

Autosomal Recessive Abnormalities and Disorders

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Tay-Sachs Disease

- This is a rare childhood disorder that is inherited from parents. A lysosomal enzyme, whose job is to break down a certain type of lipid, is misfolded and destroyed due to being miscoded by a mutated gene (Starr, 2016).
- Cells make the lipid, which is good, but cannot break it down, which is bad. High levels of the lipid accumulate in cells of the brain and spinal cord. The cells become damaged, and the cells are eventually killed (Starr, 2016).

Signs and Symptoms

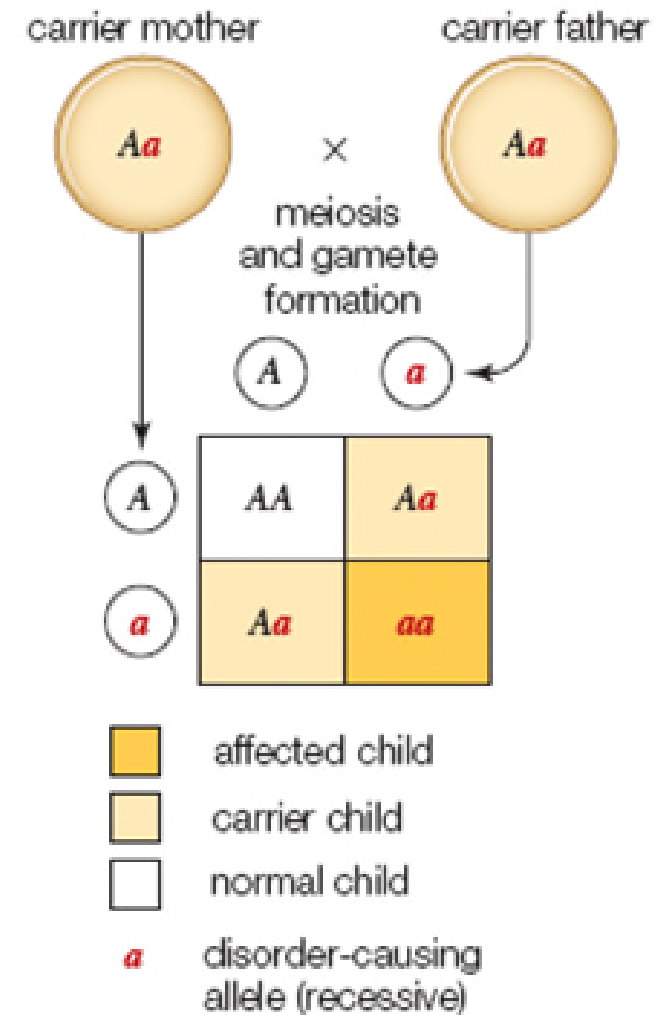
- The newborn baby seems normal (Starr, 2016).
- Within a few months the baby becomes listless and irritable and may have seizures. The seizures are caused by lipid accumulation in nerve cells (Starr, 2016).
- The baby eventually becomes blind, deaf, and paralyzed (Starr, 2016).

Inheritance and Transmission

- Alleles associated with Tay-Sachs disease are inherited in an autosomal recessive pattern (Starr, 2016).
- About 1 in 300 people is a carrier for one of the alleles (Starr, 2016).
- In some groups such as descendants of eastern European Jews, about 1 in 30 people is a carrier (Starr, 2016).
- Heterozygous people are called carriers because they have the allele but not the trait (Starr, 2016).
- Of those carriers, only people homozygous for a recessive allele on an autosome have the Tay-Sachs trait (Starr, 2016).

Inheritance Pattern

- Starr (2016) Figure 10.17
- In this example, both parents are carriers.
- Each child has a 25% chance of inheriting two alleles and having the Tay-Sachs trait.



Prognosis

- The disease is progressive (Starr, 2016).
- An affected child has deterioration of mental and physical abilities (Starr, 2016).
- The child usually dies by age five (Starr, 2016).

Additional information

- The destructive process begins in the fetus early in pregnancy (NIH, n.d.).
- A baby with Tay-Sachs disease appears normal until about six months of age when its development slows. By about two years of age, most children experience recurrent seizures and diminishing mental function (NIH, n.d.).
- The infant gradually regresses, and is eventually unable to crawl, turn over, sit or reach out. Eventually, the child becomes blind, cognitively impaired, paralyzed and non-responsive (NIH, n.d.).
- By the time a child with Tay-Sachs is three or four years old, the nervous system is so badly affected that death usually results by age five (NIH, n.d.).

Additional information

- Tay-Sachs disease results from defects in a gene on chromosome 15 that codes for production of the enzyme Hex-A (NIH, n.d.).
- The Tay-Sachs trait appears in both sexes at equal frequencies (Starr, 2016).
- The trait tends to skip generations (Starr, 2016).
- Any child of two carriers has a 25% chance of inheriting the allele from both parents and developing the trait (Starr, 2016).

References

- NIH (n.d.). *What Do We Know About Heredity and Tay-Sachs Disease?* National Human Genome Research Institute. Retrieved April 4, 2021, from <https://www.genome.gov/Genetic-Disorders/Tay-Sachs-Disease>
- Starr, C., Evers, C. A., & Starr, L. (2016). *Biology: Today & tomorrow without physiology*. Australia: Cengage Learning.