

Supplementary Table 1. RSI 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	
	c.272G>T	p.Gly91Val	1	
	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	c.325_326+21del123		1	
Exon 4-5	exon 4-5 deletion		1	
Exon 5	c.421C>T	p.Arg141Cys	1	
	c.354delCins18	p.Asp118Glufs	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	
	c.511G>A	p.Gly171Arg	1	
	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	