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Amyotrophic Lateral Sclerosis

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Signs and Symptoms

Characterization of the disease is the progression of upper and lower motor neuron degeneration. The first signs and symptoms begin in either the limbs or bulbar muscles, and then spread (Gordon, 2013, pg. 296). The loss of lower motor neuron (LMN) facilitates "fasciculation, cramps, muscle atrophy, and marked weakness" while the upper motor neuron (UMN) loss causes spasticity, hyperreflexia, and weakness (Gordon, 2013, pg. 296). Mimics of these signs include autoimmune motor neuropathy, spinal muscular atrophy, Kennedy's disease, multiple sclerosis, and cervical myelopathy, as well as several others (Lawton et al., 2014, pg. 363). ALS symptoms generally start in the arms, mostly unilaterally and focal such as weakness when lifting arms or loss of hand dexterity (Gordon, 2013, pg. 296). Bulbar-onset carries a worse prognosis compared to limb onset; first symptoms include dysarthria followed by dysphagia which progresses to sialorrhea, malnutrition, and anarthria and a sure diagnosis includes atrophied fasciculating tongue (Gordon, 2013, pg. 296). As the diagnosis progresses, eventually respiratory failure will develop, and the patient may choose to be put on a ventilator, or may opt out and go into hospice as respiratory failure advances. Electromyography confirms disease of LMN while ruling out the mimics, a brain and spinal MRI rule out conditions that could mimic UMN diseases, and occasionally the MRI will show bilateral signal changes within the corticospinal tract which is pathognomonic of ALS (Gordon, 2013, pg. 296).

Underlying Pathophysiology

ALS identification is through the upper and lower motor neuron degeneration and death, by which there is axonal loss and myelin sheath damage (Rossie, Franco, & Estevez, 2013, pg. 2). The effects of this disease are in the ventral horn and brainstem motor neurons through microglial activation and active macrophages (Rossie et al., 2013, pg. 2). Also, the disease is increasingly thought to be a disorder of the RNA metabolism (Doppelmann et al., 2013, pg. 445). Genetic basis of ALS is not known; however, frequently autosomal dominant transmission is shown, although there are instances where autosomal recessive pedigrees are seen (Wingo, Cutler, Yarab, Kelly, & Glass, 2011, pg. 1). The heritability of ALS is estimated to be 76% with the familial form (Wingo et al., 2011, pg. 1). The actual gene that causes this process to happen within the body has been evasive throughout research. Both sporadic and familial ALS have some commonalities, but not every diagnosis provides the same genetic makeup. Currently, one of the most common mutations thought to cause ALS is on the Cu/Zn superoxide dismutase gene (SOD1), which counts for roughly 20% of familial ALS and 1% of spontaneous ALS cases (Droppelmann et al., 2013, pg. 444). There has also been a discovery on a large repeat expansion in the *C9ORF72* gene that accounts for 40% of familial cases of ALS in European and North American populations (Pliner, Mann, & Traynor, 2014, pg. 391). Although the genetic makeup of ALS has not fully been discovered, advances in research are leading to more knowledge as time continues.



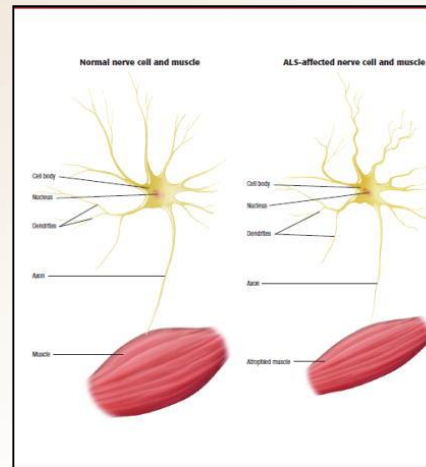
Electromyography is used to diagnose (What is ALS).

Significance of Pathophysiology

Due to the uncertainties in the pathophysiology of amyotrophic lateral sclerosis, significance is held in the current knowledge of the genes that could cause ALS, but more importantly, significance is held in response to knowing that most cases are sporadic. With 90 to 95% of ALS cases being of spontaneous origin and not understanding the genetic makeup thoroughly, physicians must focus on presenting symptoms and do the appropriate tests and evaluations to rule out any other neurodegenerative diseases (Bellomo & Cichminski, 2015, pg. 48). With the increasing development of genetic prevalence, testing could be done to see if the affected individual carries the appropriate genes for further diagnostic evaluation. However, with advanced myelin sheath damage and axonal loss, life expectancy is not long regardless of whether the patient has the gene correlation or not. The diagnosing physician must be thorough in his/her assessment, and do all the appropriate tests to rule out the mimics and give a full diagnosis of ALS, as this disease is not one to take lightly.

Why Amyotrophic Lateral Sclerosis Matters

Amyotrophic Lateral Sclerosis (ALS) is a terminal neurodegenerative disorder affecting an individual's motor neurons. With a prevalence affecting an estimated 20,000 to 30,000 people in the United States of America, the progressive disease affects the lives of many (Bellomo & Cichminski, 2015, pg. 47). Highest rates are in white males, age 60-69 (Prevalence, 2015, pg. 7). Survival rates range from a median of 37 to 49 months once the patient is diagnosed, showing this is a quick progressing disease that will lead to fatality (Malik, Lui, & Lomen-Hoerth, 2014, pg. 534). The patients affected by the disease normally present with asymmetric, painless weakness and possibly impaired speech or swallowing, spasticity, bradykinesia, pathological hyperreflexia, and Babinski signs (Malik et al., 2014, pg. 534). As the disease progresses, patients will require braces, canes, walkers, and eventually wheelchairs (Malik et al., 2014, pg. 535-536). The genetic basis of ALS is undetermined, however there is familial clustering that can suggest a genetic foundation (Wingo, Cutler, Yarab, Kelly, & Glass, 2011, pg. 1). With such uncertainty about the mechanisms of this disease, research is consistently being performed for more information.



(Porth, 2014)

Interest

As a neurological nurse, the mystery behind ALS has always been fascinating. The mechanisms in which the body deteriorates, the cognitive function of the individual suffering from the disease, and the inability to determine the exact cause has stumped science for years. As someone who sees the effects at the hospital bedside and the toll the disease has on the patients and their loved ones, an importance has always been prominent. With such complexity within ALS, interest could only be procured from this disease.

Nursing Care

- Be supportive
- Promote open communication
- Watch for depression and anxiety (Gordon, 2013, pg. 297)
- Due to the rate of progression of the disease, patient's can develop feelings of hopelessness and emotional distress about the future (Abdulla et al., 2014)
- Monitor weight, calories, and fluids to check patient's nutritional status (Gordon, 2013, pg. 300)
- Monitor patient's breathing status
- Determine if mechanical ventilation is an option for the patient-less than 5% of patients opt for mechanical ventilation, and ones that do can progress to locked in syndrome (Gordon, 2013, pg. 301).

Conclusion

- Due to the complexity, constant research must continue to be conducted
- Research should focus on finding the genetic mutations, causes, and eventually a way to avoid or cure the disease
- Presentation of signs and symptoms is important to diagnosis, and ruling out other mimic diseases is vital to diagnosis
- A caring and trusting attitude from the primary care provider is important to establish rapport.
- During early stages of disease, quality of life for the individual should be discussed for future plans, including palliative measures

References

- Abdulla, S., Vielhaber, S., Kollwe, K., Machts, J., Heinze, H., Dengler, R., & Petri, S. (2014). The impact of physical impairment on emotional well-being in ALS. *Amyotrophic Lateral Sclerosis & Frontotemporal Degeneration*, 15(5/6), 392-397. doi: 10.3109/21678421.2014.932380
- Bellomo, T. L., & Cichminski, L. (2015). ALS amyotrophic lateral sclerosis: What nurses need to know. *Nursing*, 45(10), 46-52. doi: 10.1097/01.NURSE.0000471410.04013.6d
- Droppelmann, C. A., Wang, J., Campos-Melo, D., Keller, B., Volkening, K., Hegele, R. A., & Strong, M. J. (2013). Detection for a novel frameshift mutation and regions with homozygosity with ARHGEF28 gene in familial amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis & Frontotemporal Degeneration*, 14(5/6), 444-451. doi: 10.3109/21678421.2012.758288
- Gordon, P. H. (2013). Amyotrophic lateral sclerosis: An update for 2013 clinical features, pathophysiology, management and therapeutic trials. *Aging & Disease*, 4(5), 295310. doi: 10.14336AD.2013.0400295
- Lawton, K. A., Brown, M. V., Alexander, D., Li, Z., Wulff, J. E., Lawson, R., Jaffa, M., Milburn, M. V., Ryals, J. A., Bowser, R., Cudkowicz, M. E., & Berry, J. D. (2014). Plasma metabolomic biomarker panel to distinguish patients with amyotrophic lateral sclerosis from disease mimics. *Amyotrophic Lateral Sclerosis & Frontotemporal Degeneration*, 15(5/6), 362-370. doi: 10.3109/21678421.2014.908311
- Malik, R., Lui, A., & Lomen-Hoerth, C. (2014). Amyotrophic lateral sclerosis. *Seminars in Neurology*, 34(5), 534-541. doi: 10.1055/s-0034-1396007
- Pliner, H., Mann, D., & Traynor, B. (2014). Searching for Grendel: Origin and global spread of the C9ORF72 repeat expansion. *Acta Neuropathol*, 127(3), 391-396. doi: 10.1007/s00401-014-1250-x
- Porth, C. M. (2014). *Essentials of pathophysiology: Concepts of altered health states* (4th ed.). Philadelphia, PA: Wolters Kluwer.
- Prevalence of amyotrophic lateral sclerosis-United States, 2010-2011. (2015). *American Journal of Public Health*, 105(6), e7-e9. doi: 10.2105/AJPH.2015.302747
- Rossi, F. H., Franco, M. C., & Estevez, A. G. (2013). Pathophysiology of amyotrophic lateral sclerosis. *Current Advances in Amyotrophic Lateral Sclerosis*, 1(1-15). doi:10.5772/56562
- Wingo, T. S., Cutler, D. J., Yarab, N., Kelly, C. M., & Glass, J. D. (2011). The heritability of amyotrophic lateral sclerosis in a clinically ascertained United States research registry. *PLoS ONE*, 6(11), 1-5. doi:10.1371/journal.pone.0027985
- What is ALS? (n.d.). *ALS Therapy Development Institute*. Retrieved from <http://www.alstedi.org/what-is-als/>



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