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## **ANKRD50 Antibody Blocking Peptide**

Catalog Number: bs-9751P

Activity: Not tested

Purification: HPLC

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

Background: Ankyrins are membrane adaptor molecules that play important roles in coupling integral

membrane proteins to the spectrin-based cytoskeleton network. Mutations of ankyrin genes

lead to severe genetic diseases, such as fatal cardiac arrhythmias and hereditary

spherocytosis. ANKRD50 (ankyrin repeat domain 50) is a 1,429 amino acid phosphoprotein

that contains nineteen ANK repeats. Conserved in chimpanzee,  $\operatorname{\mathsf{dog}}$  ,  $\operatorname{\mathsf{cow}}$  ,  $\operatorname{\mathsf{mouse}}$  ,  $\operatorname{\mathsf{rat}}$  ,

chicken, zebrafish, fruit fly and mosquito, ANKRD50 is encoded by a gene that maps to

human chromosome 4q28.1. Chromosome 4 represents approximately 6% of the human

genome and contains nearly 900 genes. Notably, the Huntingtin gene, which encodes an

 $expanded \ glutamine \ tract \ in \ cases \ of \ Huntington's \ disease, is \ located \ on \ chromosome \ 4.$ 

FGFR-3 is also encoded by a gene that maps to human chromosome 4 and has been

associated with thanatophoric dwarfism, achondroplasia, Muenke syndrome and bladder

cancer. Chromosome 4 is also linked to Ellis-van Creveld syndrome, methylmalonic

acidemia and polycystic kidney disease.