

Table S5. Variants identified in CHH genes.

ID	Sex <sup>#</sup>	DSD category	Clinical diagnosis	CHH Gene	Inheritance	GnomAD frequency	DNA change	Protein change	SIFT	MT	ACMG classification
6	F	DGD	CGD	<i>TACR3</i>	AR	0.03%	c.824G>A	p.Trp275*			VUS
7	F	DGD	CGD	<i>CHD7</i>	AD		c.825C>G	p.Phe275Leu	D	D	VUS
9	F	DGD	CGD	<i>SEMA3</i>	AD	0.02%	c.1450C>T	p.Arg484Trp	D	D	VUS
26	F	DAA	CAIS	<i>ANOS1</i>	XL	0.22%	c.1759G>T	p.Val587Leu	T	D	VUS
28	M	sDSD	sDSD	<i>FGF8</i>	AD	0.11%	c.77C>T	p.Pro26Leu	T	D	VUS

Abbreviations: DSD, disorder of sex development; CHH, congenital hypogonadotropic hypogonadism; GnomAD, Genome Aggregation Database; SIFT, Sorting Intolerant From Tolerant; MT, MutationTaster; ACMG, American College of Medical Genetics and Genomics; F, female; DGD, disorder of gonadal development; CGD, complete gonadal dysgenesis; AR, autosomal recessive; VUS, variant of uncertain significance; AD, autosomal dominant; D, deleterious; DAA, disorder of androgen action; CAIS, complete androgen insensitivity syndrome; XL, X-linked; T, tolerated; M, male; sDSD, syndromic disorder of sex development

<sup>#</sup>Sex of rearing