Title: Greig Cephalopolysyndactyly Syndrome *GeneReview* – Table 2
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Table 2. Selected *GLI3* Putative Normal Allelic Variants

DNA Nucleotide Change (Alias)	Protein Amino Acid Change	dbSNP Reference Number
c.124+61C>A (IVS2+61C>A)	N.A.	
c.547A>G	p.His179	rs3898405
c. 547A>G	p.Thr183Ala	rs846266
c.826+153T>C (IVS6+153T>C)	N.A.	
c.900C>T	p.Ser300	rs35961850
c.963C>T	p.Leu321	rs34965132
c.1356+11G>C (IVS9+11G>C)	N.A.	rs846273
c.1393G>C	p.Gly465Arg	rs35488756
c.1509C>T	p.Asn503	rs34020684
c.1728C>T	p.Y576	rs35128755
c.2361T>A	p.Asn787Lys	rs10259802
c.2408G>A	p.Ala803Val	rs34169786
c.2826G>C	p.Pro942	rs34245321
c.2835G>C	p.Leu945	
c.2993C>T	p.Pro998Leu	rs929387
c.3774C>G	p.Leu1258	rs35448119
c.4006G>A	p.Gly1336Glu	rs35280470
c.4020C>T	p.Pro1340	rs35139358
c.4071C>T	p.Tyr1357	rs34089404
c.4293G>C	p.Pro1431	rs28396689
c.4595C>G	p.Ser1532Cys	rs2079451
c.4609C>T	p.Arg1537Cys	rs35364414
c.4705delC	p.Leu1565X	rs35765130
c.*30G>T	N.A.	
c.*136T>A	N.A.	

Most of these variants have been seen in multiple unrelated persons and are not believed to be associated with any phenotypic effects, although they have not been rigorously analyzed for subtle effects. These are included in this table if they lie within an exon or if they are in an intron within 25 bp of an exon.
N.A.= Not applicable.
The dbSNP reference SNP cluster 'rs' ID's are at www.ncbi.nlm.nih.gov/SNP/. Readers should refer to dbSNP to confirm these data and for additional data. These SNPs are from the Human Genome build 126. Note: (1) Nomenclature for normal

and pathologic allelic variants follows recommendations of den Dunnen & Antonarakis (2001) and the updated recommendations at the Human Gene Variation Society website http://www.hgvs.org/. An asterisk indicates that the variant is located X number of nucleotides after the stop codon; (2) All protein alterations are predicted and not experimentally determined; (3) GLI3 reference sequences are NM_000168.3 and NP_000159.3.