Table 3. Polymorphisms and Non-Coding Sequence Alterations in *MKKS*

Phenotype	Sequence Alteration	Exon	Coding Frequency	Reference
BBS/MKS	c.-675_674ins [GTGGCGGCCT]	1A	2.25%	Slavotinek et al 2002
	c.-74G>A	3	0/94	
	c.117C>T; Nil	3	Not done	
BBS	p.A8T	3	Not done	Myktyn et al 2003
	p.G49V	3	8.3%	Stone et al 2002
	c.534C>T; Nil	3	Not done	Slavotinek et al 2002
	p.R517C	6	6.3%	Stone et al 2000, Hichri et al 2005
	p.G532V	6	6.4%	
BBS	p.I339V [1]	4	Not done	Slavotinek et al 2002, Hichri et al 2005
BBS	p.P39P	2	5/27 families	Hichri et al 2005
BBS	p.I178I	3	4/27 families	Hichri et al 2005
Juvenile-onset obesity (male)	p.P39P	2	In complete linkage disequilibrium with p.R517C	Andersen et al 2005
Juvenile-onset obesity (male)	p.I178I	3	In complete linkage disequilibrium with p.R517C	Andersen et al 2005
Juvenile-onset obesity (male)	p.A242S	3	2/60	Andersen et al 2005
Juvenile-onset obesity (male)	p.R517C	6	11.9%	Andersen et al 2005
Juvenile-onset obesity (male)	p.G532V	6	1/60	Andersen et al 2005
Middle-aged onset obesity (male)	p.R517C	6	9.7%	Andersen et al 2005

1. The status of this missense alteration is uncertain [Hichri et al 2005].