

## Letter to the Editor: The mode of inheritance' in differentiation of Fanconi anemia from Dyskeratosis congenita

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Dear Editor,

I read the article by Auluck A with great interest(1). A new patient with Dyskeratosis congenita (DC) who developed oral leukoplakic lesions has been described in this article. The author first has pointed out that most of the cases of DC have been described by dermatologists or pediatricians, and then emphasized the critical importance of better awareness about DC among dentists because of the oral leukoplakic lesions which take part in the classical triad of DC and have malignant transformation potential.

In the case presentation section of the article, the phases of the patient went through starting from childhood till the time of the diagnosis were described in detail. The obtained data proved to be typical for the clinical progress of DC, and will be quite useful for the target clinicians (i.e., dentists) in recognizing the disease.

In the discussion section of the article, it has been emphasized that Fanconi anemia (FA) is the major clinical condition that should be differentiated from DC. I do agree with the author that FA is a significant disease in the differential diagnosis of DC. Indeed, DC and FA resemble each other in terms of many physical features (low birth weight, growth retardation, cutaneous manifestations, head and skeletal abnormalities, hypogonadism, deafness), laboratory findings (cytopenias, increased HbF), and clinical characteristics (susceptibility to cancer in young ages), and thus they might be easily confused by the clinicians who are not professional in hematology. Right at the point where the differential diagnosis is being made between the two diseases, the author is stating that their

modes of inheritance are different and that, unfortunately, FA is inherited autosomal dominantly. This particular distinction is the one and only reason I am writing this letter. Traditionally known that FA is an autosomal recessive (AR) disorder(2,3). In fact it is the prototype of the autosomal recessively inherited disorders and has already been introduced to the medical literature as an AR disorder since its description. At this time, it is known, thought and lived to be an autosomal recessive disease. Fanconi anemia is one of the diseases that some ethnic populations, are being tested for carrier status(4,5). It is worth remembering that, premarital carrier screening tests are carried out for recessively inherited diseases. Because in autosomal recessive disorders, like FA, both parents carry one mutated allele and are referred to as 'carriers'. Each pregnancy has a 25% risk of passing on 2 mutated alleles resulting in an affected child. Interestingly, despite the AR inheritance FA has a slight male predominance (male:female 1.3). This discrepancy has been explained only recently: in 2004 Meetei et al have described a genetic subtype of FA with X-linked inheritance(6). In summary, to date, twelve genetic subtypes, also known as complementation groups, have been distinguished in FA: FA-A, B, C, D1, D2, E, F, G, I, J, L, and M. The mode of inheritance for all groups is autosomal recessive, except for FA-B, which is X-linked(7).

Obviously, for the dentists -the target clinicians of the article- the fundamental point is to be able to diagnose the DC patients and to get the precancerous oral leukoplakic lesions under control in the early stage as much as possible. Nonetheless, my position is this: It is crucial for the clini-

cians who surely will be a part of the team managing the DC patients to be armed with the right knowledge that the differential diagnosis between DC and the other disease, which is going to be primarily FA, has to do with the mode of inheritance. Specifically, the right knowledge is that of the twelve genetic subtypes of FA, 11 are AR and 1 of them is X-linked as far as inheritance is concerned.

I would like to thank to Dr Auluck both for the new case of DC described in the article, and via the present case, for the opportunity of reminding the current mode of inheritance of Fanconi anemia.

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