

Disease

Congenital adrenal hypoplasia¹

Gene

NR0B1

Inheritance	XL
Current Treatments	Hydrocortisone, 9α-fludrohydrocortisone, oral supplements of sodium chloride
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum ACTH level, cortisol level, ACTH (cosyntropin) stimulation test ¹
Disease Frequency (per 100,000)	0.8 ²
Genes Included in Disease Frequency	NR0B1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Congenital+Adrenal+Hypoplasia&term=&cntry=&state=&city=&

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1431/>

² <https://www.ncbi.nlm.nih.gov/pubmed/16684822>