DM1

DM1 is a rare neuromuscular disorder with multi-systemic involvement

Presentation

DM1 is a rare, progressive, genetic, multi-system, neuromuscular disease with high morbidity and early mortality[1–9]

Muscular symptoms involve skeletal, cardiac, and smooth muscles, and manifestations include myotonia, muscle weakness, atrophy, difficulty breathing, cardiac conduction defects, and GI symptoms[1,3]

Non-muscular symptoms mainly affect the CNS, including fatigue, daytime somnolence, and cognitive impairment, but can also include cataracts, male infertility, endocrine abnormalities, and a higher incidence of cancer[1,3,9]

The estimated global pooled prevalence of DM1 is 9.27 cases per 100,000 individuals[6]

Burden of Illness

Symptoms impair many aspects of daily life, including mobility, performing household activities, communication, cognitive functioning, and social interactions[9,10]

Individuals with DM have lower labor force participation and annual income than the general population, despite comparable educational attainment[10,11]

Diagnosis

Early diagnosis can help ensure that individuals have the opportunity to be managed by a multi-disciplinary team so that known DM1 risk factors are recognized and addressed in a timely manner[3]

The average diagnostic delay for DM1 is 6–7 years, which tends to be greater in those whose initial symptoms are non-muscular and/or non-specific in nature, such as fatigue and sleep disturbances[9,12,13]

Disease awareness is paramount to detecting DM1 cases earlier and preventing misdiagnosis[12]

Treatment

There are no approved disease-modifying therapies for DM1, with current management consisting of supportive measures and a multidisciplinary approach to manage multisystem disease manifestations[2,14–16]

Novel therapeutic agents are in development to target the underlying cause of DM1 and correct splicing[17]

CNS, central nervous system; DM1, myotonic dystrophy type 1; GI, gastrointestinal.

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