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HUMAN GENE MUTATIONS

Gene symbol: COL11A1

Disease: Marshall Syndrome

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Splicing Mutations (single base-pair substitutions)

Accession Number	IVS	Donor/ Acceptor	Relative location	Substitution	Disease state
H971266	(40)	ds	+1	G-A	Marshall syndrome

Comments:
Gene structure of COL11A1 is not completely known. Intron corresponds to approximately 40 in COL5A1.