# Letters to the Editor

The undersigned authors wish to comment on the article "Novel Dental Anomalies Associated With Congenital Contractural Arachnodactyly: A Case Report" by Drs. Kathryn Ayers and Bernadette Drummond (*Pediatr Dent.* 2003;25:501-504).

We appreciate Drs. Ayers' and Drummond's clinical description of a young girl with congenital contractural arachnodactyly (CCA). A number of their findings are consistent with our previous reports on similar features in Marfan syndrome which also is caused by fibrillin deficiency. Their findings strengthen our hypothesis that fibrillin deficiency may account for a number of developmental anomalies in the orofacial region. However, we disagree with a number of their statements and conclusions.

First, we feel that the background information provided in the article is incomplete and somehow inaccurate. Drs. Ayers and Drummond referred to our article on oral manifestations in Marfan syndrome (*Oral Surg Oral Pathol Oral Med Oral Radiol Endodontol.* 2002;93:564-572), presenting inaccurate information on several occasions. They state that "high-arched palate" and "widely spaced anterior maxillary teeth" and "central palatal clefts" were reported as cranofacial features in CCA in our article. This is not true. Reports of this nature have been made only once in a single CCA case by Sanger et al (*Oral Surg.* 1975;40:354-361). In addition, the rarity of cleft palate in Marfan syndrome (only 2 reports dating back from the 1950s) does not permit presenting this feature as intrinsic to the syndrome, as displayed in Table 1 of Drs. Ayers and Drummond's paper. To the best of our knowledge, a cleft palate is also rarely seen in CCA. As is now widely accepted, an enlargement of the palatal shelves (byzantine arch palate), as featured in a number of marfanoid syndromes, such as Shprintzen-Goldberg syndrome and Idaho syndrome type II, probably was confused with a central palatal cleft in those patients. This assignment of sporadic symptoms to the typical diagnostic spectrum of a syndrome (Table 1) may mislead the reader, especially since the authors point to the importance of determining orofacial signs in order to match an undiagnosed syndrome. The diagnostic validity of the given symptoms should be mentioned in the text (ie, the degree to which these symptoms can be assigned a high or low diagnostic specificity according to the number of reports in literature and relationship with known molecular defects). We therefore recommend that similar articles be thoroughly reviewed by an experienced geneticist.

Second, it is not clear whether the CCA case has been genetically linked to *FBN2* gene (genetic linkage or DNA sequencing). Only an unambiguous laboratory diagnosis may warrant a clinical diagnosis of CCA.

Third, we feel uncomfortable with the idea that banded enamel hypoplasia and hypomineralization on the upper anterior teeth, presenting in a single case, are considered as features of CCA. Attributing a connective tissue etiology to enamel defects may be incorrect. Over 80% of generalized developmental defects of enamel share a metabolic (non-genetic) etiology, especially when presenting in a banded pattern. Genetic enamel defects are caused by mutation in *ABMN, TUFT*, *AMELX*, or *ENAM*, which are enamel-specific genes encoding a limited number of regulatory proteins. There is no knowledge of crossover between human genetic conditions involving genes coding for collagens, fibrillins or enamel proteins. Above all, enamel is an ectodermal tissue, whereas connective tissue is descended from embryonic mesoderm. We also wonder why the authors did not include a clinical picture of the enamel defects, since this feature seems to be of diagnostic importance. In addition, we feel uncomfortable with their mentioning that our study reported on a high prevalence of structural enamel defects in Marfan syndrome, and hence might substantiate the diagnostic importance of banded enamel defects in CCA. In our paper, it was clearly stated that these enamel defects presented as hypoplastic spots in premolars, and could be related with a history of decay in the preceding primary molar (enamel defects of local infectious etiology). Please see our short communication in this issue of the journal for further details.

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# Response to Letters to the Editor

We appreciate the additional information and further discussion on Congenital Contractural Arachnodactyly (CCA) based on your extensive experience.

Our case report was based on the clinical presentation of one patient with CCA. The dental anomalies noted in this patient included long spindly tapered roots with increased curvature and abnormal pulps, thistle tube shaped pulpal chambers, pulpal obliteration, pulp stones, banded pitted enamel hypoplasia and hypomineralisation. We accept that not all of these anomalies may be related to CCA and thank you for your clarification of this subject.

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