IMAGENS EM HEMATOLOGIA/IMAGES IN HEMATOLOGY

Cytogenetic and Molecular Diagnosis of Fanconi Anemia

Diagnóstico citogenético e molecular da anemia de Fanconi

Carmen S. P. Lima [1]
Gustavo J. Lourenço [1]
Davi E. A. Rodriguez [2]
Maristela Zocca [1]
Carmen S. Bertuzzo [2]

[1] *Haematology and Haemotherapy Centre.*
[2] *Department of Medical Genetics, State University of Campinas, Campinas, São Paulo, Brazil.*

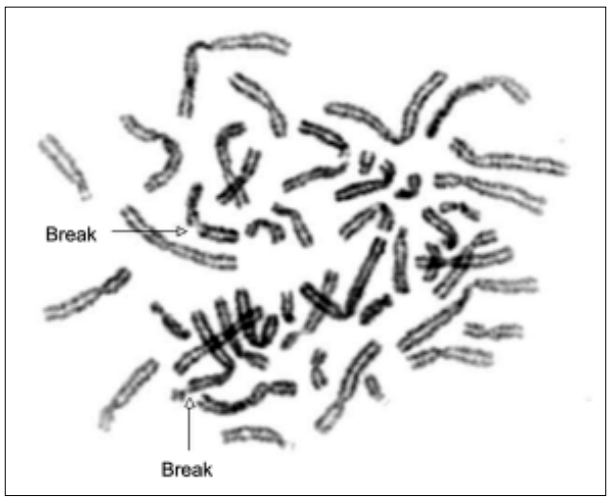

Fig. 1 – *Chromosome spreads showing typical chromosome breaks (arrows) in the Fanconi anaemia patient analysed by the diepoxybutane test*

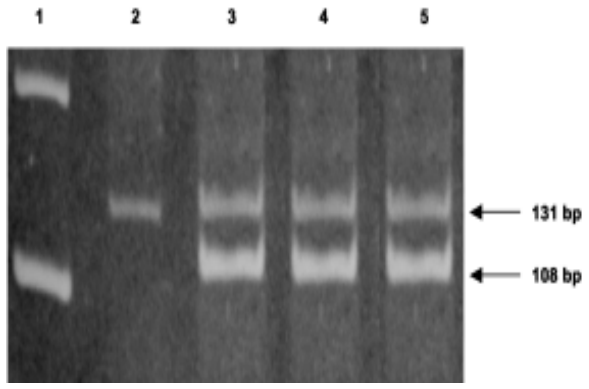

Fig. 2 – *Polymerase chain reaction and restriction endonuclease digestion for detection of IVS4+4A?T mutation of the FANCC gene. Ethidium bromide-stained 8% polyacrylamide gel showing fragments of 108 bp corresponding to the absence of the mutation and fragments of 131 bp corresponding to the presence of the mutation. Lane 1 shows the DNA size markers Ladder 100 bp. Lane 2 shows the result from an individual with homozygous mutation (Fanconi anaemia) and lanes 3, 4 and 5 show the results from heterozygous individuals of the same family*

Fanconi anaemia (FA) is an autosomal recessive disorder associated with a very high frequency of bone marrow failure, developmental abnormalities, such as aplasia of the thumb and radius, growth retardation, hyper-pigmentation, kidney and urinary tract malformations, and high risk of developing a malignant disease, particularly acute myelogenous leukaemia.[1]

Somatic cell fusion studies have shown that FA is genetically heterogeneous, resulting from mutations in at least eight complementary gene groups (FANC A, B, C, D1, D2, E, F, and G).[1]

Lymphocytes culture shows an increased sensitivity to the clastogenic agents diepoxybutane (DEB) or mytomycin (MMC). These agents induce DNA damage, mutations, chromosomal rearrangements and cell death in FA patients.[2] The DEB test is considered as the gold

Correspondence to: *Carmen Silvia Passos Lima, MD, PhD.*
Hemocentro – Unicamp – Cidade Universitária "Zeferino Vaz".
Caixa Postal 6198, Cep: 13083-970 – Campinas-SP – Brazil
Phone: + 55 19 3788-8729 – Fax: + 55 19 3788-8600 – E-mail: carmenl@unicamp.br

standard for diagnosis of disease (reference). However, its effectiveness has been questioned considering that a negative test was found in some cases diagnosed by molecular analysis.[3]

Herein, we present one patient with aplastic anaemia, who was diagnosed as FA by conventional cytogenetic[2] and molecular[4] analyses (Figures 1 and 2). It is important to comment that both analyses permitted proper management of the haematologic disease and genetic counselling for the family.

Bibliographic References

1. Owen J. Fanconi Anemia: Standards for Clinical Care. Fanconi Anemia Research Foundation Incorporation, Oregon, USA. p. 106, 1999.
2. Rosendorf J, Bernstein R, Macdougall L et al: Fanconi anemia: another disease of unusually high prevalence in the African population of South Africa. Am J Med Genet 1987;27:793-797.
3. Pearson T, Jansen S, Havenga C et al: Fanconi anemia: a statistical evaluation of cytogenetic results obtained from South African families. Cancer Genet Cytogenet 2001;126:52-55.
4. Gibson RA, Morgan NV, Goldstein LH et al: Novel mutations and polymorphisms in the Fanconi anemia group C gene. Human Mutat 1996;8:140-148.

Avaliação: Editor e um revisor externo
Conflito de interesse: não declarado

Recebido: 11/07/2003
Aceito após modificações: 15/08/2003