

## **Product datasheet for SC302225**

## OriGene Technologies, Inc.

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## GTP cyclohydrolase 1 (GCH1) (NM\_001024024) Human Untagged Clone

**Product data:** 

**Product Type:** Expression Plasmids

**Product Name:** GTP cyclohydrolase 1 (GCH1) (NM\_001024024) Human Untagged Clone

Tag: Tag Free

**Symbol:** GTP cyclohydrolase 1

Synonyms: DYT5; DYT5a; DYT14; GCH; GTP-CH-1; GTPCH1; HPABH4B

**Vector:** pCMV6 series

Fully Sequenced ORF: >NCBI ORF sequence for NM\_001024024, the custom clone sequence may differ by one or

more nucleotides

CGGGAAGAGTTCCTGACTCTCATTAGGAGCTGA

**Restriction Sites:** Please inquire **ACCN:** NM 001024024

**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 001024024.1</u>, <u>NP 001019195.1</u>

 RefSeq Size:
 1995 bp

 RefSeq ORF:
 753 bp

 Locus ID:
 2643

 UniProt ID:
 P30793

 Cytogenetics:
 14q22.2

**Protein Families:** Druggable Genome

**Protein Pathways:** Folate biosynthesis, Metabolic pathways

**Gene Summary:** This gene encodes a member of the GTP cyclohydrolase family. The encoded protein is the

first and rate-limiting enzyme in tetrahydrobiopterin (BH4) biosynthesis, catalyzing the conversion of GTP into 7,8-dihydroneopterin triphosphate. BH4 is an essential cofactor required by aromatic amino acid hydroxylases as well as nitric oxide synthases. Mutations in this gene are associated with malignant hyperphenylalaninemia and dopa-responsive dystonia. Several alternatively spliced transcript variants encoding different isoforms have been described; however, not all variants give rise to a functional enzyme. [provided by

RefSeq, Jul 20081

Transcript Variant: This variant (2), also known as Type V, differs in the 3' UTR, compared to

variant 1. Variants 1 and 2 both encode the functional enzyme, isoform 1.