Supplemental Tables: Reproductive phenotypes and genotypes in men with IHH

(Dwyer et al, *Journal of Clinical Endocrinology & Metabolism*, 2022)

Supplemental Table 1

Variants	Absent puberty (n=118)	Partial puberty (n=86)	*p* value
Variants in CHH loci (n, %)			
Protein truncating variant (PTV)	26 (22%)	10 (11%)	0.063
ANOS1	15 (13%)	1 (1%)	0.002
Oligogenic with at least 1 PTV	20 (17%)	8 (9%)	0.14

Supplemental Table 2

Variants	Undetectable LH (n=124)	Detectable LH (n=76)	*P* value
Variants in CHH loci (n, %)			
Protein truncating variant (PTV)	24 (19%)	11 (14%)	0.44
ANOS1	13 (11%)	3 (4%)	0.11
Oligogenic with at least 1 PTV	18 (15%)	9 (10%)	0.67

Supplemental Table 3

Variants	Undetectable FSH (n=113)	Detectable FSH (n=83)	*P* value
Variants in CHH loci (n, %)			
Protein truncating variant (PTV)	20 (18)	13 (16)	0.84
ANOS1	11 (10)	5 (6)	0.43
Oligogenic with at least 1 PTV	15 (13)	10 (12)	0.83

Supplemental Table 4

Variants	Undetectable LH/FSH (n=100)	Detectable LH/FSH (n=60)	*P* value
Variants in CHH loci (n, %)			
Protein truncating variant (PTV)	20 (20)	9 (15)	0.52
ANOS1	11 (11)	3 (5)	0.25
Oligogenic with at least 1 PTV	15 (15)	7 (12)	0.64

Supplemental Table 5

Variants	Apulsatile LH (n=152)	Pulsatile LH (n=52)	*P* value
Variants in CHH loci (n, %)			
Protein truncating variant (PTV)	29 (19)	7 (13)	0.40
ANOS1	15 (10)	1 (2)	0.07
Oligogenic with at least 1 PTV	23 (15)	5 (10)	0.36

Supplemental Table 6

Variants	Pure neuroendocrine phenotype (n=35)	Non-reproductive defects (n=126)	*P* value
Variants in CHH loci (n, %)			
Protein truncating variant (PTV)	4 (11)	32 (25)	0.51
Oligogenic with at least 1 PTV	2 (6)	23 (18)	0.11

Supplemental Table 7

Variants	cryptorchidism (n=64)	No cryptorchidism (n=138)	*P* value
Protein truncating variant (PTV)	14 (22)	22 (16)	0.32
Oligogenic with at least 1 PTV	12 (19)	16 (12)	0.19
	micropenis (n=49)	**No micropenis (n=153)**	
Protein truncating variant (PTV)	11 (22)	25 (16)	0.39
Oligogenic with at least 1 PTV	10 (20)	18 (12)	0.15
	cryptorchidism & micropenis (n=28)	**No cryptorchidism or micropenis (n=117)**	
Protein truncating variant (PTV)	5 (18)	16 (14)	0.55
Oligogenic with at least 1 PTV	5 (18)	11 (9)	0.19
	inhibin B <70 pg/mL (n=92)	**inhibin B >70 pg/mL (n=34)**	
Protein truncating variant (PTV)	16 (17%)	4 (12%)	0.58
Oligogenic with at least 1 PTV	13 (14%)	3 (9%)	0.55

Supplemental Table 8

Variant(s)	apulsatile LH secretion		pulsatile LH secretion		$\chi^2(3)$	*p* value
	A: absent puberty (n=108)	B: partial puberty (n=44)	C: absent puberty (n=10)	D: partial puberty (n=42)		
Variants in CHH loci (n, %)						
PTV	24 (22%)	5 (11%)	2 (20%)	5 (12%)	5.71	0.1267
standardized residuals	1.65	-1.50	1.07	-1.22		
*ANOS1**	15 (14%)	--	--	1 (2%)	34.00	< 0.0001
standardized residuals	5.77	-2.31	-2.31	-1.15		

*Values are approximated due to the small number of observations in some cells