Title: Holoprosencephaly Overview *GeneReview* Table 4

Authors: Muenke M, Gropman A

Date: October 2010

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

Table 4. Summary of Sequence Variations in *SIX3*

Mutations	**Sequence Change**	**Expected Effect**	**Type of Mutation**	**Reference**
	676C→G	L226V	Missense	Wallis et al 1999
	696-705 del	232-234 del	Deletion in frame	Wallis et al 1999
	749T→C	V250A	Missense	Wallis et al 1999
	770C→G	R257P	Missense	Wallis et al 1999
Polymorphisms	**Sequence Change**	**Expected Effect**	**Frequency**	**Reference**
	90G→T	A30A	3/326	Present study
	219C→T	P73P	1/326	Present study
	576C→T	R192R	27/351 [1]	Nanni et al 2000
	942G→A	A314A	34/315 [2]	Present study

1. Two individuals with 576C→T are homozygous T.

2. One individual with 942G→A is homozygous A; 15 individuals are heterozygous for both 576C→T and 942G→A.

References

Nanni L, Croen LA, Lammer EJ, Muenke M. Holoprosencephaly: molecular study of a California population. Am J Med Genet. 2000;90:315–9.

Wallis DE, Roessler E, Hehr U, Nanni L, Wiltshire T, Richieri-Costa A, Gillessen-Kaesbach G, Zackai EH, Rommens J, Muenke M. Mutations in the homeodomain of the human SIX3 gene cause holoprosencephaly. Nat Genet. 1999;22:196–8.