

Appendix 2. Intronic and untranslated region sequence variants found in the SKI gene.

Subject ID	Exon 1.3	Exon 3	Exon 3	Exon 3	Exon 3
	novel	novel	rs2279702	novel	deletion
	Intronic	Intronic	Intronic	Intronic	Intronic
1					
2			c.[1096-92G>A]+[=]		
3					
4					
5					
6	c.[969+53C>T]+[=]				
7		c.[1096-39T>C]+[=]			
8					
9				c.[1211+27T>C]+[=]	c.[1211+39delT]+[=]
10			c.[1096-92G>A]+[=]		
11					
12					
13			c.[1096-92G>A]+[=]		
14					
15					
16					
17					
18					
Control 1			c.[1096-92G>A]+[=]		
Control 2			c.[1096-92G>A]+[=]		
Control 3			c.[1096-92G>A]+[=]		
Control 4			G		

Exon 3	Exon 3	Exon 5	Exon 5	Exon 5
rs2256178	novel	rs11577387	rs2645073	rs2843159
Intronic	Intronic	Intronic	Intronic	Intronic
		c.[1475-76G>A]+[=]		
c.[1211+64C>T]+[=]				
c.[1211+64C>T]+[1211+64C>T]			c.[1475-70G>A]+[1475-70G>A]	c.[1475-60C>T]+[1475-60C>T]
c.[1211+64C>T]+[=]			c.[1475-70G>A]+[=]	c.[1475-60C>T]+[=]
c.[1211+64C>T]+[=]			c.[1475-70G>A]+[=]	c.[1475-60C>T]+[1475-60C>T]
		c.[1475-76G>A]+[=]		
c.[1211+64C>T]+[=]				
c.[1211+64C>T]+[=]	c.[1211+70C>T]+[=]		c.[1475-70G>A]+[=]	c.[1475-60C>T]+[=]
		c.[1475-76G>A]+[=]		
c.[1211+64C>T]+[=]			c.[1475-70G>A]+[=]	c.[1475-60C>T]+[=]
		c.[1475-76G>A]+[=]		
		c.[1475-76G>A]+[=]		
		c.[1475-76G>A]+[=]		
c.[1211+64C>T]+[1211+64C>T]			c.[1475-70G>A]+[1475-70G>A]	c.[1475-60C>T]+[1475-60C>T]

Exon 7.1	Exon 7.2	Exon 7.2	Exon 7.3
rs2230008	novel	novel	novel
UTR	UTR	UTR	UTR
c.[2187*33A>T]+[=]			
c.[2187*33A>T]+[=]			
c.[2187*33A>T]+[=]	--	--	--
c.[2187*33A>T]+[=]			
c.[2187*33A>T]+[=]			
c.[2187*33A>T]+[=]			
	c.[2187*378A>G]+[=]		c.[2187*705C>T]+[=]
c.[2187*33A>T]+[=]			
			--
		c.[2187*523G>A]+[=]	
c.[2187*33A>T]+[=]			
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