Table 6. HPE Candidate Genes and Their Chromosomal Loci

Gene	Chromosomal Location	Candidate Genes	Reference
Cytogenetically defined HPE loci			
HPE1	21q22.3	Lanosterol synthase?	Muenke et al 1995, Roessler et al 1996
HPE2	2p21	*SIX3*	Schell et al 1996, Wallis et al 1999
HPE3	7q36	Sonic hedgehog (*SHH*)	Gurrieri et al 1993; Belloni et al 1996; Roessler et al 1996; Roessler, Belloni et al 1997
Sacral agenesis	7q36	Not sonic hedgehog (*SHH*)	Vargas et al 1998
Limb anomaly	7q36	Not sonic hedgehog (*SHH*)	Vargas et al 1998
HPE4	18p11.3	Homeobox gene *TGIF*	Bertolino et al 1995, Overhauser et al 1995, Gripp et al 2000
HPE5	13q32	Zinc finger protein gene (*ZIC2*)	Brown et al 1998
HPE6	3p24-pter	Zinc finger protein gene, ZIC1 and/or ZIC4?	Aruga, Nagai et al 1996; Aruga, Yozu et al 1996
HPE7?	13q12-q14	Forkhead homolog (FKHR)? Homeobox transcription factor, CDX3?	Norman et al 1995
HPE8?	14q13	Thyroid transcription factor-1 (TTF1)?	Chen et al 1997, OMIM 600635
HPE9?	20p13	HNF3 beta	Muenke et al unpublished
HPE10?	1q42-qter	?	Norman et al 1995
HPE11?	5p	?	Norman et al 1995
HPE12?	6q26-qter	?	Norman et al 1995
***SHH* signaling pathway**			
PATCHED (PTC)	9q22.3	Role in HPE?	Hahn, Christiansen et al 1996; Hahn, Wicking et al 1996; 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			unpublished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	Stone et al 1996, Ming et al unpublished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	Roessler, Du et al unpublished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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