

bs-9013R**[Primary Antibody]****EFHC1 Rabbit pAb**

www.bioss.com.cn

sales@bioss.com.cn

techsupport@bioss.com.cn

400-901-9800

— DATASHEET —

Host: Rabbit	Isotype: IgG	Applications: IHC-P (1:100-500) IHC-F (1:100-500) IF (1:50-200) ELISA (1:5000-10000) Reactivity: (predicted: Human, Mouse, Rat, Pig, Sheep, Cow, Chicken, Dog, Horse) Predicted MW.: 74 kDa Subcellular Location: Cytoplasm
Clonality: Polyclonal		
GeneID: 114327	SWISS: Q5JVL4	
Target: EFHC1		
Immunogen: KLH conjugated synthetic peptide derived from human EFHC1: 301-400/640.		
Purification: affinity purified by Protein A		
Concentration: 1mg/ml		
Storage: 0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol. Shipped at 4°C. Store 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.		