Title: Holoprosencephaly Overview *GeneReview* Table 2

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 2. Summary of Sequence Variations in *SHH*

	Sequence Change	Expected Effect	Type of Mutation	Reference
Mutations	160-161 insGCTG	3-4 ins	Insert with frameshift	Nanni et al 1999
	189-196 del	13-15 del	Deletion with frameshift	Nanni et al 1999
	91G→A	G31R	Missense	Roessler et al 1996
	263→A	D88V	Missense	Nanni et al 1999
	298C→T	Q100X	Nonsense	Roessler et al 1996
	300G→C	Q100H	Missense	Odent et al 1999
	313A→T	K105X	Nonsense	Roessler et al 1996
	345C→A	N115K	Missense	Nanni et al 1999
	349T→G	W117G	Missense	Roessler et al 1996
	349T→C	W117R	Missense	Roessler et al 1996
	474C→G	Y158X	Nonsense	Odent et al 1999
	562G→C	E188Q	Missense	Odent et al 1999
	625C→T	Q209X	Nonsense	Nanni et al 1999
	664G→A	D222N	Missense	Odent et al 1999
	671T→A	V224E	Missense	Roessler et al 1997
	676G→A	A226T	Missense	Roessler et al 1997
	708C→A	S236R	Missense	Nanni et al 1999
	766G→T	E256X	Nonsense	Nanni et al 1999
	939-959 del	263-269 del	Deletion in frame	Nanni et al 1999
	939-959 del	263-269 del	Deletion in frame	Roessler et al 1997

Mutations	Sequence Change	Expected Effect	Type of Mutation	Reference
	850G→T	E284X	Nonsense	Roessler et al 1997
	869G→A	G290D	Missense	Nanni et al 1999
	1283-1291 del	378-380 del	Deletion in frame	Nanni et al 1999
	1147G→A	A383T	Missense	Roessler et al 1997
	1361-1375 del	404-408 del	Deletion in frame	Nanni et al 1999
	1270C→G	P424A	Missense	Nanni et al 1999
	1308C→T	S436L	Missense	Nanni et al 1999
Polymorphisms	**Sequence Change**	**Expected Effect**	**Frequency**	**Reference**
	570G→A	S190S	2/344	Nanni et al 1999
	585G→A	S195S	2/344	Roessler et al 1997
	630C→T	G210G	1/344	Nanni et al 1999
	825G→A	A275A	1/344	Present study
	876G→A	G292G	1/344	Roessler et al 1997
	885C→T	S295S	1/344	Nanni et al 1999
	1005G→A	V335V	2/344	Present study

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