

# Recombinant Human SHH

**Cat. No.** SHH-30499TH    **Lot. No.** (See product label)

## SPECIFICATION

|                         |                                                                                                                                                                                                      |
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| <b>Product Overview</b> | Recombinant Human Sonic Hedgehog fragment consisting of 174 amino acid residues, which correspond to the active N-terminal portion of the Sonic Hedgehog precursor expressed in High-5 Insect Cells. |
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| <b>Species</b> | Human |
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| <b>Description</b> | <p>This gene encodes a protein that is instrumental in patterning the early embryo. It has been implicated as the key inductive signal in patterning of the ventral neural tube, the anterior-posterior limb axis, and the ventral somites. Of three human proteins showing sequence and functional similarity to the sonic hedgehog protein of <i>Drosophila</i>, this protein is the most similar. The protein is made as a precursor that is autocatalytically cleaved; the N-terminal portion is soluble and contains the signalling activity while the C-terminal portion is involved in precursor processing. More importantly, the C-terminal product covalently attaches a cholesterol moiety to the N-terminal product, restricting the N-terminal product to the cell surface and preventing it from freely diffusing throughout the developing embryo. Defects in this protein or in its signalling pathway are a cause of holoprosencephaly (HPE), a disorder in which the developing forebrain fails to correctly separate into right and left hemispheres. HPE is manifested by facial deformities. It is also thought that mutations in this gene or in its signalling pathway may be responsible for VACTERL syndrome, which is characterized by vertebral defects, anal atresia, tracheoesophageal fistula with esophageal atresia, radial and renal dysplasia, cardiac anomalies, and limb abnormalities. Additionally, mutations in a long range enhancer located approximately 1 megabase upstream of this gene disrupt limb patterning and can result in preaxial</p> |
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|                              | polydactyly.                                                                                                                                                                 |
| <b>Tissue specificity</b>    | Expressed in fetal intestine, liver, lung, and kidney. Not expressed in adult tissues.                                                                                       |
| <b>Biological activity</b>   | Biological Activity : Determined by its ability to induce alkaline phosphatase production by C3H/10T1/2 (CCL-226) cells. The expected ED50 for this effect is 0.8-1.0 µg/ml. |
| <b>Form</b>                  | Lyophilised                                                                                                                                                                  |
| <b>Purity</b>                | >95% by SDS-PAGE                                                                                                                                                             |
| <b>Storage</b>               | Aliquot and store at -80°C. Avoid repeated freeze / thaw cycles.                                                                                                             |
| <b>Sequence Similarities</b> | Belongs to the hedgehog family.                                                                                                                                              |
| <b>GENE INFORMATION</b>      |                                                                                                                                                                              |
| <b>Gene Name</b>             | SHH sonic hedgehog [ Homo sapiens ]                                                                                                                                          |
| <b>Official Symbol</b>       | SHH                                                                                                                                                                          |
| <b>Synonyms</b>              | SHH; sonic hedgehog; HLP3, HPE3, sonic hedgehog (Drosophila) homolog , sonic hedgehog homolog (Drosophila); sonic hedgehog protein; HHG1; MCOPCB5; SMMCI; TPT; TPTPS;        |
| <b>Gene ID</b>               | 6469                                                                                                                                                                         |
| <b>mRNA Refseq</b>           | NM_000193                                                                                                                                                                    |

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| <b>Protein Refseq</b>      | NP_000184                                                                                                                                                                                                                                                                            |
| <b>MIM</b>                 | 600725                                                                                                                                                                                                                                                                               |
| <b>Uniprot ID</b>          | Q15465                                                                                                                                                                                                                                                                               |
| <b>Chromosome Location</b> | 7q36                                                                                                                                                                                                                                                                                 |
| <b>Pathway</b>             | Basal cell carcinoma, organism-specific biosystem; Basal cell carcinoma, conserved biosystem; Class B/2 (Secretin family receptors), organism-specific biosystem; FOXA1 transcription factor network, organism-specific biosystem; GPCR ligand binding, organism-specific biosystem; |
| <b>Function</b>            | calcium ion binding; glycoprotein binding; glycosaminoglycan binding; laminin-1 binding; patched binding;                                                                                                                                                                            |

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