35	c.349G>A; p.G117R (het)	0	4	Pseudohermaphroditism (46, XY)	This study
	c.878C>A; p.S293* (het)	0	10		This study

AF, allele frequency in gnomAD (http://gnomad.broadinstitute.org/); het, heterozygous; hom, homozygous; del, deletion;

* only heterozygous of p.I114F was identified in the patient, there should be another heterozygous variant in the patient, such as exon deletion.