Title: Snyder-Robinson syndrome GeneReview Table 3 Authors: Albert J, Schwartz C, Boerkoel C, Stevenson R

Date: June 2013

Note: The following information is provided by the authors listed above and has not

been reviewed by GeneReviews staff.

Table 3. SMS Allelic Variants Discussed in this GeneReview

Class of Variant Allele	DNA Nucleotide Change (Alias ¹)	Protein Amino Acid Change (Alias ¹)	Reference Sequences
Pathologic	c.449T>C*	p.I150T	
	c.983A>C [†]	p.Y328C	
	c.174T>A*	p.F58L	
	c.200G>A [†]	p.G67X	

^{*}Not published, [†]Submitted for publication.

See <u>Quick Reference</u> for an explanation of nomenclature. GeneReviews follows the standard naming conventions of the Human Genome Variation Society (<u>www.hgvs.org</u>).

1. Variant designation that does not conform to current naming conventions