Errors in Meiosis

P.161-166

Errors in Meiosis

- Often, errors in meiosis produce gametes that **do not** survive.

- Gametes that **do** survive errors in meiosis:
  - may be fertilized and produce a zygote
  - since all cells in the organism come from that one zygote, all cells will contain the error
    - E.g. all cells of a person with Down Syndrome have 47 chromosomes (instead of 46).

Errors in meiosis can result in changes to:

1. Chromosome number

(next slides)

2. Chromosome Structure

(Slide 18- 19)

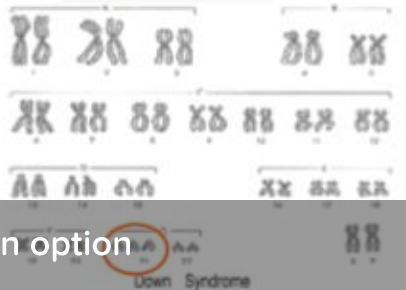
A quick overview!

ERRORS IN MEOSIS

can result in changes to

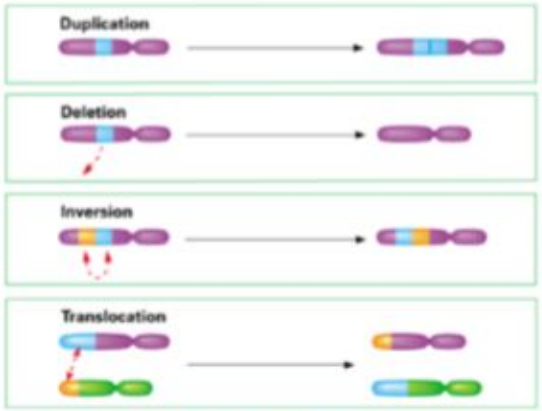
CHROMOSOME NUMBER

- Trisomy → 3 copies → too many
- Monosomy → 1 copy → too few



CHROMOSOME STRUCTURE

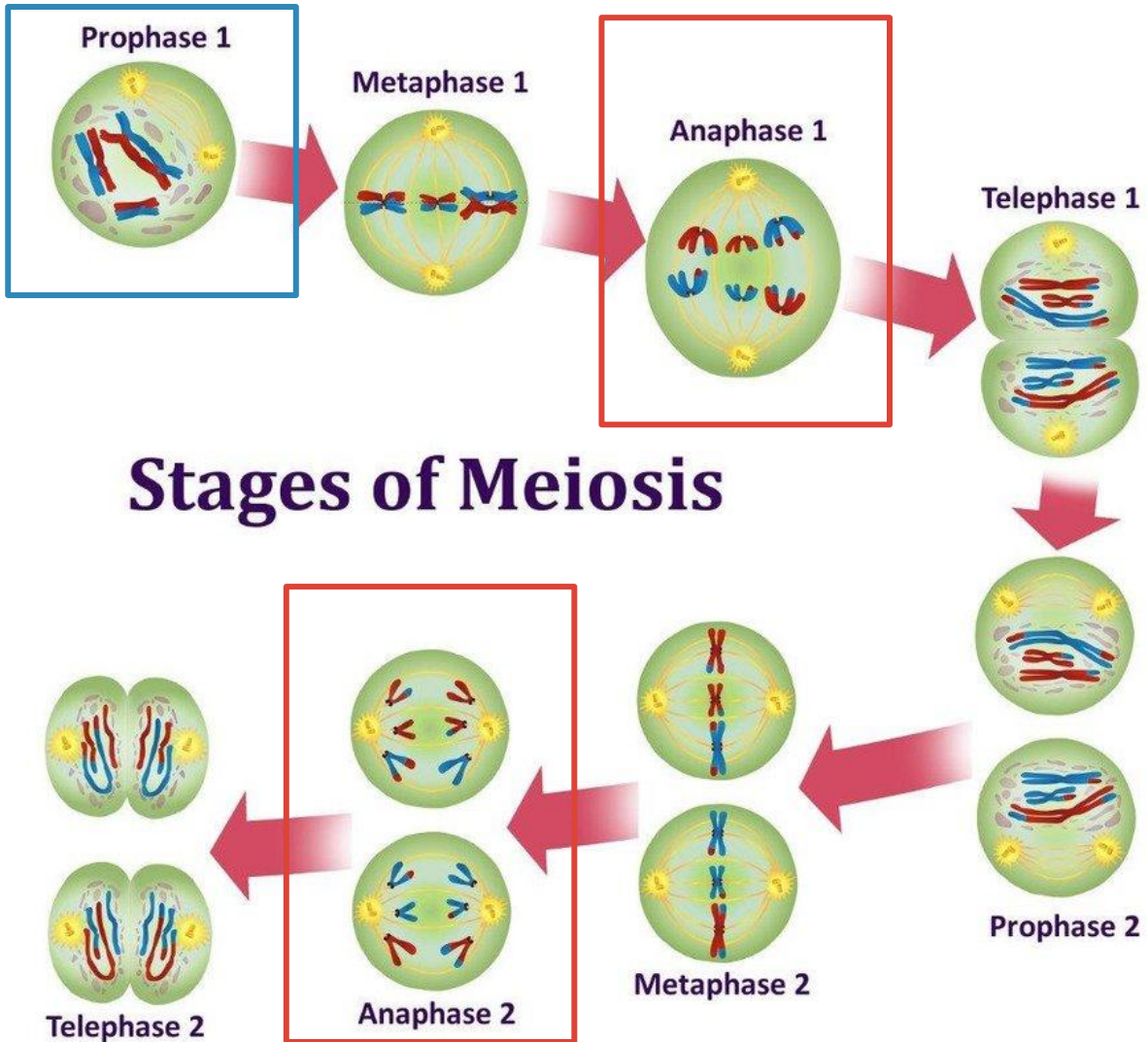
- Deletion
- Duplication
- Translocation
- Inversion



WHICH PHASE OF MEIOSIS WOULD BE RESPONSIBLE FOR THESE ERRORS?

Errors in chromosome **structure** occur here

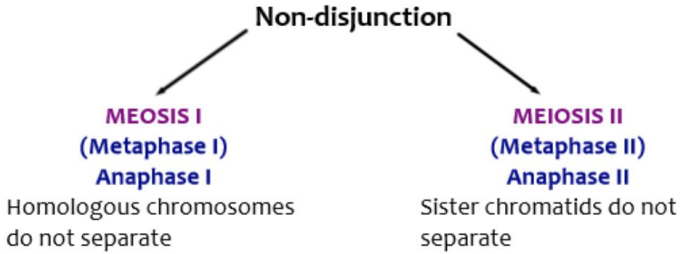
Errors in chromosome **number** occur here



1. Errors in Chromosome Number

Errors in chromosome number are a result of **nondisjunction** in meiosis and lead to **aneuploidy.**

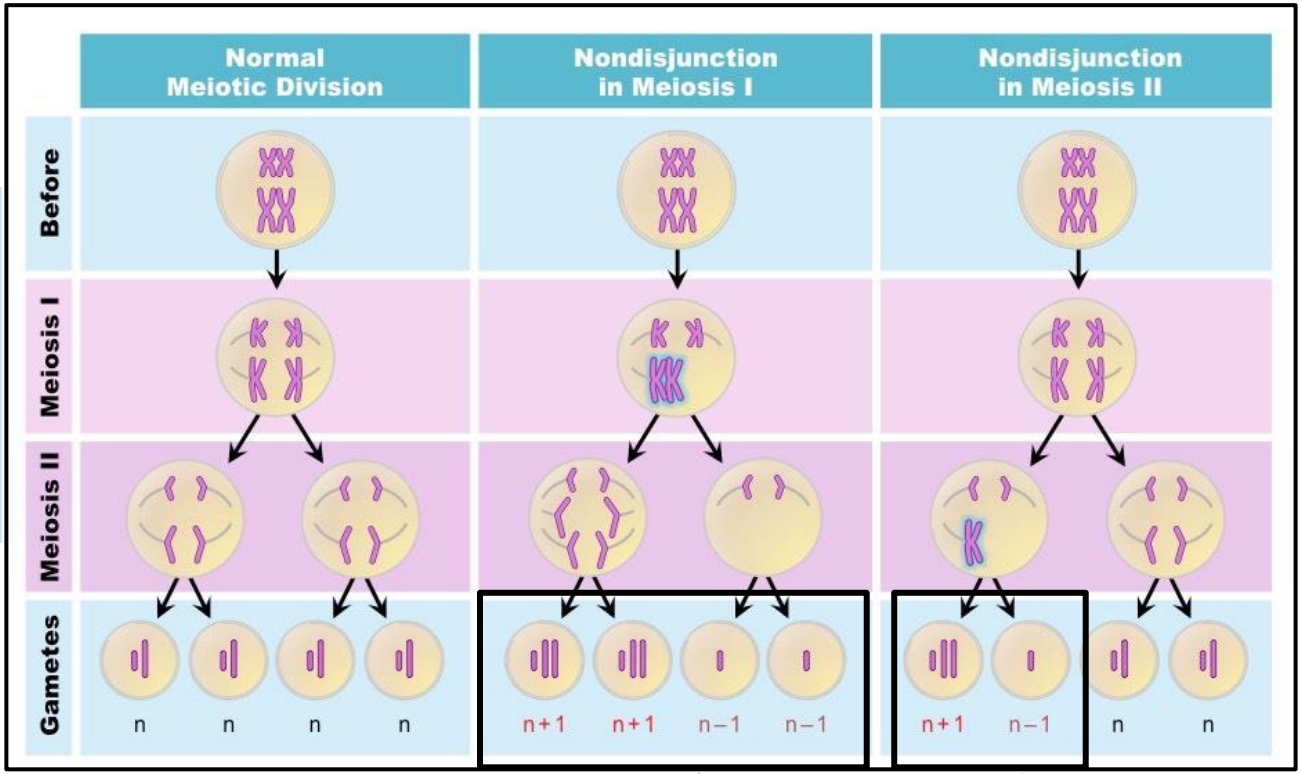
- **Nondisjunction** is the failure of homologous pairs or sister chromatids to separate properly during meiosis



- **Aneuploidy** - condition where an individual has too many or too few chromosomes in all their somatic cells

How does nondisjunction differ in meiosis 1 and 2?

These errors occur in **anaphase** when separation occurs.



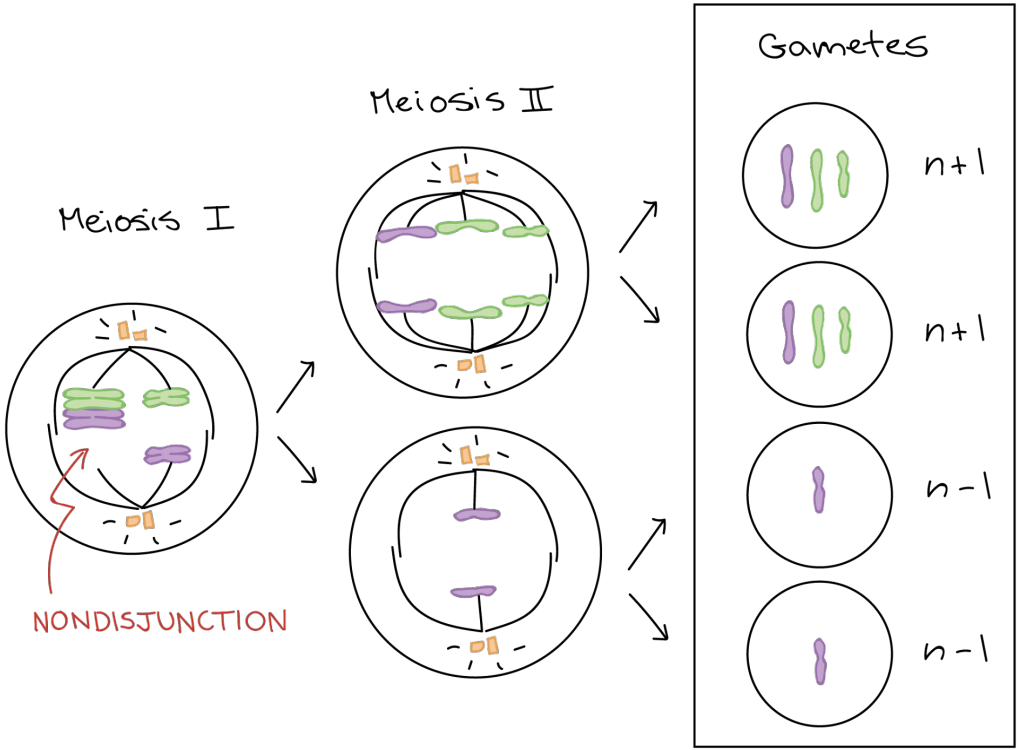
All gametes are affected and have aneuploidy.

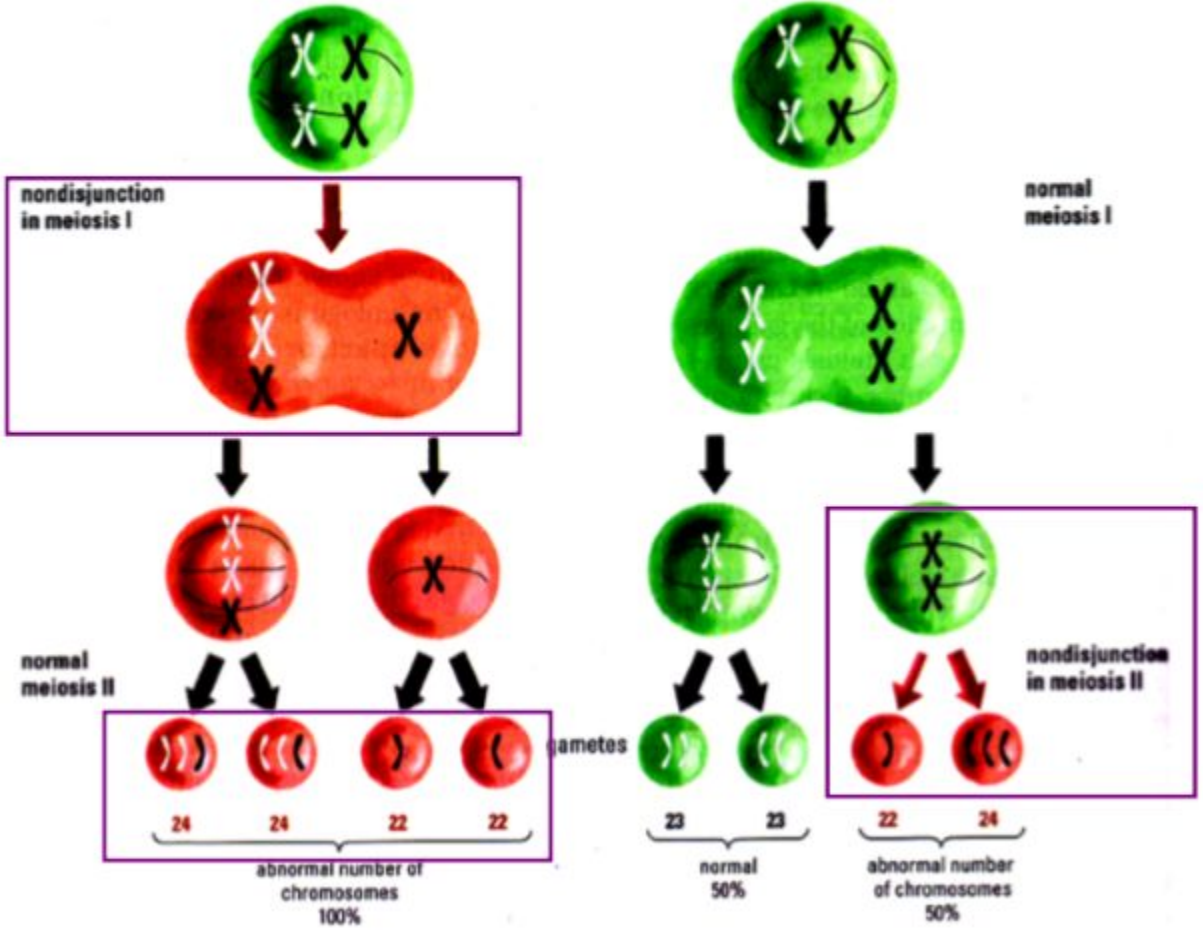
50 % of gametes are affected and have aneuploidy.

Which nondisjunction leads to more genetic problems?

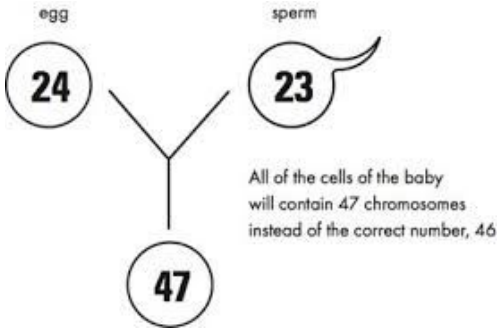
Meiosis 1

A closer look at nondisjunction in Meiosis 1

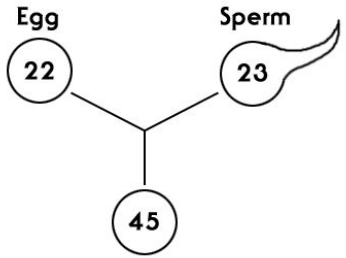



nondisjunction
in meiosis I
normal
meiosis I
normal
meiosis II
nondisjunction
in meiosis II
gametes
24
24
22
22
abnormal number of
chromosomes
100%
23
23
normal
50%
22
24
abnormal number
of chromosomes
50%

Trisomy: condition where there are **3 homologous chromosomes** in place of a homologous pair (there is a **gain of an <u>extra</u> chromosome** due to nondisjunction)



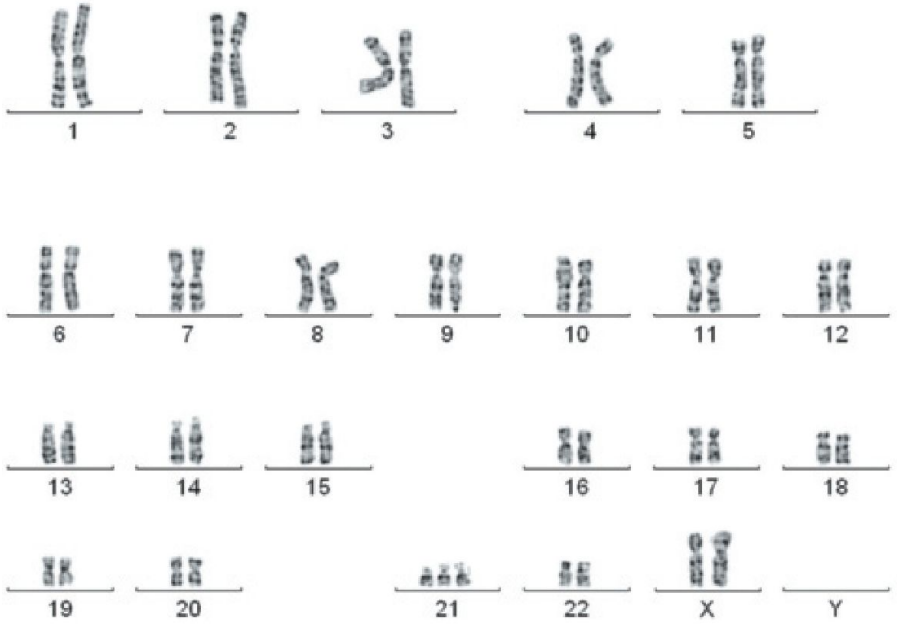
Monosomy:
condition where there is a **single chromosome** in place of a homologous pair (there is a **<u>loss</u> of one chromosome** due to nondisjunction)



Once the trisomic or monosomic zygote begins to divide (undergo mitosis and differentiation), **each cell** of the body will contain the abnormal number of chromosomes.

If the child survives one of the above two situations, they will show **effects** associated with the genetic information carried on the chromosome involved in the aneuploidy.

KARYOTYPE ANALYSIS - NONDISJUNCTION DISORDERS



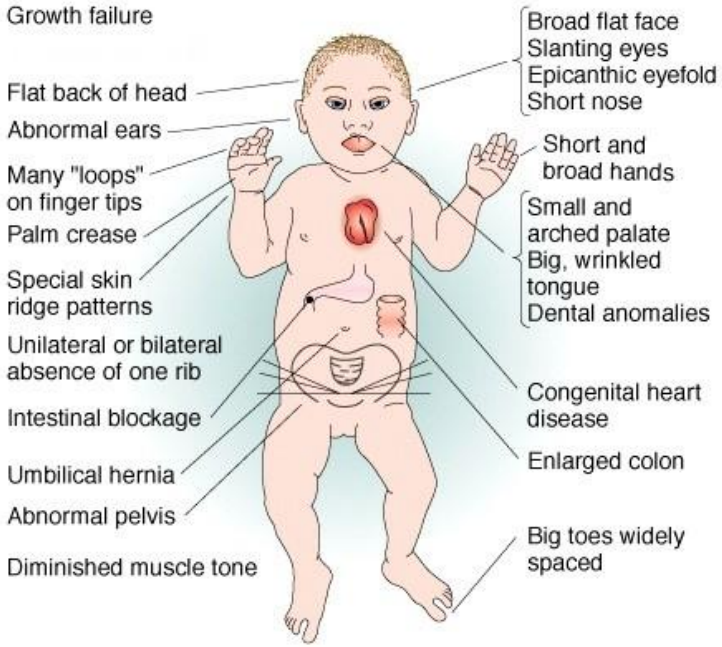
Based on their karyotype, is this person male or female?

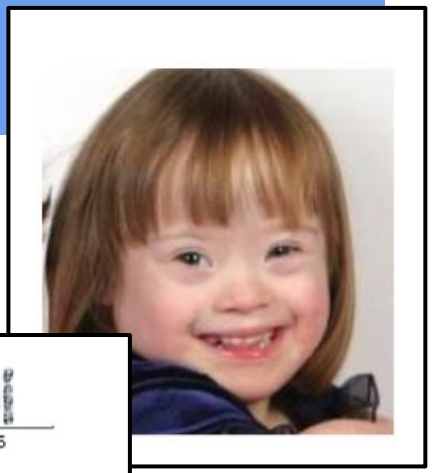
What genetic disorder is present?

How many chromosomes does this individual have in all of their cells?

Nondisjunction Disorders in Humans

Trisomy 21 – Down Syndrome





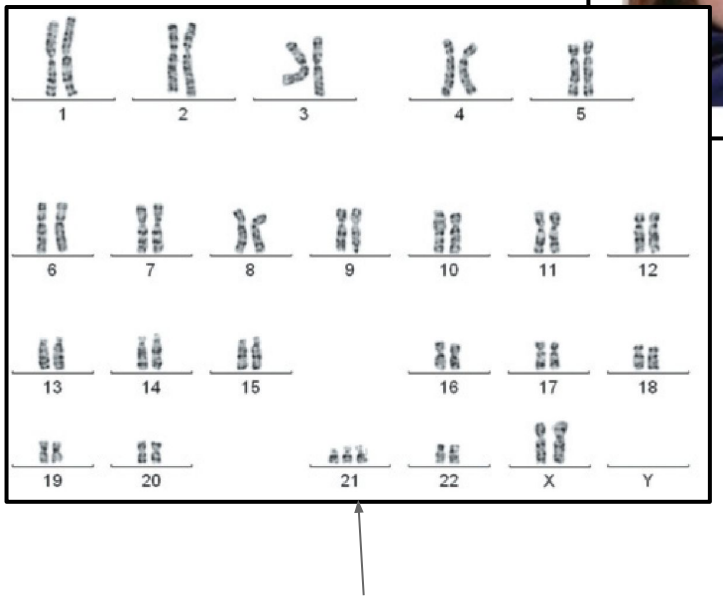
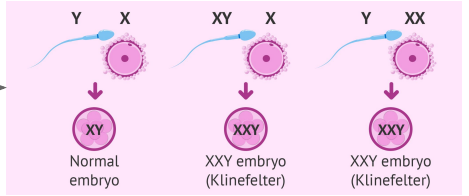
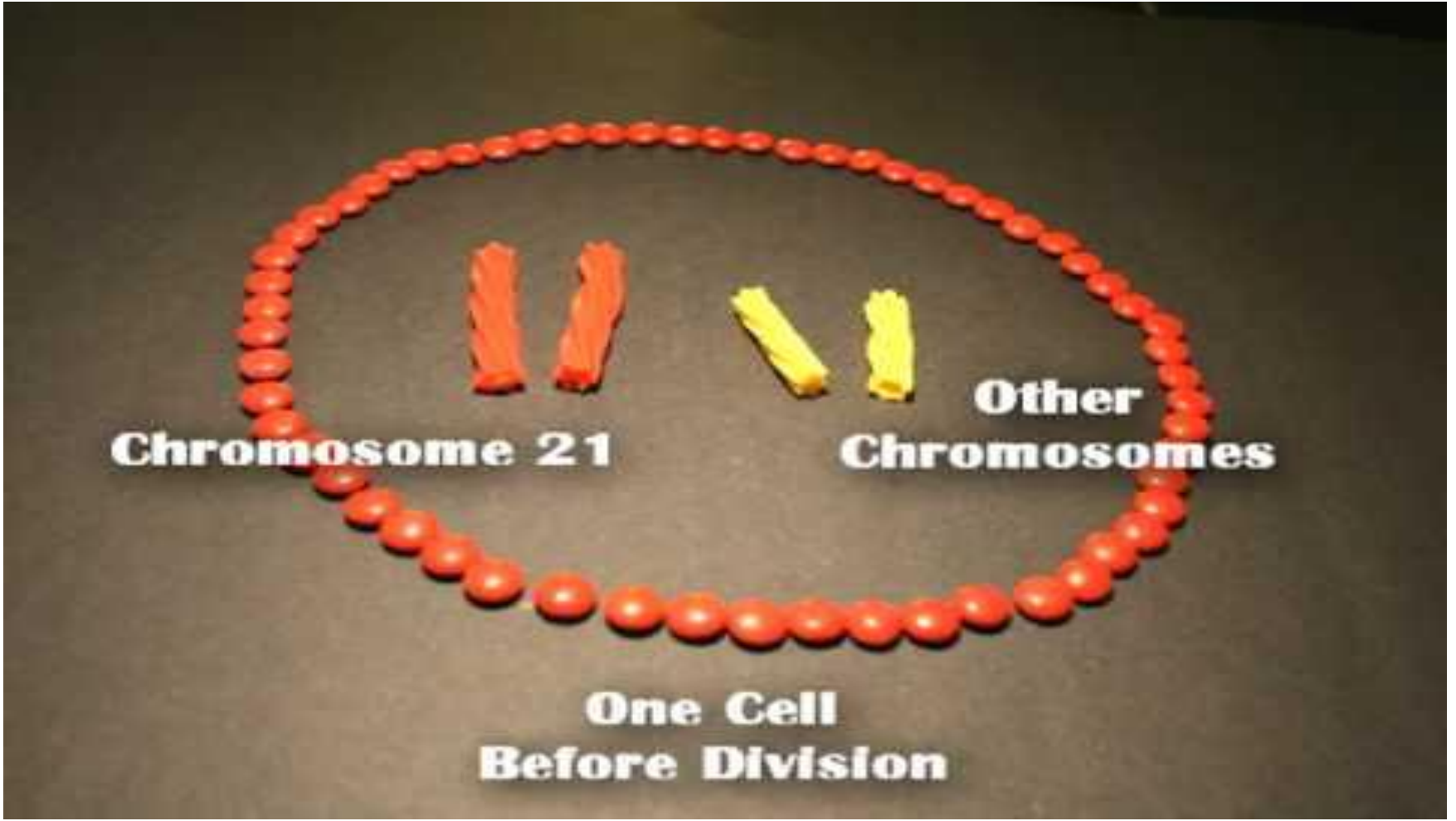


Table 1 Selected Human Non-disjunction Disorders

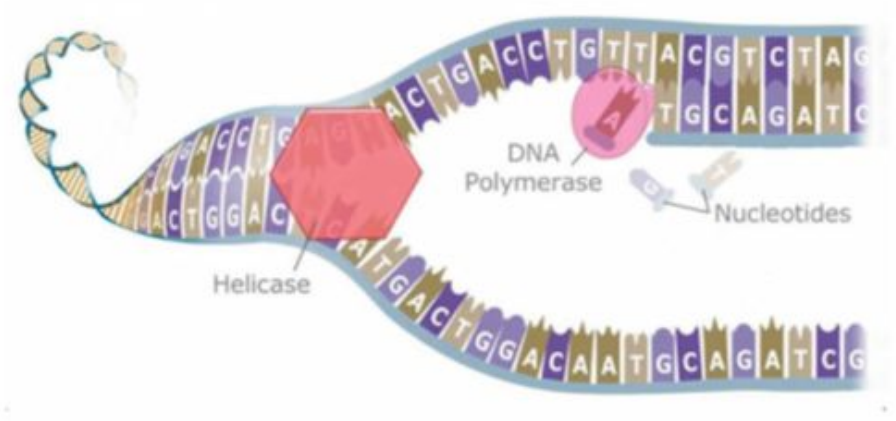
Non-disjunction disorder	Chromosome abnormality	Characteristics of the disorder
Turner syndrome	one X and no Y sex chromosome	Approximately 1:2500 female births. Female in appearance but do not mature sexually and are sterile. Most Turner syndrome fetuses are miscarried before the 20th week of pregnancy.
Klinefelter syndrome	two X and one Y sex chromosome	Approximately 1:500 male births. Males are usually sterile and exhibit some feminine body characteristics, but severity varies.
Patau syndrome	trisomy of chromosome 13	Approximately 1:25 000 live births. Many serious developmental problems, including brain, kidney, and heart defects. Children rarely live more than a few months.
Edwards syndrome	trisomy of chromosome 18	Approximately 1:6000 live births. Many organ system defects. Very low survival rate. Most fetuses die before birth. Average life expectancy of live-born infants is less than one month.




Chromosome 21
Other
Chromosomes
One Cell
Before Division

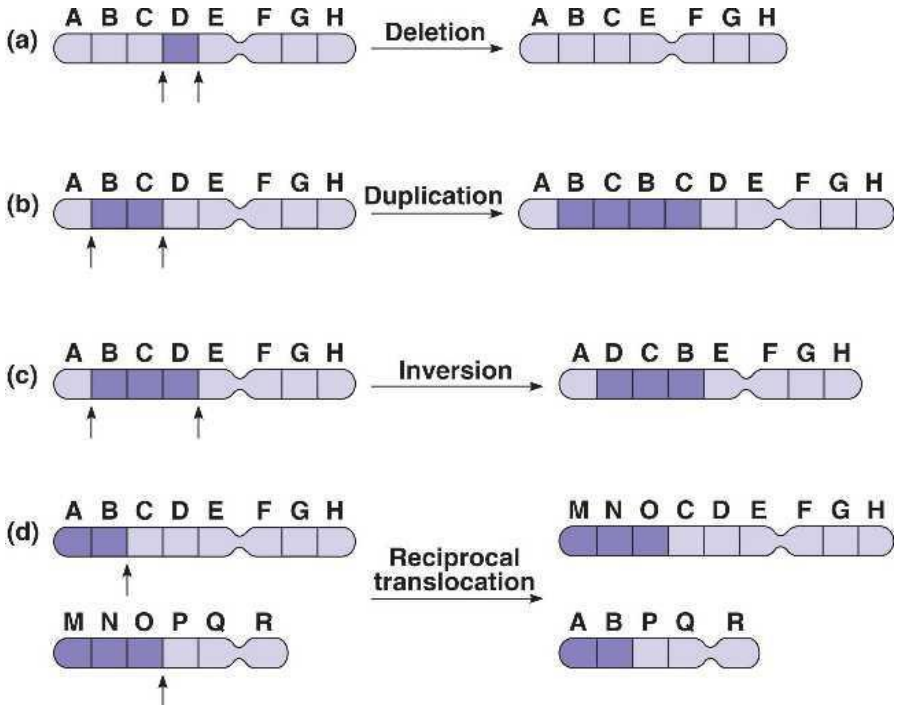
2. Errors in Chromosome Structure

- During crossing over (prophase 1), the **hydrogen bonds** that hold DNA together are broken and reformed
- Sometimes they do not reform correctly OR **non-homologous** pairs may cross over



See Table 4.1 on page 177

2. Errors in Chromosome Structure:



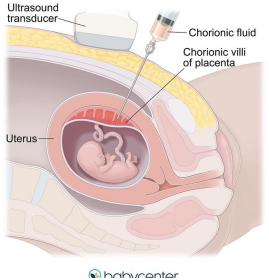
(a)deletion of chromosomal segments.

(b)chromosomal segments appear twice in a row

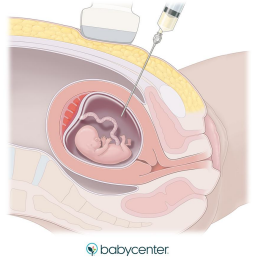
(c)chromosomal segments are reversed

(d)segments are moved between **non-homologous** chromosomes.

Prenatal Testing

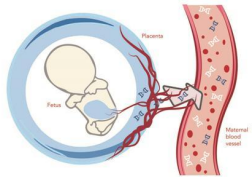


- **Chorionic Villus Sampling** (CVS): cells removed from outer membrane surrounding embryo.



- **Amniocentesis:** fluid from amniotic sac is extracted with a large syringe to prepare karyotype.

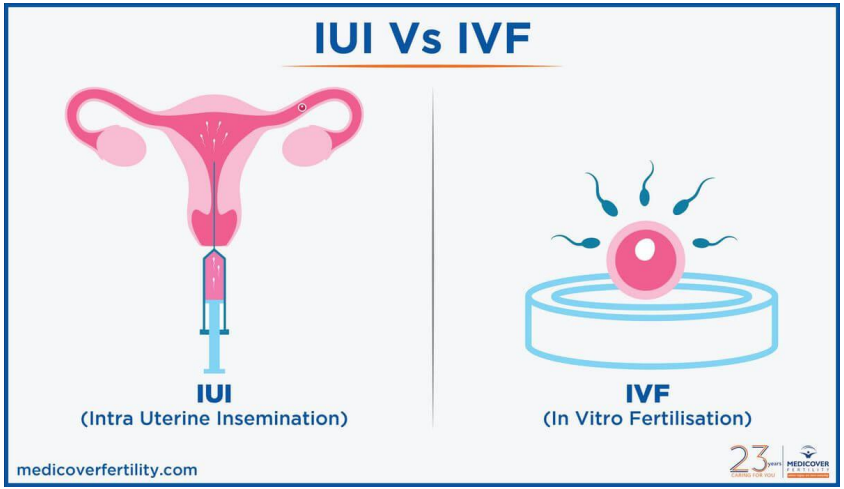
- **MMS Blood Test:** Multiple marker screening blood tests for the presence of particular hormones that may indicate increased risk of birth defects (Down syndrome and spina bifida).
  - Less invasive



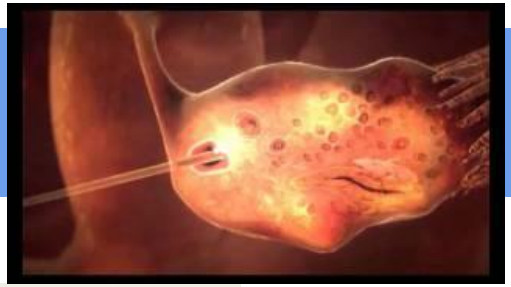
Assisted Reproductive Technologies (ARTs)

- Technologies used to enhance the chances of reproductive success as many couples have difficulty in conceiving

- **Intrauterine insemination (IUI)**
  - Placement of sperm into reproductive tract of female
- **In vitro fertilization (IVF)**
  - Stimulate ovaries using hormones to increase egg production
  - Retrieve eggs
  - Fertilize eggs in petri dish (2 ways)
    i. Place thousands of sperm in dish
    ii. Inject one sperm into egg
  - Implant fertilized egg into uterus

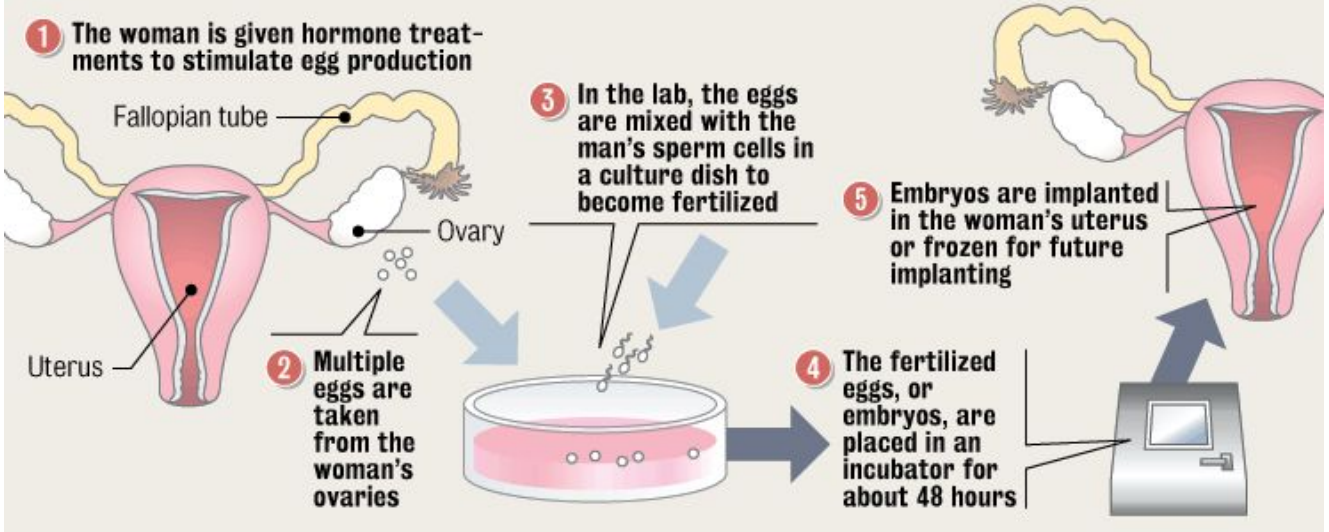


In Vitro Fertilization



THE IN VITRO FERTILIZATION PROCESS

Used as a remedy for infertility, a woman's egg cells are combined with a man's sperm cells outside the uterus. The fertilized egg is then implanted in the woman's uterus and, if successful, begins the pregnancy cycle. The first baby realized from this fertilization method was born in 1978.



4.4 Textbook Summary

4.4 Summary

- Errors in meiosis, including non-disjunction, can result in abnormal numbers of chromosomes and can cause serious genetic disorders.
- Karyotypes can be used to evaluate chromosome numbers and diagnose genetic disorders.
- Prenatal testing can be used to determine the likelihood of certain genetic disorders.
- Assisted reproductive technologies may be used when a couple is infertile in order to enhance their chances of conceiving a child.
- There are many applications of reproductive technologies in agriculture, industry, and wildlife conservation.
- The use of many reproductive technologies is highly controversial.

Genetic disorder assignment presentation

Your Task: Research a genetic disorder (list of topics on next slide) and design an informational presentation about the topic along with a one page summary of your chosen topic.

Your assignment should be attractive & eye-catching, concise (only bullet points & no length paragraphs) yet informative, and user-friendly for the audience.

You may work with one other person on this assignment. All assignments must be presented in class (5 – 8 minutes in length).

In-class Research Periods: _____

Due Date: _____. Everyone must be ready to present on the due date.

List of possible topics to research:

Mowat Wilson Syndrome

Metafemale Syndrome

Klinefelter Syndrome

Turner Syndrome

Angelman Syndrome

Cystic Fibrosis

Galactosemia

Neurofibromatosis

Amyotrophic Lateral Sclerosis

Patau Syndrome

Leber's Optic Neuropathy

Huntington's Disease

Prader-Willi Syndrome

Hemophilia

Edward Syndrome

Kabuki Syndrome

Phenylketonuria (PKU)

Cri-du-Chat Syndrome

Sickle Cell Anemia

Hemochromatosis

Spina Bifida

Hypercholesterolemia

Sever Combined Immuno Deficiency

Retinitis Pigmentosa

Albinism

Thalassemia

Rett Syndrome

Fragile X Chromosome

Tay-Sachs Disorder

Duchenne Muscular Dystrophy

Joubert Syndrome

Maple Syrup Urine Disease

Epidermolysis bullosa (butterfly syndrome)