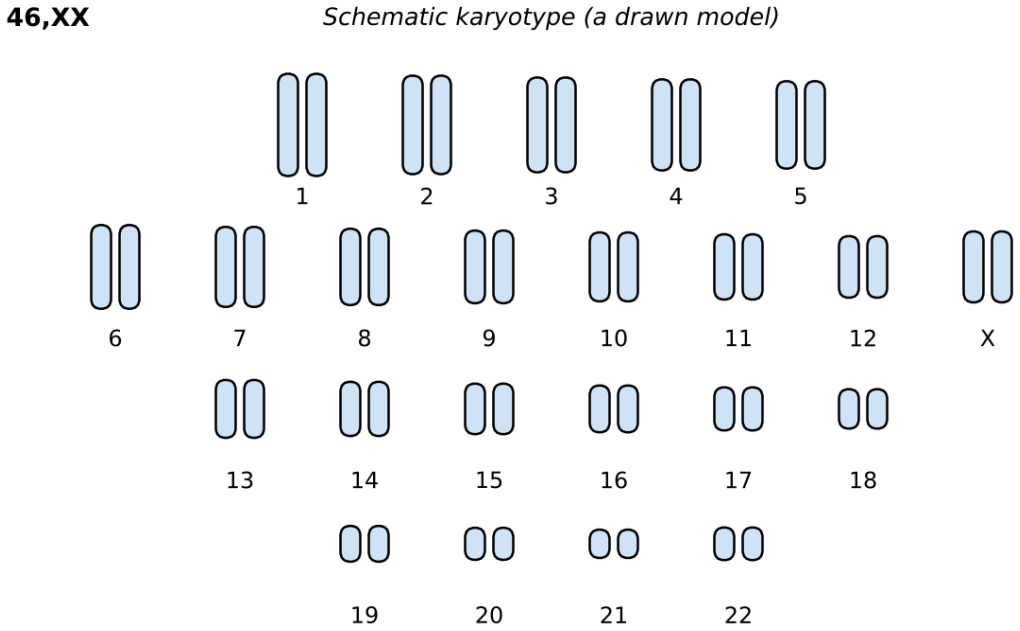
Essential Question

What happens when chromosomes do not separate correctly?

Chromosomal Disorders and Karyotypes

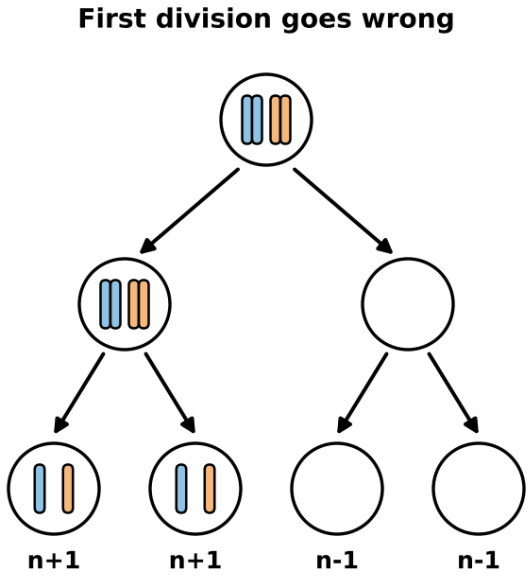
Karyotypes

- A karyotype is a picture of all the chromosomes in a cell, arranged in pairs
- Pairs 1 to 22 are autosomes, numbered from largest to smallest
- The last pair is the sex chromosomes: XX or XY in most people
- Most people have 46 chromosomes: 22 pairs of autosomes plus 2 sex chromosomes

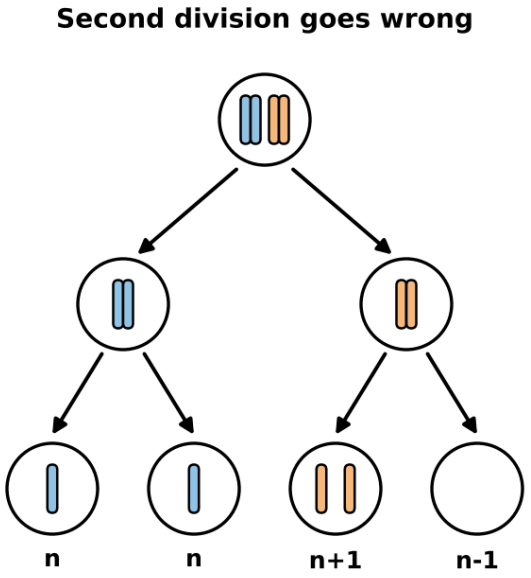


Nondisjunction

- Nondisjunction: chromosomes fail to separate during meiosis
- It can happen in the first or the second division
- A gamete ends up with an extra chromosome (n + 1) or one missing (n - 1)
- After fertilization, the zygote has 47 or 45 chromosomes



0 of 4 gametes have the usual number



2 of 4 gametes have the usual number

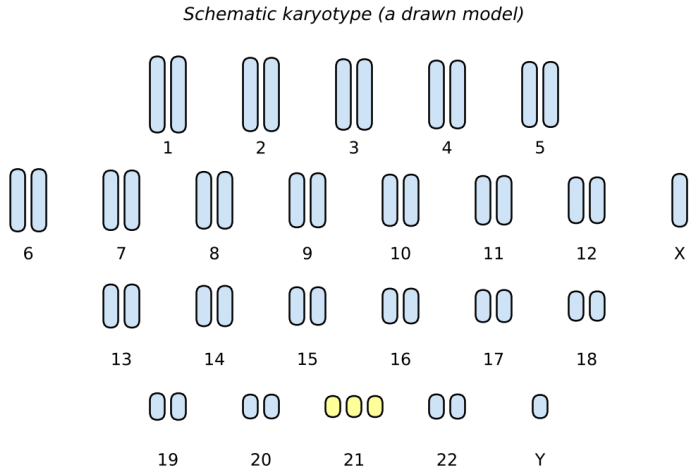
Chromosome Number Conditions

- Down syndrome: usually an extra chromosome 21, written 47,XX, +21 or 47,XY,+21
- Turner syndrome: one X chromosome and no second sex chromosome, written 45,X
- Klinefelter syndrome: an extra X chromosome with a Y, written 47,XXY
- People with the same karyotype can have very different health, abilities, and lives

Condition	Karyotype	Chromosome change
Down syndrome	47,XX,+21 or 47,XY, +21	Three copies of chromosome 21 (trisomy 21)
Turner syndrome	45,X	One X chromosome and no second sex chromosome
Klinefelter syndrome	47,XXY	An extra X chromosome along with a Y chromosome

Example 1: Read a Karyotype

The drawing shows the karyotype of one person (a drawn model). (a) How many chromosomes does the cell have? (b) Which sex chromosomes are shown? (c) Which pair is different from the others, and what is the name for having three copies of a chromosome? Write the karyotype in the form total number, sex chromosomes, change.



Solution: Read a Karyotype

(a) Count: There are 22 pairs of autosomes (44 chromosomes) plus one extra chromosome 21, which makes 45, plus the 2 sex chromosomes. The total is 47.

(b) Sex chromosomes: One X and one Y (XY).

(c) The different pair: Pair 21 has three copies instead of two. Having three copies of a chromosome is a trisomy, so this is trisomy 21.

Karyotype: 47 chromosomes, XY, extra chromosome 21 is written 47,XY,+21. This karyotype is typical of Down syndrome.

Answer: (a) 47. (b) XY. (c) Trisomy 21, written 47,XY,+21.

Example 2: Counting Chromosomes After Nondisjunction

A human egg-producing cell with 46 chromosomes goes through meiosis. Nondisjunction of chromosome 21 occurs. A sperm with 23 chromosomes fertilizes each egg. (a) How many chromosomes are in an egg that has an extra chromosome 21? (b) How many are in the zygote? (c) What fraction of the four cells made by meiosis have the usual 23 chromosomes if nondisjunction happens in the first division? In the second division?

Given	Value
Chromosomes in the egg-producing cell	46
Chromosomes in a normal sperm	23
Error in meiosis	Nondisjunction of chromosome 21

Solution: Counting Chromosomes After Nondisjunction

(a) Egg with an extra chromosome: 23 + 1 = 24 chromosomes.

(b) Zygote: 24 from the egg + 23 from the sperm = 47 chromosomes.

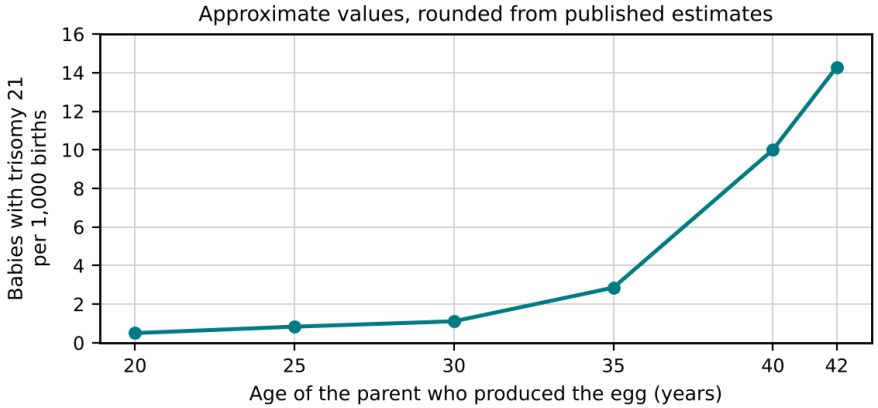
(c) First division: Both cells made by the first division have the wrong number, so none of the four final cells has 23. The fraction is 0/4 = 0.

Second division: One of the two cells divides as usual and makes 2 cells with 23. The other makes one cell with 24 and one with 22. The fraction with 23 is 2/4 = 1/2.

Answer: (a) 24. (b) 47. (c) First division: 0 of 4. Second division: 2 of 4 (1/2).

Example 3: Age and Chromosome Errors

The graph shows approximate values for the chance of a baby with trisomy 21 by the age of the parent who produced the egg. The values are rounded, and sources differ. (a) How many times more likely is a baby with trisomy 21 at age 40 than at age 30? (b) Write a claim, evidence, reasoning statement about how the chance changes with age. (c) Does the graph show that most babies with Down syndrome have older parents? Explain.



Solution: Age and Chromosome Errors

(a) Ratio: About 1 in 100 at age 40 and about 1 in 900 at age 30. 900 / 100 = 9, so it is about 9 times as likely.

(b) Claim: The chance rises with age, and rises faster after age 35.

Evidence: The chance is about 1 per 1,000 at age 30, about 3 per 1,000 at age 35, and about 10 per 1,000 at age 40.

Reasoning: Eggs are made before birth and wait for years. Errors in sorting chromosomes in meiosis become more likely as eggs age, so a larger share of eggs has the wrong number.

(c) No: The graph shows the chance per birth at each age, not how many babies are born at each age. Far more babies are born to younger parents, so most babies with Down syndrome are born to people under 35.

Answer: (a) About 9 times. (b) The chance increases with age, from about 1 to 10 per 1,000. (c) No. The graph gives chances, not the number of babies.

Chromosomal Disorders and Karyotypes

Big Ideas

- A karyotype is a picture of a person's chromosomes
- Nondisjunction is a failure of chromosomes to separate in meiosis
- It makes gametes with an extra or a missing chromosome
- Trisomy 21 is the usual cause of Down syndrome
- A karyotype tells you about chromosomes, not about a person's abilities or worth