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C12ORF61 Antibody Blocking Peptide

Catalog Number: bs-9953P
Activity: Not tested
Purification: HPLC
Storage: Shipped at 4°C. Stored at -20°C for one year. Avoid repeated freeze/thaw cycles.
Background: Encoding over 1,100 genes within 132 million bases, chromosome 12 makes up about 4.5% of the human genome. A number of skeletal deformities are linked to chromosome 12 including hypochondrogenesis, achondrogenesis and Kniest dysplasia. Noonan syndrome, which includes heart and facial developmental defects among the primary symptoms, is caused by a mutant form of PTPN11 gene product, SH-PTP2. Chromosome 12 is also home to a homeobox gene cluster which encodes crucial transcription factors for morphogenesis, and the natural killer complex gene cluster encoding C-type lectin proteins which mediate the NK cell response to MHC I interaction. Trisomy 12p leads to facial development defects, seizure disorders and a host of other symptoms varying in severity depending on the extent of mosaicism and is most severe in cases of complete trisomy. The C12orf61 gene product has been provisionally designated C12orf61 pending further characterization.