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EFHC1 Antibody Blocking Peptide

Catalog Number: bs-9013P
Activity: Not tested
Purification: HPLC
Storage: Shipped at 4°C. Stored at -20°C for one year. Avoid repeated freeze/thaw cycles.
Background: Defects in EFHC1 are the cause of juvenile myoclonic epilepsy type 1 (EJM1) [MIM:254770]. EJM1 is a subtype of idiopathic generalized epilepsy (IGE). Patients have afebrile seizures only, with onset in adolescence (rather than in childhood) and myoclonic jerks which usually occur after awakening and are triggered by sleep deprivation and fatigue.
Genetic variations in EFHC1 are the cause of susceptibility to juvenile absence epilepsy type 1 (JAE1) . JAE is a subtype of idiopathic generalized epilepsy characterized by onset occurring around puberty, absence seizures, generalized tonic-clonic seizures (GTCS), GTCS on awakening, and myoclonic seizures.