

Supplemental Table 1. PCR regions amplified, primer sequences, restriction enzymes and restricted fragments

SNP /Mutation	dbSNP rs#	Region	Primer sequence <sup>a</sup> (5'-3')	Restriction enzyme	Restricted fragments
<b>DAZL</b>					
c.34A>G (T54A)		Exon 3	F: C T A C T G G T C A T T T G G G G A T A	<i>Alu I</i>	A: 165bp
			R: C T G A A C C A T A T C T A G C A A A G A G A c G		G: 129bp, 36bp
c.211A>G (I71V)		Exon 3	F: C T A C T G G T C A T T T G G G G A T A	<i>Dpn I</i>	A: 188bp, 89bp, 12bp
			R: T C A G A A T A C C A A T T T G A A G T C		G: 277bp, 12bp
c.345A>G (R115G)		Exon 5	F: G A C T T T A G G T G T T C A T C C A A	<i>Bsc 4I</i>	A: 230bp, 24bp
			R: G C A C A T G A T A A G C A C C T T T		G: 124bp, 106bp, 24bp
c.294+222C>T	rs2217204	Intron 4	F: A A A G T T G A T C A A A A A A C T T T T C A G	<i>Rsa I</i>	C: 113bp
			R: C T T A C A T A A C T T G T T A G A A C C T G		T: 24bp, 89bp
c.570+356C>T	rs4234537	Intron 7	F: T A T T T C A C T C T A T C C C C T A	<i>Bsu RI</i>	C: 53bp, 72bp
			R: C A A C A G T T T T A T G A G A T T C G		T: 125bp
c.888+1853G>C	rs2347312	3'-UTR	F: T T G C A T A C A T G A A T A A A A A T A A g	<i>Hind III</i>	G: 211bp
			R: T C A T G C A T C T C T T A A A A T T T C T C T		C: 190bp, 21bp
<b>BOULE</b>					
c.4C>G (Q2E)		Exon 1	F: A G A T G C A A A C A G A T T C A T A T C T C g A	<i>Cvi RI</i>	C: 115bp, 46bp
			R: T T C C T C C T A C A A A G A T G C G A T T		G: 161bp
c.26C>A (S9Y)		Exon 1	F: A A A A G T G T A A A A G A A T T T A T T T T A C A G	<i>Dpn II</i>	C: 97bp, 23bp
			R: T T C C T C C T A C A A A G A T G C G A T T		A: 120bp
c.221+68C>T	rs700655	Intron 3	F: G G A G T A T C C A A A G G G T T A G T	<i>Csp 6I</i>	C: 79bp, 65bp
			R: G G T A G G A G G A A A G A A G A G A G G G		T: 144bp
c.828+7055A>C	rs700642	Intron 10	F: T A G T T G A G T A A G T G C A G A T	<i>Taq I</i>	A: 97bp, 65bp
			R: C A C G T C A T A G T T A A G A G G G A		C: 144bp
c.852+1595A>G	rs2272166	3'-UTR	F: A A T A G A T T A A G T C C C C A C A A T G A A A	<i>Dpn II</i>	A: 129bp, 22bp
			R: A G C A T C T T G T T T A A C A T T T G A A G A		G: 151bp

<sup>a</sup> The small letters in the primer sequences represent mismatched bases to produce restricted sites for distinguishing the wild from mutation sequences.

Supplemental Table 2. Pairewise linkage disequilibrium (D') of DAZL and BOULE SNPs in men with normozoospermia and spermatogenic failure

Gene/SNPs	DAZL			BOULE	
	c.34A>G	c.294+222C>T	c.570+356C>T	c.221+68C>T	c.828+7055A>C
DAZL / c.34A>G	— <sup>a</sup>	0.203	0.654	0.898	
DAZL / c.294+222C>T	0.170	—	0.151	0.148	
DAZL / c.570+356C>T	0.046	0.208	—	0.236	
DAZL / c.888+1853G>C	0.044	1.000	0.221	—	
BOULE / c.221+68C>T				—	0.263
BOULE / c.828+7055A>C				0.135	—

<sup>a</sup> Estimates in the upper right are for men with spermatogenic failure and in the lower left are for men with normozoospermia.