

Big Trouble in Small Packages

MUTATIONS



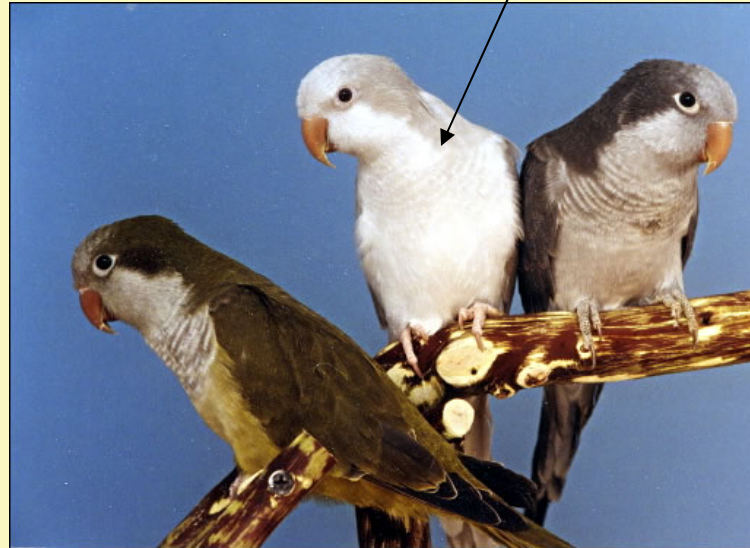
# What is a “**mutation**”?

A permanent change in the genetic code.



A “comic factor” mutant

A “grey factor” mutant



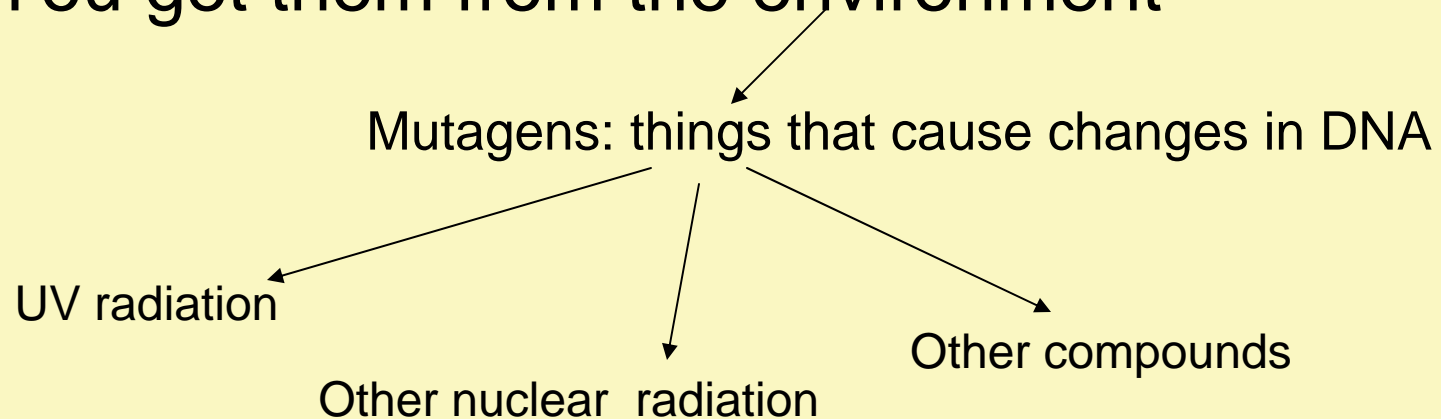
# Causing Mutations

Come from two places:

1. You inherit them (thanks mom and dad)

OR

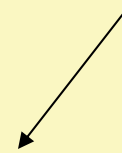
2. You get them from the environment



# Gene Mutations



Involve base-pair  
changes within a  
single gene



“What’s a gene again?”

A segment of DNA that codes  
for a polypeptide/protein

# Substitutions

These mutations involve replacing one base with another.

Only affect a single codon and, thus, a single amino acid change...maybe.

# Codons: Three-letter words

Since The DNA code is a sequence of three-base codons, we can think of a gene as a sentence made up of three-letter words

**The sun was hot but the old man did not get his hat.**

Let's make it easier to see the message

**The sun was hot but the old man did not get his hat.**

Here's a substitution.

Do you still understand some of the sentence?

**The sud was hot but the old man did not get his hat.**

Let's make it easier to see the message again.

**The sud was hot but the old man did not get his hat.**

Not sooooo bad. How 'bout with real bases?



# Silent Mutations

If the change in sequence doesn't alter the shape of the protein.

Some changes in bases occur in regions of “junk DNA” that don't code for anything,

# “Frameshift” mutations

Adding or deleting a base.

Affect all following codons.

Often have profound effects on the organism.

Lethal Mutations: Changes that are so damaging the organism cannot survive.



# Codons: Three-letter words continued

**Thesunwashotbuttheoldmandidnotgethishat.**

Let's really mess with the message

Here's a deletion.

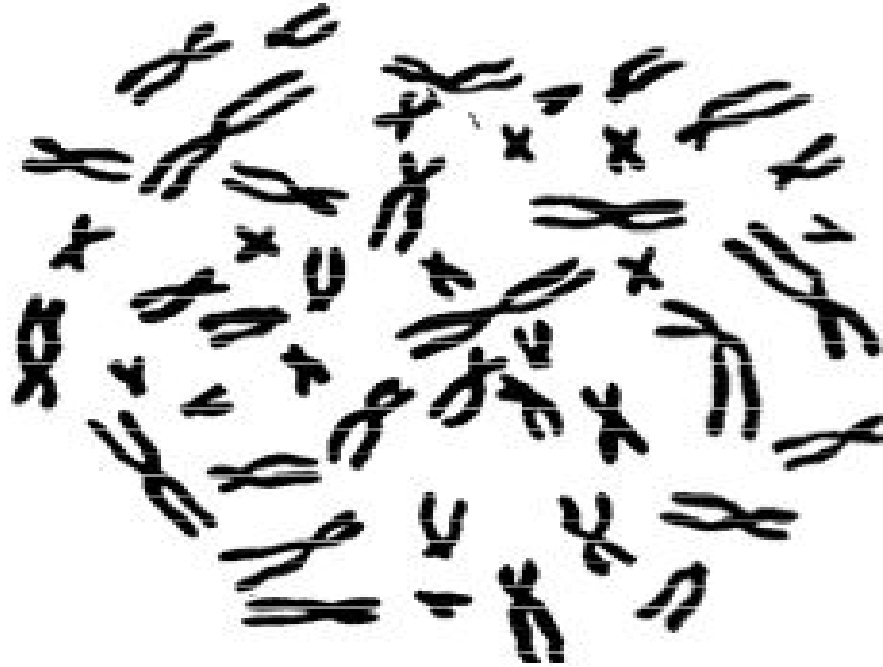
**Thesunwahotbuttheoldmandidnotgethishat.**

Now let's look at the message as three letter words.

**The sun wah otb utt heo ldm and idn otg eth ish at.**

If that makes sense to you, I cannot help you.

# Chromosomal Mutations

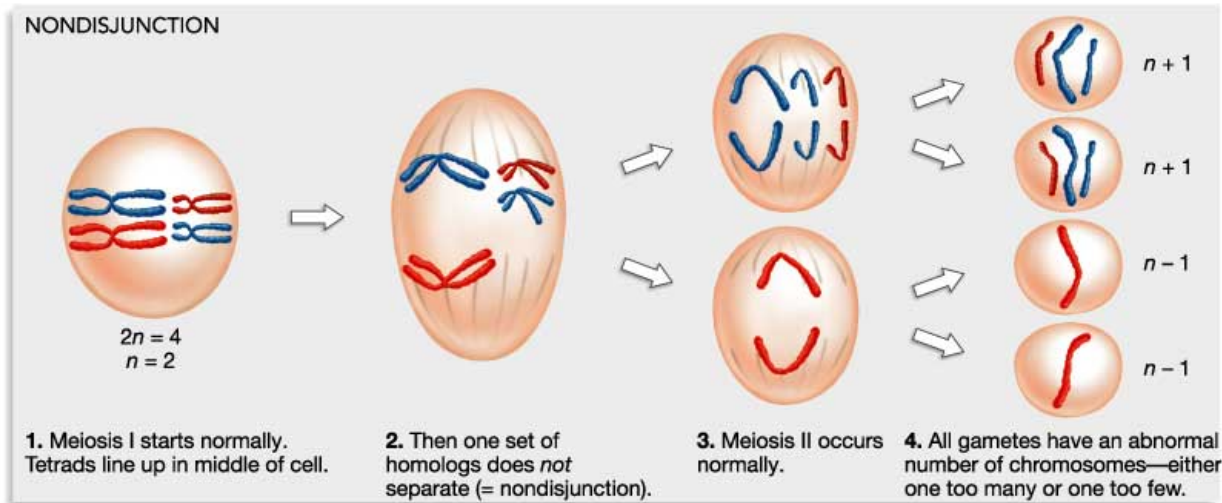


**Changes that involve whole chromosomes  
(or at least parts of 'em)**

# Chromosomal Mutations

During meiosis (gamete formation)  
homologous pairs of chromosomes fail to  
separate.

NON-DISJUNCTION



Resulting gametes  
and the zygotes  
they form will have  
the wrong number  
of chromosomes.

# Chromosomal Mutations

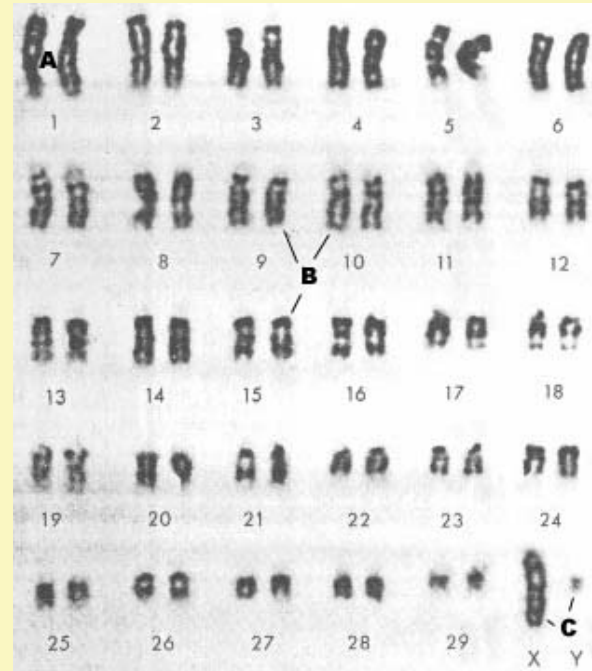
Determined in fetuses through amniocentesis and karyotyping

↓  
Withdrawing some amniotic fluid from around the fetus for analysis

↙  
Taking a look at the cell going through mitosis and taking a picture of the chromosomes. The chromosomes are counted and examined

Wait a minute.....

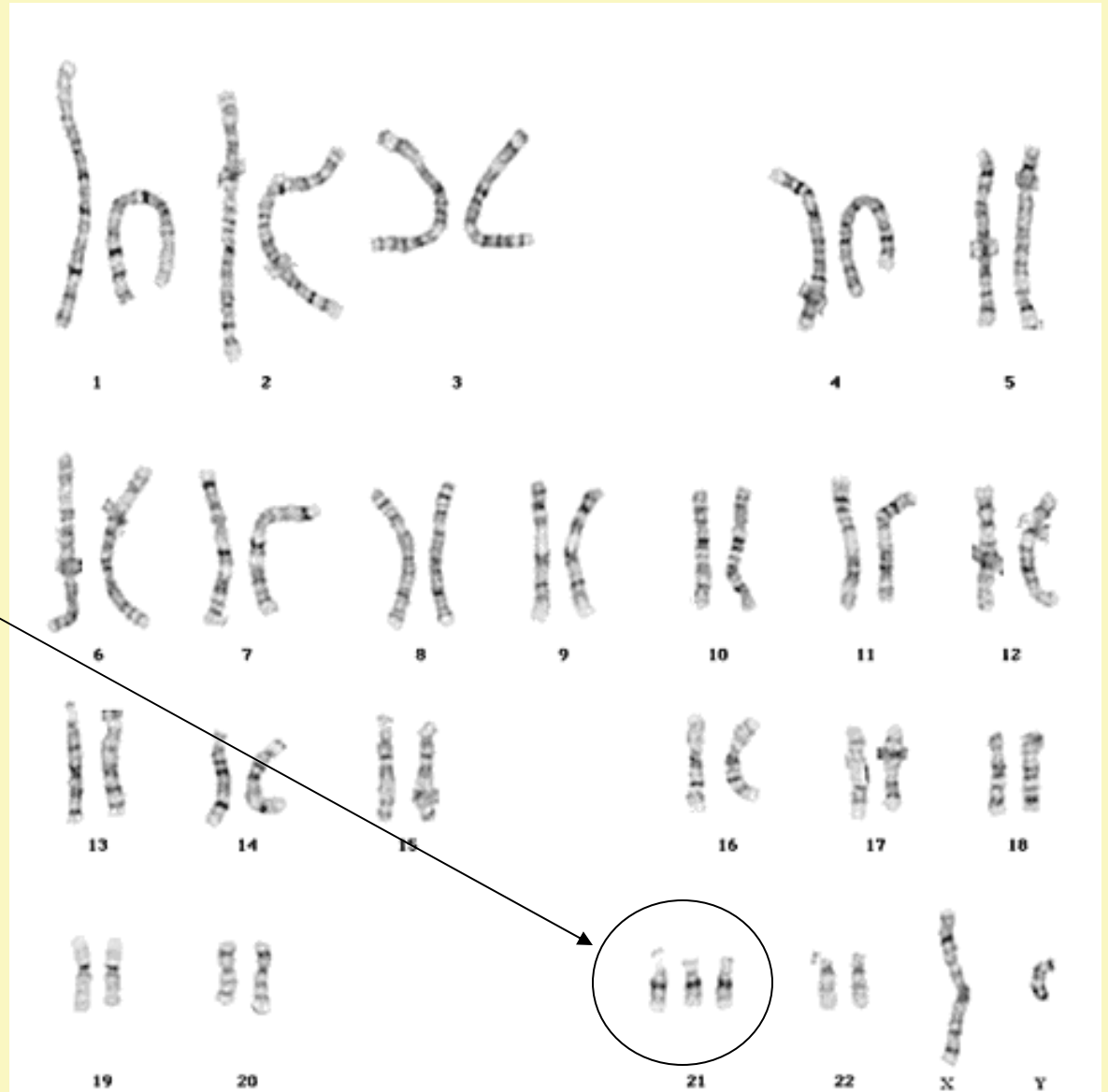
...Is this even a human karyotype?



# Chromosomal Mutations

How many chromosomes are we supposed to have?

An extra 21<sup>st</sup> chromosome!



# Chromosomal Mutations

Trisomy 21 AKA Downs Syndrome:  
An extra 21<sup>st</sup> Chromosome

Turners Syndrome::  
When a girl only has a single X chromosome



# Fixing Mutations

Mutations that are inherited from parents are in every cell of the body and therefore cannot be fixed.

They may be passed on to next generation unless repaired in the gonads.

Therapies usually supplement missing proteins (if possible).



# Fixing Mutations

Mutations that occur from exposure to mutagens often form cancers.

Wear sunscreen!!!

