Do you know this syndrome?*
*Você conhece esta síndrome?**

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CASE REPORT

Thirty-year-old white male patient, single, working as a welder, born in and coming from Congonhas, MG, who was referred from the Department of Hematoloy, where findings of macrocytosis and leukopenia were being investigated.

He complained of dark asymptomatic skin spots, with slow progression, and onset 20 years before. He also presented hypoplasia of the first right finger (Figure 1), low stature, left eye cataract (Figure 2), hypoacusis of the right ear and pelvic kidneys. He reported an excision of a skin cancer from the left infra-orbital region. He had a family history of two brothers who had died as a consequence of anemia and pneumonia.

Upon dermatological examination, he presented an intense disseminated hyperpigmentation in the face (Figures 2 and 3), neck and trunk; diffuse hyperpigmentation in the axillae, permeated by hypopigmented spots (Figure 4), besides two café-au-lait spots in the cervical region (Figure 5).

Laboratorial tests revealed both induced and spontaneous chromosome breakage.

The patient is still under medical follow-up in the Department of Hematology and remains stable, with macrocytosis and leukopenia with no clinical consequences.

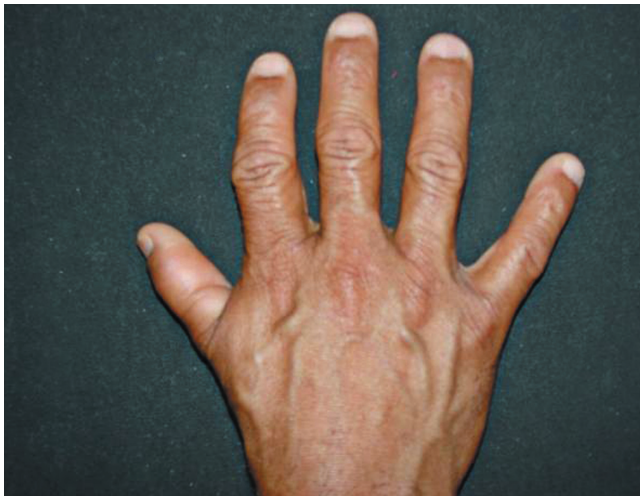

FIGURE 1: Hypoplasia of the right thumb

Reported after written consent of patient/legal guardian

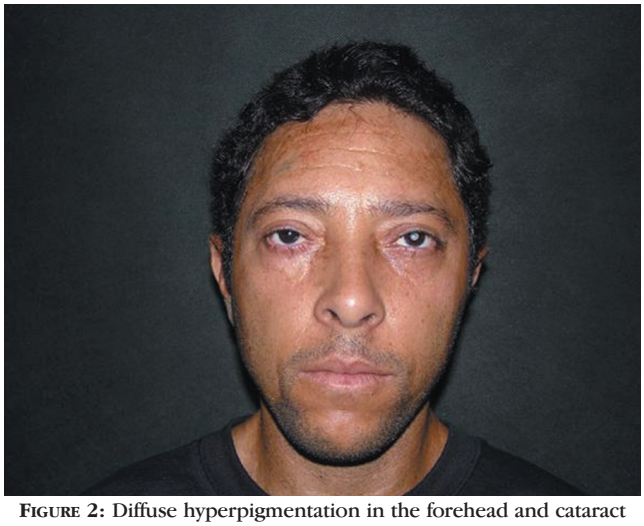

FIGURE 2: Diffuse hyperpigmentation in the forehead and cataract in the left eye

Received on July 28, 2006.
Approved by the Consultive Council and accepted for publication on July 28, 2006.
* Work done at Department of Dermatology at Hospital das Clínicas da Universidade Federal de Minas Gerais (UFMG) – Belo Horizonte (MG), Brazil.
Conflict of interests: None

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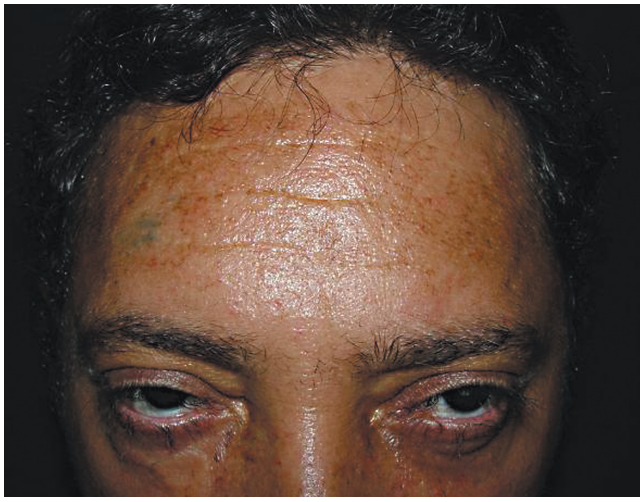

FIGURE 3: Detail of the frontal region

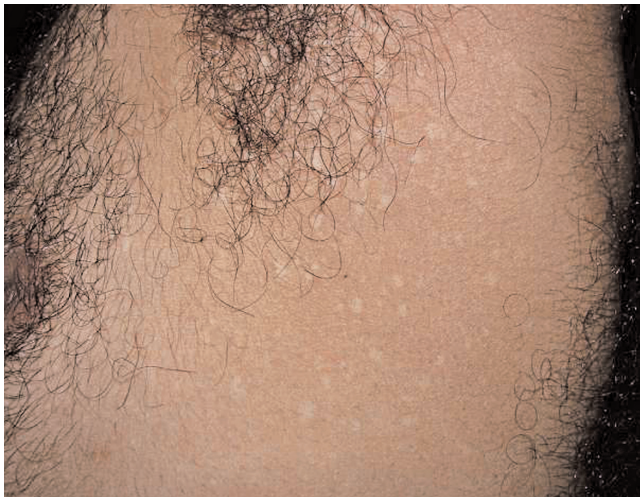

FIGURE 5: Diffuse hyperpigmentation in axillary region, permeated with hypopigmentation stains

WHAT IS THIS SYNDROME?

Fanconi Anemia

Fanconi anemia is a rare syndrome, with recessive autosomic pattern of inheritance, and whose clinical manifestations are due to chromosomic instability.[1,2] It is characterized by congenital anomalies, hematopoietic defects and high risk of developing acute myeloid leukemia and certain solid tumors.[1-5] Cells present an increase in spontaneous chromosomic breakage, as well as induced by agents such as mytomicin C, bussulfan, nitrogen mustard, cisplatin and diepoxibutane.[2,4] The high number of chromosomic breakages constitutes an essential finding for laboratorial diagnosis.[1]

Cutaneous abnormalities occur in up to 80% of the cases, and are characterized by intense and diffuse hyperpigmentation in the face and cervical regions, joints and trunk, besides café-au-lait spots and hypopigmented or achromic spots. Dermatological alterations, present at birth or started at the begging of childhood, may be the only manifestations.[4]

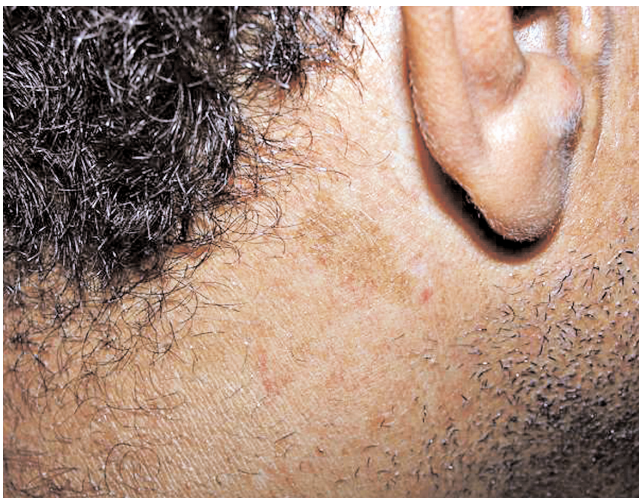

FIGURE 4: Café-au-lait spot in retroauricular region

Hematological alterations generally have their onset before age of 10, and include macrocytosis and bone marrow hypoplasia, which may evolve to aplasia.[1,2,4]

Bone malformations, such as thumb, metacarpi and radius hypoplasia, hip dislocation and scoliosis may also be part of the picture. Approximately 60% of the patients have low stature, and most of them are born pre-term.[4]

Around 28% have renal deformities – aplasia and horseshoe kidneys. Ocular abnormalities are evident in 21% of the patients, including strabismus and microophtalmy. Hypogonadism can occur in up to 20% of the cases.[4]

Central nervous system alterations and anatomical alteration of the ear can be observed in less than one fifth of patients, along with mental retardation, hyperreflexia and hypoacusis.[2,4] Incidence of neoplasias is high among Fanconi anemia patients, particularly that of myeloid leukemia. Course is generally fatal, in an early age, owing to infections, hemorrhages or neoplasias.[1,4,6] Rare are the bearers of Fanconi anemia who reach the age as the patient presented here, who is alive and clinically asymptomatic.

Treatment is based on control of occasional complications. Bone marrow transplantation is a therapeutic possibility for patients who develop aplasia.❑

Abstract: A case of Fanconi anemia is reported, with typical cutaneous manifestations of diffuse hyperpigmentation and café-au-lait spots. He also presented thumb hypoplasia, short stature, cataract, hypoacusis, pelvic kidneys and chromosome breakage. Presently 30-years-old, the patient is stable, with leukopenia and macrocytosis without clinical symptoms, in contrast to usual prognosis of this syndrome, which involves early death due to complications of bone marrow aplasia, leukemia and solid tumors.
Keywords: Adult; Chromosome breakage; Fanconi anemia; Hyperpigmentation; Leukopenia; Macrocytic anemia

Resumo: *Descreve-se caso de anemia de Fanconi com manifestações cutâneas típicas de hiperpigmentação difusa e manchas café-com-leite. Apresentava ainda hipoplasia de polegar, baixa estatura, catarata, hipoacusia, rins pélvicos e quebras cromossômicas. Atualmente com 30 anos, o paciente se mantém estável, com leucopenia e macrocitose sem repercussão clínica, contrariando o prognóstico da síndrome, usualmente letal em idade precoce, por complicações de aplasia de medula, leucemia e tumores sólidos.*
Palavras-chave: Adulto; Anemia de Fanconi; Anemia macrocítica; Hiperpigmentação; Leucopenia; Quebra cromossômica

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