

Open Peer Review on Qeios

## **EPAS1** wt Allele

National Cancer Institute

## Source

National Cancer Institute. <u>EPAS1 wt Allele</u>. NCI Thesaurus. Code C104305.

Human EPAS1 wild-type allele is located within 2p21-p16 and is approximately 93 kb in length. This allele, which encodes endothelial PAS domain-containing protein 1, is involved in the regulation of transcription in response to oxygen. Mutation of the gene is associated with familial erythrocytosis type 4.

Qeios ID: OPPCVE · https://doi.org/10.32388/OPPCVE