

C12orf40 抗原（重组蛋白）

中文名称： C12orf40 抗原（重组蛋白）

英文名称： C12orf40 Antigen (Recombinant Protein)

别名： chromosome 12 open reading frame 40; HEL-206; HEL-S-94

相关类别： 抗原

储存： 冷冻（-20℃）

概述

技术规格

Full name:	chromosome 12 open reading frame 40
Synonyms:	HEL-206; HEL-S-94
Swissprot:	Q86WS4
Gene Accession:	BC048120
Purity:	>85%, as determined by Coomassie blue stained SDS-PAGE
Expression system:	Escherichia coli
Tags:	His tag C-Terminus, GST tag N-Terminus
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.

n. 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 C12orf40 gene product has been provisionally designated C12orf40 pending further characterization.