

What is Down syndrome?

Down syndrome is a chromosomal condition that is associated with intellectual disability, a characteristic facial appearance, and weak muscle tone (hypotonia) in infancy. All affected individuals experience cognitive delays, but the intellectual disability is usually mild to moderate.

People with Down syndrome may have a variety of birth defects. About half of all affected children are born with a heart defect. Digestive abnormalities, such as a blockage of the intestine, are less common.

Individuals with Down syndrome have an increased risk of developing several medical conditions. These include gastroesophageal reflux, which is a backflow of acidic stomach contents into the esophagus, and celiac disease, which is an intolerance of a wheat protein called gluten. About 15 percent of people with Down syndrome have an underactive thyroid gland (hypothyroidism). The thyroid gland is a butterfly-shaped organ in the lower neck that produces hormones. Individuals with Down syndrome also have an increased risk of hearing and vision problems. Additionally, a small percentage of children with Down syndrome develop cancer of blood-forming cells (leukemia).

Delayed development and behavioral problems are often reported in children with Down syndrome. Affected individuals' speech and language develop later and more slowly than in children without Down syndrome, and affected individuals' speech may be more difficult to understand. Behavioral issues can include attention problems, obsessive/compulsive behavior, and stubbornness or tantrums. A small percentage of people with Down syndrome are also diagnosed with developmental conditions called autism spectrum disorders, which affect communication and social interaction.

People with Down syndrome often experience a gradual decline in thinking ability (cognition) as they age, usually starting around age 50. Down syndrome is also associated with an increased risk of developing Alzheimer disease, a brain disorder that results in a gradual loss of memory, judgment, and ability to function. Approximately half of adults with Down syndrome develop Alzheimer disease. Although Alzheimer disease is usually a disorder that occurs in older adults, people with Down syndrome usually develop this condition in their fifties or sixties.

How common is Down syndrome?

Down syndrome occurs in about 1 in 830 newborns. An estimated 250,000 people in the United States have this condition. Although women of any age can have a child with Down syndrome, the chance of having a child with this condition increases as a woman gets older.

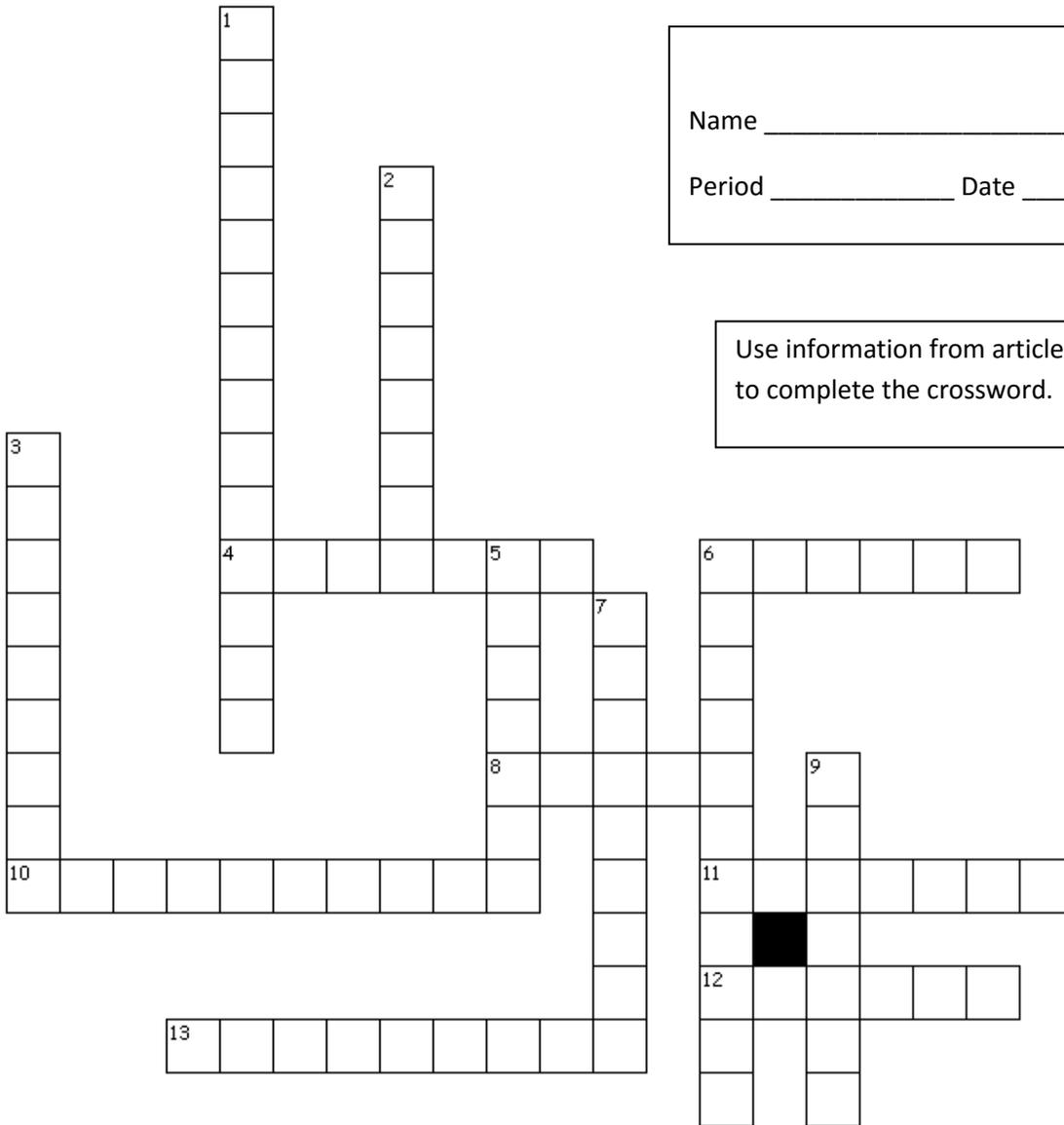
What are the genetic changes related to Down syndrome?

Most cases of Down syndrome result from trisomy 21, which means each cell in the body has three copies of chromosome 21 instead of two matching homologs. This can be detected in a karyotype: a visual depiction of all the chromosomes.

Can Down syndrome be inherited?

Most cases of Down syndrome are not inherited. When the condition is caused by trisomy 21, the chromosomal abnormality occurs as a random event during the formation of reproductive cells in a parent. The abnormality usually occurs in egg cells, but it occasionally occurs in sperm cells. An error in meiosis, called nondisjunction, results in gametes produced with an abnormal number of chromosomes. For example, an egg or sperm cell may gain an extra copy of chromosome 21. If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have an extra chromosome 21 in each of the body's cells.

Down Syndrome



Name _____

Period _____ Date _____

Use information from article on other side to complete the crossword.

Across

- 4. means "three chromosomes"
- 6. Don't eat bread with this disease!
- 8. male gamete
- 10. disease causes memory loss
- 11. refers to cells of the body
- 12. age of this parent increases risk of Down
- 13. visual representation of all the chromosomes in an individual

Down

- 1. causes abnormal number of chromosomes
- 2. matching chromosomes
- 3. muscle weakness
- 5. problem in this process can cause Down Syndrome
- 6. humans should have 46
- 7. intestinal blockage would affect this body system
- 9. produced by meiosis