Letter

Noonan syndrome with giant cell lesions

Comment on Lee and Cooper, *International Journal of Paediatric Dentistry* 2005, **15**: 140–145

Amsterdam, 23 May 2005

Dear Sir,

In the article by Lee and Cooper [1], a rare case of Noonan syndrome combined with multiple giant cell lesions is extensively described. This Noonan-like/multiple giant cell lesion syndrome was first defined by Cohen and Gorlin in 1991 [2], whereas others have reported earlier on the Noonan syndrome/cherubism association [3]. At that time, the diagnosis could only be made on clinical and radiological findings. Noonan-like/multiple giant cell lesion syndrome was seen as a separate entity from Noonan syndrome.

In 2001, the gene mutations responsible for cherubism (SH3BP2) and Noonan syndrome (PTPN11) were identified on chromosome numbers 4(4p 16.3) and 12(12q24.1), respectively [4,5]. In a recent study on three patients with a Noonan-like/multiple giant cell lesion syndrome, the Noonan mutation was found in two patients, but the cherubism gene mutation was absent in all three patients [6,7]. This was confirmed in a similar patient from our department who also missed the cherubism mutation but exhibited the Noonan syndrome mutation. This means that Noonan-like/multiple giant lesion syndrome is probably not a separate entity but part of the spectrum of Noonan syndrome.

Regrettably, in the article by Lee and Cooper no mention is made of the fact that the gene mutations causing Noonan syndrome and cherubism have already been identified several years ago. Therefore, the statement by the authors that 'it seems likely that they are independent diseases transmitted by genes closely linked on the same chromosome' is not appropriate and does not reflect current scientific knowledge. In our view, DNA analysis is now mandatory in patients with a syndrome in whom central giant cell lesions occur in the jaw(s). The presence of a specific mutation may also have therapeutic consequences. When, for instance, the cherubism mutation is identified, there is no indication for surgery, considering the tendency for spontaneous resolution in most patients. In other syndromes, including Noonan's, alternative treatment in the form of calcitonin or interferon alpha should be considered for central giant cell lesions exhibiting aggressive signs and symptoms [8–10].

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