

Product name:	GAS3 Rabbit Polyclonal Antibody
Cat number:	ABN11300
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human PMP22. AA range:111-160
Reactivity:	Human,Mouse,Rat
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:20000-1:40000
Molecular Weight:	22kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

This gene encodes an integral membrane protein that is a major component of myelin in the peripheral nervous system. Studies suggest two alternately used promoters drive tissue-specific expression. Various mutations of this gene are causes of Charcot-Marie-Tooth disease Type IA, Dejerine-Sottas syndrome, and hereditary neuropathy with liability to pressure palsies. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Jul 2013],disease:Defects in PMP22 are a cause of Dejerine-Sottas syndrome (DSS) [MIM:145900]; also known as Dejerine-Sottas neuropathy (DSN) or hereditary motor and sensory neuropathy III (HMSN3). DSS is a severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. DSS is characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome.,disease:Defects in PMP22 are a cause of hereditary neuropathy with liability to pressure palsies (HNPP) [MIM:162500]; an autosomal dominant disorder characterized by transient episodes of decreased perception or peripheral nerve palsies after slight traction, compression or minor traumas.,disease:Defects in PMP22 are the cause of Charcot-Marie-Tooth disease type 1A (CMT1A) [MIM:118220]; also known as hereditary motor and sensory neuropathy IA. CMT1A is a form of Charcot-Marie-Tooth disease, the most common inherited disorder of the peripheral nervous system. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathy or CMT1, and primary peripheral axonal neuropathy or CMT2. Neuropathies of the CMT1 group are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. CMT1A inheritance is autosomal dominant.,disease:Defects in PMP22 are the cause of Charcot-Marie-Tooth disease type 1E (CMT1E) [MIM:118300]; also known as Charcot-Marie-Tooth disease and deafness autosomal dominant. CMT1E is an autosomal dominant form of Charcot-Marie-Tooth disease characterized by the association of sensorineural hearing loss with peripheral demyelinating neuropathy.,disease:Defects in PMP22 may be a cause of inflammatory demyelinating polyneuropathy (IDP) [MIM:139393]. IDP is a putative autoimmune disorder presenting in an acute (AIDP) or chronic form (CIDP). The acute form is also known as Guillain-Barre syndrome.,function:Might be involved in growth regulation, and in myelination in the peripheral nervous system.,similarity:Belongs to the PMP-22/EMP/MP20 family.,