

## High frequency of CHD7 mutations in congenital hypogonadotropic hypogonadism

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**Supplemental Table S1.** Evolutionary conservation of CHD7 mutated amino acids (bold) across different species.

Species	G388E		H903P		T1082I	
	Codon	Alignment	Codon	Alignment	Codon	Alignment
Human	388	P S Q P Q <b>G</b> T Y A S P	903	G E P V T <b>H</b> Y L V K W	1082	F H A I I <b>I</b> T T F E M I
Mutated variant	388	P S Q P Q <b>E</b> T Y A S P	903	G E P V T <b>P</b> Y L V K W	1082	F H A I I <b>I</b> T T F E M I
<i>P.troglodytes</i>	388	P S Q P Q <b>E</b> T Y A S P	903	V T <b>H</b> Y L V K W	1082	I <b>T</b> T F E M I
<i>M.mulatta</i>	388	P S Q P Q <b>E</b> T Y A S P	903	V T <b>H</b> Y L V K W	1082	I <b>T</b> T F E M I
<i>F.catus</i>						
<i>M.musculus</i>	382	P S Q P Q <b>E</b> T Y A S P	893	P V I <b>H</b> Y L V K W	1072	I I <b>I</b> T T F E M I
<i>G.gallus</i>			141	P V T <b>H</b> Y L V K W	320	F H A I I <b>I</b> T T F E M I
<i>T.rubripes</i>						
<i>D.rerio</i>	402	A Q P P Q <b>G</b> S Y S S P	947	G E L V T <b>L</b> Y L V K W	1126	F H A V I <b>I</b> T T F E M I
<i>D.melanogaster</i>			1962	G E T T K <b>H</b> Y L V K W	2143	F N V L I <b>I</b> T T F E M I
<i>C.elegans</i>	544	P H Q H Q <b>Q</b> P A S V P	1135	G Q E F V - - L I K W	1311	I D A L I <b>I</b> T T F E T V
<i>X.tropicalis</i>	323	P S Q P Q <b>G</b> T Y A S P	830	G E P V C <b>H</b> Y L V K W	1009	F H A I I <b>I</b> T T F E M I

  

Species	V1452L		D1854G		R2065H	
	Codon	Alignment	Codon	Alignment	Codon	Alignment
Human	1452	N A T N G <b>V</b> Q Q L S K	1854	D R E D E <b>D</b> P E Y K P	2065	L L R K I <b>R</b> E Q V L H
Mutated variant	1452	N A T N G <b>L</b> Q Q L S K	1854	D R E D E <b>G</b> P E Y K P	2065	L L R K I <b>H</b> E Q V L H
<i>P.troglodytes</i>	1452	N A T N G <b>V</b> Q Q L S K	1854	D R E D E <b>D</b> P E Y K P	2065	L L R K I <b>R</b> E Q V L H
<i>M.mulatta</i>	1452	N A T N G <b>V</b> Q Q L S K	1856	D R E D E <b>D</b> P E	2067	L L R K I <b>R</b> E Q V L H
<i>F.catus</i>						
<i>M.musculus</i>	1442	N A T N G <b>V</b> Q Q L S K	1844	D R E D E <b>D</b> P E Y K P	2055	L L R K I <b>R</b> E Q V L H
<i>G.gallus</i>	690	N A T N G <b>V</b> Q Q L S K	1092	D R E D E <b>D</b> P E Y K P	1302	L L R K I <b>R</b> E Q V L H
<i>T.rubripes</i>						
<i>D.rerio</i>	1504	N A A N G <b>V</b> Q Q L S K	1906	E E E <b>D</b> P E Y K P	2149	L L R R I <b>R</b> E Q V L P
<i>D.melanogaster</i>	2512	S K D G N <b>V</b> K Q L S K	2991	K H D D G <b>D</b> E V D D D		
<i>C.elegans</i>	1683	A E G T A - - - L S K				
<i>X.tropicalis</i>	1382	N A T N G <b>V</b> Q Q L S K	1784	E <b>D</b> P E Y K P	1993	L L R K I <b>R</b> E Q V L N