Narcolepsy or Gelineau Syndrome

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Underlying Pathophysiology

The exact cause of narcolepsy is not completely understood, however, the most current research suggests a combination of genetic and environmental factors which include the following:

- Genetic predisposition:
- Genetic markers such as the generation of potential pathogenic naïve T cells
- Inadequate central tolerance
- Cross-stimulation with various environmental toxins at the wrong developmental time
- Penetration of naïve T cells into the central nervous system
- Destruction of hypocretin neurons by CD8+ T cells or other mechanisms (Mignot, 2014, p. 331).

Signs and Symptoms

Narcolepsy, or narcolepsy-cataplexy, was first described by the French neurologist Jean-Baptiste Gélineau (1859) as a disorder that “occurs once or twice in a lifetime.” Symptoms may commence at any age. Diagnosis findings are considered rare, with approximately 1 in 2,000 people being affected (Ruoff, 2010). There is no cure for narcolepsy, symptoms can typically be managed with medication and lifestyle changes that include taking short naps, avoiding caffeine or alcohol before bed, and no meals before bedtime.

Pathological Processes

Significance of Pathophysiology

There are two major classifications of narcolepsy: Type 1 (previously known as narcolepsy with cataplexy), in which the patient will have either low levels of hypocretin or report cataplexy; and Type 2 narcolepsy, which is associated with elevated levels of hypocretin. In Type 2 narcolepsy, there is considered to be secondary narcolepsy that results from the lack of hypocretin (The National Institute of Neurological Disorders and Stroke, 2018).

Auxiliary symptoms may be experienced by individuals with narcolepsy but not part of the considered diagnostic criteria.

- Strepococcal antigens
- Hypothyroidism
- Autoimmune processes
- Hypocretin deficiency
- Hypocretin antibodies, which cause REM sleep paralysis, and prevent REM sleep from occurring at inappropriate times.

Sleep Paralysis

Sleep paralysis resembles cataplexy except it occurs at the edges of sleep (The National Institute of Neurological Disorders and Stroke, 2018).

While individuals may present with EDS, they typically will experience sleep-onset paralysis, meaning they may have difficulty staying asleep at night, or sleep for short periods of time during the day. Cataplexy, characterized by sudden daytime muscular paralysis, is considered to be controlled by a key neurotransmitter produced in the hypothalamus. Hypocretin helps sustain alertness and prevent REM sleep from occurring at inappropriate times (The National Institute of Neurological Disorders and Stroke, 2018).

There are several medications currently available to treat narcolepsy, including dextroamphetamine (Metadate, Nuvigil, Vivactil), modafinil (Provigil), and stimulants such as oxybate (Xyrem). There are a number of medications recommended for the treatment of narcolepsy, including dextroamphetamine, modafinil, and stimulants such as oxybate (Xyrem). These medications are considered to be controlled by a key neurotransmitter produced in the hypothalamus. Hypocretin helps sustain alertness and prevent REM sleep from occurring at inappropriate times (The National Institute of Neurological Disorders and Stroke, 2018).

In conclusion, although considered rare, the neurological disorder narcolepsy presents itself in the family of the author of this academic poster. Narcolepsy presents a unique experience to the family and community, but it shows no significant effect on either other family members. The disease was quite debilitating for her sister. Although there is no cure for narcolepsy, symptoms can typically be managed with medication and lifestyle changes that include taking short naps, avoiding caffeine or alcohol before bed, and no meals before bedtime. Narcolepsy presents a unique experience to the family and community, but it shows no significant effect on either other family members. The disease was quite debilitating for her sister.