Title: Holoprosencephaly Overview *GeneReview* Table 5

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

Table 5. Summary of Sequence Variations in *TGIF*

Mutations	**Sequence Change**	**Expected Effect**	**Type of Mutation**	**Reference**
	83C→G	S28C	Missense	Gripp et al 2000
	188C→G	P63R	Missense	Gripp et al 2000
	451A→G	T151A	Missense	Gripp et al 2000
	485C→T	S162F	Missense	Gripp et al 2000
Polymorphisms	**Sequence Change**	**Expected Effect**	**Frequency**	**Reference**
	371C→T	S20S	1/357	Present study
	420A→G	P140P	25/357	Nanni et al 2000
	487C→T	P163S	22/357	Nanni et al 2000
	488C→T	P163L	27/357	Nanni et al 2000
	887C→T	V192V	4/357	Present study
	968T→G	T219T	6/357	Present study

References

Gripp KW, Wotton D, Edwards MC, Roessler E, Ades L, Meinecke P, Richieri-Costa A, Zackai EH, Massague J, Muenke M, Elledge SJ. Mutations in TGIF cause holoprosencephaly and link NODAL signalling to human neural axis determination. *Nat Genet*. 2000;25:205–8.

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