

Supplementary Table 1. *RS1* mutations identified in the 120 individuals (92 families) included in this review. Three novel mutations are highlighted in bold.

Location	DNA	Protein	Number of Families with Mutation	
Exon 1	c.1A>T	p.Met1Leu	1	
	exon 1 deletion		2	
Exon 2	exon 2 deletion		4	
Exon 3	c.103C>T	p.Gln35Ter	1	
	c.176G>A	p.Cys59Tyr	1	
Exon 4	c.208G>A	p.Gly70Ser	3	
	c.209G>A	p.Gly70Asp	2	
	c.214G>A	p.Glu72Lys	10	
	c.219delA	p.Glu75Argfs	1	
	c.221_237del17ins20	p.Gly74Valfs	1	
	c.253_255delAAC	p.Asn85del	1	
	c.266A>G	p.Tyr89Cys	1	
	<b>c.272G&gt;T</b>	<b>p.Gly91Val</b>	<b>1</b>	
	c.276G>C	p.Trp92Cys	1	
	c.286T>C	p.Trp96Arg	1	
	c.288G>A	p.Trp96Ter	1	
	c.288G>C	p.Trp96Cys	1	
	c.304C>T	p.Arg102Trp	4	
	c.305G>A	p.Arg102Gln	2	
	c.306_308dupGCT	p.Leu103dup	1	
Exon4/Intron 4	<b>c.325_326+21del123</b>		<b>1</b>	
Exon 4-5	exon 4-5 deletion		1	
Exon 5	c.421C>T	p.Arg141Cys	1	
	c.354delCins18	p.Asp118Gluufs	3	
	c.380T>C	p.Leu127Pro	1	
	c.390delC	p.Val132Ter	1	
	c.407T>C	p.Ile136Thr	1	
	c.416delA	p.Gln139Argfs	1	
	c.419G>A	p.Gly140Glu	1	
	c.421C>T	p.Arg141Cys	1	
	c.422G>A	p.Arg141His	2	
	c.424T>C	p.Cys142Arg	1	
	c.456delC	p.Ser152Argfs	1	
	c.461A>G	p.Gln154Arg	1	
	c.498C>A	p.Tyr166Ter	1	
	<b>c.511G&gt;A</b>	<b>p.Gly171Arg</b>	<b>1</b>	
	c.520delC	p.Arg174Glyfs	4	
	Exon 6	c.535A>G	p.Asn179Asp	1
		c.574C>T	p.Pro192Ser	4
		c.589C>T	p.Arg197Cys	1
		c.590G>A	p.Arg197His	1
c.596T>C		p.Ile199Thr	2	
c.598C>T		p.Arg200Cys	4	
c.625C>T		p.Arg209Cys	1	
c.626G>A		p.Arg209His	1	
c.631G>A		p.Ala211Thr	1	
c.637C>T		p.Arg213Trp	2	
c.638G>A		p.Arg213Gln	2	
c.657C>G		p.Cys219Trp	1	
Intron 1		c.52+1G>A		1
	c.53-34A>G		2	
Intron 4	c.326+1G>A		1	
Intron 5	c.522+1G>A		1	
	c.523-1G>C		1	