SANTA CRUZ BIOTECHNOLOGY, INC.

CYP21A2 (C-17): sc-48466

SANTA CRUZ BIOTECHNOLOGY

The Power to Question

BACKGROUND

The cytochrome P450 proteins are monooxygenases that catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids. P450 enzymes are classified into subfamilies based on their sequence similarities. CYP21A2 localizes to the endoplasmic reticulum and hydroxylates steroids at the 21 position. Activity of CYP21A2 is required for the synthesis of steroid hormones, including cortisol and aldosterone. Mutations in this gene are the primary cause of congenital adrenal hyperplasia (CAH), an autosomal recessive disorder. Gene conversion events involving the functional CYP21A2 gene (C21B) and a related pseudogene (C21A) located near the C21B gene may account for the majority of cases of steroid 21-hydroxylase deficiency.

REFERENCES

1. Nelson, D.R., et al. 1996. P450 superfamily: update on new sequences, gene mapping, accession numbers and nomenclature. Pharmacogenetics 6: 1-42.
2. Chang, S.F. and Chung, B.C. 1996. Difference in transcriptional activity of two homologous CYP21A genes. Mol. Endocrinol. 9: 1330-1336.
3. Peterson, J.A., et al. 1997. P450BM-3; a tale of two domains—or is it three? Steroids 62: 117-123.
4. Araujo, R.S., et al. 2005. Substitutions in the CYP21A2 promoter explain the simple-virilizing form of deficiency in patients harbouring a P30L mutation. Clin. Endocrinol. 62: 132-136.
5. Grigorescu Sido, A., et al. 2005. 21-hydroxylase and 11β-hydroxylase mutations in Romanian patients with classic congenital adrenal hyperplasia. J. Clin. Endocrinol. Metab. 90: 5769-5773.
6. Keen-Kim, D., et al. 2005. Validation and clinical application of a locus-specific polymerase chain reaction- and mini-sequencing-based assay for congenital adrenal hyperplasia (21 hydroxy-lase deficiency). J. Mol. Diagn. 7: 236-246.
7. Krone, N., et al. 2005. The residue E351 is essential for the activity of human 21 hydroxylase: evidence from a naturally occurring novel point mutation compared with artificial mutants generated by single amino acid substitutions. J. Mol. Med. 83: 561-568.

CHROMOSOMAL LOCATION

Genetic locus: CYP21A2 (human) mapping to 6p21.33.

SOURCE

CYP21A2 (C-17) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of CYP21A2 of human origin.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

For research use only, not for use in diagnostic procedures.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-48466 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

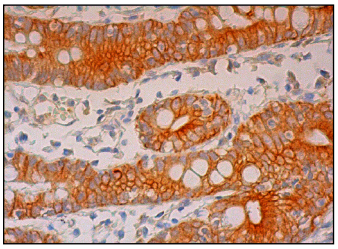
CYP21A2 (C-17) is recommended for detection of CYP21A2 of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500), immunohistochemistry (including paraffin-embedded sections) (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

CYP21A2 (C-17) is also recommended for detection of CYP21A2 in additional species, including bovine and porcine.

Suitable for use as control antibody for CYP21A2 siRNA (h): sc-60478, CYP21A2 shRNA Plasmid (h): sc-60478-SH and CYP21A2 shRNA (h) Lentiviral Particles: sc-60478-V.

Molecular Weight of CYP21A2: 52 kDa.

DATA



CYP21A2 (C-17): sc-48466. Immunoperoxidase staining of formalin fixed, paraffin-embedded human small intestine tissue showing membrane and cytoplasmic staining of glandular cells.

SELECT PRODUCT CITATIONS

1. Concolino, P., et al. 2009. Functional analysis of two rare CYP21A2 mutations detected in Italian patients with a mildest form of congenital adrenal hyperplasia. Clin. Endocrinol. 71: 470-476.
2. Concolino, P., et al. 2009. Two novel CYP21A2 missense mutations in Italian patients with 21-hydroxylase deficiency: identification and functional characterisation. IUBMB Life 61: 229-235.
3. Gaffney, D., et al. 2011. Functional characterisation of the H365Y mutation of the 21-hydroxylase gene in congenital adrenal hyperplasia. J. Steroid Biochem. Mol. Biol. 123: 109-114.
4. Concolino, P., et al. 2012. p.H282N and p.Y191H: 2 novel CYP21A2 mutations in Italian congenital adrenal hyperplasia patients. Metab. Clin. Exp. 61: 519-524.