Non-disjunction, Fertilization and Differentiation
Disjunction

How do chromosome abnormalities occur?

- Seperation of Chromosomes in Anaphase I or II of Meiosis is called Disjunction.

- Replication of DNA during “S”

- Meiosis begins with formation of tetrads in Prophase I.

- Normal Anaphase I or II results in disjunction (separation of chromosomes)
Disjunction: Chromosome Separation

(b) Metaphase I

(c) Anaphase I
Non-disjunction

When chromosomes fail to separate during Anaphase I or II we call it non-disjunction.

• Chromosomes which should split end up moving to the same pole.

• This can happen in Meiosis I or Meiosis II.

• Result: Aneuploidy—gametes are produced with either one chromosome too many or one too few.
Non-disjunction

Non-disjunction results in Aneuploidy:

- All individuals with too few chromosomes (except for the sex chromosomes) will die.
Aneuploidy

- An individual with one X will survive and exhibit:
  
  • “Turner Syndrome.” “monosomy 23” (2n-1)

- Individuals with one too many chromosomes sometimes survive. These include: chromosome #13, 18, 21 and 23.
  
  • Ex. Down Syndrome, Trisomy 21
  • Ex. Kleinfelter’s Syndrome, Trisomy 23 (XXY)
Aneuploidy

- Non-disjunction results in abnormal gametes and an aneuploid fertilized egg.

- The \((2n+1)\) or \((2n-1)\) condition will be passed on to all cells by mitosis.

Normal Gametes:
If normal Meiosis occurs, normal gametes will be produced.
- The fertilized egg will have 46 chromosomes.
- Mitosis will ensure that all the cells of the baby will have 46 chromosomes.
(a) Nondisjunction of homologous chromosomes in meiosis I

(b) Nondisjunction of sister chromatids in meiosis II
Oogenesis: Production of the Egg

- Specialized cells in the female’s ovaries will produce eggs.
- Meiosis occurs with **nonequal cytokinesis** during telophase 1.
- A non-functional “polar body” is produced.
- The larger cell continues into Meiosis II.
- Unequal cytokinesis occurs again in telophase 2.
- Another non-functional “polar body” is produced.
**Oogenesis: Production of the Egg**

• The end result is a **single, functional, haploid egg cell**.
  - The first polar body may also divide.

• Now there are **3** polar bodies.

• 4 products, but only the **egg** is useful.

• In plants the egg is called an “**ovul**” and is the precursor to a seed.
Primary Oocytes (2n) are formed in the embryo.  
They are frozen at Prophase I until puberty.  
A female is born with 2 million oocytes, but only about 400,000 survive till puberty.  
A woman will release on average around 580 eggs in her lifetime (one per month!).  
When a Primary oocyte (2n) is released it undergoes Meiosis 1 and becomes a Secondary oocyte (1n)!
Spermatogenesis
Production of sperm

• Specialized cells in the male testes will produce sperm.
• Meiosis proceeds normally, producing four haploid cells.
• The four cells then are modified to become sperm cells.
• In plants, the sperm are called “pollen”.

![Diagram of spermatogenesis process]

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Fertilization
Is when two haploid gametes (egg and sperm) join with each other to produce a diploid zygote.

• Only 1 sperm can enter the egg.

• In plants the fertilized ovul is called a “seed”.

• In animals the fertilized egg is called a zygote.
Zygote

The zygote is the diploid fertilized egg.

- In this photo you can see the two nuclei that are about to fuse.

- The egg nucleus has 23 chromosomes from Mom.

- The other nucleus from the sperm, has the 23 chromosomes from Dad.

- The result will be 2N, for a total of 46 chromosomes.
Embryo

- The embryo is the new, developing diploid individual.
- 1 cell develops into 2, both cells are 2n.
- Here the embryo has undergone mitosis again and is now in the 4 cell stage.
- All 4 cells are identical, 2n.

After day five: 70-100 cells (Blastocyst)
Differentiation

The process by which cells are directed to specialize into various different tissues.

• Differentiation allows organisms to reach their adult form and function.
• Here the developing spinal cord and brain are visible in the embryo.
• Cells become more specialized in their structure and function to be more efficient.
Cell Differentiation

- Nerve cells
- Red blood cells
- Smooth muscle
- Fat (adipose) cells
- Intestinal epithelial cells
- Striated muscle cells
- Loose connective tissue with fibroblasts
- Bone tissue with osteocytes
Differentiation

• Each cell has the same genetic information.
• The cell uses only the genetic information it needs to follow its specific pathway of development.

(All cells have the same genes, because they have the same chromosomes. Genes can be “turned on” or “turned off” to make each type of cell specific for its function.)

This is called Gene Expression!
Fetus

When the individual has developed all the major organs and structures of an adult, we call it a fetus.

For humans this is the start of the 9th week after fertilization till birth.