Title: Pallister-Hall Syndrome *GeneReview* Table 3

Author: Biesecker LG

Date: September 2012

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

Table 3. Selected *GLI3* Pathogenic Allelic Variants for Pallister-Hall Syndrome

DNA Nucleotide Change	Protein Amino Acid Change
c.1998_2001del4 [1]	p.P668_T669delinsLfsX24
c.2012delG [2]	p.Gly671GlufsX21
c.2023delG [2]	p.Glu675SerfsX17
c.2032delG [1]	p.Asp678ThrfsX15
c.2058_2059delinsAT [3]	p.Glu687X
c.2062G>T [1]	p.Glu688X
c.2110C>T [1]	p.Gln704X
c.2139delC [1]	p.Cys713X
c.2146C>T [1]	p.Gln716X
c.2149C>T [1]	p.Gln717X
c.2157delC [1]	p.Ile720SerfsX13
c.2172_2173insC [1]	p.Asn725GlnfsX13
c.2188_2206del19 [1]	p.Leu730ValfsX3
c.2197_2198delAC [1]	p.Thr733ArgfsX4
c.2346_2356del11 [1]	p.Arg782_Val786delinsSerfsX15
c.2351_2355del5 [1]	p.Lys784_Gln785delinsSerfsX15
c.2431+1G>A [1]	IVS14
c.2483delC [1]	p.Pro828ArgfsX14
c.2567C>A [1]	p.Ser856X
c.2620delC [1]	p.Arg874AlafsX16
c.2628delC [1]	p.Ser877AlafsX13
c.2770_2771ins72 [1]	p.Ala924ValfsX12
c.2799C>G [1]	p.Tyr933X
c.2935delT [4]	p.Cys979AlafsX23
c.3004delG	p.Val1002X
c.3324C>G [1]	p.Tyr1108X
c.3386_3387delT 1[4]	p.Phe1129X
c.3439G>T [5]	p.Glu1147X

DNA Nucleotide Change	Protein Amino Acid Change
c.3456G>T [1]	p.Glu1152X
c.3481C>T [6]	p. Gln1161X

Variants named according to current nomenclature guidelines (http://www.hgvs.org/). In protein nomenclature, an fsX# indicates there is a frameshift in the sequence, which terminates at a stop codon (X) that is a specific number (#) of amino acid residues downstream from the changed amino acid. GLI3 reference sequences are NM_000168.3 and NP_000159.3.

1. Johnston JJ, Olivos-Glander I, Killoran C, Elson E, Turner JT, Peters KF, Abbott MH, Aughton DJ, Aylsworth AS, Bamshad MJ, Booth C, Curry CJ, David A, Dinulos MB, Flannery DB, Fox MA, Graham JM, Grange DK, Guttmacher AE, Hannibal MC, Henn W, Hennekam RC, Holmes LB, Hoyme HE, Leppig KA, Lin AE, Macleod P, Manchester DK, Marcelis C, Mazzanti L, McCann E, McDonald MT, Mendelsohn NJ, Moeschler JB, Moghaddam B, Neri G, Newbury-Ecob R, Pagon RA, Phillips JA, Sadler LS, Stoler JM, Tilstra D, Walsh Vockley CM, Zackai EH, Zadeh TM, Brueton L, Black GC, Biesecker LG. Molecular and clinical analyses of Greig cephalopolysyndactyly and Pallister-Hall syndromes: robust phenotype prediction from the type and position of GLI3 mutations. Am J Hum Genet. 2005;76:609-22.

2. Kang S, Allen J, Graham JM Jr, Grebe T, Clericuzio C, Patronas N, Ondrey F, Green E, Schaffer A, Abbott M, Biesecker LG. Linkage mapping and phenotypic analysis of autosomal dominant Pallister-Hall syndrome. J Med Genet. 1997;34:441-6.

3. Kalff-Suske M, Paparidis Z, Bornholdt D, Cole T, Kalff-Suske M, Grzeschik KH. Gene symbol: GLI3. Disease: Pallister-Hall syndrome. Hum Genet. 2004;114:403.

4. Freese K, Driess S, Bornholdt D, Shoenle EJ, Seidel H, Tinschert S, Grzeschik KH, Kalff-Suske M. Gene symbol: GLI3. Disease: Pallister-Hall syndrome. Hum Genet. 2003;112:103.

5. Radhakrishna U, Bornholdt D, Scott HS, Patel UC, Rossier C, Engel H, Bottani A, Chandal D, Blouin JL, Solanki JV, Grzeschik KH, Antonarakis SE. The phenotypic spectrum of GLI3 morphopathies includes autosomal dominant preaxial polydactyly type-IV and postaxial polydactyly type- A/B; No phenotype prediction from the position of GLI3 mutations. Am J Hum Genet. 1999;65:645-55.

6. Freese K, Driess S, Bornholdt D, Shoenle EJ, Seidel H, Tinschert S, Grzeschik KH, Kalff-Suske M. Gene symbol: GLI3. Disease: Pallister-Hall syndrome. Hum Genet. 2003;112:103.