

# [Lorenzo’s oil essay sample](https://assignbuster.com/lorenzos-oil-essay-sample/)

1. What is the movie all about?
Lorenzo’s Oil
Lorenzo is the son of Michaela and Augusto Odone. He begins to have strange memory problems and blackouts. At age 6, he is diagnosed with the childhood cerebral form of ALD, a progressive degenerative nervous system disorder. There is no cure for this disease and his parents are told he will become totally disabled and die in a few years.

Michaela and Augusto, devastated by Lorenzo’s diagnosis, decide to research ALD even though neither has a scientific or medical background. The movie shows the Odones often working at odds with skeptical doctors, scientists, and support groups. Michaela and Augusto spend countless hours in medical libraries reading journal articles and talking to researchers and doctors. After much hard work and some inspiration, they help develop a treatment for ALD made from olive and rapeseed, which they name “ Lorenzo’s Oil.”

The movie also shows Lorenzo becoming completely bedridden and disabled as his disease progresses. At the end of the movie, he is able to communicate by a modified sign language. The story ends on a positive note as several children with ALD who have been treated with Lorenzo’s Oil are shown to be healthy. 2. a. Construct a pedigree of the characters.

Grandparent- Parent- Proband
(In the movie, Mariah is another sister of Michaela. But in real life, Mariah is actually James Emmett Murphy II, Michaela’s brother. His identity/character was hidden in the movie probably because of his career as a judge.)

-Male – Female – Male Proband – Female Carrier – Deceased Male
(Unaffected) (Unaffected) (Affected) (Affected) (Unaffected)

b. Provide examples of the case pedigree from your genetic references
Example 1

Reference: http://www. sciencedirect. com/science/article/pii/S0387760409000540
Example 2

Reference: http://www. sciencedirect. com/science/article/pii/S0006291X0801872X
c. Provide a description of the condition (1 page)
Adrenoleukodystrophy describes several closely related inherited disorders that disrupt the breakdown (metabolism) of certain fats (very-long-chain fatty acids).

Alternative names:
Adrenoleukodystrophy; Adrenomyeloneuropathy; Childhood cerebral adrenoleukodystrophy; ALD; Schilder-Addison Complex

Causes:
Adrenoleukodystrophy is passed down from parents to their children as an X-linked genetic trait. It therefore affects mostly males, although some women who are carriers can have milder forms of the disease. It affects approximately 1 in 20, 000 people from all races.

The condition results in the buildup of very-long-chain fatty acids in the nervous system, adrenal gland, and testes, which disrupts normal activity. There are three major categories of disease: \* Childhood cerebral form — appears in mid-childhood (at ages 4 – 8) \* Adrenomyelopathy — occurs in men in their 20s or later in life \* Impaired adrenal gland function (called Addison disease or Addison-like phenotype) — adrenal gland does not produce enough steroid hormones

SYMPTOMS

1. Childhood cerebral type:

\* Changes in muscle tone, especially muscle spasms and spasticity
\* Crossed eyes (strabismus)
\* Decreased understanding of verbal communication (aphasia)
\* Deterioration of handwriting
\* Difficulty at school
\* Difficulty understanding spoken material
\* Hearing loss
\* Hyperactivity

\* Worsening nervous system deterioration
\* Coma
\* Decreased fine motor control
\* Paralysis
\* Seizures
\* Swallowing difficulties
\* Visual impairment or blindness

2. Adrenomyelopathy:

\* Difficulty controlling urination
\* Possible worsening muscle weakness or leg stiffness
\* Problems with thinking speed and visual memory

3. Adrenal gland failure (Addison type):

\* Coma
\* Decreased appetite
\* Increased skin color (pigmentation)
\* Loss of weight, muscle mass (wasting)
\* Muscle weakness
\* Vomiting

The childhood form of X-linked adrenoleukodystrophy is a progressive disease that leads to a long-term coma (vegetative state) about 2 years after neurological symptoms develop. The child can live in this condition for as long as 10 years until death occurs.

The other forms of this disease are milder.
Possible Complications
\* Adrenal crisis
\* Vegetative state (long-term coma)

3. In modern medicine, what is the state of the said case/ how can it be cured or remedied?
ALD has not shown to have an increased incidence in any specific country of ethnic group. In the United States, the incidence of affected males is estimated at 1: 21, 000. Overall incidence of hemizygous males and carrier females is estimated at 1: 16, 800. The reported incidence in France is estimated at 1: 22, 000.

Treatment

Adrenal dysfunction is treated with steroids (such as cortisol).
A specific treatment for X-linked adrenoleukodystrophy is not available, but eating a diet low in very-long-chain fatty acids and taking special oils can lower the blood levels of very-long-chain fatty acids.

These oils are called Lorenzo’s oil, after the son of the family who discovered the treatment. This treatment is being tested for X-linked adrenoleukodystrophy, but it does not cure the disease and may not help all patients.

Bone marrow transplant is also being tested as an experimental treatment.

4. Considering marriage, how can choice of partner become a factor in preventing the said genetic condition?
Choosing a partner is a big factor in preventing the genetic condition because in this way, you are able to avoid diseases that might enter the family tree. In all situations, it is good to be knowledgeable of the certain genetic traits/diseases of the person you would want to spend the rest of your life with. In this way, you are aware of the consequences and ready for anything that might happen or come along.

Recommendation:
I recommend genetic counseling for married couples that are possible carriers or probands of Adrenoleukodystrophy.
Genetic counseling is the process of providing individuals and families with information on the nature, inheritance, and implications of genetic disorders to help them make informed medical and personal decisions. The following section deals with genetic risk assessment and the use of family history and genetic testing to clarify genetic status for family members. This section is not meant to address all personal, cultural, or ethical issues that individuals may face or to substitute for consultation with a genetics professional.

Result/ Discussion: Adrenoleukodystrophy
Adrenoleukodystrophy is passed down from parents to their children as an X-linked genetic trait. It therefore affects mostly males, although some women who are carriers can have milder forms of the disease. It affects approximately 1 in 20, 000 people from all races. The condition results in the buildup of very-long-chain fatty acids in the nervous system, adrenal gland, and testes, which disrupts normal activity. There are three major categories of disease: \* Childhood cerebral form — appears in mid-childhood (at ages 4 – 8) \* Adrenomyelopathy — occurs in men in their 20s or later in life \* Impaired adrenal gland function (called Addison disease or Addison-like phenotype) — adrenal gland does not produce enough steroid hormones Mode of Inheritance

X-linked adrenoleukodystrophy (X-ALD) is inherited in an X-linked manner.

Clinical Features:
1. Male Phenotypes (6)
\*
\* 48% – Childhood ALD
\* 26% – Adrenomyeloneuropathy
\* 10% – Addison Disease Only
\* 8% – Presymptomatic/Asymptomatic
\* 5% – Adolescent Cerebral ALD
\* 3% – Adult Cerebral ALD

2. Female Phenotypes (2)

\* 85-95% – Asymptomatic
\* 10-15% – Adrenomyeloneuropathy – late onset, less severe

Carrier Detection
Testing of at-risk female relatives for carrier status is a two-step process: 1. Measurement of plasma concentration of VLCFA (Very long chain fatty acids) is performed first; if abnormal, the female is a carrier. 2. Because 20% of female carriers have normal plasma concentration of VLCFA, molecular genetic testing should be used to test those females with a normal concentration if the disease-causing ABCD1 mutation has been identified in the family. Risk to Family Members

1. Parents of a male or female proband
\* Approximately 95% of individuals representing index cases have inherited the ABCD1 mutation from one parent; at minimum, 4. 1% of individuals with X-ALD have a de novo mutation. Evidence of germline or somatic/germline mosaicism is present in fewer than 1% of parents. Note: The estimated residual risk to parents of the 4. 1% of probands with an apparent de novo mutation of having germline mosaicism is at least 13%. \* It is appropriate to measure plasma VLCFA concentration in the mothers of both affected males and carrier females and in the fathers of carrier females. When the disease-causing mutation has been identified in an affected family member, molecular genetic testing of ABCD1 can be used in the evaluation of the parents.

2. Siblings of a proband
\* The risk to sibs depends on the genetic status of the parents, which can be clarified by pedigree analysis, measurement of plasma concentration of VLCFA, and molecular genetic testing. \* If the proband’s mother is a carrier, the chance of transmitting the disease-causing mutation in each pregnancy is 50%. Male sibs who inherit the mutation will be affected; female sibs who inherit the mutation will be carriers and will usually not be seriously affected. \* If the proband’s father has a disease-causing mutation in ABCD, all of the female sibs will be carriers, and none of the male sibs will be affected. \* If neither parent is a carrier, the risk to sibs of a proband is low.

3. Offsprings of a proband
\* Affected males transmit the ABCD1 mutation to all of their daughters and none of their sons. \* Carrier females have a 50% chance of transmitting the ABCD1 mutation in each pregnancy. Males who inherit the mutation will be affected; females who inherit the mutation are carriers and will usually not be seriously affected.

4. Other family members of a proband. Depending on their gender, family relationship, and the carrier status of the proband’s parents, the proband’s aunts and uncles and their offspring may be at risk of being carriers or of being affected.

Conclusion:
After gathering all necessary information about the disease, I conclude that the gene that causes ALD appears on the X chromosome. Michaela Odone, female, carries one X chromosome which makes her a carrier of the disease to Lorenzo Odone. I also conclude that it is possible for a male to have ALD even if the carrier does not have the mutated gene due to de novo mutations.

References:

http://rarediseases. about. com/cs/ald/a/041301. htm
http://health. nytimes. com/health/guides/disease/adrenoleukodystrophy/overview. html http://www. ncbi. nlm. nih. gov/books/NBK1315/
http://www. stopald. org/ald/TreatingALD. asp
http://en. wikipedia. org/wiki/Adrenoleukodystrophy
http://www. ojrd. com/content/7/1/51
http://www. fightald. org/overview. htm
http://www. orpha. net/consor/cgi-bin/OC\_Exp. php? Expert= 43
http://clasfaculty. ucdenver. edu/bstith/loren. htm
http://www. woodrow. org/teachers/bi/1994/adreno2. html
http://www. people. com/people/archive/article/0,, 20063465, 00. html http://crowleys-classroom. wikispaces. com/Lorenzo’s+Oil+page
http://news. bbc. co. uk/2/hi/health/3907559. stm
http://en. wikipedia. org/wiki/Augusto\_and\_Michaela\_Odone
http://www. thisisannouncements. co. uk/5857416
http://www. findagrave. com/cgi-bin/fg. cgi? page= gr&GRid= 23482096