESTRY ARE CARRIERS; 1 IN 50 OF GERMAN ANCESTRY)
- HAEMOCHROMATOSIS (1/200-400 INDIVIDUALS; 1 IN 9 AUSTRALIANS OF ANGLOSAXON ANCESTRY ARE CARRIERS)
- BREAST CANCER (1/8 WOMEN OF WHICH 5-10% OF WILL HAVE A INHERITED GENETIC PREDISPOSITION)

# OTHER STATISTICS

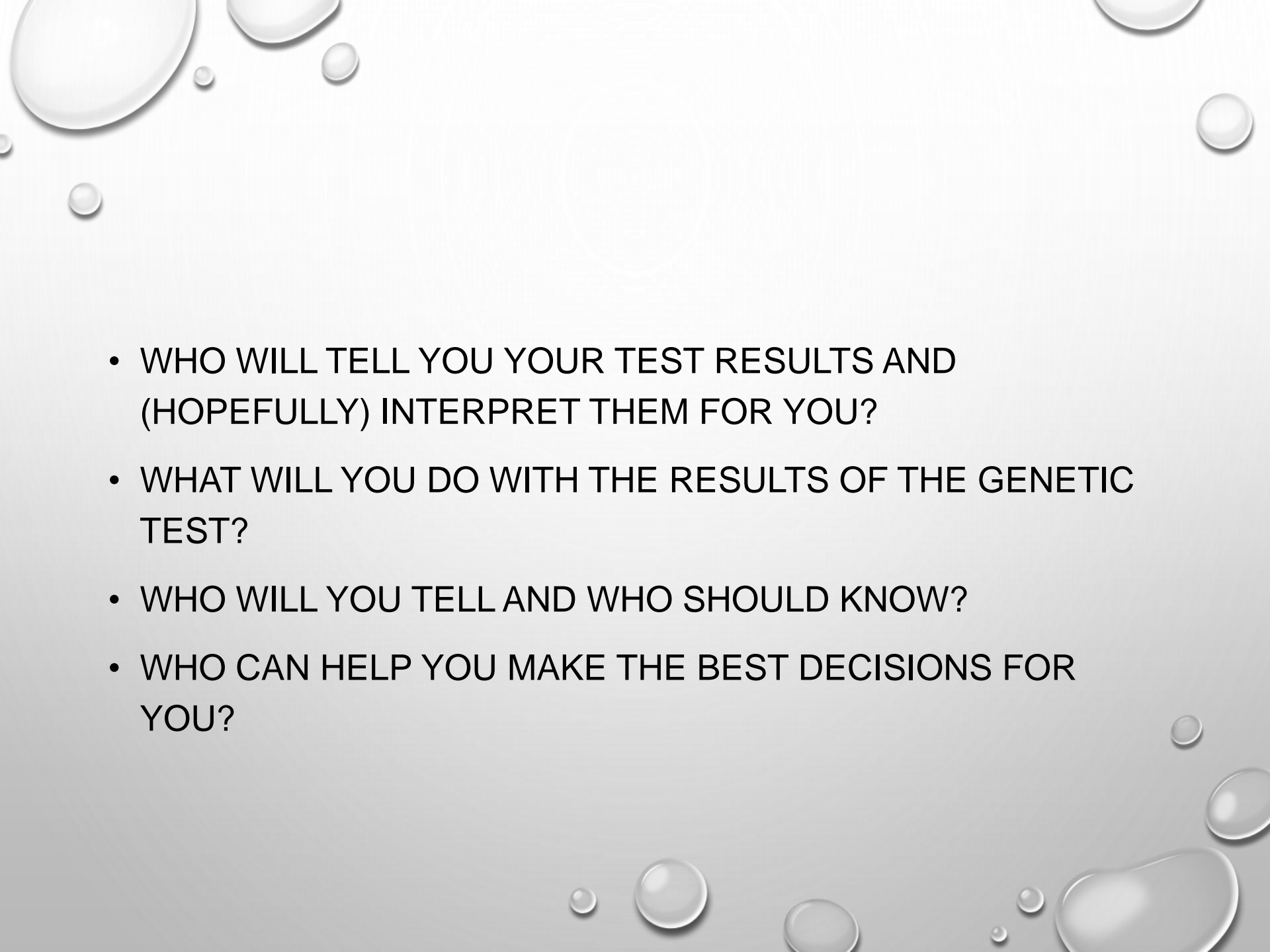
- 50% OF INTELLECTUAL DISABILITY HAS A GENETIC BASIS
- 5-10% OF ALL CANCERS ARE RELATED TO AN INHERITED SUSCEPTIBILITY GENE AND 100% OF ALL CANCERS ARE *GENETIC*
- 10% OF THE CHRONIC HEALTH CONDITIONS (INCLUDING HEART DISEASE, LATE-ONSET INSULIN DEPENDENT DIABETES AND ARTHRITIS) HAVE A GENETIC COMPONENT

# GENETIC TESTING AND GENETIC COUNSELLING

- WHY WOULD ANYONE WANT TO UNDERGO GENETIC TESTING?
- IF YOU DID WANT TO GET TESTED, HOW WOULD YOU GET TESTED? WHO WOULD PROVIDE YOU WITH INFORMATION BEFORE GETTING TESTED AND OBTAIN CONSENT?



- LOOK, LADY – YOU'RE THE ONE WHO ASKED FOR A FAMOUS MOVIE STAR WITH DARK HAIR, STRONG NOSE AND DEEP SET EYES.....

- 
- WHO WILL TELL YOU YOUR TEST RESULTS AND (HOPEFULLY) INTERPRET THEM FOR YOU?
  - WHAT WILL YOU DO WITH THE RESULTS OF THE GENETIC TEST?
  - WHO WILL YOU TELL AND WHO SHOULD KNOW?
  - WHO CAN HELP YOU MAKE THE BEST DECISIONS FOR YOU?



# GENETIC COUNSELLING

- GENETIC COUNSELLING IS THE PRACTICE OF HELPING INDIVIDUALS AND FAMILIES UNDERSTAND THE MEDICAL, PSYCHOLOGICAL, SOCIAL AND REPRODUCTIVE IMPLICATIONS OF GENETIC AND CONGENITAL CONDITIONS.

# INDICATIONS FOR GENETICS REFERRAL

- A FAMILY HISTORY OF A KNOWN GENETIC CONDITION, BIRTH DEFECT OR DEVELOPMENTAL DELAY.
- INCREASED RISK ON FIRST TRIMESTER SCREENING, NON-INVASIVE PRENATAL TEST
- ANOMALIES OBSERVED ON ULTRASOUND SCAN
- ABNORMAL CVS OR AMNIOCENTESIS RESULT
- INCREASED RISK FROM TERATOGENS
- CONSANGUINITY
- RECURRENT MISCARRIAGES
- PARENTAL BLOODS INDICATE THALASSAEMIA

# GENETIC COUNSELLING PROVIDES:

- INFORMATION AND SUPPORTIVE COUNSELLING REGARDING THE DIAGNOSIS AND RISK FOR A GENETIC CONDITION IN THE FAMILY
- DIAGNOSTIC, CARRIER, PREDICTIVE AND PRESYMPTOMATIC GENETIC TESTING WHERE APPROPRIATE (INCLUDING PRENATAL)



# ELEMENTS OF THE PRACTICE OF GENETIC COUNSELLING INCLUDE:

1. ASSESSMENT OF THE CHANCE OF RECURRENCE OR OCCURRENCE OF A CONDITION, AFTER INFORMATION GATHERING AND ESTABLISHING/VERIFYING THE DIAGNOSIS
2. EDUCATION (INFORMATION GIVING) ABOUT INHERITANCE, NATURAL HISTORY, TESTING OPTIONS, MEDICAL MANAGEMENT, PREVENTION, SOCIAL SUPPORT AND RESEARCH
3. COUNSELLING AND PSYCHOLOGICAL SUPPORT TO HELP CLIENTS ADAPT TO THEIR SITUATION AND CHOICES AND TO THE PSYCHOLOGICAL, FAMILIAL AND SOCIAL ISSUES THAT STEM FROM THE RISK OR CONDITION IN THE FAMILY

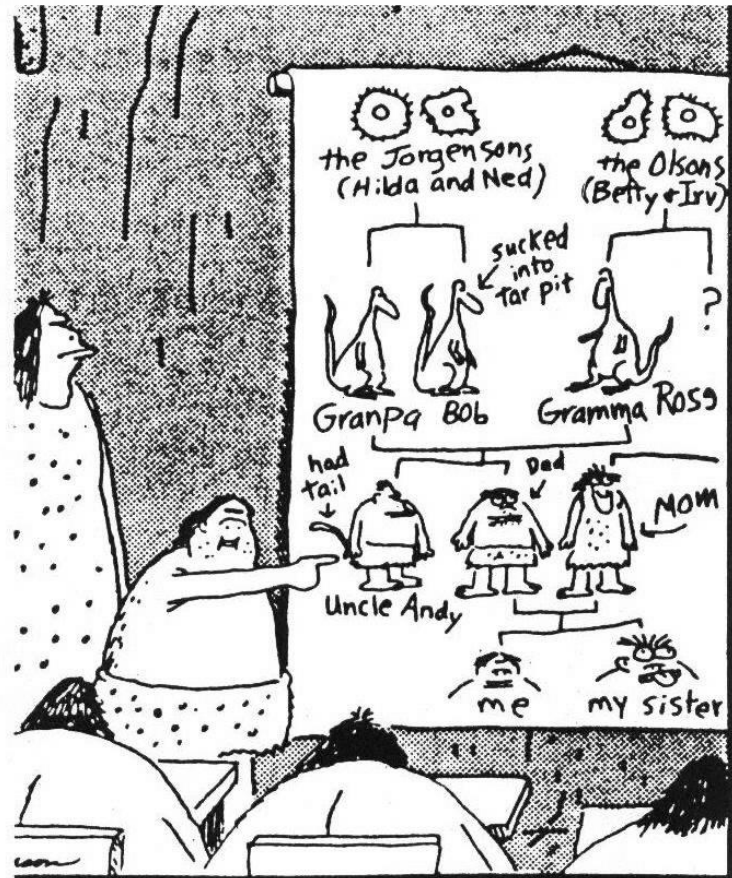
# GENETIC COUNSELLOR PROVIDERS & WORK SETTINGS

- PRENATAL CLINICS
- PAEDIATRIC CLINICS
- SPECIALTY CLINICS
- ADULT GENETIC CLINICS
- CANCER CLINICS
- GENERAL GENETICS CLINICS
- MASTERS-LEVEL GENETIC COUNSELLORS
- CLINICAL GENETICISTS
- GENETIC NURSE CLINICIANS

# INFORMATION GATHERING: MANY TYPES OF INFORMATION

- TO VERIFY/CONFIRM THE DX AND ASSESS AN ACCURATE GENETIC RISK:
  - FAMILY HISTORY/PEDIGREE
  - MEDICAL RECORDS: LAB REPORTS, PATHOLOGY REPORTS, GENETIC EXAMINATIONS
  - MEDICAL LITERATURE
  - DEATH CERTIFICATES
  - FAMILY PHOTOS





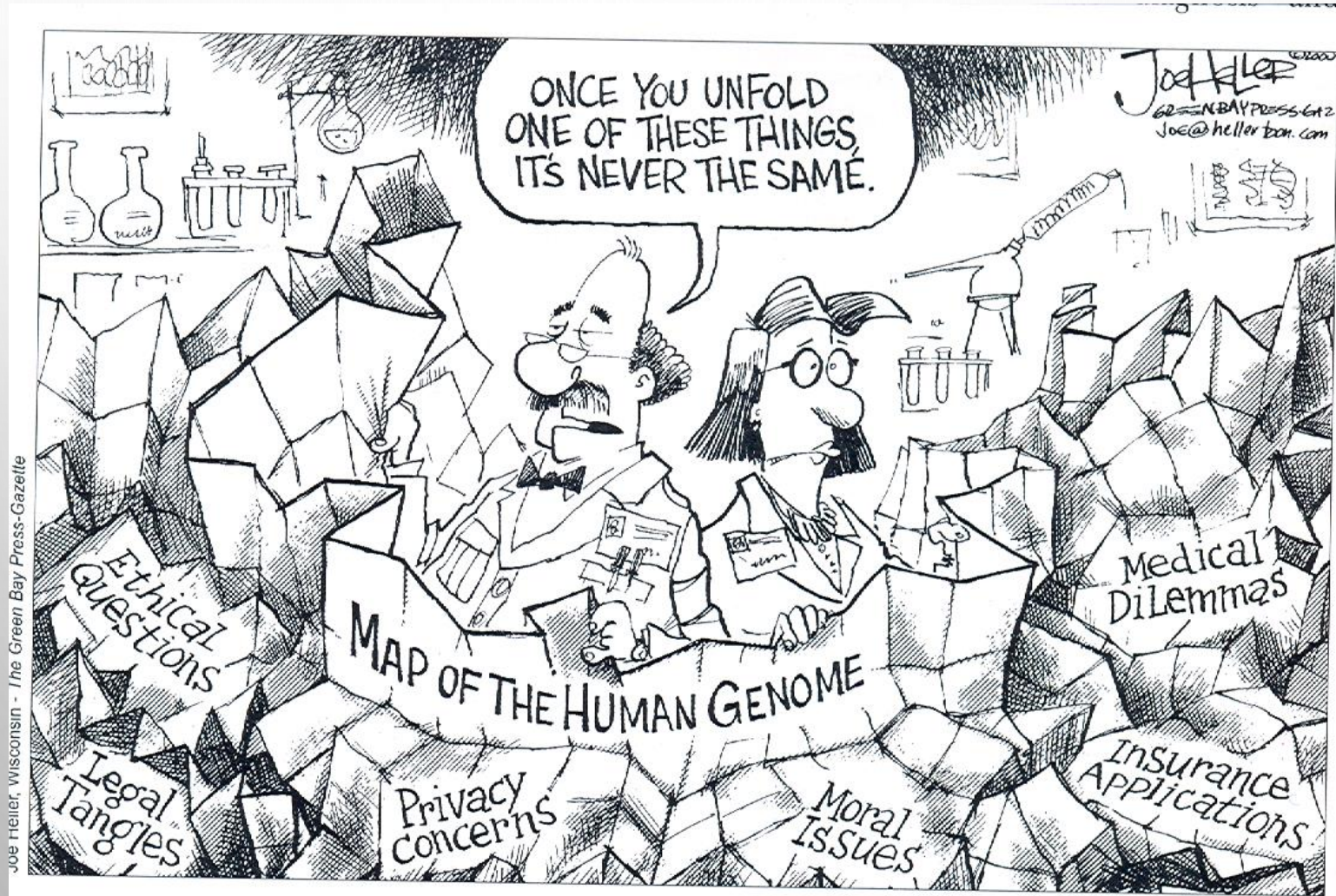
Dirk brings his family tree to class.

DIRK BRINGS HIS FAMILY TREE  
TO CLASS.


# COMMON INFORMATION OBTAINED IN A 3 GENERATION PEDIGREE

- ANY RELATIVES WHO HAVE:
  - CONGENITAL ANOMALIES (EG. CLEFT LIP, DEAFNESS, HEART DEFECT, ETC)
  - CHRONIC CONDITIONS (EG. HEART PROBLEMS, ANEURYSM, CANCER, ETC.)
  - PSYCHIATRIC DISORDERS (EG. SCHIZOPHRENIA, DEPRESSION ETC.)
  - INTELLECTUAL DISABILITY, DEVELOPMENTAL DELAY, ASD
  - DYSMORPHIC FEATURES (EG. LOW SET EARS, UNUSUAL HEAD SHAPE, SINGLE PALMER CREASE, EXTRA FINGERS OR TOES)
  - OTHER HEALTH CONCERNS
  - STILLBIRTHS, MULTIPLE MISCARRIAGES OR INFERTILITY
  - CONSANGUINITY
  - ANCESTRY/ETHNIC BACKGROUND

# BASIC GENETICS & BACKGROUND ON GENETIC TESTING



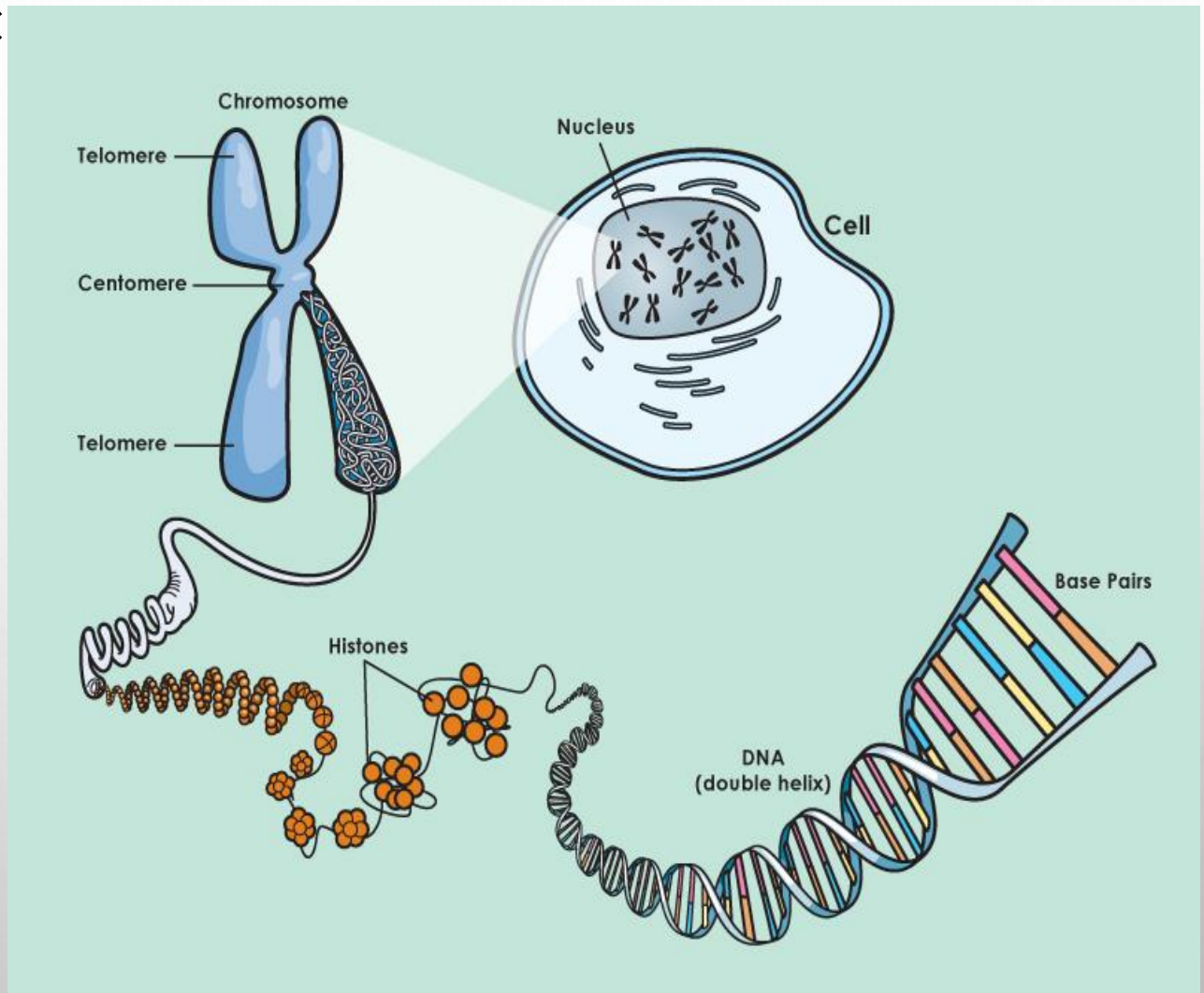


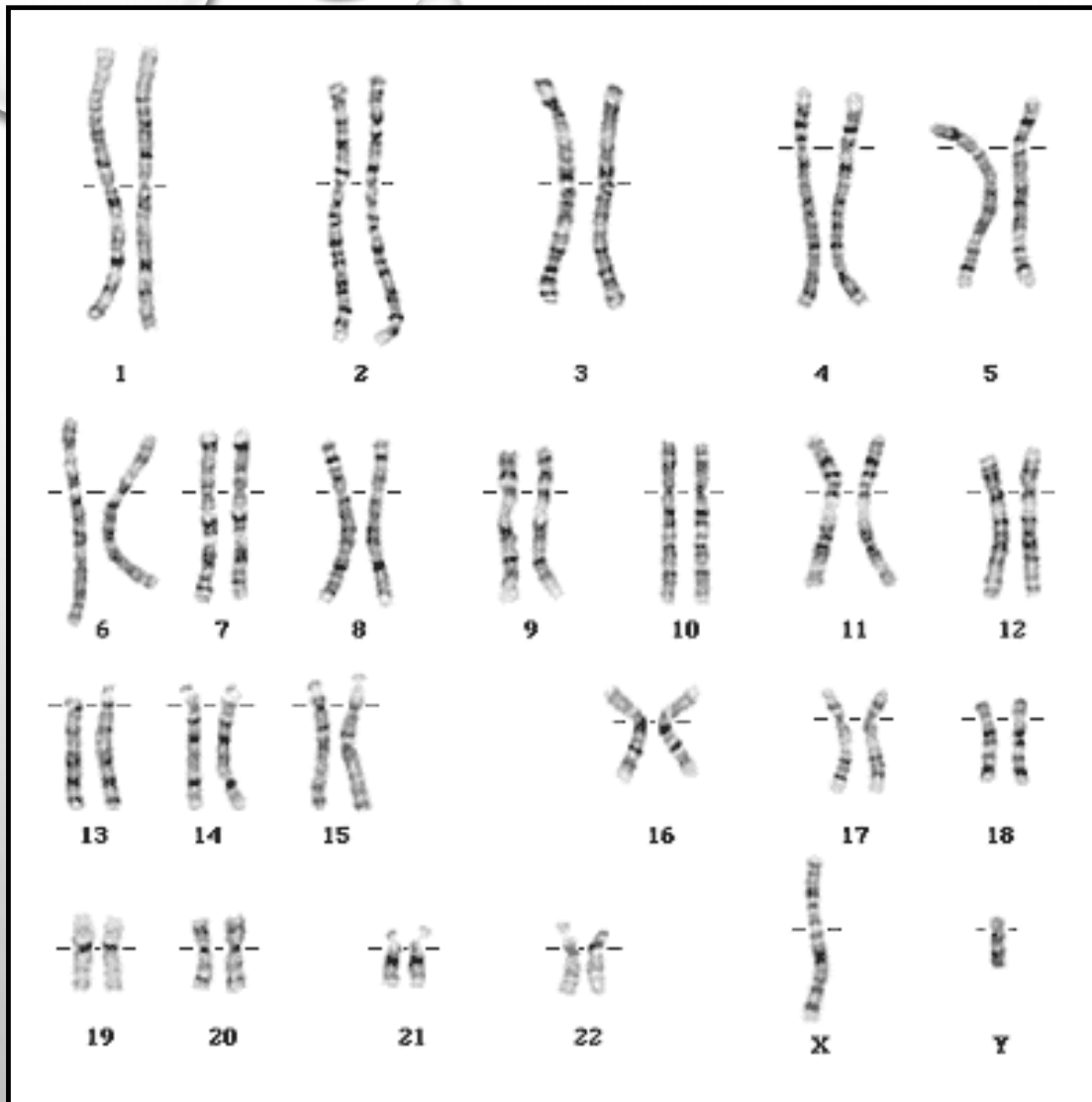
The background of the slide is a light gray gradient. In the top-left and bottom-right corners, there are several realistic-looking water droplets of various sizes, rendered with soft shadows and highlights to give them a three-dimensional appearance.

# DNA, CHROMOSOMES & GENES

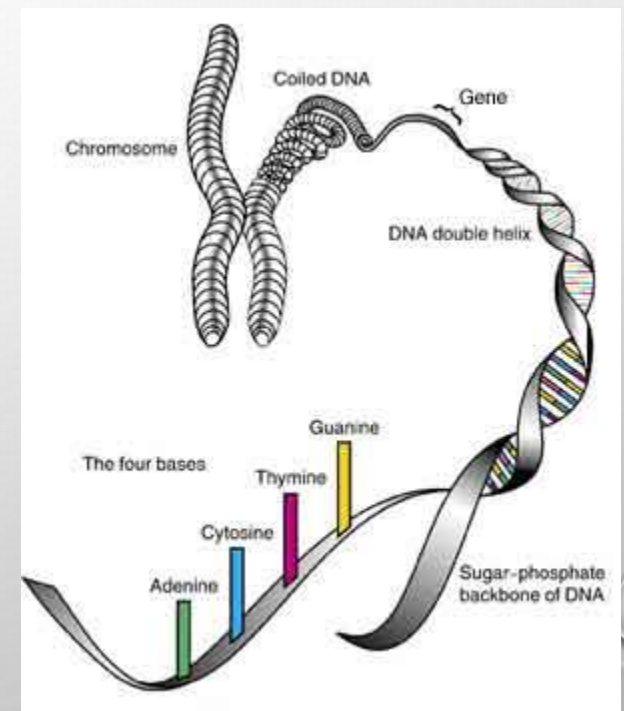
# DNA – GENETIC BLUEPRINT

- DEOXYRIBONUCLEIC ACID (DNA)
- LOCATED IN THE NUCLEUS
- RAPPED UP IN STRUCTURES CALLED CHROMOSOMES.





46 Chromosomes -23  
Pairs in every cell

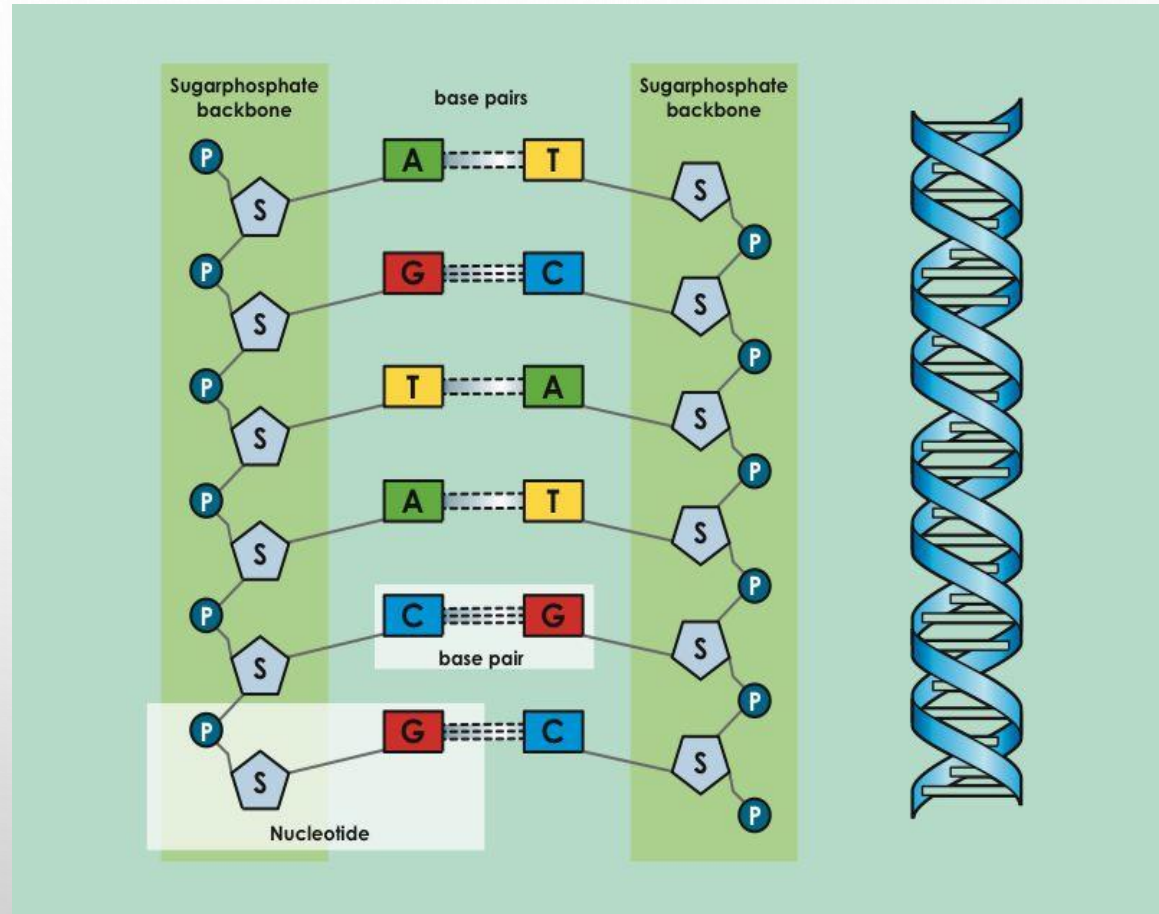


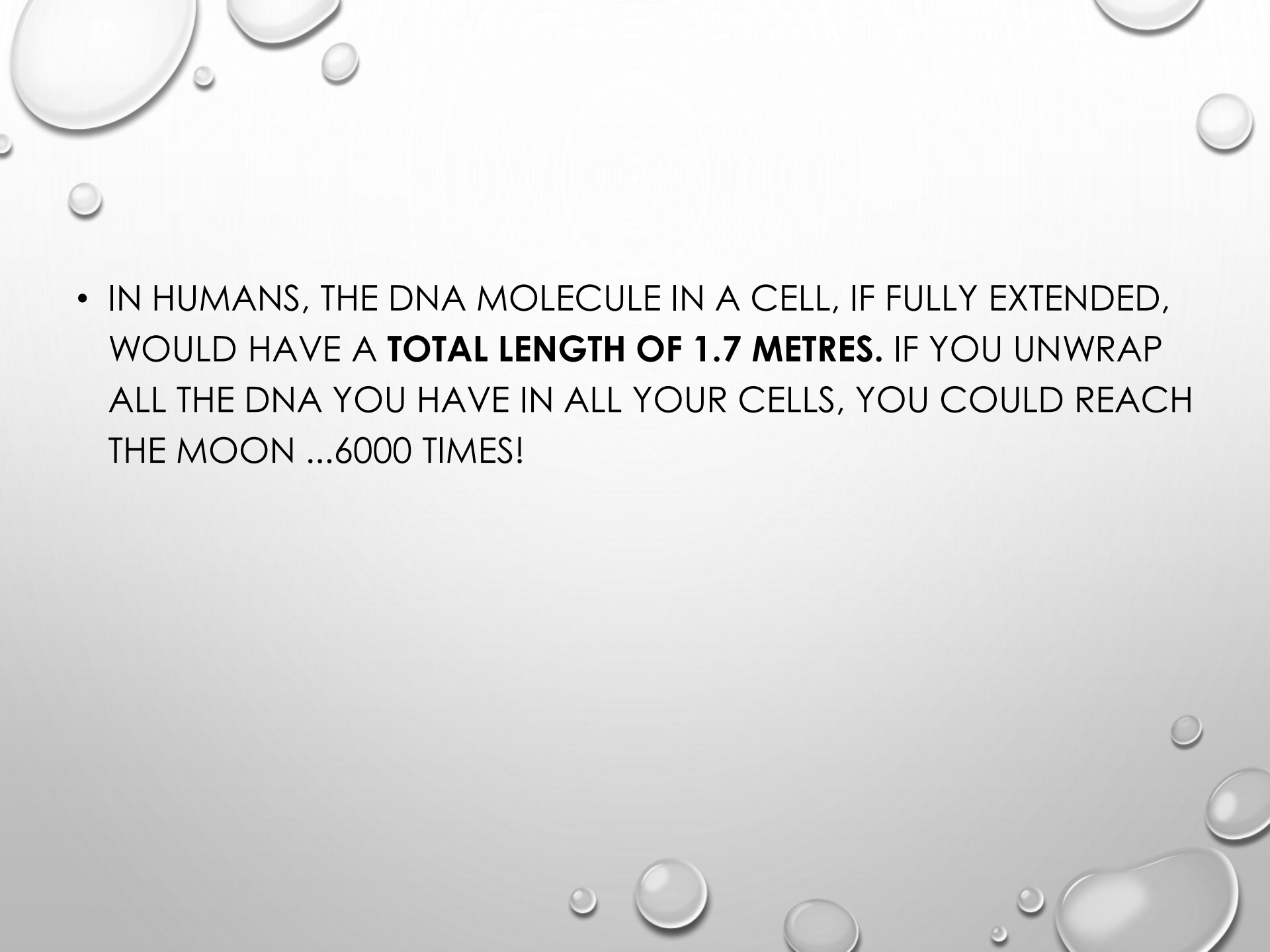
# CHROMOSOMES



# DNA IS MADE OF SEGMENTS CALLED NUCLEOTIDES

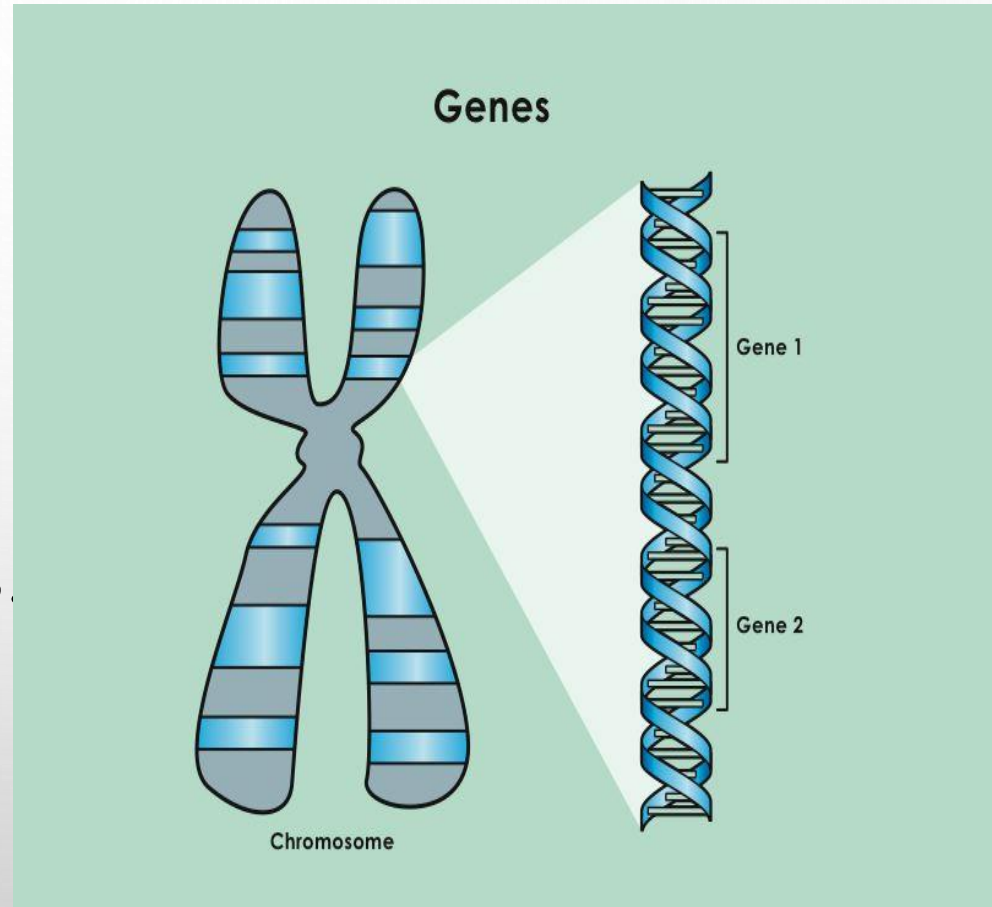
- THE BUILDING BLOCKS OF DNA ARE NUCLEOTIDES.
- THERE ARE 4 DIFFERENT NITROGEN BASES IN DNA AND THEY CAN VARY FROM ONE NUCLEOTIDE TO THE NEXT **A G T C**
- THE ALTERNATING BASES PROVIDE THE CODE



- 
- IN HUMANS, THE DNA MOLECULE IN A CELL, IF FULLY EXTENDED, WOULD HAVE A **TOTAL LENGTH OF 1.7 METRES**. IF YOU UNWRAP ALL THE DNA YOU HAVE IN ALL YOUR CELLS, YOU COULD REACH THE MOON ...6000 TIMES!

# WHAT IS A GENE?

- A PART OF THE DNA THAT CODES FOR A PROTEIN.
- NOT ALL THE DNA CODES FOR PROTEINS.
- 25,000 GENES IN THE HUMAN GENOME.



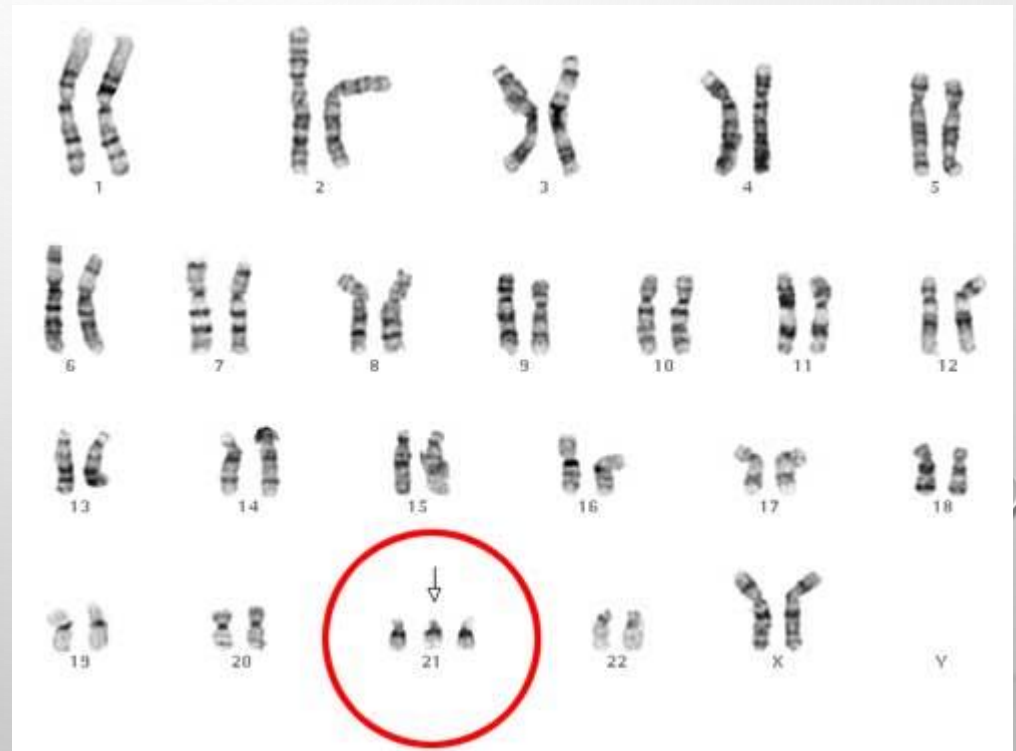
# GENETIC ALTERATIONS

- De novo
  - New change occurring at conception
- Passed from a parent
  - May be variable

# ABNORMAL NUMBER OF CHROMOSOMES

TRISOMIES - 3 COPIES RATHER THAN 2 COPIES OF A CHROMOSOME

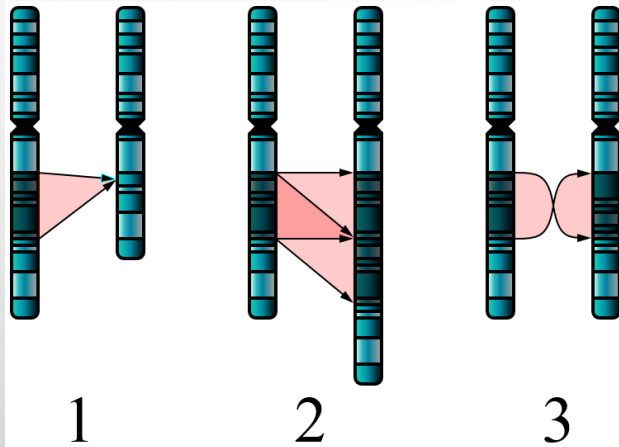
MONOSOMIES – 1 COPY RATHER THAN 2



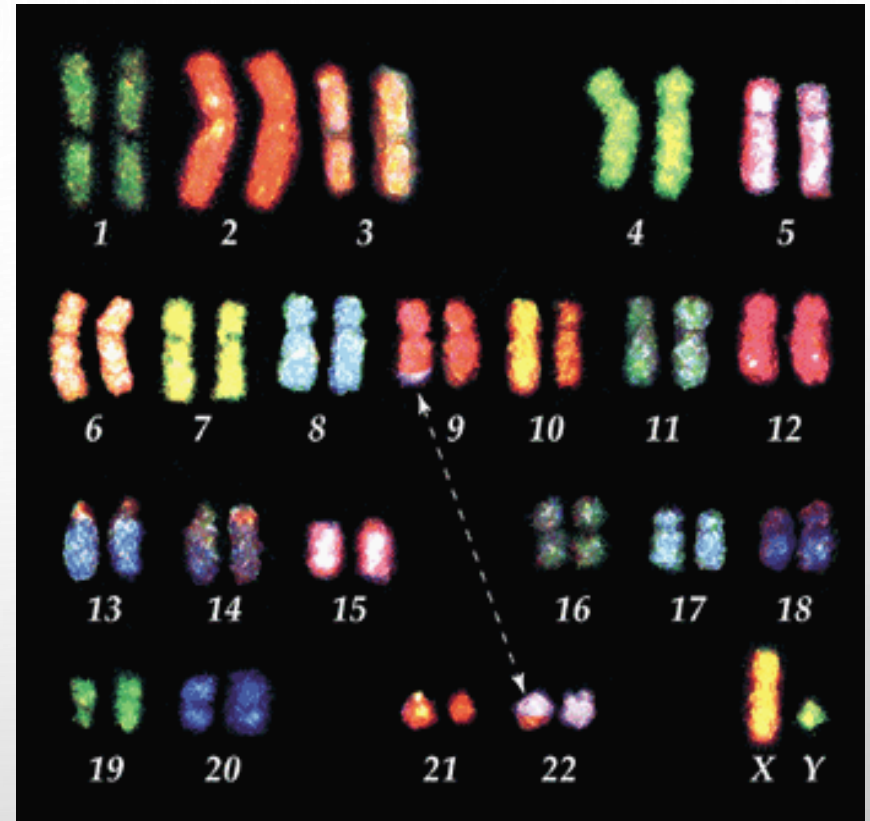


# CHROMOSOMAL ABNORMALITIES

- **DELETION:** A SECTION IS MISSING
- **DUPLICATION:** AN EXTRA SECTION OF CHROMOSOME



- **INVERSION:** A SECTION GETS SNIPPED OFF AND REINSERTED THE WRONG WAY AROUND.
- **TRANSLOCATION:** A SECTION SHIFTS FROM ONE CHROMOSOME ONTO ANOTHER
- **MOSAICISM:** THE PRESENCE OF TWO OR MORE POPULATIONS OF CELLS WITH DIFFERENT GENOTYPES IN ONE INDIVIDUAL, WHO HAS DEVELOPED FROM A SINGLE FERTILIZED EGG.



The large number of autistic individuals with unaffected family members may result from spontaneous structural variation — such as deletions, duplications or inversions in genetic material during meiosis.



# UNIQUE – UNDERSTANDING RARE CHROMOSOME DISORDERS

The image shows a screenshot of the Unique website homepage. The header features the Unique logo (a stylized DNA helix) on the left, the tagline "Understanding Rare Chromosome and Gene Disorders" in the center, and "DONATE" and "LOGIN" buttons on the right. Below the header is a dark navigation bar with links for "Families", "Professionals", "Support Us", and "Who we are", along with social media icons and a search bar. The main content area has a blue background on the left with the heading "WE ARE UNIQUE!" and a paragraph about the organization's mission. A large photo of a smiling man with tiger face paint is on the right. Below this are three yellow buttons labeled "FAMILIES", "PROFESSIONALS", and "SUPPORT US", each with a corresponding image. A white brochure titled "Trisomy 8 Mosaicism" is overlaid on the bottom right, featuring the Unique logo, the title, and photos of children. The URL "https://www.rarechromo.org/" is at the bottom left.

Unique

Understanding Rare Chromosome and Gene Disorders

DONATE LOGIN

Families | Professionals | Support Us | Who we are

Enter search text here SEARCH

## WE ARE UNIQUE!

Unique supports, informs and networks with families living with a Rare Chromosome Disorder or some Autosomal Dominant Single Gene Disorders associated with learning disability and developmental delay, among other symptoms. Sound like your family? Come and join us to help our understanding of these rare disorders. Just click on the Join Us button below

JOIN US

FAMILIES

PROFESSIONALS

SUPPORT US

Understanding chromosome disorders  
Unique

### Trisomy 8 Mosaicism

rarechromo.org

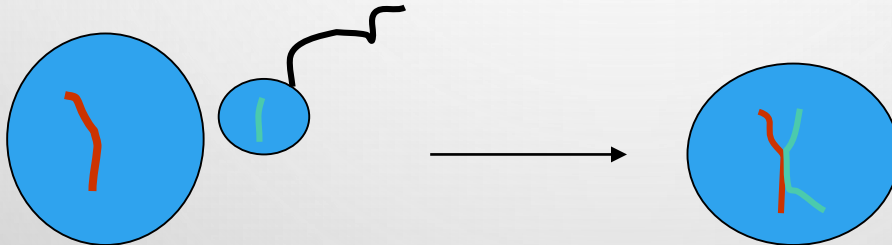
<https://www.rarechromo.org/>

# INHERITANCE

- ALL CELLS (APART FROM EGG/SPERM CELLS) HAVE 46 CHROMOSOMES (23 PAIRS).
- ONE COPY OF EACH PAIR IS INHERITED FROM THE MOTHER AND THE OTHER FROM THE FATHER.

# SEX CELLS

- SPERM AND EGG CELLS ONLY HAVE HALF THE NUMBER OF CHROMOSOMES (23)
- AT FERTILIZATION THE NUCLEUS OF A SPERM UNITES WITH THE NUCLEUS OF AN EGG TO PRODUCE A COMPLETE SET OF CHROMOSOMES (46).



# INHERITANCE

- **DOMINANT INHERITANCE**

- ONE COPY OF A GENE IS DOMINANT OVER THE OTHER

- **RECESSIVE INHERITANCE**

- A GENE IS EXPRESSED ONLY WHEN BOTH COPIES ARE THE SAME

- **X-LINKED INHERITANCE**

- A GENETIC FEATURE IS CARRIED BY THE X CHROMOSOME (FEMALES XX, MALES XY)

# SINGLE GENE DISORDERS

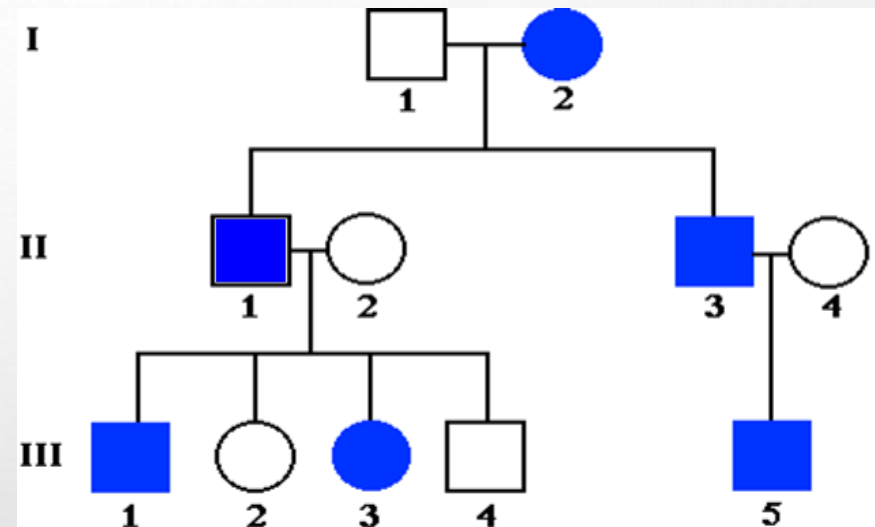
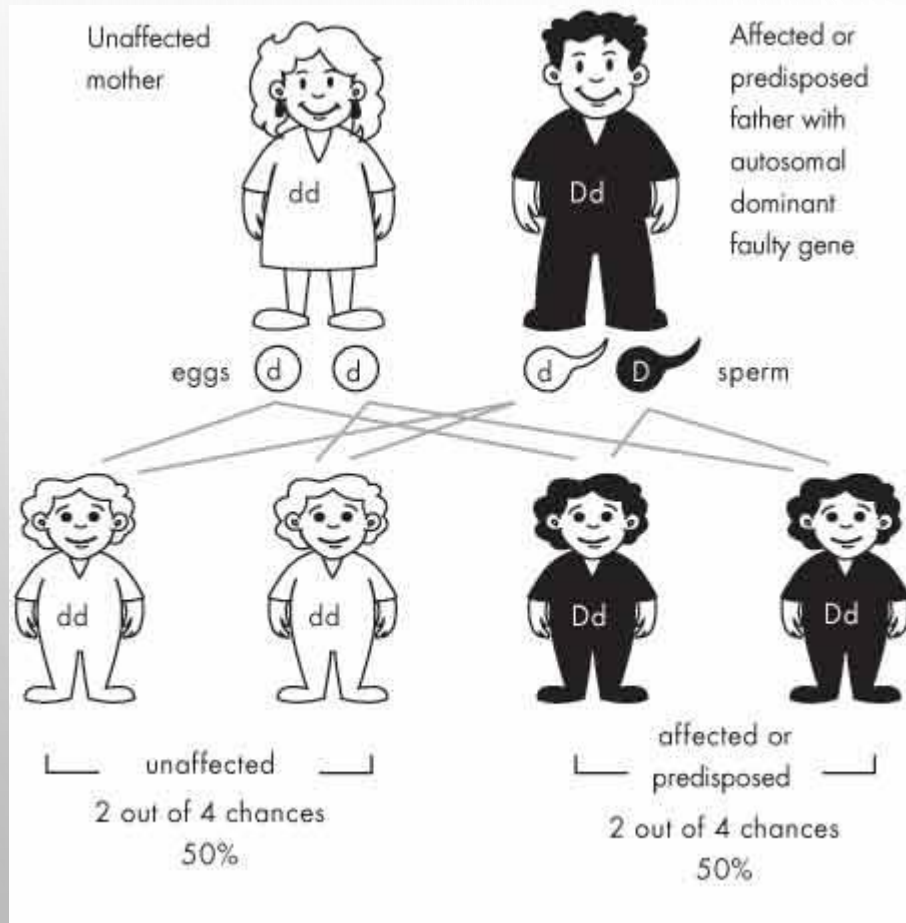
**Single gene changes:** a small nucleotide change in a segment of the DNA that codes for a gene

- AUTOSOMAL DOMINANT
  - NEW VARIANT – DE NOVO
  - FAMILIAL
- AUTOSOMAL RECESSIVE
  - CONSANGUINITY
- X-LINKED





# AUTOSOMAL DOMINANT INHERITANCE



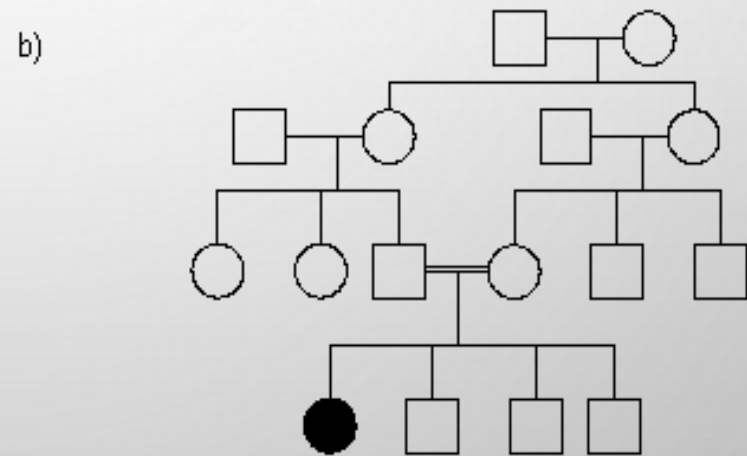
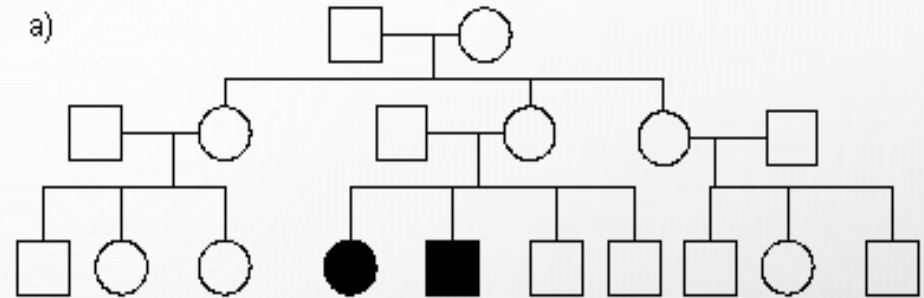
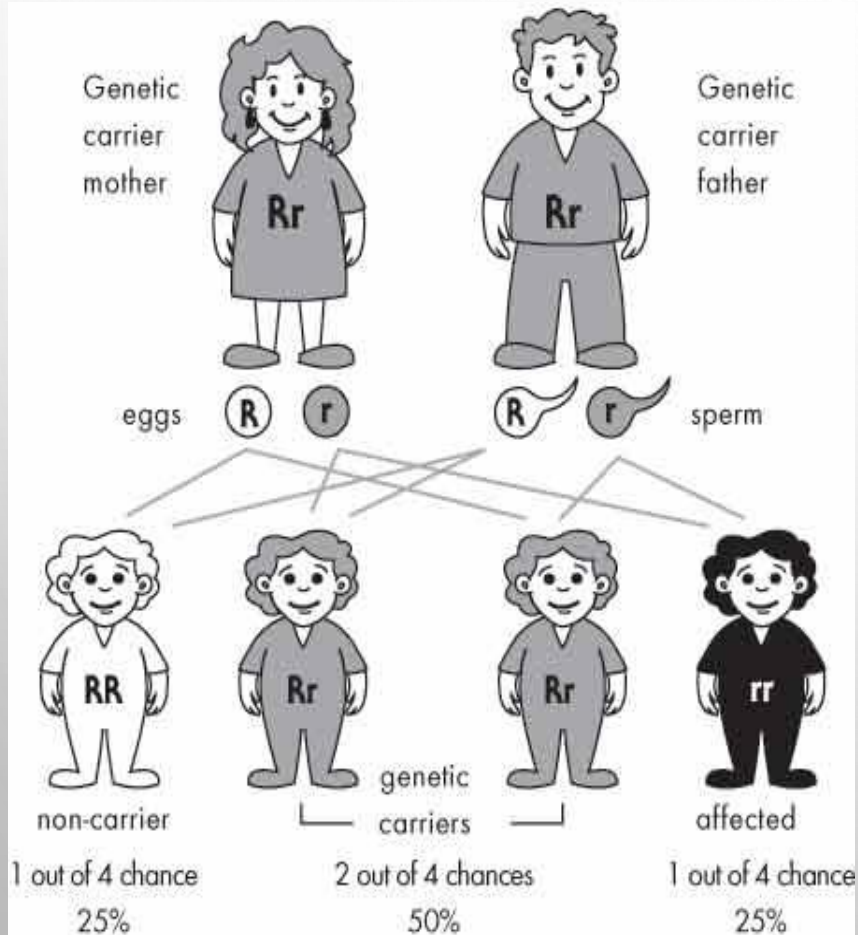
# CORNELIA DE LANGE SYNDROME

# ACHONDROPLASIA



Peter Dinklage aka Tyrion Lannister

# AUTOSOMAL RECESSIVE INHERITANCE

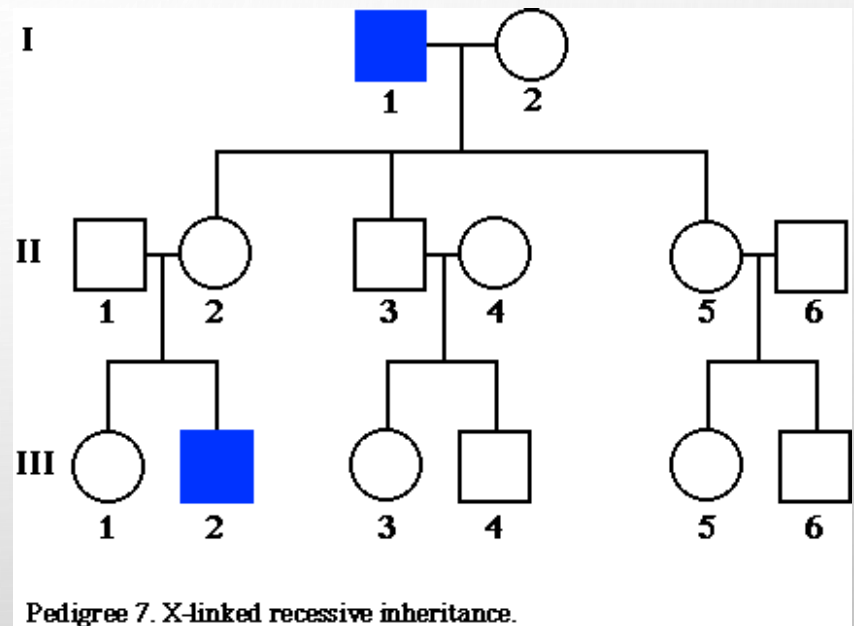
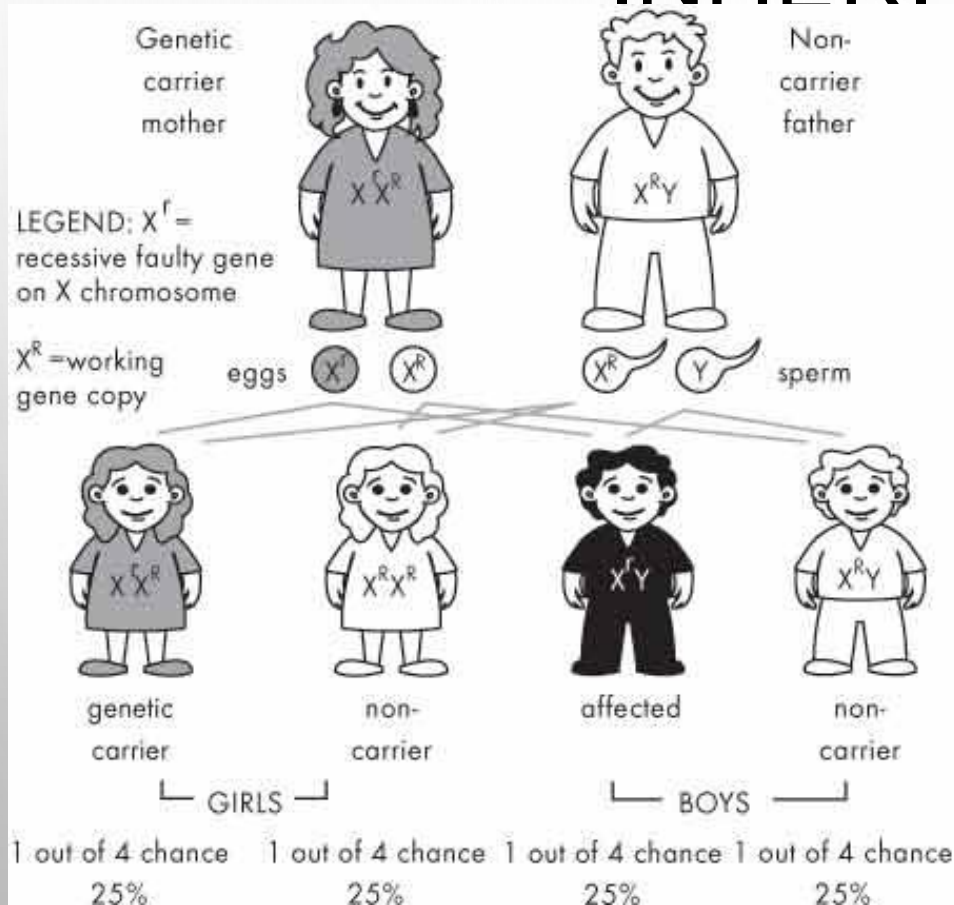




# OCULO-CUTANEOUS ALBINISM

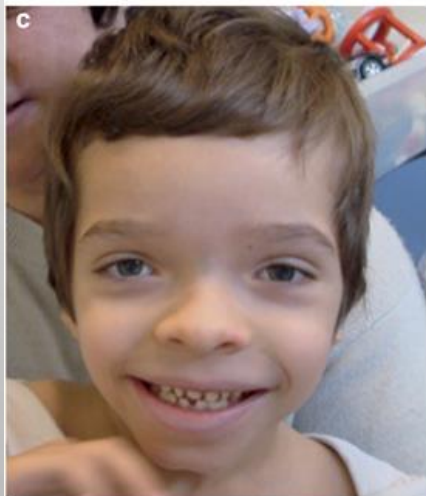


# X-LINKED RECESSIVE INHERITANCE





# COFFIN LOWRY SYNDROME

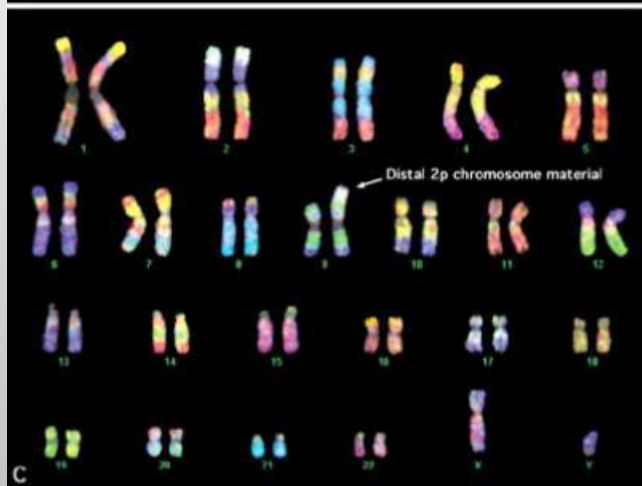
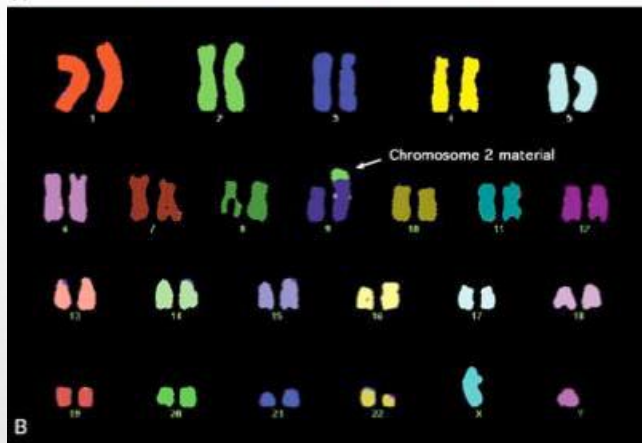
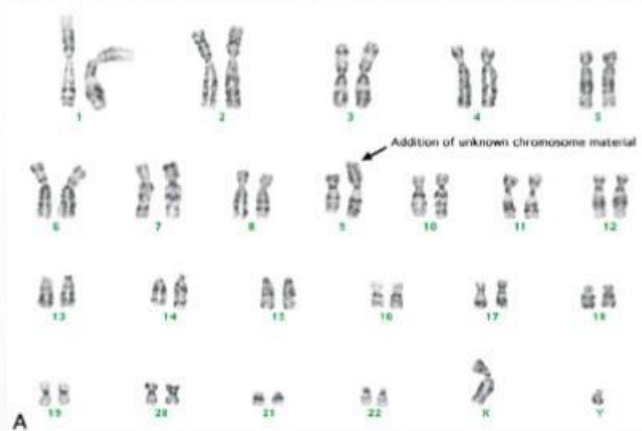


# EXAMPLES OF GENETIC CONDITIONS

- ABNORMAL NUMBER OF CHROMOSOMES
  - DOWN SYNDROME, EDWARDS SYNDROME,
- DELETION
  - CRI DU CHAT, ANGELMAN SYNDROME
- SEX CHROMOSOME ABNORMALITIES
  - TRIPLE X SYNDROME, TURNER SYNDROME, KLINEFELTER'S SYNDROME
- SINGLE GENE MUTATIONS
  - CYSTIC FIBROSIS, SICKLE CELL ANAEMIA, ANGELMAN SYNDROME, BREAST/OVARIAN CANCER SYNDROME

# GENETIC TESTING FOR SPECIFIC CONDITIONS

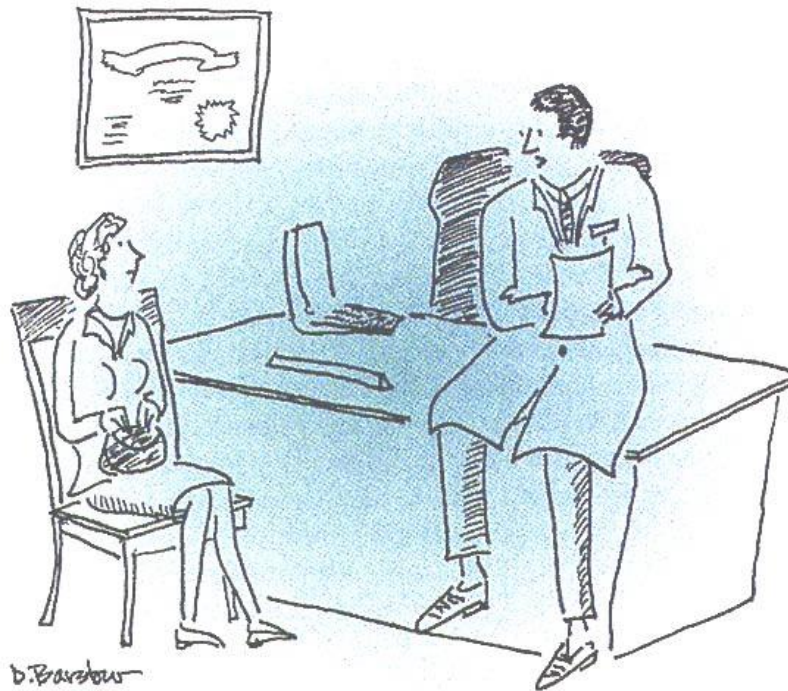
1. TAKE A SAMPLE (BLOOD/AMNIOTIC FLUID, MOUTH SWAB)
2. USE STAINING OF CHROMOSOMES TO LOCATE ANY CHROMOSOME ABNORMALITIES
3. OR USE MATCHING DNA SEQUENCES TO DETECT GENE ABNORMALITIES



# TYPES OF TESTS

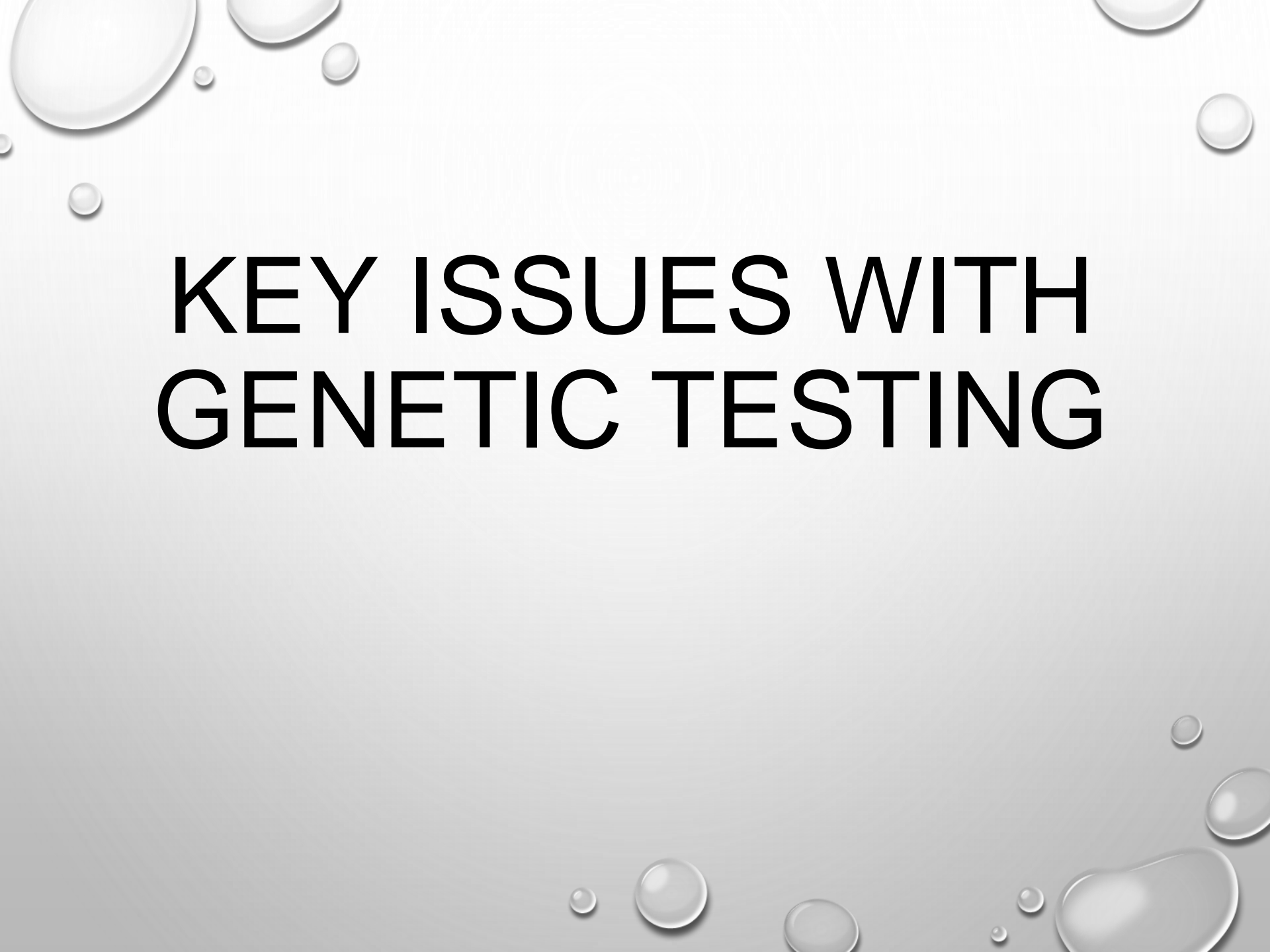
<b>Diagnostic</b>	Used to confirm a diagnosis based on physical signs <ul style="list-style-type: none"><li>•GCH microarray – chromosome deletions or duplications</li><li>•Genetic panels – single gene conditions</li><li>•Genomic testing – single gene conditions</li></ul>
<b>Predictive</b>	Used to detect gene variant associated with disorders that appear later in life <ul style="list-style-type: none"><li>•Variant must be known</li></ul>
<b>Carrier Identification</b>	Used by people with a family history of recessive genetic disorders <ul style="list-style-type: none"><li>•Variant must be known</li></ul>
<b>Prenatal</b>	Used to test a foetus when there is risk of bearing a child with mental or physical disabilities <ul style="list-style-type: none"><li>•Variant must be known</li></ul>
<b>Newborn Screening</b>	Used as a preventative health measure once the baby is born
<b>Research testing</b>	Used for finding unknown genes and identifying the function of a gene





*It's like this, Mrs. Cameron. The results are negative, but that doesn't mean not positive, exactly. Nor is it not negative, we wouldn't want a double negative there, would we ...*

It's like this, Mrs. Cameron. The results are negative, but that doesn't mean not positive, exactly. Nor it is not negative, we wouldn't want a double negative there, would we.....

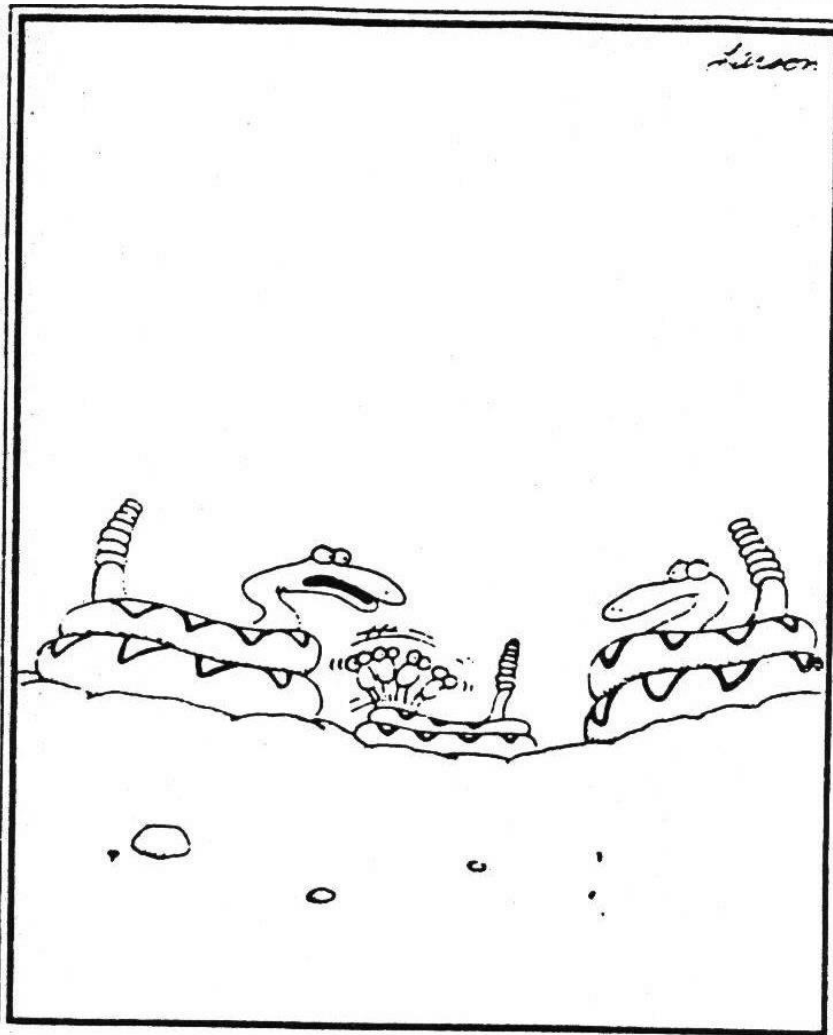
The background of the slide is a light gray gradient. It is decorated with several realistic water droplets of various sizes. Some droplets are in the top left corner, some in the top right, and a cluster of larger droplets is in the bottom right corner. The droplets have highlights and shadows, giving them a three-dimensional appearance.

# KEY ISSUES WITH GENETIC TESTING

# COUNSELLING & PSYCHOSOCIAL SUPPORT

Psychological dilemmas, emotions and reactions commonly encountered in genetic counselling:

- ANGER
- DENIAL
- DISBELIEF
- GRIEF & LOSS
- SHATTERED EXPECTATIONS OF NORMALITY
- INTELLECTUALISATION
- DISPLACEMENT (BLAME)
- ANXIETY
- GUILT
- SHAME
- FEAR
- HELPLESSNESS
- RATIONALISATION
- HOPELESSNESS
- FATALISM



"This is your side of the family, you realize."

"This is your side of the family, you realize"

# PSYCHOLOGICAL IMPLICATIONS

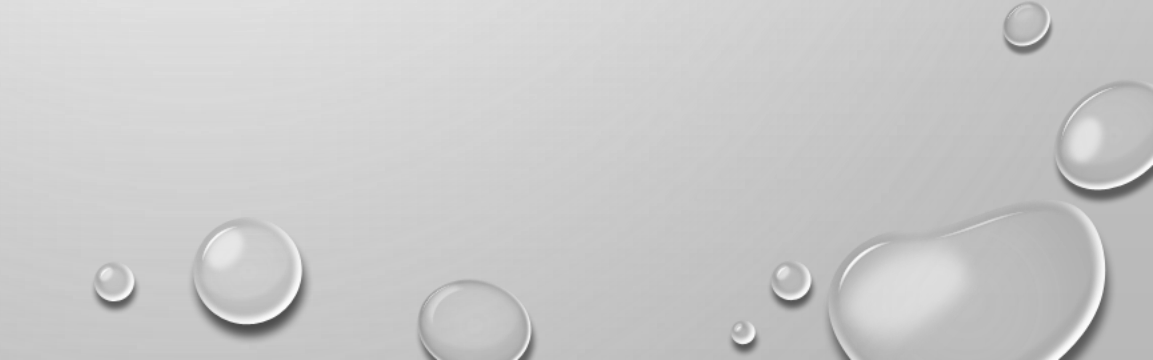


- CHANGE IN PERCEPTION OF SELF
- CHANGE IN FAMILY BELIEF SYSTEMS
- CHALLENGE TO RELIGIOUS BELIEFS
- CHANGE IN SOCIAL FUNCTIONING





# COMMON ETHICAL ISSUES FOR FAMILIES

- RIGHT TO KNOW/RIGHT NOT TO KNOW
  - SHARING OF INFORMATION
  - COERCION
  - PRIVACY
  - REPRODUCTIVE DECISION MAKING
  - TESTING OF MINORS
- 



"EUREKA! ... I'VE DISCOVERED THE GENE THAT MAKES US THINK THAT EVERYTHING'S DETERMINED BY GENES!"