

Nonclassic 21-Hydroxylase Deficiency Congenital Adrenal Hyperplasia Testing

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INDICATIONS FOR TESTING

Hirsutism, virilization, precocious puberty;
amenorrhea, irregular menses, or PCOS in females

ORDER

Serum 17-OHP test on
a specimen collected in
the early morning using
a sensitive method such
as LC-MS/MS

17-OHP
elevated

17-OHP borderline
OR
Sensitive 17-OHP
testing unavailable

17-OHP
low

ORDER

ACTH stimulation
test

17-OHP
elevated

17-OHP
low

Nonclassic 21-OHD CAH

ORDER

Adrenocortical hormone testing
Electrolyte panel test
AND CONSIDER
Genetic testing

High 17-OHP, androstenedione,
DHEA; normal electrolytes
AND/OR
Pathogenic CYP21A2 variant
identified

No pathogenic variants identified

Nonclassic 21-OHD
CAH confirmed

Nonclassic 21-OHD
CAH not excluded

Classic 21-OHD CAH excluded
CONSIDER
Other etiologies

References

- Nimkarn S, Gangishetti PK, Yau M, et al. [21-hydroxylase-deficient congenital adrenal hyperplasia](#). In: Adam MP, Mirzaa GM, Pagon RA, et al, eds. *GeneReviews*. University of Washington, Seattle. Updated Feb 2016; accessed Jul 2024.
- Speiser PW, Arlt W, Auchus RJ, et al. [Congenital adrenal hyperplasia due to steroid 21-hydroxylase deficiency: an Endocrine Society clinical practice guideline](#) [published correction appears in *J Clin Endocrinol Metab* 2019;104(1):39-40]. *J Clin Endocrinol Metab*. 2018; 103(11):4043-4088.
- Yau M, Khattab A, Yuen T, et al. [Congenital adrenal hyperplasia](#). In: Feingold KR, Anawalt B, Blackman MR, et al, eds. *Endotext*. MDText.com. Updated Nov 2022; accessed Jul 2024.

Abbreviations

17-OHP	17-hydroxyprogesterone
21-OHD	21-hydroxylase deficiency
ACTH	Adrenocorticotropic hormone
CAH	Congenital adrenal hyperplasia
DHEA	Dehydroepiandrosterone
LC-MS/MS	Liquid chromatography-tandem mass spectrometry
PCOS	Polycystic ovary syndrome