

Title: Greig Cephalopolysyndactyly Syndrome *GeneReview* – Table 2
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Table 3. Selected *GLI3* Pathologic Allelic Variants

DNA Nucleotide Change	Protein Amino Acid Change
c.1998_2001del4 ⁴	p.P668_T669delinsLfsX24
c.2012delG ¹	p.Gly671GluX21
c.2023delG ¹	p.Glu675SerfsX17
c.2032delG ⁴	p.Asp678ThrfsX15
c.2058_2059delinsAT ⁵	p.Glu687X
c.2062G>T ⁴	p.Glu688X
c.2110C>T ⁴	p.Gln704X
c.2139delC ⁴	p.Cys713X
c.2146C>T ⁴	p.Gln716X
c.2149C>T ⁴	p.Gln717X
c.2157delC ⁴	p.Ile720SerfsX13
c.2172_2173insC ⁴	p.Asn725GlnfsX13
c.2188_2206del19 ⁴	p.Leu730ValfsX3
c.2197_2198delAC ⁴	p.Thr733ArgfsX4
c.2346_2356del11 ⁴	p.Arg782_Val786delinsSerfsX15
c.2351_2355del5 ⁴	p.Lys784_Gln785delinsSerfsX15
c.2431+1G>A ⁴	IVS14
c.2483delC ⁴	p.Pro828ArgfsX14
c.2567C>A ⁴	p.Ser856X
c.2620delC ⁴	p.Arg874AlafsX16
c.2628delC ⁴	p.Ser877AlafsX13
c.2770_2771ins72 ⁴	p.Ala924ValfsX12
c.2799C>G ⁴	p.Tyr933X
c.2935delT ⁶	p.Cys979AlafsX23
c.3004delG	p.Val1002X
c.3324C>G ⁴	p.Tyr1108X
c.3386_3387delT ⁴	p.Phe1129X
c.3439G>T ²	p.Glu1147X
c.3456G>T ⁴	p.Glu1152X
c.3481C>T ³	p. Gln1161X

Variants named according to current nomenclature guidelines (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](#).

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4. Johnston JJ, Olivios-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 (2005) 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* 76:609-22.
5. Kalff-Suske M, Paparidis Z, Bornholdt D, Cole T, Kalff-Suske M, Grzeschik KH. (2004) Gene symbol: GLI3. Disease: Pallister-Hall syndrome. *Hum Genet.* 114:403.
6. Freese K, Driess S, Bornholdt D, Shoenle EJ, Seidel H, Tinschert S, Grzeschik KH, Kalff-Suske M. (2003) Gene symbol: GLI3. Disease: Pallister-Hall syndrome. *Hum Genet.* 112:103.