

**ANALYSIS OF SUPPORT GROUP AVAILABILITY FOR ADULT-ONSET  
NEUROLOGICAL DISORDERS IN THE CENTRAL APPALACHIAN REGION OF  
THE UNITED STATES**

by

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**ABSTRACT**

Huntington disease, Amyotrophic Lateral Sclerosis, Parkinson disease, and Alzheimer disease are four multifactorial, neurodegenerative, inheritable disorders affecting thousands of people collectively in the United States. The importance of public health measures such as support groups for both patients with one of these diseases, as well as caregivers, is high. Support groups are beneficial for the patients and caregivers, and are also vital to the well being of the patient. Support groups also serve to address important public health services in the United States.

I examined the number of counties with at least one support group available for each of these four diseases throughout the central Appalachian region of the United States. First, the total number of counties with at least one support group throughout each state in this region was assessed and subsequently compared to the number of counties with at least one support group within the Appalachian region of each state. The number of counties with at least one support group available to individuals living within the Appalachian region of these states was significantly lower than the rest of the state (34 vs 61%, respectively, P-value < 0.0001). In Appalachia, if the county was economically distressed, it was also significantly less likely to contain at least one support group compared to counties that were not economically distressed (19 vs 46%, respectively, p-value < 0.0001).

My results indicate the presence of health disparities regarding disease support groups, especially for diseases with lower prevalence rates such as Huntington disease and Parkinson disease. The addition of more patient support groups in this region may have many benefits, including increased awareness and education, and more patient and caretaker empowerment. These benefits may in turn impact the overall health of a region that historically has had issues with healthcare access and availability. I recommend that more research into this potential disparity is performed to investigate it further.

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## **PREFACE**

I would like to thank Dr. Durst, Dr. Kammerer, and Professor Bjerke for their valuable input and advice in constructing this essay. I would also like to thank Dr. Kammerer for advising and guiding me throughout my graduate education. I am also extending my gratitude to the Allegheny County Health Department, and particularly to Stacey Randolph and Jaime Sokol for allowing me to perform the work that helped to inspire this project.

## **1.0 INTRODUCTION AND IMPORTANCE OF DISEASES**

The purpose of this essay is to examine patient and caregiver support groups for those affected by adult-onset neurological genetic disorders. The focus is on four specific diseases that (1) display a plethora of neurological symptoms, (2) typically affect middle to advanced age adults, and (3) carries a significant risk of familial heritability. These four diseases include Huntington disease (HD), Amyotrophic lateral sclerosis (ALS), Alzheimer disease (AD), and Parkinson disease (PD). These four disorders were chosen due to their prevalence in the United States, the individual epidemiology of each disorder, the similar progression and treatment options, and the connection between these diseases and support groups.

My research focused on the central and northern states of the Appalachian region of the continental United States: West Virginia, Pennsylvania, Ohio, Maryland, Kentucky, and Virginia. This specific area was chosen to assess the potential dichotomy of health care access and support that exists between economically privileged and distressed areas of the region. An evaluation of location of support groups, and the economic status of the area in which they are located was conducted. A review of the benefits of support groups for patients and caregivers was performed.

## **1.1 DISEASE PREVALENCE AND PUBLIC HEALTH SIGNIFICANCE**

One of the many reasons that the four specific neurological diseases discussed in this essay were chosen had to do with the relatively low prevalence of some of the disorders. Support groups are a vital resource for patients and caretakers, and these groups tend to be fewer in number and lower in availability for diseases that have a lower prevalence rate (Molster, et al., 2016). This creates a disparity for those afflicted with or by these disorders.

In the U.S., the prevalence rates for Huntington disease, Amyotrophic lateral sclerosis, and Parkinson disease are 1 per 10,000 people, 2 per 100,000 people, and 13 per 100,000 people respectively (Pringsheim, et al., 2012; Ferraiuolo, et al., 2011; Samii, et al., 2004). These rates are quite low when compared to Alzheimer disease, that affects one in nine people over the age of 65, and over 5 million people in the United States have. Alzheimer disease is also the 6<sup>th</sup> leading cause of death in the United States (Gaugler, et al., 2015). This disease comparison makes for interesting results when investigating the support groups for each given disease, and further highlights the dichotomy that exists between diseases with higher prevalence and those with low prevalence.

The lack of support for disorders of lower prevalence is of major public health significance because support groups are integral to patients' and caretakers' well being, and are an important community and healthcare resource. These groups provide vital disease information to patients and their families (Locock & Brown, 2010). The problems that are associated with the lack of patient support are magnified in areas of the United States with higher poverty rates and more geographically isolated areas, like the Appalachian region. Other issues, such as healthcare

burden, also contribute to the need for patient support groups in this region. The Appalachian region has a physician and provider shortage, as well as limited hospital bed space available (Lane, et al., 2012).

In addition to the healthcare burden and lack of access, a general lack of education, combined with the low health literacy rate throughout the United States contributes to the healthcare environment of Appalachia (Kutner, et al., 2006). The healthcare challenges of this region are unique and thus an assessment of the impact of the four neurological diseases on the general well-being of the region is critical to develop possible interventions, such as increasing the number of support groups, to mitigate some of the health care burden of disease. However, it also makes this region one that is unique and interesting to research.

## **1.2 SIGNIFICANCE OF GENETIC COUNSELING AND GENETIC TESTING**

Since all the neurological disorders discussed in the essay can be inherited, it is important to touch on the aspects of genetic counseling and testing. Genetic testing is a complicated and highly personal decision, and not every patient or family may be interested in pursuing available genetic testing. However, in order for a patient or family to make an informed decision regarding genetic testing, it is advised that they seek the advice of a certified genetic counselor, if they choose to receive information about genetic testing

Genetic counselors are health professionals with training in counseling and genetics who work as part of a healthcare team including physicians, nurses, and other healthcare providers (NSGC, 2016). Genetic counselors provide information and support to families and individuals with established genetic disorders and to those who are at risk for developing these disorders.

They identify risk within families, investigate this risk, interpret information and educate patients and their families about the disorder, risks, and potential heritability (NSGC, 2016). They also review options with the individual or family, which may include genetic testing.

### **1.3 ECONOMIC BURDEN OF DISEASES**

Although HD, PD, and ALS diseases have a low prevalence rate, and are rare in the general population, their economic burden is substantial. Most of this burden is due to outpatient costs and most of these costs are borne by the affected individual and his/her family. These costs vary greatly among patients depending on disease stage, symptoms, available resources, and insurance coverage. A large encumbrance is placed on society as well.

The per patient cost of Huntington disease may be large, mostly due to outpatient services such as nursing homes. Because more late-stage HD patients utilize these services, the cost per patient increases with disease progression. Divino and colleagues (2013) assessed the individual patient costs by comparing individuals using the Thomson Reuters' MarketScan commercial insurance and Medicaid databases from the years 2002 through 2009. Among a sample size of 1,272 HD patients from across the U.S., the annual per patient costs of HD for commercial insurance users ranged from \$4,947 to \$22,582, and the Medicaid patient costs ranged from \$3,257 to \$37,495, and these are the annual per patient costs. The larger range for Medicaid patients was attributed to their usage of nursing facilities; close to 75% of late stage HD patients used nursing homes and facilities, while over 50% of commercial patients used these services. It is worth noting, however, that this sample of Medicaid HD patients was classified as late stage while the sample size who used commercial insurance contained a more

event distribution of disease stage. Because Medicaid is a government-supported program, these costs also take a toll on society (Divino, et al., 2013).

Based on the comprehensive work of Larkindale and colleagues (2014), the costs for ALS are also very high, approximately \$31,121 per patient annually. This cost includes medical, non-medical, and indirect costs, with most of the direct medical costs related to outpatient services and treatments. These costs were determined by using information from claims databases for those using private insurance plans and those using Medicare across the U.S., totaling to 1,528 ALS patients. Nonmedical costs include modification of homes, vehicles, food and travel, and caretakers. Indirect costs were defined as loss of family income determined by Cost of Illness surveys distributed to 600 households registered with Muscular Dystrophy Association (MDA). Among this sample, 124 individuals reported being an ALS patient or having a family member afflicted with ALS. The nonmedical costs came to a total of \$17,889. In assessing all of these costs and the prevalence of ALS, the annual societal cost of this disease in the United States was found to be between \$256 and \$433 million (Larkindale, et al., 2014).

Parkinson disease also results in high medical and non-medical costs. The national economic burden of PD in 2010 was estimated to be \$14.4 billion, about \$22,800 per patient. This included medical, non-medical, and indirect costs. The prevalence rates of PD are likely underestimated, thus true costs may also be underestimated. Medical and non-medical costs also increase as severity of symptoms increase (Kowal, et al., 2013).

Alzheimer's disease far and away trumps the costs of the other three disorders, due to the higher prevalence of this disease. The average annual per patient cost for Medicare beneficiaries for health care and long-term services totals to \$47,752, a figure that does not include indirect costs such as loss of income and that is based off of findings from the 2008 Medicare



Beneficiary Survey Report and Medicare Part B claims for those with dementia (Hebert, et al., 2013). This survey includes Medicare beneficiaries living in communities and in nursing homes or residential care facilities. For 2015, it has been estimated that the total costs of health care, long-term care, and hospice for individuals with Alzheimer's and other dementias in the U.S. was estimated to be \$226 billion; government health programs such as Medicare and Medicaid covered \$153 billion, or 68% of costs (Gaugler, et al., 2015). These are astronomical numbers, and a huge burden placed on society. This number is especially concerning, given that the number of patients with AD is expected to increase significantly, from 5.3 million to 13.8 million, within the coming decades (Hebert, et al., 2013; Gaugler, et al., 2015).

In summary, the substantial economic burden of these disorders demonstrates the need for further research, increased diagnosis, and more comprehensive treatments for patients. It also validates the public health significance and importance of neurological diseases, and why integral support services are needed for these patients, although some of these disorders are considered to be low prevalence. Although these financial estimates are alarming, the emotional, mental, and physical burdens faced by patients, families, and caretakers are just as meaningful, and perhaps even more worrisome.

## **2.0 DISEASE OVERVIEW AND GENETICS**

### **2.1 HUNTINGTON DISEASE (HD)**

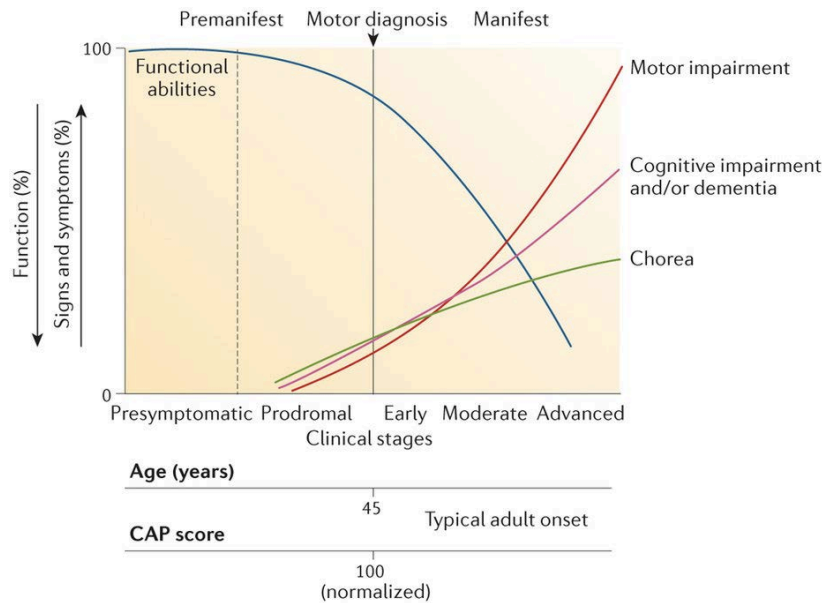
#### **2.1.1 HD OVERVIEW**

Huntington disease is a progressive, neurodegenerative disease that primarily affects the basal ganglia, a group of nerve cells at the base of the human brain. The basal ganglia are involved with different aspects of motor control, cognitive abilities, and sensory pathways. Disruption of these functions by an altered version of the huntingtin protein ultimately leads to the cellular death of the basal ganglia, producing the pathology seen with HD. Patients typically live 15 to 20 years after the onset of symptoms (Landles & Bates, 2004).

The symptoms of Huntington disease lie on a wide and varied spectrum, leading to different symptom manifestation between patients that are dependent on age of onset (typically affecting middle aged adults), general health, genetics, and the individual neurobiology of the affected patient. There are many changes that can be seen in an affected patient, and they are classified into various groups. Movement disorders may include involuntary movements, as well as impairments in existing voluntary movements. These may involve: involuntary jerking or writhing (chorea), muscle rigidity or contracture (dystonia), slow or abnormal eye movements,

impaired gait, balance, and posture, and difficulty swallowing or physically producing speech (Landles & Bates, 2004).

There are also multiple cognitive symptoms that may be seen in patients with HD, such as: difficulty with focus and organization, lack of mental flexibility (preservation), difficulties with impulse control, lack of behavioral awareness, disruptions of thought, and difficulty processing new information (Bates et al., 2015). There are also psychiatric disorders that can be attributed to HD. The most common disorder associated with HD is depression (Labbadia, Morimoto, 2013). This is not considered to be a mental reaction to a diagnosis of HD, but rather it is a physical manifestation caused by injury to the brain and disruptions of neurological function (Bates, et al., 2015). Other common psychiatric disorders that may occur in tandem with HD are obsessive-compulsive disorder and schizophrenia-like psychosis (Bates, et al., 2015). Collectively all types of these symptoms often start prior to when a formal diagnosis is received (Bates et al., 2015; Labbadia, Morimoto, 2013).



Nature Reviews | **Disease Primers**

**Figure 1: Onset of Symptom Type throughout Progression of Huntington Disease**

There are currently no treatments available to slow the progression of HD, but drug therapies do exist to treat the movement and psychiatric disorders associated with the disease (Labbadia, Morimoto, 2013). Other treatments, such as speech, physical, and occupational therapies may help the patient to handle the symptoms (Bates, et al., 2015). Psychotherapy may also help with psychiatric issues, but also with coping mechanisms, and managing expectations during the progression of disease (HDSA, 2016).

It has been estimated that there are roughly 30,000 cases of HD in the United States, 1 per 10,000 people, but there have been conflicting reports based on the ethnic groups and counties used in sample sizes (Pringsheim, et al., 2012; Rawlins, et al., 2016; Landles & Bates, 2004). However, due to a lack of incidence and prevalence studies available, it has been difficult to assess a period prevalence of this disease in the United States (HDSA, 2016). A fairly recent meta-analysis shows period prevalence of HD as 5.70 per 100,000 people starting in 1985 and ending in 2012 (Pringsheim, et al., 2012). However, this analysis included study populations from North America, Europe, and Australia, and of this analysis, only one prevalence study was from a United States population, from 1990 (Pringsheim, et al., 2012).

A more recent study has reported that prevalence rates vary widely among different populations, and shows that the prevalence in Asian populations decreasing while the prevalence in the Western Europe, North America, and Australia has been increasing over the last 50 years (Rawlins, et al., 2016). Because HD is so varied around the world, studies could be affected depending on what populations, including discreet populations, are in the cohort of a given study. The rise of HD in primarily Caucasian populations could be due to a number of reasons including better diagnostic knowledge, the wider availability of genetic testing, and potentially increased mutation rates (Rawlins, et al., 2016).

### 2.1.2 HD GENETIC ATTRIBUTES

The mutated *HTT* gene, located on 4p16.3, is known to be the cause of HD. The normal function of the *HTT* gene is to encode for the huntingtin protein, which appears to play a role in the functionality of neurons and is essential for normal fetal development, although the exact role of this protein is not yet known (Landles & Bates, 2004). While huntingtin is found in many cell types in the body, the highest level of this protein is found in the brain (Landles & Bates, 2004). Huntingtin is involved in several cellular processes including chemical signaling, cellular transport, protein binding, and protecting the cell from apoptosis (Warby, et al., 2014). A particular region of the gene contains a polyglutamine tract (CAG repeat region). Under normal circumstances, the *HTT* gene has 10-35 CAG repeats (Warby, et al., 2014).

The CAG segments contributes to the development of HD when a mutation occurs that causes these segments to expand. When they expand, a longer version of the huntingtin protein is made. This elongated protein is cleaved into smaller segments during the process of post-translational modification (Labbadia & Morimoto, 2013). When parts of this protein is in overabundance in a particular location of the brain, the polar nature of glutamine allows for protein binding (Labbadia & Morimoto, 2013). Thus, these segments will ultimately bind together in a clumped, aggregate form rather than fold into normal, functioning proteins (Labbadia & Morimoto, 2013). As more of these protein aggregates form, they coalesce into bigger forms called inclusion bodies, which disrupt normal neuronal function (Rubinsztein & Carmichael, 2003).

These aggregates can collect on the axons and dendrites of neurons, which leads to a mechanical breakdown of neurotransmitter movement (Labbadia & Morimoto, 2013). Due to this breakdown, neuronal signaling degenerates, producing the clinical symptoms of HD. This

mutated huntingtin protein also can disrupt neuronal function outside of the movement of neurotransmitters (Rubinsztein, Carmichael, 2003). The protein aggregates can disrupt the functions of chaperone proteins, caspases, cellular energy production, and the expression of other genes; this can ultimately lead to the cellular death, which is seen in the pathology of HD patients (Rubinsztein, Carmichael, 2003).

The CAG repeats in HD patients can be between 36 and 120 repeats (Warby, et al., 2014). Those with 36-39 repeats may or may not develop the disease, whereas people with 40 or more repeats nearly always do (Warby, et al., 2014). Individuals at the higher end of the “normal” CAG repeat spectrum (27-35) typically do not develop HD, but are at risk for having children who may progress to the disorder, because as the *HTT* gene passes from one generation to the next, the size of the CAG segment often increases (Warby, et al., 2014). HD is inherited in an autosomal dominant fashion, which means that only one copy of the allele needs to be present to produce disease (Rubinsztein, Carmichael, 2003). Thus, a patient has a 50% chance of inheriting the altered *HTT* allele from an affected parent. It is rare that a patient will develop HD without inheriting the altered version of the *HTT* gene (Warby, et al., 2014).

## **2.2 AMYOTROPHIC LATERAL SCLEROSIS**

### **2.2.1 ALS OVERVIEW**

Amyotrophic lateral sclerosis (ALS), commonly known as Lou Gehrig’s disease, is a neurodegenerative disease that attacks neurons, specifically those that are responsible for voluntary motor movements (NINDS, 2013). It is also a rapidly progressive and often deadly

disease (NINDS, 2013). This disease targets the motor neurons in the upper and lower parts of the motor cortex of the brain, as well as the neurons in the brain stem and spinal cord (Ferraiuolo, et al., 2011). Prior research has indicated that ALS was simply a motor neuron disease, affecting only physical voluntary movements (Ferrari, et al., 2011). However, in the last two decades, research has indicated that about 20 percent of ALS patients are also affected by frontotemporal dementia (FTD) (Ferrari, et al., 2011). This combination of ALS and FTD can lead to changes in personality, behavior, and communication skills (Ferrari, et al., 2011). There is also a rare form of ALS known as ALS-parkinsonism-dementia complex (ALS-PDC) (Waring, et al., 2004). Along with the signs and symptoms of ALS, there are additional movement abnormalities present, such as slow movements, stiffness, and tremors, as well as a progressive loss of intellectual function (ALSA, 2016).

The physical symptoms of ALS include: difficulty walking, tripping, weakness in extremities, speech slurring, difficulty swallowing, muscle cramps, twitching in arms, shoulders, and tongue, and difficulty holding up the head or maintaining good posture (NINDS, 2013). The disease usually begins in the auxiliary limbs, and then spreads to other areas of the body (Ferrari, et al., 2011). In addition to difficulties in speaking and swallowing, the neurons that are also responsible for breathing are eventually affected (NINDS, 2013). The disease typically begins between the ages of 55 and 60 (Ferraluolo, et al., 2011). Disease progression varies widely among those affected, but most patients diagnosed with the disease will die from respiratory failure 3-5 years after the onset of symptoms (Ferrari, et al., 2011).

There are some risk factors for ALS, stemming from environmental triggers. Survey module analysis performed with 1,647 participants from the National ALS registry (Mehta, et al., 2014) have also indicated that smoking and workplace exposure to lead and a history of military

service may correlate with ALS development (Mehta, et al., 2014; NINDS, 2013). However, it is important to note that this is a correlation and there are no conclusions that smoking, lead exposure, or military service triggers ALS (Mehta, et al., 2014). It is not truly known why the latter is a trigger, but it has been postulated that exposure to certain metals or chemicals, traumatic injury, exertion, and viral infections could have something to do with this relationship (Mehta, et al., 2014).

There is no cure for ALS, thus treatment focuses on slowing the progression of the disease, and prevention of complications (Ferrari, et al., 2011). In 1995, the Food and Drug Administration (FDA) approved the only drug shown to help with slowing disease progression, riluzole (Rilutek) (ALSA, 2016). This drug reduces the release of glutamate in the brain, a chemical messenger that appears to be significantly higher in ALS patients (NINDS, 2013). In clinical trials, this drug has been shown to prolong life expectancy, and extends the time before respiratory distress becomes a major issue for patients (NINDS, 2013). Combination drug therapies may be used for symptoms specific to the patient, in order to make him or her as comfortable as possible (NINDS, 2013). There are also many therapies that can help improve the quality of life of ALS patients. These include: breathing care, physical, occupational, and speech therapies, dietary support, and psychological support (NINDS, 2013).

The exact prevalence of ALS is hard to determine, given that historical records have not been kept, and that ALS is not a reportable disease on a national level (Mehta, et al., 2014). Recent reports have estimated that there are more than 12,000 people living with ALS in the United States, with approximately 6,000 new diagnoses each year (ALSA, 2016). The incidence rate of the disease is 2 per 100,000 people (Ferraiuolo, et al., 2011). Most of these cases are



considered to be sporadic (90-95%), with only 5 to 10% of ALS patients having an inherited form of the disease (Ferraiuolo, et al., 2011).

In 2009, the federal Agency for Toxic Substances and Disease Registry (ATSDR) began the first registry for ALS patients (Mehta, et al., 2014). This registry helps to provide information about prevalence and incidence rates, and environmental triggers that may have a role in causing ALS (Mehta, et al., 2014). This has been a successful public health measure. In little more than a year (October 2010-December 2011), over 12,000 people in the United States with ALS joined the registry (Mehta, et al., 2014). The registry was also able to provide valuable information on risk related to sex, ethnic background, health history, and occupation. Hopefully this will continue to be utilized by patients and physicians to gain more knowledge about ALS (Mehta, et al., 2014).

### **2.2.2 ALS GENETIC ATTRIBUTES**

Although environmental triggers play an important role of disease development, there are several genes that are related to, and may be responsible for, ALS (DeJesus-Hernandez, et al., 2011). One of the major genes that account for familial cases of ALS is the chromosome 9 open reading frame 72 gene (*C9orf72*) (DeJesus-Hernandez, et al., 2011). This gene is located on the short arm of chromosome 9p21.2 (DeJesus-Hernandez, et al., 2011). Under normal conditions, this gene provides instructions for making a protein that is in the cerebral cortex, and in motor neurons (DeJesus-Hernandez, et al., 2011). This protein also is believed to have a heavy impact on the functionality of RNA. *C9orf72* also contains a hexonucleotide repeat segment composed of four guanines and two cytosines (GGGGCC) (DeJesus-Hernandez, et al., 2011). Studies

suggest that 30 or less of these repeats will result in normal gene function (Todd & Petrucelli, 2016).

It is unclear by what mechanism the disease is caused (DeJesus-Hernandez, et al., 2011). The two primary theories are that either the expansion reduces *C9orf72* function or leads to the production of an abnormally functioning protein that both disrupts RNA function and protein production in the cell, contributing to the growth of protein aggregates (DeJesus-Hernandez, et al., 2011; Todd & Petrucelli, 2016). Overall disruption to the *C9orf72* protein function can lead to premature motor neuron death, which contributes to the pathology of ALS (Todd & Petrucelli, 2016).

Another gene that contributes to ALS cases is the superoxide dismutase 1, soluble (*SOD1*) gene. This gene is located on 21p22.11 (Andersen & Al-Chalabi, 2011). The normal function of the *SOD1* gene is to make an enzyme called superoxide dismutase, which attaches to copper and zinc molecules in order to breakdown toxic superoxide radicals (Andersen & Al-Chalabi, 2011). These radicals are the result of normal cellular processes, and must be broken down to avoid damage in the body (Andersen & Al-Chalabi, 2011). Most of the mutations that are associated with the disease are missense mutations, which can occur when one amino acid is substituted for another (Bertolin et al, 2014). It is not clear why motor neurons are affected by *SOD1* mutations, but researchers have proposed that this could be due to an increase in superoxide and other toxic radicals, increased levels of apoptosis, and aggregates of misfolded superoxide dismutase (Andersen & Al-Chalabi, 2011).

The FUS RNA binding protein (*FUS*) has also been shown to have many mutations that can lead to ALS (Ferraiuolo, et al., 2011). The *FUS* gene (located on 16p11.2) is normally responsible for making a protein that helps to regulate transcription, the processing of messenger

RNA (mRNA) via alternative splicing, and repairing mistakes in DNA (Ferraiuolo, et al., 2011). Most of these mutations are also missense and primarily affect the parts of the protein involved in DNA binding and mRNA processing, particularly hindering mRNA transport out of the cells, which causes mRNA to become trapped in the cells and form aggregates (Andersen & Al-Chalabi, 2011). Patients who have ALS caused by *FUS* mutations tend to have a shorter life expectancy and the onset of the disease occurs at a younger age (Bertolin, et al., 2014).

The TAR DNA binding protein gene (*TARDBP*), is responsible for making a protein called transactive response DNA binding protein 43 kDa (TDP-43) (Borrioni, et al., 2010). It is located on 1p36.22 (Borrioni, et al., 2010). This protein has several functions relating to protein production such as, regulating transcription, RNA binding, and mRNA processing via alternative splicing (Borrioni, et al., 2010). This protein is particularly active during fetal development and it influences production of proteins involved in the nervous system and organ development (Borrioni, et al., 2010). Many missense mutations of the *TARDBP* gene have been found to cause ALS (Andersen & Al-Chalabi, 2011). The majority of these mutations affect mRNA processing, which disrupts the production of other proteins and leads to mRNA aggregates within the cell (Borrioni, et al., 2010).

The pattern of inheritance associated with ALS is dependent on what gene is responsible for causing the disease. Most mutations are inherited in an autosomal dominant fashion, in which a mutation in one copy of the gene is enough to cause the disease (Ferraiuolo, et al., 2011). However, there is also reduced penetrance in many of these mutations, meaning that, a person can inherit a disease causing mutation, but never progress to illness (Andersen & Al-Chalabi, 2011). Although it is not as common, ALS can also be inherited in an autosomal recessive pattern, in which a person would need two copies of the mutated allele, one from each parent, in

order to progress to a diseased state. In this case, the parents do not show symptoms of the disease, so the affected patient's diagnosis of ALS is typically classified as sporadic (Conforti, et al., 2008). In very rare cases, it is also possible for ALS to be inherited in an X-linked dominant pattern, in which females would only need to inherit a mutation on one of their two X chromosomes to be affected, and males would be affected if they inherit a mutation on their one X chromosome, which they always receive from their mother (Deng, et al., 2011).

## **2.3 ALZHEIMER DISEASE (AD)**

### **2.3.1 AD OVERVIEW**

Alzheimer disease is a progressive, neurodegenerative disorder that affects memory and cognitive abilities. It is the most common cause of dementia among older adults, as well as the sixth leading cause of death in the United States (Cummings, 2004). With AD, the patient's neurons progressively lose function and die off (Cummings, 2004). This occurs in multiple parts of the brain, including the cerebral cortex, temporal and parietal lobes, frontal cortex, cingulate gyrus, and in parts of the brainstem (Mattson, 2004). Survival times can range anywhere from 1 to 25 years after symptom onset, with most patients surviving an average of 8 to 10 years (NIA, 2015; Mattson, 2004). The first symptoms that patients usually experience are loss of memory and mild confusion (NIA, 2015). These start when the patient experiences short term memory loss, and as the disease progresses, the memory problems get steadily worse, leading to the inability to identify objects, express thoughts, and remember the names or lives of friends, family members, or themselves (Cummings, 2004).

Thought processes and reasoning abilities can become impaired, especially those involving more complex concepts such as numbers (Gaugler, et al., 2015). The ability to multitask also declines, and both of these aspects can usually manifest themselves in a patient's finances, which may ultimately further decrease their quality of life (Gaugler, et al., 2015). Judgment and decision making are also affected, which could lead to the patient putting themselves or others in dangerous situations (NIA, 2015). Planning and performing familiar tasks are also abilities that may be adversely affected, affecting the patient's daily routine greatly (NIA, 2015). There are also psychiatric issues that can occur in conjunction with AD such as changes in personality and behavior. These include: depression, apathy, mood swings, distrustfulness, irritability and aggressiveness, changes in sleeping habits, loss of inhibitions, wandering, and delusions (NIA, 2015; Cummings, 2004). As AD progresses into later stages, the changes in the brain can often manifest into physical symptoms. These include swallowing, balance, and bowel and bladder control (Gaugler, et al., 2015).

There are many risk factors that pertain to AD, with age being the most significant factor (Harmen, 2006). AD usually affects patients 65 years of age and older, although some people with rare genetic mutations can begin to see symptoms as young as 30 (Harmen, 2006). Family history and genetics plays an important role in the development of AD, with risk especially increasing if a person has a first degree relative (a member of the immediate family) who has the disease (Gaugler, et al., 2015). Having Down syndrome is also a major risk factor for AD development, with patients developing symptoms of AD 10 to 20 years earlier than the general population (Lott & Head, 2005). This is believed to occur due to a gene inherited on chromosome 21, the extra chromosome that causes Down syndrome (Lott & Head, 2005).

Other risk factors for AD include a patient's health history, particularly if that person has had a past head trauma, both a single incident as well as repetitive head injuries (Sivanandam & Thakur, 2012). This factor is believed to increase the risk of amyloid protein deposition in the brain (Sivanandam & Thakur, 2012). Certain aspects of heart health can also contribute to the development of AD (Cummings, 2004). Some of these factors include: lack of exercise, obesity, smoking, high blood pressure, high cholesterol levels, poor diet, and poorly controlled type 2 diabetes (Mattson, 2004).

While there is no cure for AD, there are drugs available to help with the management of cognitive changes. Cholinesterase inhibitors work to help replenish depleted acetylcholine, a neurotransmitter that boosts cellular communication (Cummings, 2004). This inhibitor can also help some of the psychiatric symptoms of AD, such as agitation or depression (Cummings, 2004). Another pharmaceutical, memantine (Namenda) can improve cellular communication and this helps to slow the progression of the cognitive symptoms, particularly in patients with moderate to severe AD (Cummings, 2004; NIA, 2015). Other psychiatric drugs, such as antidepressants or anti-anxiety medications, can be used in combination with those listed above to help alleviate some symptoms and make the experience easier on the patient, but only when behavioral healthcare measures have been taken first (Cummings, 2004).

Arguably, one of the most important parts of the treatment plan for AD is creating a safe environment for the patient to live, with support from both a medical team and loved ones (Mattson, 2004). Exercise and nutrition are also important aspects of care; these not only work to improve physical health, but can also help to alleviate some psychiatric symptoms such as mood changes and depression (Cummings, 2004). These treatment plans are also helpful in allowing

the patient and the caregivers to establish a regular daily routine (Cummings, 2004; Gaugler, et al., 2015).

In 2015, approximately 5.3 million Americans were living with AD; 5.1 million people out of this number are 65 years and older (Gaugler, et al., 2015). Approximately 500,000 people in the United States are diagnosed with AD each year (Gaugler, et al., 2015). From these numbers it is quite easy to tell how risk of having AD increases with age. This is especially a concern with the aging population of the United States. People are living longer, but they are also at risk for more diseases as they age (Riedel, et al. 2015). Due to these increased prevalence and incidence rates, the number of Alzheimer's patients in the United States is expected to double by 2025 (Gaugler, et al., 2015).

Women have also been found to be most at risk, with an estimated two-thirds of current American AD patients being women (Riedel, et al. 2015). There are also racial and ethnic differences in AD patients. African Americans and Hispanics are twice as likely and one and a half more times as likely to develop AD when compared to non-Hispanic whites, respectively (Riedel, et al. 2015). Both the sex and racial differences can be attributed to a number of factors. Age is the most important factor for the differences between the sexes because women tend to live longer than men, thus having a higher risk of developing AD (Ghebremedhin, et al., 2001). For the differences by both sex and race, variations in lifestyle, health, and socioeconomic status can be related to risk of AD (Ghebremedhin, et al., 2001; Riedel, et al. 2015).

Due to the increased surveillance for and incidence of AD, compared to other neurological disorders, researchers have the opportunity to compare the number of AD patients by state in the United States, and make future predictions using these numbers (Gaugler, et al., 2015). The states within the central Appalachian region of the United States have different rates

or projected increases of AD (Gaugler, et al., 2015). This is important given the focus on support groups for this essay. The figure below shows projected increases of Alzheimer's disease between 2015 and 2025. It is important to note that every state in the United States is increasing by some amount; none are staying the same or decreasing (Gaugler, et al., 2015).

### **2.3.2 AD GENETIC ATTRIBUTES**

Although AD is considered to be a multi-factorial disease, there are genetic attributes that play a role in disease pathology, especially in the cases of early onset AD (Riedel et al., 2015). These genetic attributes are inherited in an autosomal dominant fashion in the cases of early onset AD, whereas the cases of late onset AD have inheritance patterns that are difficult to determine (Mattson, 2004). There are three genes that contribute to early onset AD. There are potentially many genes that could have an effect on the development of late onset AD, and these are usually population specific (Bird, 2008). One specific gene related to late onset AD that has been extensively studied in relation to late onset AD is the apolipoprotein E gene (*APOE*).

Under normal conditions, *APOE* provides instructions for making apolipoprotein E (Riedel et al., 2015). This protein produces lipoproteins by combining with fat molecules (lipids). Lipoproteins package and carry fats throughout the bloodstream, which helps the body to maintain a normal and healthy amount of cholesterol in the blood (Riedel et al., 2015). There are three alleles known in the human population: e2, e3, and e4, with e3 being the most common, found in more than half of the world's population (Riedel et al., 2015). The *APOE* gene is located at 19p13.2 (Riedel et al., 2015).

The e4 allele of the *APOE* gene has been shown to increase a person's chances of developing AD, with the amount of risk increasing if two e4 alleles are inherited (Bird, 2008). It



is not known exactly how the e4 allele is related to AD, but there have been significant associations between the APOE e4 genotype and the presence of neurofibrillary tangles and amyloid plaques characteristic of AD (Bird, 2008). Neurofibrillary tangles are aggregates of tau protein that has been hyperphosphorylated and amyloid plaques are aggregates of misfolded proteins (Mattson, 2004). These both work to physically disrupt neuronal function, eventually leading to neuron death and contributing to the pathobiology of AD (Mattson, 2004).

The amyloid beta precursor protein gene (*APP*) is located on chromosome 21q21.3 (Bird, 2008). This gene provides instructions to make the amyloid precursor protein, a protein found throughout the body (Bird, 2008). The normal function of this protein is still uncertain, although some studies have suggested that it may aid in the movement of neurons (Rogaeva, et al., 2007). This protein is also cut into smaller fragments by enzymes (Mattson, 2008). Two of these fragments, soluble amyloid precursor protein (sAPP) and amyloid beta ( $\beta$ ) play an important role in the functions of neurons (Mattson, 2008). sAPP aids in the formation of nerve cells and protein inhibition, while amyloid beta aids in the adaptability of neurons (Mattson, 2008). *APP* contains many mutations that are related to early-onset AD, although these mutations are responsible for a small number of cases (Bird, 2008). The mutations cause an abnormal amount of amyloid beta protein to be created or can cause longer proteins that are more likely to form aggregates (Harmen, 2006). The increased amount and altered version of amyloid beta can accumulate and stick together, forming the amyloid plaques that contribute to neuron death in AD (Sivanandam & Thakur, 2012). Since *APP* is also on chromosome 21, those with Down syndrome inherit an extra copy of this gene, increasing the production of amyloid beta protein, as well as risk of AD development (Bird, 2008).

Presenilin 1 (*PS1*) is a gene responsible for making the presenilin 1 protein. This protein is part of a larger complex called gamma- ( $\gamma$ -) secretase (Gómez-Isla, et al. 1999). Presenilin 1 carries out the major function of the complex, which is to cleave other proteins into fragments (Gómez-Isla, et al. 1999). This is the protein complex responsible for cutting the amyloid precursor protein into sAPP and amyloid beta protein (Mattson, 2008). This gene is located at 14q24.3. Many of the mutations related to this gene that can be related to early onset AD (Gómez-Isla, et al. 1999). The defective presenilin 1 protein disrupts the function of the gamma secretase complex, leading to processing errors in APP that aids in producing the amyloid plaques seen in AD (Gómez-Isla, et al. 1999).

The presenilin 2 gene (*PS2*) makes the presenilin 2 protein. This gene is located at 1q42.13. This protein works to help process chemical signaling proteins (Gómez-Isla, et al. 1999). These chemical signals activate genes that are important for cellular growth and maturation (Gómez-Isla, et al. 1999). Presenilin 2 also processes amyloid precursor protein (Gómez-Isla, et al. 1999). Mutations in this gene lead to overproduction of amyloid beta protein, increasing the formation of protein aggregates (Gómez-Isla, et al. 1999).

## **2.4 PARKINSON DISEASE (PD)**

### **2.4.1 PD OVERVIEW**

Parkinson disease is a progressive disorder that affects the nervous system, especially an area in the middle of the brain called the substantia nigra that controls movement (Obeso, et al., 2008). The disease begins gradually, usually on one side of the body, but over time will affect

all movements (Samii, et al., 2004). The disease can progress to include cognitive and psychiatric symptoms (Samii, et al., 2004). PD can be either late or early onset, but the majority of patients develop the disorder after the age of 50 (Samii, et al., 2004).

The symptoms of PD can vary from person to person, and many of the early signs can be mild causing them to go unnoticed during clinical evaluation (Samii, et al., 2004). The typical first sign of PD is a tremor, usually beginning in a specific limb, when the body is at a complete rest (Nuytemans, et al., 2010). Slowed movement, as well as muscle rigidity, begins to occur as the disease progresses (Obeso, et al., 2008). Other symptoms may include impairments in posture and balance, loss of automatic movements, speech changes and motor changes. Some individuals may experience psychiatric symptoms such as depression and hallucinations (Samii, et al., 2004). Those affected with PD also have an increased risk of developing dementia (Samii, et al., 2004).

PD symptoms begin when the neurons in the substantia nigra begin to die due to a lack of dopamine, an essential chemical messenger that controls communication between the brain and the muscle cells of the body (Obeso, et al., 2008). Lewy Bodies, which are protein aggregates that coalesce around neurons, are also a hallmark of Parkinson disease. They can be made up of a variety of proteins, and their role in PD is not fully understood (Kalinderi, et al., 2016). PD cannot be cured, but treatment is available to alleviate some of the symptoms (Samii, et al., 2004). The majority of medications used for the treatment of PD act to affect the dopamine insufficiency. (Samii, et al., 2004). These medications work by replacing dopamine, replicating the effects of dopamine, or by preventing the breakdown of already existing dopamine (Samii, et al., 2004). There are also medications available to help control involuntary movements and tremors (Samii, et al., 2004). In addition to drug therapy, there is

also an operation that PD patients can have called deep brain stimulation, which involves the implantation of electrodes in a specific part of the brain (Samii, et al., 2004). These electrodes send electrical pulses to the brain to control PD symptoms (Samii, et al., 2004).

PD occurs in 13 people per 100,000 (Samii, et al., 2004). The prevalence of PD is estimated to be one to two percent of the population older than 60, worldwide (Nuytemans, et al., 2010). While this disease may not be fatal, these numbers and the effects that PD has on a person's everyday life signal the need for more research, as well as more medical and social support for patients. (Samii, et al., 2004).

#### **2.4.2 PD GENETIC ATTRIBUTES**

Although PD is considered to be a multi-factorial disease, there are several genes that can play a significant role in the pathology of the disorder. Of the cases of PD that are considered to be inherited, mutations in one of five known genes may have caused the disorder (Klein & Westenberger, 2012). However, there are two other genes that do not directly cause PD, but may modify the risk of PD development in some families (Kalinderi, et al., 2016). While the basic, normal functions of these genes are readily known, the mechanisms by which they cause PD remain inconclusive (Klein & Westenberger, 2012). The five genes that are believed to cause PD are all within the PARK family of genes (Klein & Westenberger, 2012).

The leucine-rich repeat kinase 2 gene (*LRRK2*) is responsible for producing the protein dardarin (Hardy, et al., 2006). The exact biological role of dardarin is not yet known (Nuytemans, et al., 2010). *LRRK2* is located at 12q12 (Hardy, et al., 2006). It is unclear how this gene may cause PD, but it has been shown to be associated with families that have a history of late-onset PD (Nuytemans, et al., 2010). It is hypothesized that *LRRK2* may have an effect on the

phosphorylation of proteins central to PD, such as  $\alpha$ -synuclein and Tau, which can be found in the protein aggregates called Lewy bodies (Cookson & Bandmann, 2010). There have been over 100 mutations in this gene that have been found in families afflicted with PD (Kalinderi, et al., 2016).

The parkin RBR E3 ubiquitin protein ligase gene (*PARK2*), makes the protein parkin under normal conditions (Nuytemans, et al., 2010). This protein has a role in the breakdown of other proteins by tagging them with ubiquitin, a signaling molecule designed to move unneeded or damaged proteins to the proteasome of the cell, where they are degraded (Nuytemans, et al., 2010). Studies have also indicated that parkin may have a role in the maintenance of mitochondria (Kalinderi, et al., 2016). Most of these mutations lead to the production of abnormal parkin protein (Nuytemans, et al., 2010). The loss of parkin activity may affect the breakdown of damaged proteins, leading to a buildup of aggregates that can potentially disrupt neurons that transmit dopamine (Kalinderi, et al., 2016). The *PARK2* gene is located between 6q25.2 and 6q27 (Nuytemans, et al., 2010).

The gene Parkinsonism associated deglycase (*PARK7*) is located on 1p36.23 (Kalinderi, et al., 2016). The function of this gene is to make the DJ-1 protein. The DJ-1 protein has several functions including the protection of neurons and other cells from oxidative stress, serving as a chaperone molecule to assist in protein folding and degradation, and RNA processing (Kalinderi, et al., 2016). Although it is unclear how the DJ-1 protein affects PD pathobiology, studies have suggested that mutations in the *PARK7* gene can cause a breakdown of the chaperone qualities of DJ-1, leading to a build up of protein (Hardy, et al., 2006). Also, the loss of oxidative stress protection could affect the neurons that transmit dopamine (Klein & Westenberger, 2012).

PTEN induced putative kinase 1 is made by a gene of the same name (*PINK1*). This protein appears to protect mitochondria from malfunctioning during times of stress, when there are high energy demands within the cell, although this is not well understood (Brooks, et al., 2009). There are two different segments of this protein that appear to be vital for basic functionality; these are the mitochondrial-targeting motif, which ensures that the protein moves to the mitochondria after it is made, and the kinase domain, which performs the protective functions (Klein & Westenberger, 2012). Mutations in the *PINK1* gene are associated with the early-onset form of PD (Brooks, et al., 2009). These mutations work to disrupt or remove the kinase domain, which leads to loss of protein function, and eventual cell death. The *PINK1* gene is located on 1p36 (Klein & Westenberger, 2012).

The synuclein alpha gene (*SNCA*) is located on 4q21, and is responsible for making the alpha synuclein protein (Nuytemans, et al., 2010) The function of this protein is to supply synaptic vesicles within the presynaptic terminals, and also may regulate the release of dopamine (Nuytemans, et al., 2010). The mutations in *SNCA* can either lead to a misfolded alpha synuclein protein, or extra copies of the normal protein (Nuytemans, et al., 2010). The excess amount of protein can form aggregates that can impair neuronal function and lead to problems with the regulation of dopamine (Klein & Westenberger, 2012).

The gene glucosidase, beta, acid (*GBA*) functions to make an enzyme called beta-glucocerebrosidase, which is a digestive enzyme used to breakdown toxic substances within the lysosome of the cell (Blanz & Saftig, 2016). *GBA* is located on 1q21. Mutations in this gene and their association with PD are not yet clear, although it has been proposed that these genetic alterations may contribute to lysosomal dysfunction and collection of protein aggregates (Blanz

& Saftig, 2016). These genetic changes have also been seen in those with Gaucher disease, and individuals with this condition have an increased risk of PD (Blanz & Saftig, 2016).

Ubiquitin C-terminal hydrolase L1 (UCHL1) is a gene responsible for making an enzyme called arboxyl-terminal esterase L1, which is found in nerve cells throughout the brain (Samii, et al., 2004). This enzyme has a similar function to beta-glucocerebrosidase, which breaks down toxic substances (Samii, et al., 2004). One particular polymorphism in the UCHL1 gene actually reduces the risk of developing PD, and this is primarily found in Chinese and Japanese populations (Samii, et al., 2004). It is not quite clear by what mechanism this variation affects risk. The UCHL1 gene is found on 4p14 (Samii, et al., 2004).

### **3.0 CENTRAL APPALACHIAN HEALTHCARE AND SUPPORT GROUP CONSIDERATION**

#### **3.1 HEALTHCARE BURDENS AND DISEASE SIGNIFICANCE IN APPALACHIA**

##### **3.1.1 ACCESS TO HEALTHCARE IN APPALACHIA**

The Appalachian region of the United States is a region that encompasses the range of the Appalachian Mountains in the eastern part of the country (ARC, 2016). It comprises approximately 205,000 square miles and contains the entire state of West Virginia, and parts of twelve other states: Alabama, Georgia, Kentucky, Maryland, Mississippi, New York, North Carolina, Ohio, Pennsylvania, South Carolina, Tennessee, and Virginia (ARC, 2016).

The differences we see in the demographics, poverty, and healthcare access of the Appalachian region contribute to the unique healthcare burdens that these states experience. In Appalachia, there is a higher rural population compared to the rest of the United States, (42% versus 20%, respectively) (OCC, 2013). Also, the poverty rate is slightly increased above the national average (15.4% versus 14.8%) (ARC, 2014). It has also been established that there is a healthcare provider shortage in the Appalachian region and this is due in part to the low healthcare reimbursement (Lane, et al., 2012). These provider shortages in turn lead to fewer clinics and hospitals being opened in the rural areas of Appalachia (Lane, et al., 2012). This is



especially an issue for the Appalachian region, as the region as a whole suffers from a higher mortality rate due to chronic disease (Halverson, et al., 2004).

There is also the issue of the low health literacy rate throughout the country (Kutner, et al., 2006). Health literacy, defined as “the degree to which individuals have the capacity to obtain, process, and understand basic health information and services needed to make appropriate health decisions” (Selden, et al., 2000), is a problem throughout the United States. Currently, only twelve percent of adults in the United States are considered health literate, and those living in lower income areas and among populations that have a lower educational level overall, are more at risk to experience low health literacy (Kutner, et al., 2006).

This lack of health literacy has left its mark on the people of this region. Knowledge deficiency often leads to fear of physicians and healthcare procedures, which in turn breeds distrust among patients (Behringer, et al., 2006). The poor patient-provider relationship can often lead to issues with keeping appointments, medication adherence, poor health behaviors and suffering of treatment plans (Liang, et al., 2013). With all of these other factors, it is easy to see how the issues of healthcare access in Appalachia stretches far beyond dollars and cents.

### **3.1.2 HEALTHCARE ACCESS IN CENTRAL APPALACHIA**

Due to the lack of patient registries and potential under diagnosis it is difficult to determine the significance of the four adult-onset neurological disorders outlined above in central Appalachia. However, due to the provider shortage that this region is suffering from (Lane, et al., 2012), and given that these disorders also have fairly generalized symptoms that can easily be overlooked and misdiagnosed, (Bates et al., 2015; NINDS, 2013; NIA, 2015;

Samii, et al., 2004), it stands to reason that there may be many undetected cases of these diseases throughout the region.

Because of the attributes of these disorders and the provider shortage in this region the disparities already experienced by these populations are more emphasized. The healthcare burdens faced by patients and family members who are affected by disease are numerous in nature (Halverson, et al., 2004). Because these individuals may have limited access to basic healthcare needs, as well as patient and caregiver support, these patients may face a more difficult time with their diagnosis and treatment (Lane, et al., 2012). To address access to patient and caregiver support groups, an assessment of current services followed by comprehensive research to determine patient needs should be completed, and then be used to provide services and support in an economically and geographically sound way that makes sense for the given region.

## **3.2 SUPPORT GROUP EXPLORATION**

### **3.2.1 BENEFITS AND DRAWBACKS OF SUPPORT GROUPS**

Support groups are important to those afflicted with long term chronic illness because they provide a forum for social interaction, peer support, and discussion of the illness in question (Locock & Brown, 2010). These groups are also important for the caretakers involved with those who have diseases, as the lifestyle and duties of caretakers can produce a great deal of stress, which can in turn affect the patient (Schulz & Martire, 2004). The benefits and harms to the patients and caretakers involved in disease support groups rests on the individuals (Locock &

Brown, 2010), but may also be due to the participants, and the organization and professionals facilitating the support groups. The effects of support groups rest heavily on social comparison theory, in which an individual has a drive to gain accurate self-evaluations and awareness, by drawing comparisons to others with their predicament (Locock & Brown, 2010).

For those who suffer from disease, especially diseases that can produce cognitive or psychiatric symptoms, emotions play a rather large role in treatment and management (Attard & Coulson, 2012). This is a fickle aspect of support groups, as some people may derive benefits from them, while in others, social support may beget emotional harms (Locock & Brown, 2010). Many benefits of support groups have been studied using questionnaires disseminated to patients and family members affected with various health conditions (Ziebland & Wyke, 2012), some of which can include: information exchange, the sharing of experiences, social connection, increased optimism, improved emotional well-being, and potentially better disease management and health outcomes (Mo & Coulson, 2014).

In contrast to this, based on interviews of 48 patients with Motor Neuron Disease and 22 caregivers, followed by literature review and analysis, it was found that support groups can foster negative effects such as the fear of meeting others, fear of interaction, fear of judgment, misinformation regarding the disease, and potential damage to the patient-provider relationship (Locock & Brown, 2010). Again, drawing from social comparison theory, a patient may also begin to identify as just a person with a disease, and thus, sense of self can eventually be lost (Locock & Brown, 2010).

These benefits and harms can also change depending on the environment of the support group in question. With the advent of the Internet, online support groups for both patients and caretakers have blossomed, especially for long term or chronic illness (Coulson, et al., 2007).

Patients or caregivers often choose online support groups for the same reasons that they would choose in-person groups, but there are benefits that in-person groups cannot provide, such as connecting with a large amount of people from around the world, as well as the benefit of being anonymous (Mo & Coulson, 2014). However, with new advances in healthcare technology, there are almost always some shortcomings unique to the specific advance that should be improved upon.

Some of the disadvantages of online disease support groups include: unidentifiable presences online, inappropriate behaviors, risk of personal information being detected and stolen, over exaggerated claims, information overload, and misinformation (Malik & Coulson, 2010). Apart from these distinctive limitations, there is also the major problem of the impersonal nature of the Internet (Mo & Coulson, 2014). The inability to connect physically to another person or groups of people can be a major setback for all patients, but particularly for those who do not feel important or heard by others, which can lead to increased anxiety and depression (Mo & Coulson, 2014). Another issue with online support groups is attendance because many people tend to drop out of these groups whether it is due to a loss of interest, loss of connectivity, or simply life getting in the way (Sandaunet, 2008).

Another problem with online support groups is the specific demographics that can fall by the wayside, such as older populations. In a study involving 230 adults diagnosed with lung cancer, it was found that online support groups are more conducive to younger populations (Xu, et al., 2013). It was also found that older generations are less likely to use online resources due to a lack of knowledge, as well as habitual practices of having in person interaction (Xu, et al., 2013). This study used questionnaires disseminated to participants to gather data, and this data

organized predisposing, enabling, and need factors across various demographics (Xu, et al., 2013).

Another problem with online support groups is the specific demographics that can fall by the wayside. Online support groups are more conducive to younger populations, and those that can afford an Internet connection (Xu, et al., 2013). Older generations are less likely to use online resources due to a lack of knowledge, as well as habitual practices of having in person interaction (Xu, et al., 2013). Those in economically disadvantaged areas may not believe they are able to afford an Internet connection, may not put high importance on it, or may not want to invest in technology (Porter & Donthu, 2006).

The positive aspect of online support groups is that they often have the same outcomes that in person meetings produce. A study assessing 1,313 messages for evidence of social support accessed via a public Huntington disease support message board showed that online support groups for both patients and caretakers reduce anxiety and give them a medium in which they can communicate and draw support (Coulson, et al., 2007). While information overload and misinformation can be a problem in online communities, the plethora of information and sources online lends a good deal of support and opportunities to learn about disease management and new treatments (Attard & Coulson, 2012).

### **3.2.2 SIGNIFICANCE OF SUPPORT GROUPS FOR NEUROLOGICAL DISORDERS**

Having more support groups that cater to patients and caretakers of Huntington's disease, Amyotrophic lateral sclerosis, Alzheimer's disease, and Parkinson's disease play an important role in disease management and social support, as evidenced by prior work (Attard & Coulson, 2012; Coulson, et al., 2007; Schulz & Martire, 2004; Locock & Brown, 2010). The unique

aspects of these neurological conditions include the cognitive and psychiatric symptoms present in these disorders, the progressive, sometimes fatal nature of the disease, the associated genetic attributes, and the prevalence throughout the United States. Due to these qualities, support groups may fulfill the needs and improve the lives of patients, families, and caretakers afflicted by the four disorders characterized in this essay.

Coping with a disease that often results in loss of cognitive functions and the initiation of psychiatric disorders can be extremely difficult. Caretakers of these patients must go through the rigors of these symptoms with the patient, which can and does take a deep emotional toll on them (Schulz & Martire, 2004). Having support groups available to both the patient and caretaker can be important to helping them remain emotionally steadfast and mentally capable as long as it is possible or necessary (Schulz & Martire, 2004). Support groups have been identified by patients to be socially stimulating, helpful in managing day-to-day tasks, and integral to their self-esteem (Locock & Brown, 2010).

Often, due to the rapid progression of some neurological disorders such as Huntington's disease, patients will experience high levels of stress and anxiety (Coulson, et al., 2007). Many support groups for diseases such as this will frequently discuss stress and disease management, and social support for patients who feel as though no one can understand their experiences (Coulson, et al., 2007). Having access to both information and support is critical for the well-being of the patient, as learning about and utilizing new treatments and management techniques can reduce symptoms and perhaps prolong life expectancy (Mo & Coulson, 2014).

The low prevalence of these neurological conditions could also have an effect on the amount of information available, as well as access to healthcare specialists that work with these disorders. This can lead to decreased availability of support groups, due to the lack of trained

individuals available to moderate them, especially in the rural areas of the United States, where hospital and physician shortages already exist. Of course, support groups can never replace the role of hospital facilities and medical care that patients need. However, if more support groups are available within rural areas, physicians, nurses, therapists, and genetic counselors would have an additional way to reach out to these patients and could perhaps use support groups as an environment to speak to people about their various concerns, while providing valuable education.

## **4.0 METHODS**

### **4.1 DATA SOURCING**

In determining the number and locations of disease support groups for the four neurological conditions outlined previously, I used an online search engine query using Google to find the primary organizations that facilitated in-person support groups for each disorder. An example of a phrase used in the search was “Huntington disease support groups”. My queries tabulated many search results; however, many of these results often linked back to national organizations offering a variety of resources, and this occurred for each disease. I chose to use one organization for each of the four neurological diseases in order to best compare the support availability for each disorder, as some diseases had multiple organizations to choose from, whereas others had only one or two organizations that offered support groups. I chose the organizations based on the amount of data present on each website, and on the number of support groups that the website listed. Although patient and caregiver support groups operated by local organizations or private citizens exist, assessing and comparing the number of these that are available for each state and each disease is difficult, primarily because they are not as accessible via online searches. Because part of my goal was to determine how patients and caregivers identify resources, I focused on the national organizations



The organizations that I chose were The Huntington’s Disease Society of America, The ALS Association, The National Parkinson Foundation, and The Alzheimer’s Association. Each of these organizations had different types of online tools that one could use to locate support groups. However, each of these online tools was structured differently. Some of these websites required an input of an address or area code and the nearest support groups were shown based on a chosen mileage, while some simply gave a listing of all support groups available within each state. The structures of these online tools are detailed below in Table 1. The exact number of support groups per city was difficult to determine from the organization data, given that entire regions of some states were missing. Therefore, I used towns and cities that had available support groups in them to determine county-level data.

**Table 1: Support Group Organization Listing and Online Tool Description**

Disease	Organization	Online tool description
Huntington disease	Huntington’s Disease Society of America <a href="http://hdsa.org/about-hdsa/locate-resources/">http://hdsa.org/about-hdsa/locate-resources/</a>	Address or area code input and distance indicator (25-1000 miles); also included interactive map with locations of support groups indicated
Parkinson disease	The National Parkinson Foundation <a href="http://www.parkinson.org/find-help/resources-in-your-community">http://www.parkinson.org/find-help/resources-in-your-community</a>	Area code input or state selector can be used in tandem with a distance indicator (10-250 miles) and a selection of PD Care or Community Organizations is offered; when selections are made, a list of community organization that offer support services is tabulated
Amyotrophic Lateral Sclerosis	The ALS Association <a href="http://www.alsa.org/community/support-groups/?referrer=https://www.google.com/">http://www.alsa.org/community/support-groups/?referrer=https://www.google.com/</a>	State selector available; once a state was chosen, a list of organization chapter websites given, which linked to a tabulated listing of support groups offered by the chapter
Alzheimer disease	The Alzheimer’s Association <a href="http://www.communityresourcefinder.org/">http://www.communityresourcefinder.org/</a>	Address or area code input or name input along with other search options such as date of event, delivery, audience served, language; a listing of support groups with locations, dates, and times is tabulated

To determine the county that each support group is located in, I performed a Google search of each city or town indicated in the support group location information. Next, I used the Appalachian Regional Commission webpage (ARC, 2016), which contains a listing of counties in Appalachia for each state that falls within the Appalachian region to determine if a county was located in the defined Appalachian region of the United States.

Using the Appalachian Regional Commission webpage, I determined each Appalachian county's economic status. The Appalachian Regional Commission uses an index-based county economic classification system to determine a county's status. This system uses three economic indicators to compare counties to the national averages. These indicators are three-year average unemployment rate, per capita market income, and poverty rate. The counties are then ranked by different levels of economic distress level based on a composite index value assigned to the county by this comparison. The levels of economic distress are: distress (worst 10 percent of the nation's counties), at-risk (between the worst 10 percent and 25 percent of the nation's counties), transitional (between the worst 25 percent and the best 25 percent of the nation's counties), competitive (between the best 25 percent and the best 10 percent of the nation's counties are classified competitive), and attainment (best 10 percent of the nation's counties are classified attainment) (ARC, 2014).

## **4.2 DATA ANALYSIS**

I used Microsoft Excel for data entry and analysis and created a worksheet for each disease and listed the number of support groups for each state, their town or city location, the county that the location was in, and the economic status of each county. I then determined the

total number of counties represented by support group availability in each state, as well as the number of Appalachian counties, and the number of economically distressed or at risk counties in Appalachia. I then split this Excel workbook into two separate workbooks, one for Appalachian counties and one for the total number of counties with support groups. I arranged this information into bar charts organized by state and disorder to produce a visual representation of the descriptive statistics of support groups for each disease by state (See Figures 4 and 5).

I tabulated the total number of counties with and without support groups (across all four diseases) for each of the six states (Table 2). Within each state, I also tabulated the number Appalachian counties with and without support groups, as well as the number of non-Appalachian counties with and without support groups (Table 3). To assess whether distressed counties had fewer support groups than non-distressed counties within Appalachia, I also compiled, by state, the number of distressed counties with and without support groups and the number of non-distressed counties with and without support groups (Table 4).

To test for differences in numbers of support groups between Appalachian and non-Appalachian counties, and between distressed versus non-distressed counties, I performed chi-squared tests. Calculations were done in Microsoft Excel using the function =chitest.

## 5.0 RESULTS

### 5.1 PRESENCE OF SUPPORT GROUPS BY STATE AND APPALACHIAN REGION

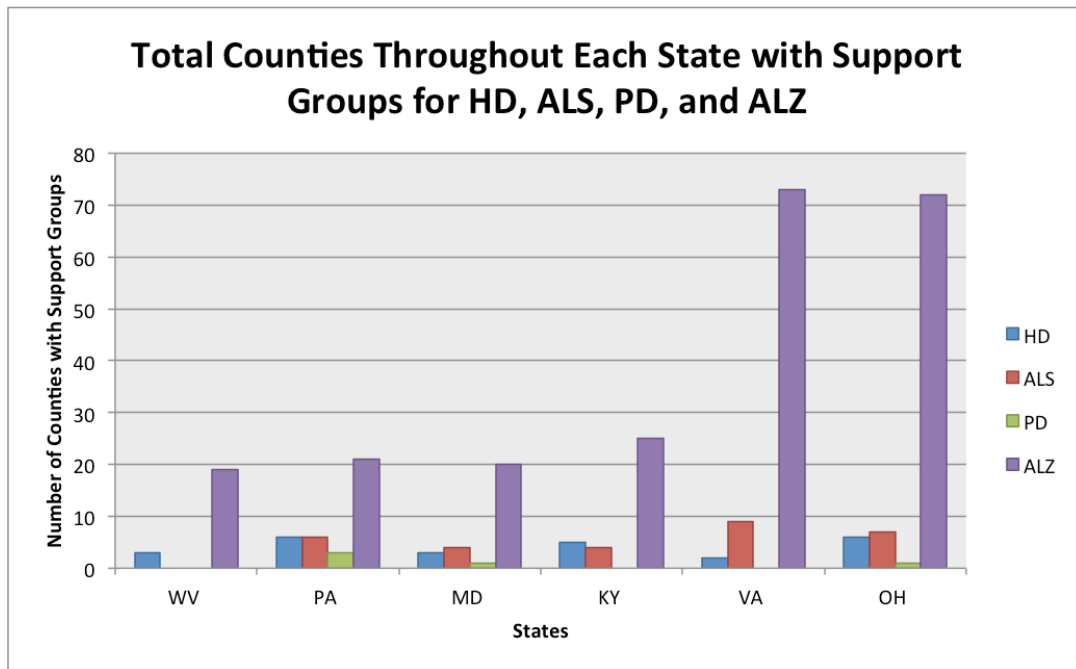
Using online databases (Table 1), a total of 449 counties in six states were assessed to determine whether they did or did not contain at least one support group for at least one of the four neurological conditions: Huntington Disease (HD), Amyotrophic lateral sclerosis (ALS), Parkinson Disease (PD), and Alzheimer Disease (ALZ). Both patient and caretaker support groups were included in the assessment.

**Table 2: Number of Counties by State With and Without Support Groups for Four Neurological Diseases**

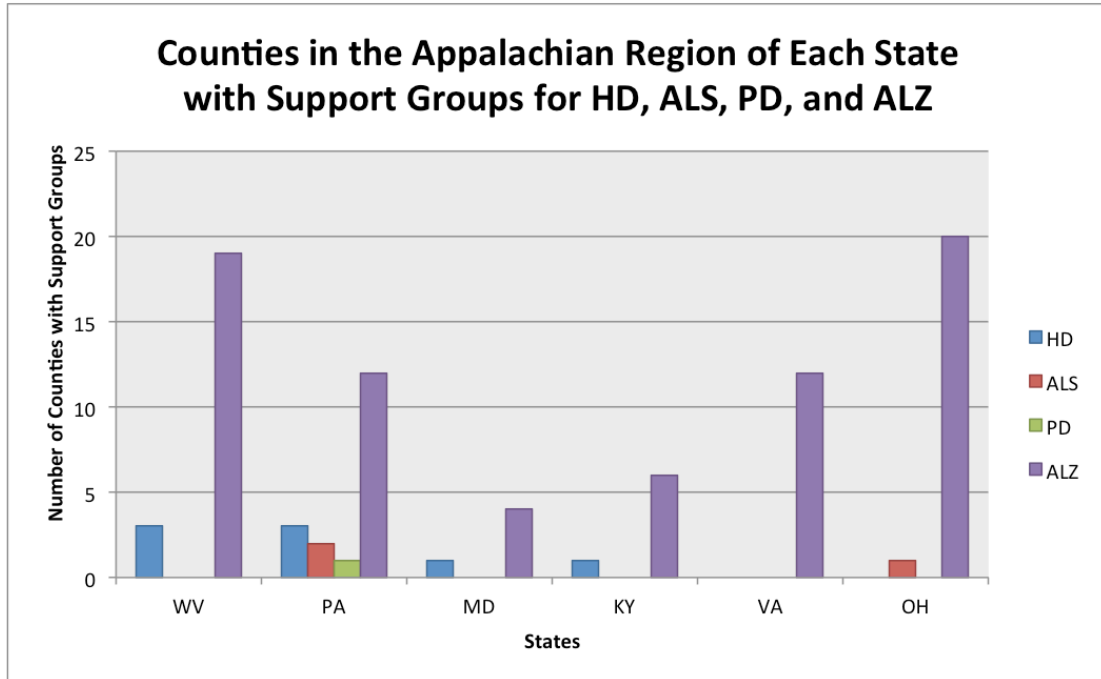
States	Number (percent) of counties with support groups	Number (%) of counties without support groups	Population Size (millions)	Area (1000 sq mi)
WV	20 (36%)	35 (63%)	1.85	24.2
PA	24 (36%)	43 (64%)	12.79	46.1
KY	27 (23%)	93 (77%)	4.41	40.4
MD	19 (79%)	5 (21%)	5.98	12.4
OH	51 (58%)	37 (42%)	11.59	44.8
VA	73 (77%)	22 (23%)	8.33	42.8
Total	214 (48%)	235 (52%)		

Across the six states, the number of counties varied widely from 24 in MD to 120 in KY (Table 2). The difference in the number of counties among the states was not related to the size of the population of the area. Across the six states, the proportion of counties with at least one support group ranged from 23% in KY to 77% in VA. However, over all states (Table 2), the proportion of counties with support groups (48%) was comparable to the proportion of counties without support groups (52%),

The following clustered column charts illustrate the difference of the number of support groups for each disease, designated by both the disorder and state (See Figures 3 and 4).

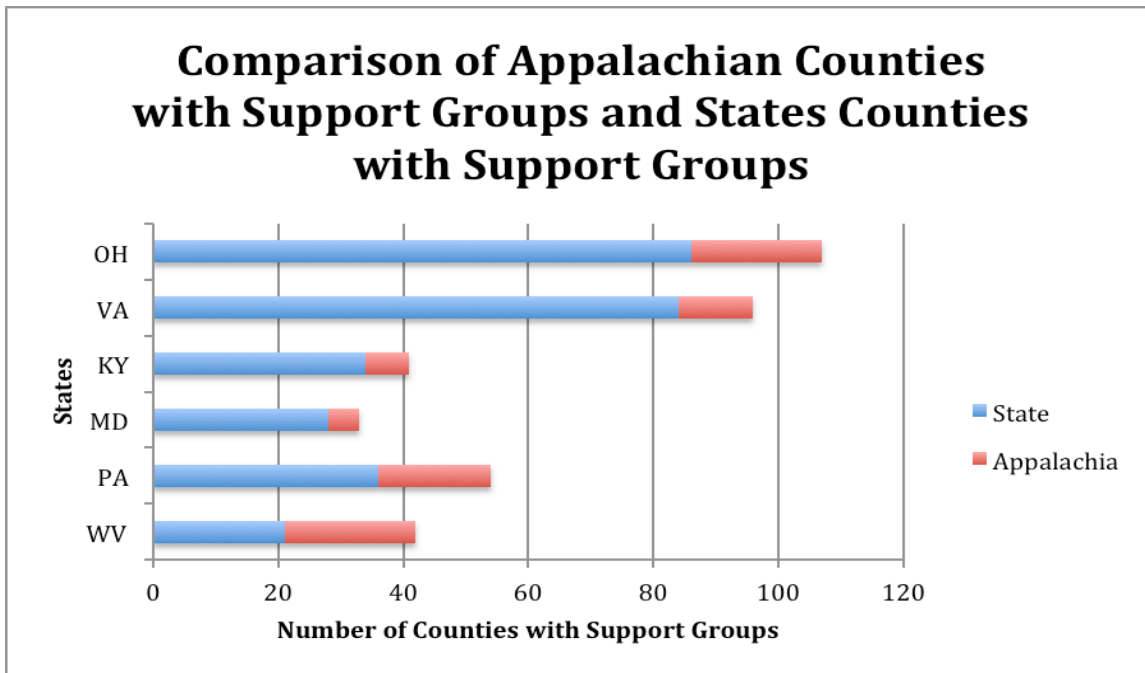


**Figure 2: Count of Counties in each State with Support Groups**



**Figure 3: Count of Counties in the Appalachian Region of each State with Support Groups**

From this data, a disparity can clearly be seen regarding location of support groups. Some states do not have support groups for all diseases within Appalachia, forcing those who do live in this region to travel to utilize these in person resources. This regional disparity can be more clearly seen in Figure 5, which compares the total number of counties with support groups throughout a state (blue) with the number of counties in the Appalachian regions of a state with support groups (red). The support groups for each condition are combined in this graph for visual simplicity (See Figure 5).



**Figure 4: Comparison of Support Groups Availability: Total State Counties and Appalachian Region Counties**

One of the goals of this essay was to assess whether the number of support groups available in Appalachian counties differs from the number of support groups in non-Appalachian counties. Across all states, the proportion of counties with support groups was lower within Appalachian regions compared to the non-Appalachian regions, 34% versus 61%, respectively (Table 3). However, these proportions differ by state. In West Virginia, Maryland, and Ohio, the proportion of counties with support groups is higher within the Appalachian regions than outside the Appalachian region (Table 3). These results are skewed for West Virginia and Maryland because all of the counties in West Virginia are within the Appalachian region, whereas only three counties in Maryland are in Appalachia. For the other three states, the proportion of counties with support groups is lower in the Appalachian region versus the non-Appalachian

region (Table 3). Thus, among these six states, Ohio may be the only one that has similar support group representation both in and out of the Appalachian counties.

**Table 3: Appalachian and Non Appalachian Counties with Support Groups Present and Absent Represented by Count and Percentage**

State	Appalachian Counties with Support Groups Present	Appalachian Counties with Support Groups Absent	Non-Appalachian Counties with Support Groups Present	Non-Appalachian Counties with Support Groups Absent
WV	20 (36%)	35 (64%)	0 (0%)	0 (0%)
PA	15 (29%)	37 (71%)	9 (60%)	6 (40%)
KY	7 (13%)	47 (87%)	20 (30%)	46 (70%)
MD	3 (100%)	0 (0%)	16 (76%)	5 (24%)
OH	21 (67%)	11 (33%)	30 (54%)	26 (46%)
VA	9 (36%)	16 (64%)	64 (91%)	6 (9%)
Total	75 (34%)	146 (66%)	139 (61%)	89 (39%)

I next tested whether the number of Appalachian counties with at least one support group differed from the number of non-Appalachian counties with at least one support group (Table 4). As can be seen, the proportion of counties (34%) with support groups is statistically significantly lower in Appalachian counties compared to the proportion of counties (61%) outside of this defined region ( $X^2 = 32.86$ , 1 df, p value =  $9.881 \times 10^{-9}$ ).

**Table 4: Chi Square Analysis Table (Appalachian Counties and Non Appalachian Counties)**

	Counties with Support Groups	Counties without Support Groups	Total
Appalachian Counties	75 (34%)	146 (66%)	221
Non-Appalachian Counties	139 (61%)	89 (39%)	228
Total	214	235	449



## 5.2 PRESENCE OF SUPPORT GROUPS IN APPALACHIAN REGION BY ECONOMIC STATUS

Most of the support groups were located in major metropolitan areas, and very few of these metropolitan areas were close to counties in Appalachia. I determined the urban (metropolitan) and rural classification of the counties by using the most updated listing of the Rural-Urban Continuum Codes provided by the United States Department of Agriculture Economic Research Service, which classifies counties by population and proximity to metropolitan areas. These codes classify all counties within the United States and organize them into nine different categories (See Figure 2) (Parker, 2013).

<b>2013 Rural-Urban Continuum Codes</b>	
Code	Description
<b>Metro counties:</b>	
1	Counties in metro areas of 1 million population or more
2	Counties in metro areas of 250,000 to 1 million population
3	Counties in metro areas of fewer than 250,000 population
<b>Nonmetro counties:</b>	
4	Urban population of 20,000 or more, adjacent to a metro area
5	Urban population of 20,000 or more, not adjacent to a metro area
6	Urban population of 2,500 to 19,999, adjacent to a metro area
7	Urban population of 2,500 to 19,999, not adjacent to a metro area
8	Completely rural or less than 2,500 urban population, adjacent to a metro area
9	Completely rural or less than 2,500 urban population, not adjacent to a metro area

**Figure 5: Rural-Urban Continuum Code Description for County Categorization (Parker, 2013)**

The proportion of counties within Appalachia that do or do not contain support groups by economic status (distressed or non-distressed) was assessed to answer the question: “Is the economic status of the Appalachian county associated with the presence or absence of support groups? Overall, the proportion of counties with support groups is lower for economically distressed counties (19%) versus economically non-distressed counties (46%) in Appalachia. When examined on a state-by-state basis, this trend applies to every state except Ohio (Table 5). This overall difference in proportions between economically distressed and non-distressed counties was statistically significant ( $X^2 = 17.39$ , 1 df, p-value =  $3.043 \times 10^{-5}$ ).

**Table 5: The Number (Percent) of Distressed and Non Distressed Counties in Appalachia With and Without Support Groups**

State	Distressed Counties with Support Groups Present	Distressed Counties with Support Groups Absent	Non-Distressed Counties with Support Groups Present	Non-Distressed Counties with Support Groups Absent
WV	5 (21%)	19 (79%)	15 (48%)	16 (52%)
PA	0 (0%)	1 (100%)	15 (29%)	36 (69%)
KY	5 (10%)	45 (90%)	2 (50%)	2 (50%)
MD	0 (0%)	0 (0%)	3 (100%)	0 (0%)
OH	9 (69%)	4 (31%)	12 (63%)	7 (37%)
VA	0 (0%)	11 (100%)	9 (64%)	5 (36%)
Total	19 (19%)	80 (81%)	56 (46%)	66 (54%)

)

**Table 6: Chi Square Analysis Table (Distressed Counties and Non Distressed Counties in Appalachia)**

	Counties with Support Groups	Counties without Support Groups	Total
Distressed Counties	19 (19%)	80 (80%)	99
Non-Distressed Counties	56 (46%)	66 (54%)	122
Total	75	146	221

These results indicate that a significant disparity may exist regarding the availability of support groups in the Appalachian regions of these states. Furthermore, the availability of support groups is lower in the distressed counties of the Appalachian region of these states. The only state that had approximately equal availability of support groups in and out of Appalachia was Ohio, but the reasons for this difference are unclear. Nevertheless, more research should be done to further assess these apparent disparities.

## **6.0 DISCUSSION**

### **6.1 PUBLIC HEALTH SIGNIFICANCE OF SUPPORT GROUPS**

The results of the online search for local support groups of the four national support organizations examined in this essay (see Table 2 and Figures 5-7) suggest that there may be a need for more support groups for these disorders in the Appalachian region of the United States, particularly in distressed and at-risk counties. While the results do seem to indicate disparities between the amount of support groups found in Appalachian counties compared to non Appalachian counties, and the amount of support groups in economically distressed Appalachian counties compared to non distressed Appalachian counties, there could be barriers that are limiting support group availability in these areas. These include: lack of resources, lack of facilitators, a smaller number of people that are affected with these disorders, or those who may not feel as though they need support groups. Ohio was the state that seemed to be significantly different, indicating that there is a more equal distribution of support groups throughout the

counties. Every state is different of course, but examining the state of Ohio more thoroughly could be useful in learning lessons about support group distribution.

The four neurological disorders considered in this essay are ideal examples of patient and caregiver populations that may benefit from support groups due to the progressive nature of the diseases, the resulting loss of normal functionality, the lack of research on the prevalence and incidence of some of these disorders, and the genetic features of each disease. Support groups can be a helpful resource to both patients and caretakers, and while there may be online support groups and other resources that they can utilize, having the option of in-person support groups should be an option for those who may not be comfortable with online environments, those who would like more personal interaction, those who do not have access to the internet, and those who are worried about online safety (Malik & Coulson, 2010; Xu, et al., 2013).

Disease awareness is an issue of major public health significance. It is the opinion of the author that support groups serve to exemplify two of the ten essential services of public health, which are 1) “To inform, educate, and empower people about health issues,” and (2) “Link people to needed personal health services and assure the provision of health care when otherwise unavailable” (CDC, 2013). Disease support groups can act as forums that can be used to educate patients, caregivers, and families about the disease, and also to help make disease management better, by promoting connections to other people going through similar circumstances (Locock & Brown, 2010; Mo & Coulson, 2013). Through education and empowerment, support groups can provide group members with more information about various therapies as well as foster partnerships with healthcare professionals, which may lead to better care (Mo & Coulson, 2013). Though support groups can potentially exemplify another essential service of public health, which is mobilizing community partnerships and action to identify and solve health problems,

(CDC, 2013), there has not been sufficient research performed in this area to show that support groups truly provide this service.

For future actions to be taken on the subject of support group placement in central Appalachia, more comprehensive research is needed. If future research suggests a disparity, then a needs assessment can be performed in this region to determine if and/or how support groups would meet the needs of Appalachian communities. A needs assessment is public health tool that determines and analyzes a gap or a “need” that exists between current conditions and desired outcomes (Grant, 2002). This gap is something that must be measured before any public health intervention can take place (Grant, 2002).

## **6.2 ETHICAL CONSIDERATIONS OF SUPPORT GROUP PLACEMENT**

After reviewing the information about support group locations presented in this essay, one may question why there is a disparity that exists in support groups, and why they are placed more often in metropolitan areas. There could be many good reasons for the placement, such as proximity to hospitals and health care providers, placement in areas that are more populous, and placement in areas where vital services for patients are available, but this is speculation. While support groups are needed in major metropolitan areas for these reasons and more, there could potentially be many patients in more rural areas such as Appalachia that are not getting to experience the benefits that these groups can provide.

When considering healthcare ethics and disease support groups, the ethical principles of autonomy, beneficence, and justice can play a role. The concept of beneficence refers to an action, or idea that benefits another person or group of people (Beauchamp, 2013). There is evidence that support groups help both patients and caretakers (Locock & Brown, 2010; Mo &

Coulson, 2013). The true relation between the principle of beneficence and support groups has more to do with ensuring that these services are located in areas where they are needed and can be easily accessed.

Furthermore, beneficence must be a guiding principle for facilitators of support groups to ensure to the best of their abilities that the groups act to benefit participating patients and caregivers. There are a number of barriers that can affect the location of support groups, such as a lack of facilitators, a small number of individuals affected by a particular condition in specific areas, or a lack of interest in a support group by affected individuals, their caregivers, and/or their family members in a specific area. While these barriers are often outside of the control of national support organizations, healthcare facilities, or group facilitators, it would be important to carefully consider approaches that would allow support groups to benefit as many people as possible.

Autonomy is defined as “the measure of the person's independence from influences that control the person's preferences, thoughts, and behavior” (Beauchamp, 2013). Promoting patient and caregiver autonomy is very important, and support groups can help to do this by providing education, knowledge of resources, and social support from others in a similar situation (Locock & Brown, 2010; Mo & Coulson, 2013). A patient or a caregiver who is knowledgeable about a condition and the resources that are available to them may gain additional autonomy in managing and treating that condition.

Justice is also an ethical principle that is important in support group placement. Justice means that there is an equal allocation of resources and their benefits, as well as burdens. A limit of face-to-face support groups and the fact that support groups are not available in every community may lead to patients and caregivers feeling that their choices are limited. The

addition of more online support groups may help, but only one organization for the four disorders facilitated online support groups directly, and with the dangers that online support groups can impose, (Malik & Coulson, 2010) it may be better if online groups were organized and monitored by organizations and sources that patients and caretakers can trust, like the four organizations that are discussed in this essay. However, the barriers described above still exist and more research on these location-based barriers and the methodology of support group placement is needed, though the preferences of support group participants should also be considered.

### **6.3 LIMITATIONS**

This study has several limitations. One of the primary detriments was the general lack of epidemiologic information about the diseases. There is limited data on the prevalence and incidence rate for all four neurological conditions discussed in this paper. This limitation may be due to underreporting or under diagnosis of the diseases, as well as limited education of both patients and healthcare professionals. Another limitation is the lack of research on disease support groups for genetic neurological diseases. Although information regarding the experiences of support groups for other diseases is useful, these neurological disorders are unique, and the same benefits and drawbacks of support groups may not be similar.

There was also a lack of information in certain aspects of the support group investigation. The organizations that provided the support group locations may not have always had an updated website or the best online search tools, and this could have lead to discrepancy in the number of support groups in each state or the counties that were represented in each state. I also did not take into account support groups that are operated by other organizations on a national, state or local

level, or those operated by private citizens. While some disorders had multiple national organizations that facilitated support groups (Parkinson disease), others had one primary national organization that did the same (Huntington disease). The organizations that might have facilitated support groups on a state, local, or private level were difficult to identify and assess, therefore, for consistency across disorders, I decided to compare data obtained from one national organization for each disorder

Discrepancies may have also occurred when information was gathered on the different counties and their economic status. When examining different states, there also must be a consideration of how a state divides up cities and towns into counties. Some cities are located entirely within one county, whereas others may encompass multiple counties. These counties can of course have different economic statuses. The economic status of the counties and the population sizes of the counties also may not be up to date. Furthermore, I did not assess the number of support groups per capita, based the state (or county) population, nor did I assess the support groups by geographic area (of counties, cities, etc). These limitations , also may affect the results.

The various differences between the states and their Appalachian region counties was also a limitation in regards to the research findings of this study. Some states had a majority of their counties as located within the Appalachian region, while other states had very few counties in Appalachia. This difference may have affected the findings listed in the investigation discussion in regards to the differences between the number of counties with support groups within Appalachia versus the number found statewide.

Finally, I did not include the entire Appalachian region of the United States. Data on these additional states might have had an effect on the research findings listed in this essay. More



research that includes all states of the Appalachian region is necessary before actionable conclusions can be drawn.

## **6.4 RECOMMENDATIONS**

The purpose of this essay was to examine a potential disparity in the locations of patient and caretaker support groups for progressive, neurological disorders with genetic components, available through four major, national support organizations specific to these conditions. Drawing data from the Appalachian region of the United States, and choosing to focus on central Appalachia, provided important information regarding differences in access to support groups that exists between economically privileged and distressed areas of the Appalachian region. From the information and data presented in this essay, this disparity does seem to exist in some regard with only 33% of Appalachian counties overall having support groups and only 19% of distressed Appalachian counties having support groups. Noting the many benefits of support groups, and the integral part that this resource can play in disease management, there should be more research and potentially a needs assessment done for central Appalachia to determine if a discrepancy truly does exist and if there is a public health need for support groups for individuals with these disorders. If an established need does exist, a focus group investigation of patient and caretaker support groups similar to the basic focus group investigation described in the Appendix should be undertaken.

Including supplements to formal healthcare services such as support groups can provide valuable information, and empower participants to help themselves and their caretakers to cope with aspects of progressive disease. Today, the importance of patient centered care cannot be overlooked in the United States. Healthcare services that promote patient involvement will

become useful to strengthen the patient-provider relationship, and to promote the well-being of patients, their families, and their caretakers.

## **APPENDIX: PROPOSED FOCUS GROUP INVESTIGATION**

### **A.1: INVESTIGATION GOAL**

This is a proposed project to conduct focus groups on participant opinions about adult-onset neurological disease support group implementation in the Central Appalachian region of the United States. The project will focus on four diseases: Huntington disease (HD), Amyotrophic lateral sclerosis (ALS), Alzheimer disease (AD), and Parkinson disease (PD). The states of the Appalachian region that will be included in this project will be West Virginia, Pennsylvania, Ohio, Kentucky, Maryland, and Virginia. The goal of this investigation will be to gain insight on the level of support available for these disorders, participant perceptions and opinions of support groups, and the type(s) of support groups that they would prefer. This project aims to gather information on disease support groups that could possibly be utilized in the establishment of new support groups, and to inform the actions of stakeholders. The stakeholders of the project are patients, families of patients, caregivers of patients, healthcare providers, disease organizations that facilitate support groups, and government entities such as the local and state health departments. The questions below were developed based on my prior focus group facilitating experience. Audio recordings from each focus group would be transcribed, themes identified, and reports created to be disseminated to stakeholders.

## **A.2: FOCUS GROUP OUTLINE**

There will be a focus on each state for this project, given that every state has different populations, and different healthcare needs. There will be thirty focus groups, five per state taking part in the most Appalachian region of the given state. The focus groups will last two hours, and will be composed of 8-10 participants with two moderators. These moderators will be public health specialists who have experience moderating focus groups, and are familiar with the aspects of the diseases as well as the benefits and drawbacks of support groups.

The participants will include affected patients, the families of affected patients, and the caretakers of affected patients. It is understood that families and caretakers may be one in the same, and this will be an important dynamic to assess during the sessions. The participants will be recruited through advertisements in local healthcare facilities, health departments, nursing facilities and local areas of interest that are highly populous, such as grocery stores and churches. The advertising will be in print and Internet medium. The funding for the project may come from a variety of sources including but not limited to, health departments, organizations for each disease, or national and regional organizations such as the Center for Disease Control and Prevention or the Appalachian Regional Commission. This information can be used to further assess patient, family, and caretaker needs for support groups by any of the funding organizations.

### **A.3: FOCUS GROUP QUESTIONS**

One goal of this focus group project is that each participant has the ability and the opportunity to have their opinions heard, all within a timely manner for others participating in the groups. Therefore, the following open-ended, non leading questions will be asked of the participants, in no particular order.

Do you feel that you have adequate support in this area in managing your illness, the illness of your family member, or patient?

What is your perception of disease support groups?

Do you feel that a support group would be helpful to you?

How would you describe the ideal support group to for your needs?

Would you prefer to meet in person with a support group or use online support groups?

Is it easy for you to participate in the in person support groups that are available in your state?

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