

ORAL AND DERMATOLOGICAL FEATURES IN FANCONI'S ANAEMIA PATIENTS

Aspectos dermatológicos e bucais em pacientes com anemia de Fanconi

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Abstract

Objective: The objective of this study was to evaluate the oral and dermatologic findings in a group of patients with Fanconi's anemia. **Patients and Method:** 30 patients with Fanconi's anemia under treatment at the Clinics Hospital of the Federal University of Paraná and at the Associação Paranaense de Apoio à Criança com Neoplasia em Curitiba, PR, Brazil were evaluated. The research was an exploratory type study and the focus was in the oral mucosa and in the skin were clinically evaluated. Patients submitted to bone marrow transplantation were excluded from this study. **Results:** 18 patients were female and 12 male; mean age 10 years and 10 month. The dermatological findings were present in 68% of the patients. ($p \leq 0.05$) (*t* test) (skin pigmentation, *cafe-au-lait* spots, petechia and purple). Melanine pigmentation, hypochromatic areas, nodules, spots and gingivitis were found in the oral mucosa of all patients (100%). **Conclusion:** There is a high prevalence of oral and head and neck dermatological and oral alterations in patients with Fanconi's anaemia.

Keywords: Fanconi's anaemia; Oral lesions; Head and neck skin lesions.

Resumo

Objetivo: O propósito deste trabalho foi estudar as alterações bucais e dermatológicas em pacientes portadores de anemia de Fanconi. **Método:** Os pacientes foram examinados no Hospital das Clínicas da UFPR e na Associação Paranaense de Apoio à Criança com Neoplasia, em Curitiba, PR, Brasil. A pesquisa foi realizada como um estudo do tipo exploratório, utilizando abordagem quantitativa, avaliando as características de pele e mucosa bucal, com uma amostra do tipo intencional de pacientes portadores de anemia de Fanconi. Foram avaliados 30 pacientes, sendo 18 do sexo feminino e 12 do sexo masculino, com idade média de 10 anos e 10 meses. Os pacientes submetidos a transplante de medula foram excluídos do estudo. **Resultados e conclusão:** 18 pacientes eram do sexo feminino e 12 do masculino. A idade média foi de 10 anos e 10 meses. Os achados dermatológicos estavam presentes em 68% dos pacientes (pigmentação cutânea, manchas *café com leite*, petéquias e púrpura). Pigmentação melânica, áreas hipocromáticas, pontos pigmentados e gengivite foram encontrados na mucosa bucal de todos os pacientes (100%). **Conclusão:** Há uma prevalência alta de alterações bucais e dermatológicas em pacientes com anemia de Fanconi.

Palavras-chave: Anemia de Fanconi; Lesões bucais; Lesões de pele; Cabeça e pescoço.

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Introduction

Fanconi's anaemia is an autosomal recessive disorder described in 1927 by Guido Fanconi (1). Since this description, more than 1000 cases have been reported, and the disease is now better understood. It is characterized by severe constitutional aplastic anemia with pancytopenia associated with brown pigmentation of the skin along with congenital abnormalities such as microcephaly, short stature, absence of the radii and the thumb and malformations of the kidney and the heart (2)

The diagnosis of Fanconi's anaemia is based on detection of chromosome breakages in cultured blood lymphocytes following exposure to clastogenic agents. Some studies have demonstrated involvement of at least eight genes. Fanconi's anemia patients are prone to develop leukemia and squamous cell carcinoma in the head and neck and anogenital region (3).

The mean age of patients at the time of diagnosis is 8-years with 75% of cases diagnosed between 4 and 14 years of age. However, there are reports of cases diagnosed from birth up to 48 years of age (4). Evolution of the disease is usually fatal within five years of the onset of anaemia and bone marrow transplantation offers the only possibility of a cure (5)

Congenital anomalies and the later haematological disturbances are responsible for the clinical manifestations of Fanconi's anaemia. Patients may present for clinical examination appearing to be in good general health. Considering this possibility, it is very important to know the clinical aspects of Fanconi's anaemia in order to facilitate early diagnosis of the disease, before haematological disturbances develop (4, 6).

A case of early diagnosis of Fanconi's anaemia in an 11-years-old boy was described by Ogilvie et al (5). The patient presented *cafe-au-lait* spots, which are the most common features in patients with Fanconi's anaemia (present in 63-79% of patients (7)

Malignant transformation in the mucosa and mucocutaneous sites (oral and urogenital) have been reported. Since squamous cell carcinomas of these sites is rare in patients of this age, it was concluded Fanconi's anaemia may predisposes to this malignancy (8)

Patients with Fanconi's anaemia may present with elfin faces with delicate features, microg-

nathia, a broad nasal base and epicanthal folds. Skin and mucosas may present with a pale aspect, proportional to the anaemia. Purpuric phenomena may be present, but are not prevalent (6). The aim of this study is to evaluate the oral and dermatologic features in a group of patients with Fanconi's anaemia.

Materials and Methods

Thirty patients with Fanconi's anaemia were examined. All were under treatment at the University Hospital of the Federal University of Paraná, at Curitiba, Brazil. The research was approved by the Ethical Committee of the Pontifical Catholic University of Paraná. Patients who had submitted to bone marrow transplantation were excluded from this study. The selected patients were clinically evaluated and the clinical findings were registered. Eighteen of the patients were females and twelve were males. The mean age was 10 years and 10 months. The focus of the examination was the oral mucosa and the skin.

Results

The dermatological findings were present in 68% of the patients. ($p \leq 0.05$) (*t* test) (skin pigmentation, *cafe-au-lait* spots, petechia and purpura) (Table 1)

Table 1 - skin alterations (color)

Coffee (<i>cafe-au-lait</i>)	18 %
White	2 %
Purple	10 %
Others	32 %
Hair	38 %

Melanine pigmentation, hypochromatic areas, nodules, spots and gingivitis were found in the oral mucosa of all patients (100 %) (Table 2)

Table 2 - oral mucosa alterations

Pigmentation	37 %
Nodules	21 %
Spots	20 %
Gingivitis	18 %
Other	14 %

Discussion

Patients previously submitted to bone marrow transplantation were excluded from the study because the results would be altered, as demonstrated in previous research.(9) Radiotherapy and host versus graft disease increase the risk for squamous cell carcinomas (10,11).

Of the 30 thirty patients, 68 % presented with dermatological signs of the head and face. Petechia and purpura were the prevalent dermatological alterations, resulting from anaemia and medullary deficit. Other studies and literature reviews have supported these findings. Sagaseta et al (4) and Gianpetro (7) found that skin is affected in 60% of cases presenting with *cafe-au-lait* spots or hyperpigmentation, showing a "tan skin" characteristic due to melanin deposition.

However, excessive body and facial hair as well the low head hair implantation are characteristics related to genetic and endocrinal alterations, as shown by Ogilvie (12). Common areas of either skin hyper or hypopigmentation of the skin are equally characteristic of the disease.

There is a significant higher incidence of head and neck carcinomas in patients with Fanconi's anaemia compared with observations of the general population (5,13)

All thirty patients in this study (100%) presented estomatological findings. Hyperpigmentation was the prevalent sign, with higher prevalence in the buccal mucosa and gingiva. Nodules were prevalent in the buccal mucosa. Smalls brown pots in the floor of the mouth and other signs such as purpura, petechial hemorrhage and coating of the tongue were found in 14 % of cases. A swollen gingiva leading to a frequent spontaneous gingival bleeding was an important finding in 18 % of the patients.

Conclusion

General dentists are in a special position to detect Fanconi's anaemia even before haematological disturbances develop. Thus, knowledge of the skin and specially of the oral features of Fanconi's anaemia may facilitate such early diagnosis of the disease.

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