

Disease

Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete^{1, 2, 3}

Gene

CYP11A1

Inheritance	AR and AD
Current Treatments	Hydrocortisone, 9α-fludrohydrocortisone
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum cortisol, aldosterone and adrenocorticotrophic hormone (ACTH) levels ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=cyp11a1&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/613743>

² <https://www.ncbi.nlm.nih.gov/pubmed/30817990>

³ <https://pubmed.ncbi.nlm.nih.gov/29995203/>