Abstract

For the purpose of this essay, research was conducted to provide a clearer picture of what Lissencephaly is. Information was presented in a format suitable for an “outside” audience to understand. Research found only a few sources, only one of which was very dense. Topics covered in the paper were the disease itself, symptoms, progression, cause, related syndromes, treatment options, social issues relating to the disease, and an anecdote of the Thompson Family whom is directly affected by Lissencephaly. The conclusion of the paper leads one to believe that more research needs to be done, and more people need to be made aware of the Lissencephaly.
Paul Taylor, the newborn of Peter and Katherine Thompson, will never learn to walk. He will never talk or go to school and must always be under the careful supervision of his parents and doctors. More than likely, he will not spend more than two years on Earth. Paul suffers from a rare malformation of the brain called Lissencephaly, which leaves most of his upper level brain functions inoperative. With few resources containing information about the disease, research must persist in order to help patients of Lissencephaly and those affected by it indirectly.

Lissencephaly originates from the Greek words "lissos" meaning smooth, and "enkephalos" which means brain (1). Paul’s brain is smooth instead of containing the usual ridges and canyons. Basically, the brain is a ping-pong ball instead of a golf ball. In the case of Lissencephaly, the nerves do not reach the surface of the brain, but settle in irregular positions. The abnormalities of the nerves contribute to the “smoothness” of the brain and the lack of nerves reaching the edge of the brains is the cause of the symptoms associated with Lissencephaly.

Children with Lissencephaly possess serious mental retardation, and very restricted physical movement. Months after birth, patients develop repetitive seizure episodes and facial irregularities including large foreheads, depressed temples, and small jaws. Children seem to always be drowsy or in a state of “hypnosis.” Doctors classify the disease in two types depending on the manifestation of the disease on the child. Type I is the less serious kind with mild cases of the aforementioned symptoms. Type II has more distinct symptoms and birth defects including hydrocephalus (severe enlargement of the head) (1). Because Lissencephaly affects mental abilities first, it is a difficult disease to detect in infants.
Lissencephaly arises during pregnancy, but is undetectable by parents and doctors until about two months after birth. This is because the lower, unaffected section of the brain controls most of a newborn’s functions. Therefore, most babies with Lissencephaly seem completely normal to doctors and parents. Sometimes it can be detected on either a CT or a MRI scan, but this is highly unlikely because newborns usually do not undergo such exams unless disorders are suspected in the first place. Problems appear when the child fails to develop good feeding habits, visual dexterity, and physical coordination. Symptoms of the fully developed disorder include seizures, abnormal face development, lack of motor skills, and serious mental retardation.

Lissencephaly is also associated with several syndromes including Isolated Lissencephaly sequence (ILS), Miller-Dieker syndrome (MDS) and Walker-Warburg syndrome (WWS) (1). Isolated Lissencephaly is characterized as moderate mental retardation, with very little appearance abnormalities. Subtle instances of emotion can be seen such as smiles and brief visual tracking. Isolated Lissencephaly does not completely restrict movement, although only one patient has ever been recorded as learning how to walk. Seizure episodes start later than the other Lissencephaly-caused syndromes, but they still begin within the first year. Because of malnutrition from poor feeding habits, ILS produces repeating cases of pneumonia and this is often the cause of death within the child’s first few years of life (1).

The Miller-Dieker syndrome (MDS) possesses more physical manifestations of Lissencephaly than ILS. Facial abnormalities are more common, and there is practically no movement in patients. The mental disabilities are also more serious. A missing piece on chromosome seventeen in addition to Lissencephaly’s effect on the brain causes MDS (5). Many children are born with heart defects, cataracts, and ear abnormalities. The risk of having another child with MDS (twenty-five to fifty percent) exceeds ILS (seven to twenty-five percent) (1).
Lifetime is considerably shorter when compared to ILS, as most children die before the age of two.

Walker-Warburg syndrome (WWS) differs from Isolated Lissencephaly and Miller-Dieker in that lower brain defects, eye abnormalities, and muscular dystrophy accompany it. Unlike Lissencephaly itself, and syndromes associated with it, WWS can be detected very early as the newborn struggles with coordination and breathing control. Because the lower brain remains mostly inoperative, life-dependent functions, such as obtaining oxygen, are difficult for the child. The eyes possess no color, but appear cloudy and gray. WWS rarely occurs, because it is a recessively inherited trait. Very simply, this means the parents have one working part and one abnormal part of the gene; therefore they are normal, but their child inherited both non-working sections of the gene and hence has WWS. Because of breathing complications, many die within a few weeks and never exhibit the symptoms of Lissencephaly that would have developed later in the child’s life (1).

The source of Lissencephaly is unknown, but theories include genetic complications, insufficient blood supply to the brain, or viral infections during early fetal development. Although extensive therapy has been developed, there is little hope for any kind of normal growth. This is because of the disease causes severe lack of coordination and shortened lifetime. Infants are very limited in movement and play (most never learn to roll over or grab objects) and hardly live past the age of puberty (2). The unfortunately grim progress of Lissencephaly sparks the will to continue research and funding to organizations such as NORD (National Organization for Rare Diseases) in hopes of finding a cure for this unforgiving disease.

What is a parent to do when struck with the staggering news that their child has Lissencephaly? A long and tough road lies ahead of the Thompson’s as they struggle to care for
their sick child. Many nights of panic await them as Paul will suffer through sudden seizure episodes, risk of illness and pneumonia, and constant discomfort. Peter and Katherine will sleep uneasy in worry of their child’s life. For this reason, the caretakers are often in just as much need of emotional support as the patients themselves. Living with and taking care of a physically and mentally disabled person is one of the biggest challenges parents can face in their lifetime. Denial, depression, and frustration can take over leaving parents lost in a state of doubt and confusion. Counseling services help support those indirectly affected by Lissencephaly. Such services can be obtained from organizations such as NORD. Parents must have a proper sense of direction and compassion in order to fully care for their child.

Unfortunately, the treatment options for Lissencephaly remain slim: seizure medication to control episodes, and a gastronomy tube if feeding becomes too difficult. If researchers discover Lissencephaly to be a hereditary disease, genetic counseling (altering genes to repair faults) looks to be a hopeful treatment option if future technology makes such procedures possible. Consequently, moral questions rise in the wake of this proposed option. Should gene therapy be allowed? In effect, scientists change the baby to a completely different person. Political leaders, citizens and religious groups usually demote such “god-like” practices. Also, when doctors detect Lissencephaly early, the issue of abortion arises. Certain parents and doctors may believe that instead of putting a child and themselves through the pain of the disease, they would rather let the inevitable happen before such suffering can occur. Abortion, like genetic engineering, is a highly debatable issue. On one hand, the doctors know that Lissencephaly will eventually take the life of the baby, but on the other hand, as abortion protesters argue, the baby becomes “living” at conception and nothing should be done to change the forces of nature. As for now, the most widely used solution for parents is hope: hope that
their child will overcome Lissencephaly, or that a cure will be developed while the baby is still living.

Existing with Lissencephaly reminds one how fragile life really is. Research should be intensified in order to discover more treatment options or, hopefully, a cure. Parents need to know how to live and cope with Lissencephaly. Currently, loving their child and providing the proper medical and emotional support is the most parents can do. Making the child as happy as possible during their short life on this planet is the main goal of “treatment.” Paul Thompson’s life is a gift. Peter and Katherine’s gift may be short-lived, but the joy and excitement of embracing him will last forever.
References


