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Development of a standard of care for patients with valosin-containing protein associated multisystem proteinopathy

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Valosin-containing protein (VCP) associated multisystem proteinopathy (MSP) is a rare inherited disorder with a reported incidence of 0.66/100,000 based on population data from the UK. VCP-associated MSP may result in multisystem involvement of varying presentations and phenotypes including inclusion body myopathy, Paget's disease of bone, frontotemporal dementia, parkinsonism, and amyotrophic lateral sclerosis, among others. An international multidisciplinary consortium of 50+ experts in neuromuscular disease, dementia, psychology, cardiology, pulmonology, physical therapy, occupational therapy, speech and language pathology, nutrition, genetics, and endocrinology were convened by the patient advocacy organization, Cure VCP Disease, in 2021 to elicit collaboration and development of a standard of care for this rare and under-diagnosed disease. To achieve this goal, working groups were established to generate best evidence recommendations in 10 key areas: genetics and diagnosis, myopathy, frontotemporal dementia, Paget's disease of bone, ALS and CMT, parkinsonism, cardiomyopathy, supportive therapies, pulmonology, nutrition and supplements, and mental health. In April 2021, facilitated discussion of each working group's conclusion with consensus building techniques enabled final agreement on the recommended standard of care for patients with VCP-associated MSP. Timely referral to a specialty neuromuscular center is recommended to aid in efficient diagnosis of VCP-associated MSP via single-gene testing in the case of a known familial VCP variant, or multi-gene panel sequencing in undifferentiated cases. Additionally, regular and ongoing multidisciplinary team follow up is essential for proactive screening and management of the secondary complications. The goal of our consortium is to raise awareness of VCP-associated MSP, to expedite the time to accurate diagnosis and elevate the recommended best practices guidelines for multidisciplinary care internationally.

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