Amyotrophic Lateral Sclerosis

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Recommended Citation
Stitzel, Emily, "Amyotrophic Lateral Sclerosis" (2016). Nursing Student Class Projects (Formerly MSN). 153.
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Underlying Pathophysiology

Signs and Symptoms

ALSI identification is through the upper and lower motor neurons-degeneration and death, by which there is an animal loss and killing neuron damage (Ronsse, Franco, & Estévez, 2013, pg. 2). The effects of this disease are in the ventral horn and brainstem motor neurons through microglial and active macrophages (Ronsse, 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, autoimmune dominant transmission is shown, although there are instances where autosomal recessive pedigrees are seen (Wingo, Curd, Yang, Kelly, & Glass, 2013, pg. 3). The heritability of ALS is estimated to be 76%, with the familial form (Wingo, et al., 2013, pg. 1). The actual cause that gene causes this process to happen within the body has been elusive throughout research. Both sporadic and familial ALS have some commonalities, but not every disease proven to have gene mutations (Doppelmann et al., 2013, pg. 446). Currently, some of the most common mutations thought to cause ALS in 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. 446). There has also been a discovery on a large-repeat expansion in the C9ORF72 gene that accounts for 40% of familial cases of ALS and 10% of spontaneous cases (Wingo, Curd, Yang, Kelly, & Glass, 2013, pg. 1). With such uncertainty about the mechanisms of this disease, research is consistently being performed for more information.

Significance of Pathophysiology

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 that has stumbled science for years. As someone who sees the effects at the hospital bedside and the tell the disease has on the patients and their loved ones, an importance has always been present. With such complexity within ALS, interest could only be procured from this disease.

Why Amyotrophic Lateral Sclerosis Matters

Antomyotrophic Lateral Sclerosis is a terminal neurodegenerative disorder affecting an individual's motor neurons. With a prevalence estimated at 20,000 to 30,000 people in the United States of America, the progressive disease affects the lives of many (Belloccio & Cichmaniski, 2015, pg. 67). Highest rates are in white males, age 60-69 (Prevalence, 2015, pg. 7). Survival rates range from a median of 37 to 49 months after the patient is diagnosed, showing this as a quick progressing disease that will lead to fatality (Malik, Lai, & Lomen-Hoerth, 2014, pg. 534). The patients affected by this disease normally present with asymmetric, pesudoseizures, 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, Curd, Yang, Kelly, & Glass, 2013, pg. 1). With such uncertainty about the mechanisms of this disease, research is consistently being performed for more information.

Conclusion

Nursing Care

Nursing Care

Due to the complexity, constant research must continue to be conducted

- 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 can the progress to locked in syndrome (Gordon, 2013, pg. 301)

References

Wingo, T. S., Curd, D. J., Yang, N., Kelly, C. M., & Glass, J. D. (2013). The heritability of amyotrophic lateral sclerosis in a clinically ascertained United States research registry. PLoS ONE, 8(11), 1-5. doi:10.1371/journal.pone.0079792