Title: Hypochondroplasia *GeneReview* Table 3

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

Table 3. FGFR3 Pathologic Allelic Variants that cause Hypochondroplasia

Amino acid substitution	Base pair change	Domain
Gly65Arg	Not reported	IgI
Ser84Leu	c.250C>T	Ig I
Gln115Leu	Not reported	I-II linker
Arg200Cys	c.597C>T	Ig II
Asn262His	c.783A>C	II-III linker
Thr264Met	Not reported	II-III linker
Gly268Cys	c.801G>T	II-III linker
Tyr278Cys	c.829A>G	Ig IIIa
Leu324Val	c.970C>G	Ig IIIa
Asn328Ile	c.983A>T	Ig IIIa
Gly342Cys	c.1024G>T	Ig IIIa
Val381Glu	c.1142T>A	TM
Ile538Val	c.1612A>G	TK1
Asn540Thr	c.1619A>C	TK1
Asn540Ser	c.1619A>G	TK1
Asn540Lys*	c.1620C>A or G	TK1
Lys650Gln	c.1948A>C	TK1
Lys650Thr	c.1949A>C	TK1
Lys650Asn	c.1950G>T or C	TK1