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Cornelia de Lange Syndrome: In the Realm of Caregivers and Surviving

Honors Project

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Abstract

Cornelia de Lange Syndrome (CdLS) is a rare congenital disorder that occurs in 1 in 10,000 live births. The goal of this research project was to further understand the impact of caregivers on children with disabilities, specifically CdLS. The purpose of this research project was to critically analyze the caregivers of those with Cornelia de Lange Syndrome, using the social model of disabilities as a theory and Derrida’s “structure” as a method; I argued caretakers of those with disabilities, specifically CdLS need to be given support, as well as resources, in order to maintain a healthy environment and family for the individual with special needs. In the study, it was found that caregivers would like more resources for their child, whether that be awareness, respite, or long-term care facilities.

Key words: caregiver, Cornelia de Lange Syndrome (CdLS), disability, resources, respite
Introduction

My sister, Emily, was diagnosed with Cornelia de Lange Syndrome (CdLS) when she was just a few days old. Her life expectancy was to be no longer than a year, but she beat those odds and today she is 23 years old. Emily has a severe case of CdLS, and as a result is about 18 months developmentally. She has a multitude of physical impairments as well, including, two fingers on one hand and three on the other. She cannot walk or talk and is fed through a G-tube placed in her stomach. Needless to say, Emily has had a difficult yet rewarding life thanks to her caregivers. However, it is the life of a caregiver that guides this study. Since 1 in 10,000 live births result in Cornelia de Lange Syndrome, the goal of this study is to further understand the impact of caregivers on children with disabilities, specifically Cornelia de Lange Syndrome. The purpose of this research paper is to critically analyze the caregivers of those with Cornelia de Lange Syndrome, using the social model of disabilities as a theory and Derrida’s “structure” as a method; I argue caretakers of those with disabilities, specifically CdLS need to be given support as well as resources in order to maintain a healthy environment and family for the individual with special needs.

I begin with a background disability and on Cornelia de Lange Syndrome, more specifically those who are caring for individuals with CdLS, whether that be a mother, father, sister, brother, or respite provider. then explain the social model of disabilities and Derrida’s “structure.” Individuals with disabilities experience many problems in their lifetime, but many of the problems that they may face are caused by the society’s views of that individual. The attitude and behavior that a society has towards an individual (Atinay, 2018, p. 3) impacts how the person may view themselves. In turn, enforcing a negative stereotype placed on them because of the attitudes of how people think that they should act. Those who are mentally handicapped need
support and for that reason this study looks into the support that a caregiver needs and provides to an individual with Cornelia de Lange Syndrome.

**Literature Review**

**History of Disability**

Intellectual disability is defined by the World Health Organization as “presence of incomplete or arrested mental development, principally characterized by the deterioration of concrete functions at each stage of development” (Katz, 2008, p. 2). Intellectual disabilities may be present from birth, but many occur later on in life, e.g. Alzheimer’s, dementia. Physical disabilities follow the same pattern. Historically, because of a person’s disability, his or her disability outlined who can and cannot become a citizen. Specifically, in the United States “disability was called on to clarify and define who deserved, and who was deservedly excluded from, citizenship” (Longmore, 2001, p. 34). The rights of citizenship led to an inequality for those with disabilities similar to the inequality faced by women and people of Color. Those with disability were seen as “monstrous” (p. 35) because they were not considered “typical” in society, instead, viewed as defective. Often, individuals with disability were considered “abnormal,” making them less than human. In this, the question of what exactly normal or typical is, remains. Furthermore, African Americans were seen as subhuman, classifying them in the same group as those with disabilities. The only way a Caucasian person would be viewed as an equal to an African American is if they had a disability. In other parts of the world, specifically Cambodia, physical disability is prominent because of the war that ran rampant decades prior. In Cambodia, a developing country, life with a physical disability can be tough because it is a developing country. Living with a disability in Cambodia comes with “a social
stigma of worthlessness, and discrimination is widespread” (Bergman, 2001, p. 25). Disability is found all over the world, regardless of race, gender, or wealth.

One of the most marginalized groups of individuals are children with disabilities. The issue of marginalization comes not from the disability specifically, but instead the lack of knowledge of how to treat individuals with disabilities. Within this, the lack of a person comes into play. Persons with disabilities are often seen as less than because they “lack,” whether that be intellectually or physically. This does not make a person with disabilities any less of a person (Unicef, 2013, p. 4). It has been said that,

“children who are poor are more likely to become disabled through poor healthcare, malnutrition, lack of access to clean water and basic sanitation, dangerous living and working conditions. Once disabled, they are more likely to be denied basic resources that would mitigate or prevent deepening poverty. Poverty and disability reinforce each other, contributing to increased vulnerability and exclusion” (p. 5).

Those with disabilities are not only facing poverty, but also prejudice over something he or she may be unable to change. To work against these factors the United Nations Convention convened on the Rights of Persons with Disabilities and Mental Health. This convention has international law and within the conventions there are multiple references stating the “rights to life, equal recognition before the law, access to justice, liberty and security of the person, privacy, […] education, health, work and employment […] with freedom from torture or cruel, inhuman or degrading treatment” (Barlett, 2012, p. 755). The statements above are considered the basics of human rights, also applying to those with disabilities. Historically, individuals with disabilities have been treated differently and sometimes even tortured because of their impairment. The Convention on the Rights of Persons with Disabilities (CRD) works to outline
the rights of disabled individuals, because they are often treated differently. In having a
disability, persons are forced into the position of the other, (Hughes, 1997, p. 326). Society looks
at people differently in he or she may look different or speak differently. This causes an
individual to be forced into a completely separate group, the other, because that individual does
not fit society’s ideals of perfection. It is because of grouping individuals separately that there
became a need for the CRD. Humanity should not need guidelines on how to treat a human
being, but humanity is competitive and being seen as less than whole, puts the individual with
disabilities at a disadvantage.

**About CdLS**

Cornelia de Lange Syndrome (CdLS), also known as Brachman de Lange Syndrome, is
classified as a congenital syndrome and is present from birth. It is not a disease that can be
cought and passed from one individual to the other. Those with CdLS are diagnosed often within
a few days of being born because of common characteristics of the syndrome such as a small
upturned nose, missing limbs, a small chin, thin lips, and low birth weight (Characteristics, 2018,
p. 1). Genetic testing may also confirm a positive mutation on a gene known to contain CdLS.
CdLS does affect many areas of the body. The syndrome ranges from mild to severe and does
often contain some sort of intellectual disability. Around 65% of those with CdLS have a
mutation on the NIPBL gene, 5% have a mutation on either the SMC1A, SMC3, HDAC8, and
RAD21 gene. In other 30% of the cases, it is unknown where the mutation lies (Cornelia, 2018,
p. 1). CdLS can be inherited, but the majority of the time the mutation causing CdLS occurs
sporadically. Parents of a child with CdLS could potentially have another child with CdLS, but
the chance of this is less than 1%. Rarely there have been cases of parents actually having two
children with CdLS. If a person who has CdLS, has a child, there is a 50% chance that any of their children will also have CdLS (Cookman, 2001, p. 47).

Cornelia de Lange Syndrome was discovered in 1933 by a Dr. Cornelia de Lange, but Brachmann described a child with similar features in 1916. The syndrome occurs anywhere from 1 in 10,000 to 30,000 to 50,000 to 100,000 live births (Kalal, 2009, p. 1). The disease is so rare that it is hard to say just exactly how often it occurs and, as a result, there are a multitude of different numbers out there. Most of the individuals are not expected to make it past two years of life “[…] the main cause of death recorded is pneumonia along with cardiac, respiratory, and ocular anomalies like myopia, ptosis, and nystagmus, and gastrointestinal abnormalities, including colonic duplication and nonfixation of the colon” (p. 1). Individuals with CdLS often have feeding issues as well and need to be fed through a G-Tube because of inability to swallow. In diagnosing an individual with CdLS, the case will be sorted into classical or mild. In a mild case, the individual shows “less retardation of growth and intellect” (p. 1). Most of the cases of Cornelia de Lange Syndrome are sporadic and are due to a new mutation that occurs. There is a risk of reoccurrence in siblings, but that risk is less than 1 percent.

**Disabilities and Legal Rights**

Those with disabilities have rights, though sometimes those rights are overlooked. Human rights are applicable to all individuals regardless of having a disability or not. Those with disabilities are “entitled to human rights; in theory, everyone is entitled to their basic human rights, but “in both law and practice they [those with disabilities] have often been denied” (Mécret, 2008, p. 500). Though one may have a disability, that does not mean that they are any less of a human being. All individuals with disabilities have rights, though sometimes he or she may not be able to exercise those rights, e.g. voting, driving. Disabled people were not included
in the Civil Rights Act in 1964 (Fletcher, 2008, p. 523), further enforcing discrimination and positioning those with disabilities as a separate group of people, again as the other. Civil rights needed to become social rights, as the rights of a human being are the same across the spectrum. It was not until the 1990s that the civil rights of disabled people were challenged. Treating special needs people differently separates them from the rest of society; in turn creating a large inequality gap. Within human diversity there are disabled people, but, not all disability is the worst possible scenario for an individual. Disability can have a negative effect on the individual’s well-being, but in some cases, it can enrich his or her life “Someone might have been a lonely shut-in, with no friends and no community, before she became disabled. She goes to rehabilitation, where she makes a lot of friends” (Barnes, 2014, p. 92). All individuals deserve to be happy, regardless of classifications, whether that be disabled, African American, or woman; treat all as equals.

The 1960s were important for the history of disability. Without the Civil Rights Movement, disabled individuals would have been left as those still considered less than human. Within disability it is key to define the difference between disability and impairment “Disability then is the social extension and implications of a particular impairment or bodily configuration. Impairment refers to the injury, lack, or loss of particular bodily or mental abilities” (Livingston, 2013, p. 563). Socially, disability is the way an individual is seen by those around them, while impairment refers to an actual lack of something. Using the word ‘lack,’ makes the individual who may have an impairment seem less than human. Lacking changes how the world views a person with special needs. Often, pity is felt because of the circumstances of a disabled individual, but even then, disability is a social construct. In this context, one must see the individual as a person living with a disability, not just a disabled person. Harriet McBryde
Johnson, a disability rights activist, argued that though one may be disabled, he or she does not have any less quality of life, it is just a different life than many may have around that person (Weinberg, 2009, p. 1). On the other side of this, there are individuals such as Peter Singer, a philosopher, believes that there are certain circumstances where it is acceptable kill individuals who he believes are not whole, “individuals with cognitive impairment” (Johnson, 2003, p. 91). Although someone may be different than those around them, it does not make them any less worthy of life. Harriet argues that although Singer is advocating for genocide, Singer does not look at it as such. Instead, he sees it as a way of giving parents a choice when dealing with a child who may be impaired. It comes down to morality, Singer will not reinvent morality at this point in time, but he already is having an effect. Disabled fetuses are aborted in today’s society, often because the child will likely die before birth or shortly after because of a disability.

The Americans with Disabilities Act (ADA) was passed in 1990. The act forbids employers from discriminating against those who may have a disability. One may not be considered disabled to fall under the ADA, but he or she may have other impairments (Hoffman, 2017, p. 786). The ADA was a huge step for the protection of those with disabilities. It has allowed those with disabilities to gain more independence by allowing them to hold their own jobs if they are able to do so. However, the ADA is out of date and needs to be amended. Employers are able to get around some of the guidelines of the ADA because of ambiguous wording.

In 2005, it was said that nearly 33 million people in America were disabled, roughly 1 in 5 Americans (Verville, 2006, p. 455). The rate of individuals with disabilities has increased in recent years because of better medical care and living conditions within the United States. Almost 20% of America’s population is disabled. Yet, the health care of non-elderly disabled
adults is not where it should be, the focus is on disabled elderly people (Hiranandani, 2012, p. 2). Many children within the United States experience some sort of severe impairment and are in need of quality healthcare to increase their quality of life. Medicaid is one of the main insurance agencies for disabled adults. It is expensive to be disabled; often individuals need special things, such as wheel chairs or beds and none of this comes cheap. Without insurance, the quality of life of an individual with special needs in the United States would be quite low. Medicaid is actually providing eight million people with disabilities, under 65, with healthcare in the United States (p. 3). There are financial as well as educational struggles when one has a disability as can be seen in several Supreme Court cases.

In the next three paragraphs, three different court cases regarding special needs children and education will be analyzed. In 1982, Rowley versus the Board of Education of the Hendrick Hudson Central School District went to court regarding a deaf student, Amy Rowley. Rowley went to school in New York and prior to her first year of public school, her parents met with the school district in order to form an individual education plan (IEP) for Rowley. In the IEP, it was stated that Rowley would receive a sign-language interpreter in order to help her understand everything in the class. For the first two weeks of the class, Rowley had the interpreter until he decided that she was able to function well enough on her own, without the help of an interpreter because she had a hearing aid and was able to read lips, but her understanding was only limited. So, for the rest of the school year Rowley depended solely on herself for her learning. The next year of school, her parents went back for another IEP and her parents requested a sign language interpreter once again and this time the school district refused. Rowley would only be able to understand 60 percent of what was being said to her without the interpreter (Britannica, 2014, p. 1). Her parents then went to court against the school district, eventually ending up at the United
States Supreme Court. Under the Education of the Handicapped Act of 1974, disabled individuals have the right to “free appropriate public education” (p. 1). In smaller courts leading up to the Supreme Court it was ruled that Rowley was not actually receiving “free appropriate public education.” Once Rowley reached the Supreme Court, the Court overthrew this decision and said that Rowley was receiving “free and appropriate education” because “the intent of the Act was more to open the door of public education to handicapped children on appropriate terms than to guarantee any particular level of education once inside” (p. 1). The door was opened for Rowley to receive an education, but it was not the school districts job to ensure that she had as a good of an education as the rest of the students.

In 1985, another case entered the Supreme Court dealing with the Education for All Handicapped Children Act (EAHCA). Michael Panico was a young boy who was considered handicapped by the guidelines in the EAHCA. As a result of this, an IEP was created for Panico, but after a few years in school, it was decided that the school he was at in Burlington, Massachusetts was not equipped to deal with Panico. As a result of this, Panico’s parents decided to enroll him in a private special education school that would allow Panico to receive a better education. Because of this, there was a series of hearings that were held in order to see where Panico should be attending school. In 1980, the Bureau of Special Education Appeals decided that the best place for Panico was at the private school and that the Burlington school district needed to pay his tuition and pay Panico’s parents back for the money that they had already spent on the school. The Burlington school district was unhappy with this ruling and went to the federal district court to look at the case at hand. In 1985, the case made it to the Supreme Court. The court decided that the parents were correct in moving Panico to a private school since the public school was not fulfilling his needs and that the parents of Panico did not violate the
EAHCA. With this, it was decided, once again, that the parents should be reimbursed (Rehberg, 2014, p. 1). Michael’s parents acted as advocates for their child and they made the right decision.

In past years, it has been debated whether it is the parents or the schools job to prove that a child needs an IEP. In a 2006 Supreme Court case, Schaffer v. Weast, it was decided that it is the parent’s responsibility to look at the IEP given to them and decide whether or not it works for their child and if it does not work, it is the parent’s responsibility to prove why the IEP does not fit. The parents in the Supreme Court case tried to argue that it is the schools job to “justify the appropriateness of a disabled child’s IEP” (Parents, 2018, p. 1). This idea and the parents were overruled at the Supreme Court. Within this, the Individuals with Disabilities Act, previously known as the Education for All Handicapped Children Act, has become an important guideline in helping individuals with special needs get the education that they deserve.

**Education for those with Disabilities**

Education of those with disabilities is often disregarded as less of a priority than that of the education of typical functioning individuals. This is a common occurrence all over the world, therefore, it is important to remember and focus on the rights of disabled children in regard to their education. Today, 85% of children with disabilities, specifically classified as mentally retarded (MR), are unable to make use of the normal educational resources (Altinay, 2018, p. 2). The world is not designed for disabled people. Special education for those with disabilities is needed in order to cater to the specific individual’s needs, allowing them to succeed later in life. If the mentally handicapped child cannot succeed in a mainstream classroom, a special education classroom is needed. Awareness is key in order to treat all students equally based on his or her needs. Many low-income countries do not treat disabled children with equality as they should “disabled children and youth appear to have very unequal access to education compared to non-
disabled peers” (Croft, 2013, p. 233). In a Multiple Indicator Cluster Survey by UNICEF, it was found that children who did not attend school tested more likely to have a disability than children who attended school. The education of all people helps not only the individual, but society as well.

In foreign counties, such as Bangladesh, those with disabilities are not as well off as normal functioning individuals in categories such as “employment, material wealth, education, health, access to development assistance and poverty relief, and in social well-being” (Ahmed, 2015, p. 94-5). It is in categories such as these, that put an already disadvantaged individual at even more of a disadvantage. There is a lack of education for disabled individuals all over the world, specifically in developing countries, resulting in neglect. In Bangladesh, 18% of teachers in a research study believe that if a child with disabilities gains an education, he or she will be able to get a job, allowing them to be productive members of society, (p. 95). Humanity, as a whole, runs on the contributions by working members of society and in working towards the education of disabled, even more members of society will become working members. In the education of the group of disabled people, education will allow them to find jobs and in turn become financially independent (Cherevyk, 2016, p. 779). In Kyrgyzstan, in the Constitution, there is a law that states there may be no discrimination and there is a right to education for all of the people of Kyrgyzstan. Disabled individuals “should” be able to receive higher educational parts, such as vocational and technical education, within this, it is vital to understand that only there are around 155,000 disabled people in Kyrgyzstan and a small number of college students in the country are disabled because many disabled people in the country do not receive education at the secondary level, “children with physical disabilities who could well be successful at school are practically cut off from access to education and opportunities to socialize with their peers”
The government is willing to work for the education of disabled people, but only on paper and in theory. These individuals are left to defend for themselves because they are different.

In Ghana, disabled individuals are more likely to be illiterate than the typical children because of “disabling social and physical environments” (Gregorius, 2016, p. 323). It is thought that 98 percent of disabled young people in Ghana do not attend school and, for that reason, the education of disabled young people needs to reach the forefront of international agenda. Around the world, whether a developed or developing country, disabled individuals are not treated equally, although laws may state otherwise. There have been different initiatives to try and combat the lack of education for disabled people such as the Education for All agenda, but agendas like this do not always work in practice (p. 323). Education is necessary for the bettering of humanity, regardless of being disabled or not. In Vietnam in 1986, the doi moi policy was passed which led to a transformation and debate on the special education system in the country at the time. In the country, there is not necessarily a lack of understanding, but lack of ambition on how to teach children with special needs. In the Inclusive Education program, there are a wide range of students who deal with things such as brain injuries, Down’s syndrome, and attention deficit hyperactivity disorder (Rydstrom, 2010, p. 82). In a study by Helle Rydstrom, she discovered that there were different levels of education depending upon the severity of the special needs individual. Vietnam has a literacy rate of 94 percent, making it easy to see how important education is to the country. In 1995 the responsibility of the education of the disabled transferred from the Ministry of Labor, Invalids, and Social Affairs (MOLISA) to the Ministry of Education and Training (p. 84). Disabled children were no longer classified as “invalids” under the MOLISA, but instead as another group of individuals who needed an education. The Inclusive Education became the only way to send a child with disabilities to school from a low
socio-economic family. The Inclusive Education program works to integrate special needs children into the education system, but within this, the teachers also need to be trained on how to educate a disabled child. North Korea on the other hand, does not accept disabled individuals, although their laws may say they do. In the country, it is thought by many North Koreans that it is just better for the disabled to be left to die, resulting in infanticide. In 2006, it was found that “all children who were severely disabled were left to die without being nurtured” (Won, 2017, p. 163). North Korea is just one example of disabled individuals all over the world.

In 1975, in the United States, the Individuals with Disabilities Act (IDEA) passed (What is, 2018, p. 1). This Act was created in order to allow disabled children to receive special education services. It is a federal law, but it is thought that the Individuals with Disabilities Act cannot reach its full potential until parents or caretakers of individuals with special needs are given more attention. The connection between the child and the education system is the parent, and it is important for the parent to know what resources he or she has in regard to the child (Czapanskiy, 2014, p. 733). Within the Individuals with Disabilities Education Act, children with disabilities are entitled to “free and appropriate public education.” Though the child may be entitled, he or she may not actually get that public education they have been guaranteed. If a parent has a child with special needs it is their responsibility to establish that their child does need a special education. The parent is the child’s advocate and in being the advocate the parent has to manage the child’s life (p. 736). Although it is the parent’s responsibility to take care of the child with special needs, they still need support, whether that be in the form of knowledge or just another set of hands. The special education system is important for those with special needs, but in a book by Ruth Colker, a parent of a child with special needs, she found that the “special education system does not work particularly well for any children, but works particularly badly...
for poor and minority children” (Yell, 2015, p. 495). Although Colker is adamant that the special education does not work well for any special needs children; it becomes important to understand that prior to the Individuals with Disabilities Act children with disabilities were basically excluded from education or a part of the education, even though it was not catered to them in any way, shape, or form. The Individuals with Disabilities Act has helped dramatically with the education of special needs children and 86 percent to 90 percent of parents are satisfied with the special needs education system at this point in time (p. 497).

**Caretakers of the Disabled**

Often, parents or caregivers of special needs do not put the individual into a home or facility and instead take care of them for as long as they possibly can. There are a multitude of challenges that a caregiver faces when taking care of an individual with disabilities. The challenges only become greater as both the caregiver and individual with special needs grows older “challenges include the extended duration of their caregiving role, lifespan health issues that arise from their own and their care recipient’s often complex aging process, and anxiety for the future well-being of their care recipient after they die or discontinue their caregiving role” (Perkins, 2010, p. 409). Because of medical advancements many adults with intellectual disabilities are living longer, whereas in previous years the caregiver was outliving the individual with intellectual disabilities. As a result of this, many caregivers, are caregivers their entire lives and do not receive a break. In having the same caregiver for a large portion of the individual with disabilities life, the individual does not have to become comfortable with a new person tending to them. Caregivers are extremely important to the lives of disabled individuals because many would not be able to survive without a caretaker. Many caregivers experience higher levels of “stress, anxiety and depression compared to other parents” (Lunsky, 2017, p. 1374). For most
parents, they have a child or two and then have them for around eighteen years until the young adult can successfully be on their own and independent from their parents. This is not the case for individuals with disabilities; they will constantly need a caregiver and as a result of this, parents are not just focused on their own future and what they will do after their typical children get out of the house. These parents will have to plan ahead for their child with disabilities in case something happens to them.

Parents or caregivers of those with disabilities often do not just want to put the child or adult in an institution, but instead would like a little bit of extra help in taking care of the individual. In 1963, the Netherlands created a day care just for those with intellectual disabilities. The role of these centers was to give the parents a break from their child while allowing for development for the child with disabilities (De, 2006, p. 204). Children with disabilities will become attached to their caregiver at their day care or other center. This is a good thing because it allows the child to accept other people in his or her life. Typically, as time goes on, families are always changing to deal with different things that may be happening in their lives. Children with disabilities are often dependent on the caregiver, but individuals with disabilities are not always just passive recipients. Parents of children with disabilities frequently experience sorrow and loss because they do not have a typical child, but this is not always the case “children with disabilities can grow as active agents in their own lives […] their children can also become partners with their mothers and self-advocates who see to take charge of their own lives in the midst of life challenges” (Lee, 2015, p. 3673). It is not always a bad thing to take care of an individual with special needs because he or she can help the caretaker grow as a person, showing strength and courage.
Respite care is a type of care that many primary caregivers take advantage of because it is a type of temporary relief in which an individual with special needs attends some sort of institution. It is difficult to take care of a person with special needs and eventually there will be consequences on the caregivers physical and mental health if they are with the same person continuously, day in and day out. There are benefits to having a child with special needs attend respite care “access to regular, high-quality respite care is known to have a beneficial impact on a carer’s ability to continue in their caring role” (Mansel, 2009, p. 256). Typically, couples, siblings, and even parents cannot be with each other all of the time. The same thing goes for caregivers and the individual with a disability. It is important to have time apart for the mental stability of both individuals; it can be enjoyable and beneficial for the two. Respite can be difficult to come by because there has to be a large need in order for there to be respite providers in the area in which one lives. Though caregiving for an individual with special needs is a demanding process, it is not necessarily the only job a caregiver may have “Many experience conflicting demands between paid work and caregiving and have to miss days at work or reduce their paid work hours” (Fast, 2015, p. 1). As a result of missing work, the caregiver may be at risk for losing their job, further resulting in “poor social, economic, physical, and mental health outcomes” (p.1). This is difficult for families because the caregiver needs to care with the individual with disabilities, as well as keep a job in order to support the family. For this reason, respite care is extremely important to families with disabilities.

Often, caregivers of an individual with disabilities often have other children to take care of as well. It can be difficult for a sibling growing up with a brother or sister with special needs, and it is important to remember that they often also have a caregiver type role in the family. Siblings of those with disabilities cannot be counted out, these individuals often grow up to fight
for disability rights “siblings of individuals with disabilities reported providing daily support, facilitating employment opportunities, and advocating for disability rights” (Burke, 2016, p. 736). In having a sibling with a disability there is often more contact and closer relationships between the siblings; the relationship shared is a positive one. Sisters of siblings with disabilities versus brothers, have a stronger relationship with the disabled sibling (p. 736). Individuals with disabilities are now outliving their parents thanks to medicine and “the adult service delivery system lacks the capacity to meet the needs of aging individuals with disabilities” (p. 737). As a result of this, families are taking care of the individuals with special needs long term and siblings are taking over the role of caregiving when the parents die. People do not think of the caregivers of special needs individuals and if they do, many only think of parents and not other family members.

The issue with siblings becoming the main caretaker after parents die, is that the individual with special needs is not their child and taking care of a disabled sibling should not be the only option that he or she has for the individual with special needs. If the siblings share a positive relationship with another it becomes expected by both parents and the sibling that they will soon be the one in charge of care of the individual with special needs “[…] positive relationships in childhood and adulthood are not only expected by their parents, but also have self-imposed expectations to coreside [sic] and provide future caregiving” (Saxena, 2015, p.214). In taking in a sibling with a disability, there are also other circumstances to consider, such as, the sibling getting married and the individual with special needs becomes a responsibility to not only the sibling, but the new family as well. Many individuals do not want to willingly step into the role of caregiving, and may give the sibling a difficult time about taking in their sibling with a disability. Financial resources are often strained in a family with an individual with special needs
because “mothers were more likely to be employed part-time and earned less than mothers with typically developing children” (p. 215). Children with disabilities are expensive because of doctors’ appointments and other resources the individual may need. The burden of an individual with disabilities would be passed from parents to siblings, which may in turn cause siblings to give up on some of their life goals, as well as, personal interests.

Research Question

Do caretakers of those with disabilities, specifically CdLS need to be given support as well as resources in order to maintain a healthy environment and family for the individual with special needs?

Theoretical Framework

Mark Oliver, a disability advocate, first introduced the social model of disability following the Fundamental Principles of Disability document, (Oliver, 2013, p. 2). The social model of disability was first expressed during the 1970s by the Union of the Physically Impaired Against Segregation. In 1976 disability was defined as:

“a situation, caused by social conditions … In our view, it is society which disables physically impaired people. Disability is something imposed on top of our impairments by the way we are unnecessarily isolated and excluded from full participation in society. Disabled people are therefore an oppressed group in society.” (Fletcher, 2008, p. 527)

The social model of disability has been a big step in helping to define disability. Within the model there has been discussion between civil and social rights. Often, disability is thought of as a social construct further pushing the bounds of just what a disability entails. The social model of disability has worked in past years to help create an equality of understanding in disabled individuals. Those with disabilities are often oppressed in today’s society and the social model
has done great things to bring light to oppression that is faced. Oppression faced by those with impairments directly relates to caregivers who are dealing with the one being oppressed. If the individual is oppressed, it makes life on the caregivers more difficult because they are forced to work around what may be oppressing the individual. Although disabilities are often seen as something negative, that could not be farther from the truth. Like any human being, a person with a disability does struggle in life at different points, but they are still a human being. There are positives that come out of living life as a disabled person, often those positives are the impacts those individuals are able to make on those around them. Caretakers of those with disabilities are also affected by what happens to the disabled individual and they are the ones who get the highest reward of spending time with a disabled person.

Derrida’s deconstruction method discusses humans trying to make meaning out of the world. Derrida focuses on the binaries and presence without hinting of what it is not. Within deconstruction there is structure. Structure focuses on play. Play is about relationships and how without play there is no meaning, yet play is variable. This means that there is nothing concrete in life and nothing is relational. Within the structure there is a center, but the center is ever changing. A center organizes the world, but the center is always collapsing in on itself creating a new center. The caretaker of an individual with special needs is the center of that individual’s life. Without the center, everything around collapses releasing play. When the individual with special needs loses their caretaker, their center, a new center is created, but causes disruption in the individual’s life. The structure of the individual’s life depends on the social constructs of disability created by society. If society does not look at the caretaker as a caretaker, then the center collapses, but the center will collapse no matter what, it is only a matter of time. An individual with a disability can also be looked at as their own center. If the views they have of
themselves changes, then the center collapses and a new one is created, releasing play. Play is what creates meaning, creating a center. This is a never-ending circle brought to light by Derrida

**Statement of Purpose**

The purpose of this research project was to critically analyze caregivers of individuals with Cornelia de Lange Syndrome. There is a gap in knowledge regarding caregivers of those with disabilities because research is mostly focused on the disability itself. If research does focus on caregivers, the research is on the “sexy” diseases that are more common, such as ADHD, ALS, and Down’s Syndrome. Disease ranges across cultures, genders, and socioeconomic status and because of that there are a wide range of caregivers. Each disease has certain characteristics that make the disease the disease it is. There is not a universal caregiver that can just take care of anyone with any disease, each caregiver has to accommodate the individual they are taking care of. I have a sister with Cornelia de Lange Syndrome, making me a caregiver of the syndrome.

**Justification**

Cornelia de Lange Syndrome is a disease that crosses cultures, race, gender, and income. Disease does not discriminate, and it is important for all education levels, regardless of specific interest, to be aware of disease. This topic is important to study in the English field because diseases affect all of humanity, regardless of education or status. Often the study of English is thought to contain only literature, but all documents allowing for further research on specific areas, such as CDLS, contribute to the field as a way to broaden the English canon. There is a discrepancy in knowledge regarding caregivers of Cornelia de Lange Syndrome. This research specifically looks at caregivers of CdLS. In the study, *Cornelia de Lange Syndrome: In the Realm of Caregivers and Surviving*, it was found that caregivers would like more resources for their child, whether that be awareness, respite, or long-term care facilities. The caregivers are the
center of the individual with disabilities life and the caregivers need support, as well as resources, in order to reach full potential in society, while taking care of an individual with special needs.

**Methods**

This study was conducted as a qualitative content analysis of caregivers of Cornelia de Lange Syndrome answers to the research questions mentioned in the preview section. This method was used to analyze caregiver’s opinions on their abilities and access resources to take care of an individual with Cornelia de Lange Syndrome. A survey titled, *Cornelia de Lange Syndrome: In the Realm of Caregivers and Surviving*, was placed on the Cornelia de Lange Syndrome Facebook page, asking for caregivers of CdLS to participate in the study.

Different is often viewed as wrong or bad in society, therefore, those with disabilities are looked at as less than human. When one sees a disabled individual in society, he or she will often just look at the individual and the thought of who takes care of the individual will never cross his or her mind. It takes a village to take care of an individual with special needs, specifically an individual with Cornelia de Lange Syndrome. In taking care of a special needs person, one will face hardships and difficulties because their child is not “typical” and will not have the same milestones that a typically developing child will have. There is quite a lot of research dealing with specifics of disabilities and the different types of disabilities, but there is a small amount of research that has been done regarding caretakers of those with disabilities. This study was conducted in order to gain a better understanding of the needs of a caretaker with CdLS, and what other types of support that he or she could benefit from. Cornelia de Lange Syndrome is a rare congenital disorder, with little research regarding caregivers of the syndrome. It is for this reason that I conducted this research and put a survey on the Cornelia de Lange Syndrome
Before entering the survey, the potential participants read a short intro into why this research was being conducted. The intro was as follows,

“Hello! My name is Madissen Hester and I am an undergraduate student at the University of Wyoming. My sister, Emily, has CdLS and I am currently working on a project/thesis, so I can receive my Honors Minor from the University. The purpose of this research project is to critically analyze the caregivers of those with Cornelia de Lange Syndrome. It is for this reason that I come to the Cornelia de Lange Support Facebook page. I think that this group will give me the best feedback regarding taking care of an individual with CdLS. At the end of this post, I have attached a link to a survey that I would like any of you who are caregivers of an individual with CdLS to complete. I know surveys can be tedious and time consuming, but the survey is only 10 short questions. I would greatly appreciate your feedback and I would like to thank all of you who have stepped up and taken care of an individual with CdLS. It is not always an easy task. Once again, thank you and thank you for your feedback!”

Survey questions can be found in Appendix A (see p. 32).

**Results**

In the research study, there were 32 participants and on average 25 of the participants answered each question, with only 7 participants skipping questions. With the first question, “I feel like I am capable of taking care of an individual with CdLS,” 23 of the participants answered Agree or Strongly Agree, while only one participant answered Disagree. In the second question, “I feel like I am not capable of taking care of an individual with CdLS,” 23 participants answered Strongly Disagree and Disagree, with 56 percent of participants answering Strongly Disagree. It was expected that these two questions would have similar data, specifically with the first
question being positive and the second being negative, the answer was ultimately the same; the study asked the same question, twice, in a different manner each time. In the third question, “I feel like I do not have resources,” there appeared to be hesitancy in answering the question. Eight participants answered Neutral for this question, leaving the other participants to be spread out across the board. Three participants Strongly Agreed that they do not have resources and three also Agreed. The last eleven participants answered in the Disagree, Strongly Disagree section, with only one participant answering strongly disagree. Similar numbers followed for like question, “I feel like I do have resources.” Seven participants, answered Neutral, down one from the previous question. Sixty percent of participants answered in the Agree, Strongly Agree section, while only 12 percent answered Disagree. When asked if the participants felt like they did not have enough resources, 24 percent agreed, but when asked if the participants felt like they did have resources, only 12 percent disagreed. In the following question, “Extended family is helpful with the individual with CdLS,” 76 percent Agreed/Strongly Agreed, while only 2 percent, Strongly Disagreed/Disagreed, with no participants answering, Neutral. In the sister question, “Extended family is not helpful with the individual with CdLS,” 3 participants selected neutral. 16.67 percent answered Agree/Strongly Agree while 70.83 percent answered Disagree/Strongly Disagree. In the two questions, 2 percent of participants said that they did not agree that extended family was helpful when taking care of an individual with CdLS, but when asking the question in a different manner, by saying that extended family is not helpful with taking care of an individual with CdLS, 16.67 percent agreed.

The last two Likert Scale questions were as follows, “The government is helping me financially take care of an individual with CdLS,” and “The government is not helping me financially take care of an individual with CdLS.” Similar numbers did follow for the two
questions. Fifty-six percent of participants either agreed or strongly agreed that the government is helping them and 28 percent either disagreed or strongly disagreed, with 16 percent neglecting the answer. Fifty percent of participants disagreed that the government was not helping them take care of an individual with CdLS, while 33.33 percent agreed or strongly agreed, while 16.67 percent answered neutral.

The final question was an open-ended question allowing for individualized responses. A total of 21 participants participated on the question, while 11 participants skipped. The question was as follows, “How do you think you could receive better assistance in taking care of an individual with CdLS? What kind of support would be beneficial for you and your family regarding the individual with CdLS?” The answers to the open-ended question can be found in Appendix B (see p. 33).

The answers to the open-ended questions, though varied, do have commonalities mentioned with in the answers. The most prominent mention was to have some sort of residential option for the caretaker of the individual with CdLS, with five respondents making a reference if not directly, indirectly in their answer. In the categories, therapy access, respite, and a better understanding of professionals, e.g. doctors, therapists, teachers; a combined twelve participants mentioned each category, four, four, and four, respectively. Three participants mentioned awareness in some way and another three participants mentioned support in one way or another. Though only two participants mentioned speech, it cannot be counted out because speech is such an important part of the social hierarchy. All of these categories can be combined into one overhead, resources. Caretakers of individuals with CdLS need help, whether that be respite, more awareness, or even knowledge.
Discussion/Analysis

In this section, I will prove that there is a gap in knowledge regarding caretakers of the disabled, specifically, Cornelia de Lange Syndrome. In this research, I critically analyzed caretakers of individuals with CdLS and was able to prove my research question, that the caretakers of individuals with CdLS need to be given support, as well as resources, in order to better support the individual with CdLS. Individuals with disabilities are often oppressed in today’s society and the social model of disability has worked to combat that oppression. In my research, it was found that the caretakers, although not disabled, are oppressed by the disabled individual in their care. The social model of disability takes on disability, but not the caretakers behind disability who are the ones that the individual with disability could not live without. In Derrida’s “structure,” he discusses a center, the center being what organizes the world, yet the center is always collapsing. The caretaker of an individual with special needs, is the center of that individual’s life. So, if the caretaker was lost, or interchanged with another caretaker, a new center is created, which causes chaos or disruption in the individuals life. Multiple participants in the research shared that their child is in their twenties and that they are looking for options for their child, whether that be different “living arrangements” or just better access. When and if the parents of these children do reach the spot where the child moves to a new location, with a different caregiver, their center will collapse. This will be traumatic in the child’s life, but it is something that the child will have to deal with. Within this, these same participants as caretakers, are being oppressed in their search for options for their child. Most parents are responsible for their child until they reach the age of eighteen and then the child can be on their own. This is not the case for special needs parents or caretakers, in a harsh way, these parents are stuck with the child who can never be fully independent. This leads to a break in the natural cycle of parenting,
causing the parents to feel stuck and without options, proven in the research study. Within the study, it was found that many parents often struggled with resources for their child because of living in a rural area, “we live in a small town in Ohio so anything that we need we have to go to Cleveland for medical issues and sometimes dentist appts.” This in turn, caused another form of oppression for both the caregiver and the individual with CdLS. The caretaker is unable to fully help the individual with CdLS because of a lack of resources and the individual may miss out on certain therapies as a result of rurality. Two participants in the study mentioned living in a “rural” or “small” town and having to travel anywhere from 30-45 minutes just to reach therapy options that are necessary for the individual, “I live in a rural area. My daughter is intellectually disabled and would benefit from tutors, art classes and more therapy options. I have to drive 30-45 minutes for all of those options.” Many of the participants who answered the open-ended question cited issues within therapy and one participants said that their son ran out of speech therapy by age 3. In the area where that individual lives, each child has a small allotment for the amount of therapy that they can receive through the school district. In the case of a particularly severe child, he or she does not receive as much therapy as they should because of a lack of resources by the school district, another form of oppression for the child. Without the correct resources, an individual with CdLS cannot succeed, further their caretaker cannot succeed in allowing them to reach their potential.

In the study, a few of the participants were from foreign countries, this is known by the way of spelling certain words such as, “furtunate, programmes, cheque, proloquo” (errors are in original). One participant specifically cited that they currently live in India with their daughter with CdLS, “In India, support groups are very rare and people don’t want to seek support or disclose these kind of rare diseases of the loved one. I have personally tried to get in touch with
families to extend/ seek and share knowledge gained raising my daughter with CdLS.” This participant mentions not being able to seek support or discuss a rare disease like CdLS. The reason for this is because India is a collectivist society. This means that the society focuses on the needs of the group, rather than the needs of an individual, “In collective cultures, the individual is emotionally dependent on organizations and institutions, and group membership is emphasized,” (Samovar, 2017, p. 186). America, on the other hand, is an individualistic society, meaning that the individual is more important than the social setting. This is why the participant from New York, is able to have resources and services, as it is more acceptable to be disabled in United States over places like India,

New York is a great state of any resources or services. I have come to realize we are able to get many more things to help my son that I unfortunately not been available to other families and outside states. The only thing wish [sic] we had more access to your doctors that were more familiar with this engine. We do travel once a year to see doctors that specialize in it, but wish we had access to these doctors. (interview participant, 2018)

In having a disability, an individual is considered an outcast and not a part of the group any longer in places like India. This is a way of saving face. In 1990, a study was conducted in Taiwan dealing with the concern over human feelings. In expressing feelings, individuals in Taiwan, another collectivist society, experience fear in expressing emotion, “fear of expressing love; fear of talking about sex; fear of talking with authority figures; fear of revealing one’s own shortcomings” (Jia, 2003, p. 50). The key part of this statement is the fear of shortcomings. An individual with a disability, specifically CdLS, would be considered an individual with shortcomings because they do not fit into the typical model of a human being. Since the individual does not fit the typical model, he or she will have issues fitting into a collectivist
society, which is why the participant from India is unable to find support. The participant and child with CdLS from India would not be completely shunned, but they would not be looked upon as accepted and would be faced with particularly “cold faces of community members” (p. 51) because “face concern still seems to overcome the self” (p. 51). An individual who looks different or has separate needs from the society, is no longer collectivist, but instead, individualistic. India is a collectivist society that values the group over all else, so it would seem that they would be accepting and welcoming of someone who is different, but that is not the case. Face remains most important in a collectivist society.

There is a gap in knowledge regarding caretakers of individuals with CdLS. Often the caretakers are left on their own with the child, with few resources, causing a form of oppression on not just the child, but the caretaker. It is difficult to take care of a disabled individual, no matter where one may be in the world. However, in places like India it may be even more difficult. The participant in India did not have support in taking care of their child, which causes unnecessary stress both physically and mentally on the caretaker. Individuals with CdLS are considered to have shortcomings, and in places like India, disabled people are often marginalized. Caretakers need resources in order to fully be able to support the individual with special needs and the individual also needs resources, in order to reach their full potential as a human being. The gap in knowledge by the public is prominent, caretakers and the disabled alike, need support and resources in order to be successful in life.

Conclusion

It is safe to say that caregivers of Cornelia de Lange Syndrome need support as well as resources in order to fully fulfil their role as caregiver. Caregivers cannot be left to only rely on themselves, regardless of country, status, or wealth. Those with disabilities, specifically CdLS
deserve caregivers who are equipped and feel equipped to care for them, but it is also important to not forget that individuals with CdLS need their own resources, such as therapy, regardless of how much they are allotted, they still need it. Although society may make it appear that those with disabilities are oppressed, it is truly a case of oppression on both sides, on the disabled and the caretaker. This is not just a national issue, but an international epidemic that is disabling, disabled human beings. Ultimately, disability is a matter of perception, it is not simply just a disease, but a way of allowing society to oppress human beings.
Appendix A

Survey questions 1-8 were on a Likert Scale ranging from 1 – Strongly Agree to 5 – Strongly Disagree. Survey question nine was an open-ended question that allowed for feedback by the caregiver of an individual with Cornelia de Lange Syndrome. The questions are as follows:

Survey Question 1: I feel like I am capable of taking an individual with CdLS

Survey Question 2: I feel like I am not capable of taking care of an individual with CdLS

Survey Question 3: I feel like I do not have resources

Survey Question 4: I feel like I do have resources

Survey Question 5: Family is helpful with the individual with CdLS

Survey Question 6: Family is not helpful with the individual with CdLS

Survey Question 7: The government is helping me financially with the individual with CdLS

Survey Question 8: The government is not helping me financially with the individual with CdLS

Survey Question 9: How do you think you could receive better assistance for your child?

What kind of support would be beneficial for you and your family regarding your child with special needs
Appendix B

1. “My daughter has mild CdLS so we are very fortunate [sic]. We get let's of help from our government in providing home support and share a break programmes [sic]. Access to SLT and OT is unfortunately very limited, these are the only areas where we wish we could have more support.

2. “More nursing hours.”

3. “Respite care before age 3.”


5. “Support from surrounding people, especially my parent…”

6. “In India, support groups are very rare and people don’t want to seek support or disclose these kind of rare diseases of the loved one. I have personally tried to get in touch with families to extend/seek and share knowledge gained raising my daughter with CdLS”

7. “More resources for siblings and how to tell their friends about their sister (who has CdLS) at various ages.”

8. “Respite”

9. “My daughter will be 22 next year and will be out of the school system. We need more programs for kids when they reach this age.”

10. “I live in a rural area. My daughter is intellectually disabled and would benefit from tutors, art classes and more therapy options. I have to drive 30-45 minutes for all of those options.”

11. “Not specific to a child w[ith] cdls, more to the overall behavioral special needs population; if there were more resources and workshops available at good pricing.”

12. “Family counseling would be great.”
13. “New York is a great state of any resources or services. I have come to realize we are able to get many more things to help my son that I unfortunately not been available to other families and outside states. The only thing wish we had more access to your doctors that were more familiar with this engine. We do travel once a year to see doctors that specialize in it, but wish we had access to these doctors”

14. “Therapists need to be more aware of the syndrome”

15. “We live in a small town in Ohio so anything that we need we have to go to Cleveland for medical issues and sometimes dentist appts. But I would say everything we have had and have now is pretty beneficial”

16. “For us speech is our biggest issue. The school board in our area only has so many hours allowed per child. We ate through that therapy by grade 3. Nicholas is 18 and should start receiving a disability cheque [sic] which will help a great deal and maybe we can use some of that to pay for private therapy. He is non verbal, has few words, using proloquo [sic] and some signs. Understands way more than he lets on! He is he light of our lives and has brought our family so much joy!!”

17. “Full time care facilities would be nice”

18. “More training for teachers and professionals at school to help with developmental outcomes.”

19. “Long term care is my greatest concern. With the elimination of many group homes we are finding it difficult to find living arrangements for our 20yr old son.”

20. “Increase awareness of CdLS. Increased awareness of how Medicaid assists families of individuals with needs. Respite care is essential. And just an overall understanding that
most parents of special needs kids are doing their absolute best to manage, but that just about anyone would need help - whether financial, emotional or physical.”

21. “It would be wonderful if specialty doctors had better knowledge of CdLS. I always have to bring special growth charts to my daughter’s appointments to show that she is growing, but at a different pace”
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