# Neuromuscular Disease Symposium 15 Nov 2020

**Abstract**

**Topic: (1). The role of serology in the modern era of myositis. (2) Diagnosis, treatment and pathogenesis of immune-mediated necrotising myopathy.**

**Speaker: Prof. Andrew Mammen (USA)**

**The role of serology in the modern era of myositis**

The inflammatory myopathies (IM) are a heterogeneous family of systemic autoimmune diseases that includes dermatomyositis (DM), the antisynthetase syndrome (ASyS), immune-mediated necrotizing myopathy (IMNM), and inclusion body myositis (IBM). Many IM patients have myositis-specific autoantibodies (MSAs) that define distinct phenotypic subtypes. In addition, some IM patients have one or more myositis-associated autoantibodies (MAA) that are associated with certain clinical features. In this presentation, Dr. Mammen will discuss how MSAs and MAAs can be used to diagnose and manage IM patients.

**Diagnosis, treatment, and pathogenesis of immune-mediated necrotizing myopathy.**

Immune-mediated necrotizing myopathy (IMNM) is now recognized as a distinct type of inflammatory myopathy (IM) characterized by muscle biopsies with myofiber necrosis and regeneration but without prominent endomysial inflammation or perifascicular atrophy. IMNM patients can be further subdivided into those with anti-HMG-CoA reductase autoantibodies, those with anti-SRP autoantibodies, and those without a known myositis-specific autoantibody. Each of these three subtypes of IMNM is characterized by unique immunogenetic risk factors, prognosis, and optimal treatment strategies. In this presentation, Dr. Mammen will discuss the diagnosis, prognosis, treatment, and pathogenesis of each IMNM subtype.

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**Topic: Spinal Muscular Atrophy: New Treatment and Challenge**

**Speaker: Dr Sophelia Hoi Shan Chan (HK)**

Spinal muscular atrophy (SMA) is a hereditary neuromuscular disorder with an autosomal recessive inheritance and a spectrum of clinical presentations from the severe lethal infantile-onset SMA type I to the adult-onset SMA type IV. The disease is caused by mutations of the *SMN1* gene leading to deficiencies of SMN protein. Additionally, the number of copies of *SMN2* gene, a disease-modifier gene, which produces a small quantity of SMN protein, plays a major role in determining the clinical severity of the disease. Without disease-modifying drugs, all patients have progressive weakness and deterioration of health over time. The impact on the patients and families are profound. This presentation will explain how the improved understandings of the molecular basis and natural history of SMA support the development of novel therapeutic strategies; as well as recognise the impact of the standardized care on outcomes. Therapeutic strategies in the pipeline, ranging from the modulation of *SMN2* encoded transcripts to *SMN1* gene replacement therapy, will be evaluated. This presentation will also share the local experience of the Hong Kong SMA treatment programme with nusinersen, which was started in 2018. The changes in motor outcome, health-related quality of life, and cerebrospinal fluid neurofilament level, pre-and-post-treatment, will be appraised. The way forward, which includes advancing the pre-symptomatic diagnosis and screening programme, establishing a transition of care programme, and setting up monitoring programme to determine the long-term impacts of the disease-modifying treatments and further improvements in the supportive care, will be discussed.