

Product datasheet for **SC302227**

GTP cyclohydrolase 1 (GCH1) (NM_001024071) Human Untagged Clone

Product data:

Product Type:	Expression Plasmids
Product Name:	GTP cyclohydrolase 1 (GCH1) (NM_001024071) Human Untagged Clone
Tag:	Tag Free
Symbol:	GTP cyclohydrolase 1
Synonyms:	DYT5; DYT5a; DYT14; GCH; GTP-CH-1; GTPCH1; HPABH4B
Vector:	<u>pCMV6 series</u>
Fully Sequenced ORF:	>NCBI ORF sequence for NM_001024071, the custom clone sequence may differ by one or more nucleotides ATGGAGAAGGGCCTGTGCGGGACCGCGGAGAAGCCGCGGGCGCCAGGTGCAGCAAT GGGTTCCCGAGCGGGATCCGCCGCGGCCCGGGCCAGCAGGCCGGCGAGAAGCCCCG CGGCCGAGGCCAAGAGCGCGCAGCCCGCGGACGGCTGGAAGGGCGAGCGCCCCGAGC GAGGAGGATAACGAGCTGAACCTCCCTAACCTGGCAGCCGCTACTCGTCCATCCTGAGC TCGCTGGGCGAGAACCCCGCGCAAGGGCTGCTCAAGACGCCCTGGAGGGCGGCCTCG GCCATGCAGTTCTTACCAAGGGTACCAGGAGACCATCTCAGATGTCCTAAACGATGCT ATATTTGATGAAGATCATGATGAGATGGTGATTGTGAAGGACATAGACATGTTTTCCATG TGTGAGCATCACTTGGTTCCATTTGTTGAAAGGTCCATATTGGTTATCTTCTAACAAG CAAGTCCTTGGCCTCAGCAAACCTTGCGAGGATTGTAGAAATCTATAGTAGAAGACTACAA GTTTCAGGAGCGCCTTACAAAACAAATTGCTGTAGCAATCACGGAAGCCTTGCGGCCTGCT GGAGTCGGGGTAGTGGTTGAAGCAACGTCTGCAGAACCATAG
Restriction Sites:	Please inquire
ACCN:	NM_001024071
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).



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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: [NM_001024071.1](#), [NP_001019242.1](#)

RefSeq Size: 1852 bp

RefSeq ORF: 642 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 2008]

Transcript Variant: This variant (4), also known as Type II, lacks an alternate in-frame exon in the 3' coding region and uses a downstream stop codon, compared to variant 1. The resulting non-functional protein (isoform 3) is shorter and has a distinct C-terminus, compared to variant 1.