bs-7854R **[Primary Antibody]**

AHI1 Rabbit pAb

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DATASHEET

Host: Rabbit **Isotype:** IgG

Clonality: Polyclonal

GeneID: 54806 **SWISS:** Q8N157

Target: AHI1

Immunogen: KLH conjugated synthetic peptide derived from human AHI1: 801-900/1196.

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: Highly expressed in the most primitive normal hematopoietic cells. Expressed in brain, particularly in neurons that give rise to the crossing axons of the corticospinal tract and superior cerebellar peduncles. Expressed in kidney (renal collecting duct cells) (at protein level).
Involvement in disease:Defects in AHI1 are the cause of Joubert syndrome type 3 (JBTS3) . JBTS is an autosomal recessive disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. JBTS3 shows minimal extra central nervous system involvement and appears not to be associated with renal dysfunction.

Applications: **WB** (1:500-2000)
IHC-P (1:100-500)
IHC-F (1:100-500)
IF (1:100-500)
ELISA (1:5000-10000)

Reactivity: (predicted: Human, Mouse, Rat, Rabbit, Pig, Sheep, Cow, Dog, Horse)

Predicted MW.: 137 kDa

Subcellular Location: Cell membrane ,Cytoplasm