

Appendix 1. Intronic and untranslated region sequence variants found in the STRA6 gene.

Subject ID	Exon 1B	Exon 1B	Exon 1B	Exon 1B
	rs61466937	rs62003660	rs11639369	Novel
	Intronic 2 Base Pair Deletion	Alternate UTR (Exon 1B)	Intronic	Intronic 5 Base Pair Insertion
1				
2	c.[-949C>A]+[=]			
3		c.[-766T>G]+[-766T>G]	c.[-338C>A]+[-338C>A]	
4	c.[-949C>A]+[=]			
5		c.[-766T>G]+[=]	c.[-338C>A]+[=]	
6	--	--		
7	c.[-949C>A]+[=]			c.[-288C>A]+[=]
8		c.[-766T>G]+[-766T>G]	c.[-338C>A]+[-338C>A]	
9				
10	c.[-949C>A]+[=]	c.[-766T>G]+[=]	c.[-338C>A]+[=]	
11				
12				
13	c.[-949C>A]+[=]			
14				c.[-288C>A]+[=]
15				
16				
17		c.[-766T>G]+[=]	c.[-338C>A]+[=]	
18	c.[-949C>A]+[=]			
Control 1		c.[-766T>G]+[=]	c.[-338C>A]+[=]	
Control 2	c.[-949C>A]+[=]	c.[-766T>G]+[=]	c.[-338C>A]+[=]	
Control 3	c.[-949C>A]+[=]			
Control 4	c.[-949C>A]+[=]	c.[-766T>G]+[=]	c.[-338C>A]+[=]	

Exon 2	Exon 6	Exon 7	Exon 13
rs12912258	rs971756	rs971757	Novel snp
Intronic	Intronic	Intronic	Intronic
c.[113+108G>A]+[113+108G>A]	c.[406+24T>A]+[406+24T>A]	c.[407-37C>T]+[407-37C>T]	
c.[113+108G>A]+[=]	c.[406+24T>A]+[=]	c.[407-37C>T]+[=]	
--	--	--	--
			c.[1091-118G>C]+[=]
c.[113+108G>A]+[113+108G>A]	c.[406+24T>A]+[406+24T>A]	c.[407-37C>T]+[407-37C>T]	
c.[113+108G>A]+[=]	c.[406+24T>A]+[=]	c.[407-37C>T]+[=]	
c.[113+108G>A]+[=]	c.[406+24T>A]+[=]	c.[407-37C>T]+[=]	
--	c.[406+24T>A]+[=]	c.[407-37C>T]+[=]	
c.[113+108G>A]+[=]	c.[406+24T>A]+[=]	c.[407-37C>T]+[=]	
--	c.[406+24T>A]+[=]	c.[407-37C>T]+[=]	

Exon 13	Exon 13	Exon 13	Exon 14	Exon 18
rs351237	Novel snp	rs351238	rs2277608	rs12912578
Intronic	Intronic	Intronic	Intronic	Intronic
c.[1166+82C>T]+[1166+82C>T]		c.[1166+105C>T]+[1166+105C>T]		
c.[1166+82C>T]+[=]		c.[1166+105C>T]+[=]	c.[1167-10C>G]+[=]	
			c.[1167-10C>G]+[=]	c.[1840+50T>C]+[=]
c.[1166+82C>T]+[1166+82C>T]		c.[1166+105C>T]+[1166+105C>T]		
			c.[1167-10C>G]+[=]	c.[1840+50T>C]+[=]
--	--	--	--	c.[1840+50T>C]+[=]
			c.[1167-10C>G]+[=]	
				c.[1840+50T>C]+[1840+50T>C]
c.[1166+82C>T]+[=]		c.[1166+105C>T]+[=]	c.[1167-10C>G]+[=]	
c.[1166+82C>T]+[=]		c.[1166+105C>T]+[=]		c.[1840+50T>C]+[=]
			c.[1167-10C>G]+[=]	
			c.[1167-10C>G]+[=]	
c.[1166+82C>T]+[=]		c.[1166+105C>T]+[=]	c.[1167-10C>G]+[=]	
c.[1166+82C>T]+[=]		c.[1166+105C>T]+[=]	c.[1167-10C>G]+[=]	
c.[1166+82C>T]+[1166+82C>T]		c.[1166+105C>T]+[1166+105C>T]		
c.[1166+82C>T]+[1166+82C>T]	c.[1166+98G>A]+[=]	c.[1166+105C>T]+[1166+105C>T]		
c.[1166+82C>T]+[=]		c.[1166+105C>T]+[=]		c.[1840+50T>C]+[=]
c.[1166+82C>T]+[1166+82C>T]		c.[1166+105C>T]+[1166+105C>T]		
c.[1166+82C>T]+[=]		c.[1166+105C>T]+[=]		c.[1840+50T>C]+[=]
c.[1166+82C>T]+[=]				c.[1840+50T>C]+[=]
c.[1166+82C>T]+[1166+82C>T]		c.[1166+105C>T]+[1166+105C>T]		
				c.[1840+50T>C]+[=]

Exon 19	Exon 19	Exon 19	Exon 19
rs11631944	rs10910	novel	rs28447005
UTR	UTR	UTR	UTR
	c.[2004*222G>A]+[2004*222G>A]		
	c.[2004*222G>A]+[2004*222G>A]		
c.[2004*127T>C]+[=]			
	c.[2004*222G>A]+[2004*222G>A]		
c.[2004*127T>C]+[2004*127T>C]	c.[2004*222G>A]		
c.[2004*127T>C]+[=]			
			c.[2004*384T>C]+[=]
c.[2004*127T>C]+[2004*127T>C]			
	c.[2004*222G>A]+[2004*222G>A]		
c.[2004*127T>C]+[2004*127T>C]	c.[2004*222G>A]		
	c.[2004*222G>A]+[2004*222G>A]		
	c.[2004*222G>A]		
	c.[2004*222G>A]	c.[2004*284G>A]+[=]	
	c.[2004*222G>A]+[2004*222G>A]		
	c.[2004*222G>A]+[2004*222G>A]		
c.[2004*127T>C]+[2004*127T>C]	c.[2004*222G>A]		
	c.[2004*222G>A]+[2004*222G>A]		
c.[2004*127T>C]+[2004*127T>C]	c.[2004*222G>A]		
c.[2004*127T>C]+[2004*127T>C]	c.[2004*222G>A]		
	c.[2004*222G>A]+[2004*222G>A]		
c.[2004*127T>C]+[=]			

Exon 19
Novel
UTR
c.[2004*628G>A]+[=]
c.[2004*628G>A]+[=]
c.[2004*628G>A]+[2004*628G>A]
c.[2004*628G>A]+[=]
c.[2004*628G>A]+[=]
c.[2004*628G>A]+[=]
c.[2004*628G>A]+[=]
c.[2004*628G>A]+[=]