

Supplementary Table 1. Mutations detected and their predicted effects  
(reference NM\_005989) in a cohort of patients with the  $\Delta$ 4-3-oxosteroid  
5 $\beta$ -reductase

cleotide hange	Predicted effects	HGMD or SNP ID	Mutation Taster prediction	PolyPhen-2 Prediction (HumVar)	SIFT prediction	Allele counts in patients
148C>T	p.Arg50*	CM097184	disease causing	NA	NA	1
158A>G	p.Asp53Gly	NA	disease causing	probably damaging	affect protein function	1
178delA	p.His93LeufsTer1 9	NA	disease causing	NA	NA	1
196C>A	p.Tyr132*	NA	disease causing	NA	NA	1
193C>T	p.Pro198Leu	CM032543	disease causing	NA	NA	1
214delT	p.Leu205ProfsTer 2	NA	disease causing	NA	NA	1
222A>T	p.Asp241Val	NA	disease causing	probably damaging	affect protein function	1
297G>A	p.Arg266Gln	<a href="#">rs18282035</a> <u>3</u>	disease causing	probably damaging	affect protein function	7
319G>T	p.Arg307Cys	NA	disease causing	possibly damaging	affect protein function	4
333delG	Deletion of 1 or 2AA	NA	disease causing	NA	NA	1