
C9orf117 Antibody Blocking Peptide

Catalog Number: bs-9495P

Activity: Not tested

Purification: HPLC

Storage: Shipped at 4°C. Stored at -20°C for one year. Avoid repeated freeze/thaw cycles.

Background: C9orf117 is a 520 amino acid protein that exists as two alternatively spliced isoforms. The gene encoding C9orf117 maps to human chromosome 9q34.11. Chromosome 9 consists of about 145 million bases, represents 4% of the human genome and encodes nearly 900 genes. Thought to play a role in gender determination, deletion of the distal portion of 9p can lead to development of male to female sex reversal, the phenotype of a female with a male X,Y genotype. Hereditary hemorrhagic telangiectasia, which is characterized by harmful vascular defects, is associated with the chromosome 9 gene encoding endoglin protein, ENG. Familial dysautonomia is also associated with chromosome 9 though through the gene IKBKAP. Notably, chromosome 9 encompasses the largest interferon family gene cluster. Chromosome 9 is partnered with chromosome 22 in the translocation leading to the aberrant production of BCR-ABL fusion protein often found in leukemias.