

# BIOMDB Mutations Causing BH4-Deficiency

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## REGISTRATION FORM BIOMDB DATABASE

<http://www.bh4.org/BH4DatabasesBiomdb.asp>

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**Genes: *GCHI*, *PTS*, *SPR*, *PCBD*, *QDPR***

Patient ID#			
BIODEF ID#			
Country			
Gene	<input type="checkbox"/> <i>GCHI</i>	<input type="checkbox"/> <i>PTS</i>	<input type="checkbox"/> <i>SPR</i> <input type="checkbox"/> <i>PCBD</i> <input type="checkbox"/> <i>QDPR</i>
Phenotype	<input type="checkbox"/> HPA	<input type="checkbox"/> DRD	<input type="checkbox"/> severe <input type="checkbox"/> mild
	Mutation name* (amino acid change)	Systemic name** (nucleotide change)	Exon / Intron
Allele 1			
Allele 2			
Literature:			
Comments:			

\*Example: p.E390G; \*\*Example: c.1169A>G (use HGVS nomenclature)

Name (print):

Address (print):

Date:

Signature: