

## **Product datasheet for SC325589**

## OriGene Technologies, Inc.

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## HSD3B7 (NM\_001142778) Human Untagged Clone

## **Product data:**

**Product Type:** Expression Plasmids

**Product Name:** HSD3B7 (NM\_001142778) Human Untagged Clone

Tag: Tag Free
Symbol: HSD3B7

Synonyms: CBAS1; PFIC4; SDR11E3

**Vector:** pCMV6 series

Fully Sequenced ORF: >NCBI ORF sequence for NM\_001142778, the custom clone sequence may differ by one or

more nucleotides

CCTGGATGCACGTGCTGGCAGCCCGGGAGCTGGAGCAGCGGGCAACCC

**Restriction Sites:** Please inquire ACCN: NM\_001142778

**OTI Disclaimer:** Our molecular clone sequence data has been matched to the reference identifier above as a

point of reference. Note that the complete sequence of our molecular clones may differ from the sequence published for this corresponding reference, e.g., by representing an alternative

RNA splicing form or single nucleotide polymorphism (SNP).

OTI Annotation: This TrueClone is provided through our Custom Cloning Process that includes sub-cloning

into OriGene's pCMV6 vector and full sequencing to provide a non-variant match to the expected reference without frameshifts, and is delivered as lyophilized plasmid DNA.

**Components:** The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube

containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).





**Reconstitution Method:** 

- 1. Centrifuge at 5,000xg for 5min.
- 2. Carefully open the tube and add 100ul of sterile water to dissolve the DNA.
- 3. Close the tube and incubate for 10 minutes at room temperature.
- 4. Briefly vortex the tube and then do a quick spin (less than 5000xg) to concentrate the liquid at the bottom.

5. Store the suspended plasmid at -20°C. The DNA is stable for at least one year from date of shipping when stored at -20°C.

RefSeq: <u>NM 001142778.1</u>, <u>NP 001136250.1</u>

 RefSeq Size:
 2040 bp

 RefSeq ORF:
 591 bp

 Locus ID:
 80270

 UniProt ID:
 Q9H2F3

 Cytogenetics:
 16p11.2

**Protein Families:** Transmembrane

**Protein Pathways:** Metabolic pathways, Primary bile acid biosynthesis

**Gene Summary:** This gene encodes an enzyme which is involved in the initial stages of the synthesis of bile

acids from cholesterol and a member of the short-chain dehydrogenase/reductase

superfamily. The encoded protein is a membrane-associated endoplasmic reticulum protein which is active against 7-alpha hydrosylated sterol substrates. Mutations in this gene are associated with a congenital bile acid synthesis defect which leads to neonatal cholestasis, a form of progressive liver disease. Multiple transcript variants encoding different isoforms

have been found for this gene. [provided by RefSeq, Dec 2008]

Transcript Variant: This variant (3) lacks an exon in the 3' coding region which results in a frameshift and an early stop codon, compared to variant (1), resulting in a shorter protein

(isoform b), compared to isoform a. Variants 2 and 3 encode the same protein.