Special Issue

Molecular Genetics of Malignant Hyperthermia Susceptibility and Related Diseases

Message from the Guest Editor

Malignant hyperthermia (MH) is a pharmacogenetic disorder of skeletal muscle that manifests as a hypermetabolic reaction in susceptible individuals upon exposure to halogenated inhalational anesthetic or succinylcholine. Susceptibility to MH has been associated with pathogenic variants in RYR1, CACNA1S, and STAC3 genes. Dominant variants in RYR1 account for about 70% of MH cases. RYR1 encodes the skeletal muscle calcium-release channel, which plays a central role in muscle Ca2+ regulation, linking surface membrane potential to muscle contraction. Non-anesthesia-related environmental factors such as exercise, heat, or both may trigger MH. This led to finding RYR1 variants in environmentally triggerable conditions such as exertional rhabdomyolysis and exertional heat illness. Dominant and recessive variants in RYR1 have also been associated with various non-dystrophic myopathies that include central core disease, multi-minicore disease, centronuclear myopathy, and congenital fiber type disproportion. These disorders are considered to be RYR1-related disorders. This Special Issue is focused on the genetic contribution to MH susceptibility, and RYR1-related disorders.

Guest Editor

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