Amyotrophic Lateral Sclerosis (ALS)

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How to study ALS

To study ALS, I would begin with a 5 minute video to get a quick overview and lay the mental foundation for the future topics such as: Khan academy ALS video.

The textbooks I recommend for further research or reference is:

- (PT Specific) Physical rehabilitation - O'Sullivan[1]
- Neuromuscular disorders by Amato[2]
- Umphred's neurological rehabilitation[3]
- Continuum - AAN[4]

Lastly, I would then recommend going into true evidence based practice by using scientific articles to find up to date information on the topic.

introduction

What does "Amyotrophic Lateral Sclerosis" even mean?

- a –> no
- myo –> muscle
- Trophic –> Nourishment Thus Amyotrophic means "no muscle nourishment"

Amyotrophic lateral sclerosis (ALS), AKA Lou Gehrig's disease, is the most common and devastatingly fatal motor neuron disease (MND) among adults[1]. ALS is characterized by the degeneration and loss of motor neurons in the spinal cord, brainstem, and brain, resulting in UMN and LMN clinical signs and symptoms[1]. Recently, ALS is being recategorized as a multisystem disorder/syndrome with variable pathological involvement of extra-motor networks and connections, in addition to the LMNs and UMNs[1]

Epidemiology

Age

ALS can occur at any age but onset generally occurs in the mid-to-late 50s[1].

Gender

Most studies have found that the disease affects men slightly more than women, with an approximate ratio of 1.7:1. After age 65, the gender difference decreases[1].

Family History

About 5% to 10% of individuals have a family history of ALS (familial ALS, [FALS])[1]. Familial ALS is phenotypically and genetically heterogeneous[1].

Most cases of FALS are autosomal dominant[1]. Regardless, recessive and X-linked forms have been described[1]. For example, the rare juvenileonset ALS is reported to be inherited in an autosomal recessive pattern[1].

FALS Categorization

FALS is categorized by mode of inheritance and further subcategorized by specific gene or chromosomal locus@osullivanPhysicalRehabilitation2019.

The very large majority of adult individuals with ALS have no family history of the disease (sporadic ALS)[1].

> **Note**
>
> A very small percentage of individuals with sporadic ALS do have a mutation in SOD1

Over 20 chromosomal regions and a number of identified genes have been linked to ALS. 20% of hereditary ALS cases are attributed to one of 100+ mutations in superoxide dismutase 1 (SOD1)[1]. ~50% of individuals with an SOD1 ALS variant are symptomatic by 46yrs, and 90% are symptomatic by 70 years of age[1].

Onset

- ~70% to 80% of individuals develop limb-onset ALS, with initial involvement in the extremities[1].
- 20% to 30% develop bulbar-onset ALS, with initial involvement in the bulbar muscles[1].
- Bulbar-onset ALS is more common in middle-aged women, and initial symptoms may include difficulty speaking, chewing, or swallowing[1]

References

1. O'Sullivan SB, Schmitz TJ, Fulk GD, eds. *Physical Rehabilitation*. 7th ed. F.A. Davis Company; 2019.
2. Amato AA, Russell JA. *Neuromuscular Disorders*. 2nd ed. McGraw-Hill Education; 2016.
3. Lazaro RT. *Umphred's Neurological Rehabilitation*. 7th ed. Elsevier, Inc; 2019.
4. Goutman SA. Diagnosis and Clinical Management of Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders. *CONTINUUM: Lifelong Learning in Neurology*. 2017;23(5):1332-1359. doi:10.1212/CON.0000000000000535