

Title: Holoprosencephaly Overview *GeneReview* Table 6

Authors: Muenke M, Gropman A

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

Table 6. HPE Candidate Genes and Their Chromosomal Loci

Gene	Chromosomal Location	Candidate Genes	Reference
Cytogenetically defined HPE loci			
<i>HPE1</i>	21q22.3	Lanosterol synthase?	Muenke et al 1995, Roessler et al 1996
<i>HPE2</i>	2p21	<i>SIX3</i>	Schell et al 1996, Wallis et al 1999
<i>HPE3</i>	7q36	Sonic hedgehog (<i>SHH</i>)	Gurrieri et al 1993, Belloni et al 1996, Roessler et al 1996, Roessler et al 1997
Sacral agenesis	7q36	Not sonic hedgehog (<i>SHH</i>)	Vargas et al 1998
Limb anomaly	7q36	Not sonic hedgehog (<i>SHH</i>)	Vargas et al 1998
<i>HPE4</i>	18p11.3	Homeobox gene TGIF	Bertolino et al 1995, Overhauser et al 1995, Gripp et al 2000
<i>HPE5</i>	13q32	Zinc finger protein gene (<i>ZIC2</i>)	Brown et al 1998
<i>HPE6</i>	3p24-pter	Zinc finger protein gene, <i>ZIC1</i> and/or <i>ZIC4</i> ?	Aruga et al 1996a, Aruga et al 1996b
<i>HPE7?</i>	13q12-q14	Forkhead homolog (FKHR)? Homeobox transcription factor, <i>CDX3</i> ?	Norman et al 1995
<i>HPE8?</i>	14q13	Thyroid transcription factor-1 (TTF1)?	Chen et al 1997, OMIM 600635
<i>HPE9?</i>	20p13	HNF3 beta	Muenke et al unpublished
<i>HPE10?</i>	1q42-qter	?	Norman et al 1995
<i>HPE11?</i>	5p	?	Norman et al 1995
<i>HPE12?</i>	6q26-qter	?	Norman et al 1995

Gene	Chromosomal Location	Candidate Genes	Reference
SHH signaling pathway			
PATCHED (PTC)	9q22.3	Role in HPE?	Hahn et al 1996a, Hahn et al 1996b, Ming et al unpublished
SMOOTHENED (SMO)?		Role in HPE?	Stone et al 1996, Ming et al unpublished
GLI family	(various)	Role in HPE?	Roessler, Du et al unpublished
WNT family	(various)	Role in HPE?	OMIM 164820, 147870, 165330, 164975, 601570, 601967, 601396, 601906, 601968
BMP family	(various)	Role in HPE?	Golden et al 1999, OMIM 112264, 112262, 112265
Cholesterol metabolism			
Smith-Lemli-Opitz (SLO gene)	11q13	7-dehydro cholesterol reductase	Kelley et al 1996, Moebius et al 1998

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