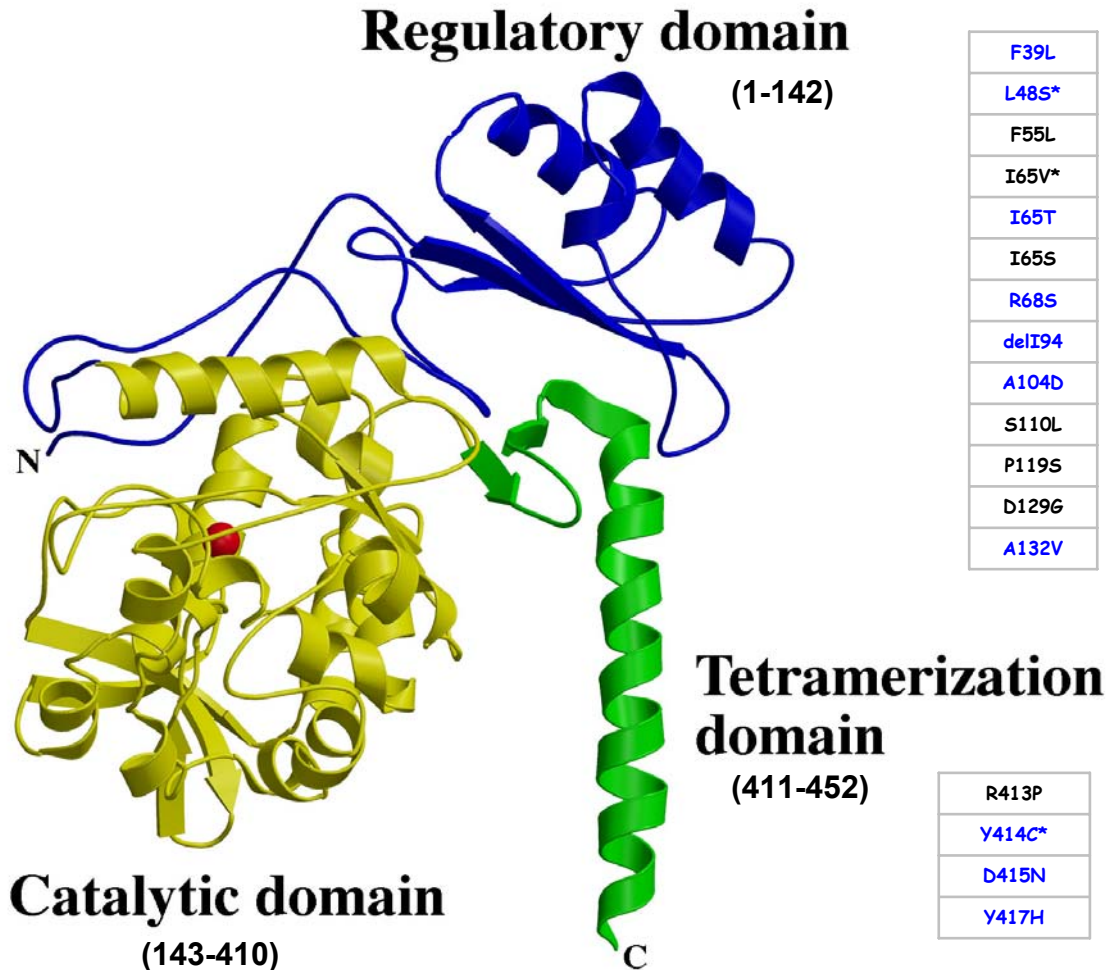


Mutations Detected in Patients with BH4-responsive PAH Deficiency

R158Q
R158W
H170Q
V177M
V190A
P211T
R241C
R241H
R243Q
R243X
V245A
R252W
R261Q*
R261X
G272X
K274fsdel11bp

P275L
P278I
E280K
P281A
P281L
P281S
A300S
S310Y
A313T
P314S
K320N
L367fsinsC
A373T
V388M
P390G
E390G
A395P
A403V
P407S
R408Q
R408W
P470S



F39L
L48S*
F55L
I65V*
I65T
I65S
R68S
delI94
A104D
S110L
P119S
D129G
A132V

R413P
Y414C*
D415N
Y417H

PAH Source: Erlandsen H and Stevens RC, *Mol Genet Metab.* 1999 68:103-125

blue = mutations with residual activity (measured or proposed)
* inconsistent phenotype