Cyto genetic and Molecular Diagnosis of Fanconi Anemia
Diagnóstico citogenético e molecular da anemia de Fanconi

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 mitomycin (MMC). These agents induce DNA damage, mutations, chromosomal rearrangements and cell death in FA patients.4 The DEB test is considered as the gold standard for the diagnosis of FA, as it can detect the hallmark chromosome breaks in FA cells.1

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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 cytogenetic2 and molecular4 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**