

Supplementary Table.

Complete list of all mutations described within the LMX1B coding region. The nomenclature has been corrected to fit the current guidelines [39] and to allow for the additional 69bp at the 5' end of the coding region. The reference numbers refer to the citations in the manuscript; the absence of a reference indicates description in the attached manuscript.

Original Designation	Corrected Nomenclature		Ref.
	Mutation	Nucleotide	
	p.G38fs	c.113-114delGGinsT	
71-37del17	c.140-38_140-22del	c.140-38_140-22del	49
71-2A→G	c.1401-2A>G	c.1401-2A>G	17
71-1G→A	c.140-1G>A	c.140-1G>A	8
85del27	p.H52_Q60del	c.155_181del	17
	p.C56S	c.167T>A	
	p.E57X	c.169G>T	
C36R	p.C59R	c.175T>C	17
C36S	p.C59S	c.176G>C	17
C36Y	p.C59Y	c.176G>A	18
	p.Q60X	c.177C>T	
115delC	p.P62fs	c.184delC	19
R47X	p.R70X	c.208C>T	19
	p.N72fs	c.214_321del	
S51X	p.S74X	c.221C>A	19
154del9	p.S75_H77del	c.223_231del	17
	p.W76R	c.226T>C	
	p.W76X	c.227G>A	
W53X	p.W76X	c.228 G>A	17
H54N	p.H77N	c.229C>A	19
H54Y	p.H77Y	c.229C>T	17
H54L	p.H77L	c.230 A>T	18
H54Q	p.H77Q	c.231C>R	17
	p.E78X	c.232G>T	
C57R	p.C80R	c.238T>C	17
C57X	p.C80X	c.239delGinsAA	19
L58W	p.L81W	c.242T>G	19
Q59X	p.Q82X	c.244C>T	9
C60G	p.C83G	c.247T>G	17
C60R	p.C83R	c.247T>R	19
C60Y	p.C83Y	c.248G>A	17
C60F	p.C83F	c.248G>T	19
C63R	p.C86R	c.255T>C	17
187insT	p.C86fs	c.256_257insT	17
187del46	p.C86fs	c.256_301del	17

Original Designation	Corrected Nomenclature		Ref.
	Mutation	Nucleotide	
Q64X	p.Q87X	c.259C>T	17
199insTC	p.L90fs	c.268_269insTC	17
200del8	p.L90fs	c.270_277del	17
200ins57	p.L90_Q109dup	c.270-326ins	19
	p.C94W	c.282C>G	
232delTG	p.L101fs	c.302_303del	9
C80W	p.C103W	c.308C>G	19
	p.K104fs	c.310_312del1	
	p.Q105fs	c.313del	
D83G	p.D106G	c.317A>G	17
	p.Y107fs	c.320del	
Y84X	p.Y107X	c.321C>G	17
	p.Q108fs	c.321_322del1	
Q85X	p.Q108X	c.322C>T	19
Q86X	p.Q109X	c.325C>T	19
	c.326+1G>A	c.326+1G>A	
257+2T→C	c.326+2T>C	c.326+2T>C	8
	c.326+3T>C	c.326+3T>C	
	c.327-2_334del	c.327-2_334del	
258-2A→G	c.327-2A>G	c.327-2A>G	17
	p.C115R	c.343T>C	
C95Y	p.C118Y	c.353G>A	17
C95F	p.C118F	c.353G>T	9
	p.E120fs	c.356_357del1	
320delG	p.A131fs	c.389del	8
C111X	p.C134X	c.402C>A	17
Y113X	p.Y136X	c.408C>G	17
H114Y	p.H137Y	c.409C>T	19
	p.H137R	c.410A>G	
C117Y	p.C140Y	c.419G>A	19
C120Y	p.C143Y	c.428G>A	19
C120S	p.C143S	c.428G>C	17
C123Y	p.C146Y	c.437G>A	17
C123F	p.C146F	c.437G>T	17
	p.R148P	c.443G>C	
F133X	p.F156X	c.466_467insGAAT	17
Q139X	p.Q162X	c.484C>T	17
C142W	p.C165W	c.495C>G	8
Y146X	p.Y169X	c.507C>A	8
485ins8	p.S186fs	c.553_554ins8	8
	p.V187_K247del	c.560-1848_741+3del	
534delG	p.Q202fs	c.600_603del1	8
553del3insA	p.G208fs	c.622_624delinsA	19

Original Designation	Corrected Nomenclature		Ref.
	Mutation	Nucleotide	
571delA	p.K214fs	c.641_642del1	19
	p.R221fs	c.661del	
	p.P222fs	c.662_664insA	
R198X	p.R221X	c.661C>T	7
R200Q	p.R223Q	c.668G>A	8
606del6	p.L226_T227del	c.676_681del	19
611del7	p.T228fs	c.681_687del	8
R208X	p.R231X	c.691 C>T	9
A213P	p.A236P	c.706G>C	8
E216X	p.E239X	c.715G>T	8
ΔS218	p.S241del	c.721_723del	19
S218P	p.S241P	c.721T>C	8
S218X	p.S241X	c.722C>A	17
	p.S242X	c.725 C>A	
	p.K243fs	c.727_728del1	
	p.C245X	c.735_736CC>AA	
R223X	p.R246X	c.736 C>T	8
672+1G→A	c.741+1G>A	c.741+1G>A	8
672+1G→T	c.741+1G>T	c.741+1G>T	8
672+1G→C	c.741+1G>C	c.741+1G>C	19
672+2T→A	c.741+2T>A	c.741+2T>A	17
	p.V248_Q265del	c.742-9_785del51	
	c.742-2A>G	c.742-2A>G	
	c.742-1G>A	c.742-1G>A	
	c.742-1G>C	c.742-1G>C	
676insC	p.17250fs	c.743_746insC	19
	p.R249G	c.745C>G	
R226X	p.R249X	c.745C>T	8
R226P	p.R249P	c.746G>C	8
L229P	p.L252P	c.755T>C	17
A230V	p.A253V	c.758C>T	8
694del17	p.V260fs	c.763_768del	19
697insA	p.T256fs	c.765_767insA	7
	p.T256R	c.767C>G	
699delG	p.L258fs	c.768_770del1	10
	p.L258F	c.772C>T	
	p.S259fs	c.774_775del	
706delAG	p.S259fs	c.775_776del	10
708delT	p.V260fs	c.777del	17
	p.R261C	c.781C>T	
V240D	p.V263D	c.788 T>A	18
	p.V263A	c.788T>C	

Original Designation	Corrected Nomenclature		Ref.
	Mutation	Nucleotide	
Q241X	p.Q264X	c.790C>T	17
	p.V265L	c.793G>C	
	p.V265F	c.793G>T	
W243X	p.W266X	c.797G>A	8
W243C	p.W266C	c.798G>C	19
	p.F267L	c.799T>C	
	p.F267I	c.799T>A	
Q245X	p.Q268X	c.802C>T	8
	p.Q268H	c.804 G>T	
N246K	p.N269K	c.807C>A	7
A249P	p.A272P	c.814G>C	50
750+1G→A	c.819+1G→A	c.819+1>→A	8
	c.819+2T→G	c.819+2T>G	
	c.820-2del	c.820-2del	
	c.820-1G>A	c.820-1G>A	
	p.L277fs	c.829del	
	p.R280fs	c.838_847del,856_857ins(845_856,845_876,853_856,854_856)	