Title: Holoprosencephaly Overview *GeneReview* Table 3

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Date: October 2010

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

Table 3. Summary of Sequence Variations in *ZIC2*

	Sequence Change	Amino Acid Location	Type of Mutation	Reference
Mutations	1 bp del G	7	Frameshift	Brown et al 1998, 2001
	56 bp ins	60	Frameshift	Brown et al 1998, 2001
	1 bp del G	312	Frameshift	Brown et al 1998, 2001
	7 bp del	348	Frameshift	Brown et al 1998, 2001
	2 bp del AG	364	Frameshift	Brown et al 1998, 2001
	1 bp ins C	440	Frameshift	Brown et al 1998, 2001
	30 bp ins	468-478	Alanine expansion [1]	Brown et al 1998, 2001

1. Alanine expansion found in five unrelated individuals

References

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