

Title: Congenital Contractural Arachnodactyly *GeneReview* Table 2

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

Table 2. *FBN2* Mutations Known to Cause Congenital Contractural Arachnodactyly

Amino Acid Change	Exon	Reference
p.Gly1056Asp	24	Park et al 1998
p.Ile1092Thr	25	Park et al 1998
p.Ala1114His	25	Babcock et al 1998
p.Cys1141Phe	26	Belleh et al 2000
p.Cys1252Tyr	29	Putnam et al 1995
p.Cys1252Trp	29	Belleh et al 2000
Deletion of exon 29	29	Putnam et al 1997
Deletion of exon 31	31	Maslen et al 1997, Park et al 1998
p.Cys1433Ser	33	Putnam et al 1995
Deletion of exon 34 ¹	34	Wang et al 1996

1. Found in an individual with severe/lethal CCA and in her mother, who was mosaic for the mutation [Wang et al 1996]

References

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