

## Recombinant Human PCBD1 Protein (His Tag)

**Catalog Number:** PKSH032981

**Note:** Centrifuge before opening to ensure complete recovery of vial contents.

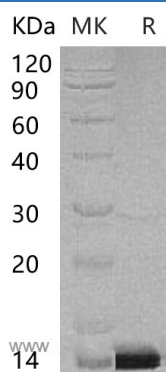
### Description

<b>Species</b>	Human
<b>Source</b>	E.coli-derived Human PCBD1 protein Ala2-Thr104, with an N-terminal His
<b>Calculated MW</b>	14.2 kDa
<b>Observed MW</b>	14 kDa
<b>Accession</b>	P61457
<b>Bio-activity</b>	Not validated for activity

### Properties

<b>Purity</b>	> 95 % as determined by reducing SDS-PAGE.
<b>Concentration</b>	Subject to label value.
<b>Endotoxin</b>	< 1.0 EU per µg of the protein as determined by the LAL method.
<b>Storage</b>	Store at < -20°C, stable for 6 months. Please minimize freeze-thaw cycles.
<b>Shipping</b>	This product is provided as liquid. It is shipped at frozen temperature with blue ice/gel packs. Upon receipt, store it immediately at < - 20°C.
<b>Formulation</b>	Supplied as a 0.2 µm filtered solution of 20mM Tris-HCl, 150mM NaCl, 1mM DTT, pH 8.0.

### Data



> 95 % as determined by reducing SDS-PAGE.

### Background

Pterin-4- $\alpha$ -Carbinolamine Dehydratase (PCBD1) is the founding member of the Pterin-4- $\alpha$ -Carbinolamine Dehydratase Family. PCBD1 is involved in Tetrahydrobiopterin biosynthesis. It seems to prevent the formation of 7-Pterins and accelerate the formation of Quinonoid-BH2. Furthermore, PCBD1 regulates the homodimerization of the transcription factor Hepatocyte Nuclear Factor 1 (HNF1) and enhances its transcriptional activity. Defects in PCBD1 are the cause of BH4-Deficient Hyperphenylalaninemia Type D (HPABH4D). HPABH4D is characterized by the excretion of 7-substituted Pterins in the urine of affected patients.