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REVIEW

Fanconi Anemia: a lethal genetic challenge with therapeutic hope

Anemia de Fanconi: un desafío genético mortal con esperanza terapéutica

Ileana Beatriz Quiroga López[1], Dionis Ruiz Reyes[1], Diana García Rodríguez[1], Madyaret Águila Carbelo[1], Adriel Herrero Díaz[1]

[1]Universidad de Ciencias Médicas de Villa Clara. Facultad de Medicina. Hospital Provincial Clínico Quirúrgico "Arnaldo Milián Castro". Villa Clara. Cuba.

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Corresponding author: Ileana Beatriz Quiroga López

ABSTRACT

Introduction: Fanconi anemia (FA) is a rare autosomal recessive inherited disorder characterized by genomic instability, congenital malformations, and progressive bone marrow failure. It presents a high predisposition to hematologic malignancies and solid tumors, making it a significant clinical and social concern.
Aim: to comprehensively characterize the clinical progression, diagnosis, and treatment of Fanconi anemia through an updated literature review.
Method: a narrative review was conducted using 24 documents; including articles, clinical guidelines, and specialized manuals were reviewed, focusing on recent and relevant data on FA's diagnosis, pathophysiology, and treatment.
Results: FA results from mutations in over 22 genes affecting the DNA repair pathway. Diagnosis is confirmed by chromosomal breakage tests using diepoxybutane or mitomycin C. Clinically, it presents with pancytopenia, multiple congenital anomalies, and cancer predisposition. The most effective treatment is hematopoietic stem cell transplantation, although supportive therapies like androgens and growth factors are also used. Lifelong multidisciplinary follow-up is essential to improve life quality and expectancy.
Conclusions: Fanconi anemia is a severe and complex genetic disease. Prognosis has improved significantly due to advances in hematopoietic transplantation. Early diagnosis, interdisciplinary management, and continuous follow-up are key to optimizing clinical outcomes. Establishing regional disease registries is urgently needed to enhance care and research in Latin America.

Keywords: Fanconi Anemia; Bone Marrow Failure; Bone Marrow Transplant; Chromosomal Instability; Medical Genetics.

RESUMEN

Introducción: la anemia de Fanconi (AF) es una enfermedad hereditaria rara, autosómica recesiva, caracterizada por inestabilidad genómica, malformaciones congénitas y fallo progresivo de la médula ósea. Esta condición presenta una alta predisposición a neoplasias hematológicas y sólidos tumores, siendo un problema clínico y social significativo.
Objetivo: caracterizar de forma integral el desarrollo clínico, diagnóstico y terapéutico de la anemia de Fanconi mediante una revisión bibliográfica actualizada.
Método: se realizó una revisión narrativa consultando 24 fuentes; se incluyeron artículos, guías clínicas y manuales especializados, priorizando información actual y relevante sobre diagnóstico, fisiopatología y tratamiento de la AF.
Resultados: la AF es causada por mutaciones en más de 22 genes que comprometen la vía de reparación

del ADN. El diagnóstico definitivo se basa en la prueba de ruptura cromosomal utilizando diepoxibutano o mitomicina C. Clínicamente se manifiesta con pancitopenia, malformaciones múltiples y predisposición al cáncer. El tratamiento más efectivo es el trasplante de células madre hematopoyéticas, aunque se exploran terapias complementarias como andrógenos y factores de crecimiento. El seguimiento multidisciplinario de por vida es fundamental para mejorar la calidad y esperanza de vida.
Conclusiones: la anemia de Fanconi es una enfermedad genética grave y compleja, cuyo pronóstico ha mejorado significativamente gracias a los avances en el trasplante hematopoyético. La detección precoz, el manejo interdisciplinario y el seguimiento continuo son claves para optimizar los resultados clínicos. Urge la implementación de registros regionales para mejorar la atención y la investigación en América Latina.

Palabras clave: Anemia de Fanconi; Fallo Medular; Trasplante de Médula Ósea; Inestabilidad Cromosómica; Genética Médica.

INTRODUCTION

Anaemia is when the number of red blood cells or the concentration of haemoglobin within them is lower than usual. Haemoglobin is necessary to transport oxygen, and if a person has too few red blood cells, if they are abnormal, or if there is not enough haemoglobin, this will decrease the blood's ability to transport oxygen to the body's tissues.[1]

Scientists divide aplastic anaemia into two categories: "acquired" and "hereditary" (genetic) aplastic anaemia. The causes of 'acquired' aplastic anaemia can include exposure to excessive radiation, toxic chemicals, certain drugs, infections, and several environmental agents that damage the bone marrow. In many cases of acquired aplastic anaemia, the specific cause is never discovered. These cases are known as 'idiopathic aplastic anaemia'. Fanconi anaemia is a hereditary anaemia. It is one of the rare genetic conditions that develop into aplastic anaemia.[2]

Fanconi anaemia is a rare inherited disorder encompassing various abnormalities related to the bones, ears, kidneys, and other physical features. Some patients may develop leukaemia or bone marrow failure. In addition, patients with Fanconi anaemia have a very high risk of developing multiple types of cancer throughout their lives.[3] It is characterised by genomic instability and hypersensitivity to DNA cross-linking agents, such as diepoxybutane and mitomycin C (MMC). The abnormal response to these substances is a unique cellular marker and manifests as an increase in the frequency of chromosomal breaks.[4]

Fanconi anaemia was described in 1927 by paediatrician Guido Fanconi,[5] who described three cases of fatal anaemia in three siblings aged between five and seven years. Later, in 1929, Vehliger proposed the name familial hypoplastic panmyelopathy, and in 1931, Nageli gave it its definitive name, Fanconi anaemia. In 1964, Schroeder, Aushnetz, and Knapp described chromosomal abnormalities in the lymphocytes of patients with FA.[6]

Anaemia is a serious public health problem worldwide, particularly affecting young children and pregnant women. The WHO estimates that 42 % of children under the age of 5 and 40 % of pregnant women worldwide are anaemic.[7]

The global incidence of Fanconi anaemia at birth is approximately 3 per million births. Still, this figure varies because the distribution of the mutated genes differs depending on the population being studied. Fanconi anaemia can reach a frequency of up to 1 in 181 in some ethnic groups.[8] Children in southern Africa and Ashkenazi Jews seem to be particularly affected.[3]

The carrier frequency is 1/300 in Europe and the United States. Diagnosis occurs between the ages of 4 and 14 in 75 % of cases, with the average age of eight.[5]

Standardised collection of information through registries of patients with rare diseases is essential to improve patient diagnosis, treatment, and follow-up, as well as for research purposes. Several registries worldwide have proven their importance in producing knowledge about Fanconi anaemia and supporting patients and their families. Unfortunately, there are none in the Americas, which limits the adequate diagnosis, management, and research of Fanconi anaemia.[9]

In Cuba, a study of chromosome breaks was conducted in 32 patients. In six (20 %) of the cultures with results, the MMC assay at 300 nM showed a higher number of chromosomal breaks than the controls, allowing them to be diagnosed as FA. Of these six, only two had various dysmorphic features typical of the condition, and the rest had only aplastic anaemia.[4]

Survival rates vary from person to person. The prognosis is poor in people with low blood counts. Survival has probably improved with new and better treatments, such as bone marrow transplantation.[10]

These types of diseases are currently a significant clinical and socio-health problem. Early diagnosis will allow for reasonable control of the haematological involvement and enable the necessary surgical treatments to be carried out before the onset of thrombocytopenia. It will also be possible to identify affected siblings

before the onset of symptoms or, during pregnancy, to determine whether the foetus could be a potential haematopoietic progenitor donor for an affected sibling.[5] It is a disease in which the manifestations vary significantly from one person to another.[11]

The research team was motivated to write this article due to the importance of knowledge about anaemia, especially Fanconi anaemia, one of several fatal hereditary anaemias.

Objective: To characterise the development of Fanconi anaemia.

DEVELOPMENT

Haematological abnormalities are triggered by progressive bone marrow failure, as the bone marrow is responsible for the production of all blood lines or haematopoiesis. In the bone marrow, we find STEM CELLS or HCT (haematopoietic stem cells), which can self-replicate and differentiate; these are stem cells. CTH gives rise to a multipotent progenitor (UFC-LM), which differentiates into a common lymphoid progenitor (UFC-L) or a common myeloid progenitor (UFC-M), from which the different differentiated blood lines will emerge. Usually, pancytopenia, which is the simultaneous deficiency of all three blood cell lines (red blood cells, white blood cells, and platelets), will already be present in the first decade.[12,13]

No one has yet been able to explain why patients with FA develop bone marrow failure. This can only be understood once the FA genes have been isolated and studied. However, scientific studies show that almost all patients with FA will eventually suffer from bone marrow failure.[2]

Fanconi anaemia (FA) is an autosomal recessive syndrome characterised by chromosomal fragility leading to congenital malformations in various organs, progressive bone marrow failure, and a tendency to malignant diseases.[14,15]

For an individual to have the disease, both parents must be carriers, and the individual must receive the defective AF gene from both parents. If the individual receives only one non-functional gene, they will only be a carrier. The main characteristic of AF cells is chromosomal instability and sensitivity to agents that induce cross-links in DNA, such as mitomycin C or diepoxybutane. Up to 22 genes involved in the disease have been described, corresponding to different complementation groups. Each complementation group has a distinct phenotype and, in some cases, the nature and severity of the pathology can be determined based on which group it belongs to. For example, the FANCA, FANCC, and FANCG groups account for 85 % of FA cases. There is evidence that all proteins encoded by FANC genes act on a shared pathway responsible for DNA damage recognition and repair, so that when one of the links fails, chromosomal damage develops and the corresponding cell failure is triggered.[16]

People with Fanconi anaemia have lower than normal levels of white blood cells, red blood cells, and platelets (cells that help blood to clot). The normal red blood cell count is 4,5 to 6 million per cubic millimetre for men and 4 to 5,5 million per cubic millimetre for women. The total white blood cell count (neutrophils, eosinophils, basophils, lymphocytes, and monocytes) is 5 000 to 10 000 per cubic millimetre of blood. Normal platelet counts in blood are 150 000 to 450 000 per cubic millimetre of blood. The absence of red blood cells causes anaemia and fatigue, while a decrease in white blood cells makes infections more likely. A lack of platelets causes bleeding.[17]

Early diagnosis of AF allows other diseases to be ruled out and prevents inappropriate management of haematological diseases (aplastic anaemia [AA], myelodysplastic syndrome [MDS], acute myeloid leukaemia [AML]), stem cell transplantation, androgens, haematopoietic growth factors, or supportive care. Furthermore, knowing the diagnosis of AF optimises surgical intervention for orthopaedic, renal, or other abnormalities. Genetic counselling is essential, given the 25 % risk of AF in each subsequent pregnancy. Opportunities for family planning, prenatal diagnosis, and even pre-implantation genetic diagnosis should be available.[18]

The clinical picture is usually asymptomatic during early childhood, four to seven years in boys and six to ten years in girls. Symptoms progress gradually and are due to the development of progressive pancytopenia. Initially, clinical manifestations attributable to thrombocytopenia appear: petechiae (small spots on the skin, formed by the escape of blood), haematomas (cystic blood collection), severe episodes of epistaxis (nosebleeds) and gastrointestinal bleeding; later, signs of anaemia become evident: pallor, easy fatigue, weakness and hyporexia (decreased appetite).[19]

A paediatric patient, in most cases, is between 13 and 14 years old at the time of diagnosis. Subsequently, findings that are characteristic of the disease are found, such as Fanconi facies, which consists of small eyes, microcephaly, and abnormal position, size, and shape of epicanthic folds and auricles—dermatological abnormalities such as hyperpigmentation in areas of folds, neck, and trunk. There may also be coffee-coloured macules—endocrine abnormalities, such as short stature due to impaired growth hormone secretion or hypothyroidism. Musculoskeletal abnormalities include absence of the radius, thumb abnormalities (absence, hypoplasia, bifid, supernumerary), congenital hip dislocation, and abnormalities of the feet and legs. Genitourinary disorders such as vaginal, uterine and ovarian disorders in women; in men, there may be testicular atrophy, cryptorchidism, hypospadias and phimosis. Renal disorders such as renal ectopia, renal

hypoplasia, and renal agenesis. Some other abnormalities to look for are cardiac and gastrointestinal, as well as mental retardation, which can occur in up to 10 % of patients.[20,21]

The primary method for confirming the diagnosis of Fanconi anaemia is the chromosomal breakage test, which the Fanconi Anaemia Guidelines recommend performing in patients with congenital abnormalities accompanied by findings suggestive of this disease. This test is based on the cellular hypersensitivity of affected individuals to DNA cross-linking agents, such as mitomycin C and diepoxybutane, to which lymphocytes or fibroblasts from patients are exposed to identify chromosomal breaks and radial figures, which are highly characteristic of the disease. Techniques without alkylating agents have proven insensitive and non-specific, so it is advisable to perform the procedures with mitomycin C and diepoxybutane, the latter being the most specific. Due to the strong genetic component of Fanconi anaemia, molecular diagnostic strategies, such as gene panels or sequencing, have been proposed to enable rapid, accurate, and cost-effective diagnosis in routine clinical practice. The determination of pathogenic family variants is necessary to perform prenatal or pre-implantation molecular testing, as well as for carrier detection.[8]

The research team believes that cytometry can be used to detect bone marrow failure in the first instance, in findings such as thrombocytopenia and macrocytosis, until anaemia is finally found in advanced stages. However, for a definitive diagnosis, a study called chromosome breakage is performed on lymphocytes, which can be done with substances such as diepoxybutane (DEB) or mitomycin C (MMC).

The treatment of Fanconi anaemia seeks to prolong life expectancy and improve patients' conditions as much as possible. It therefore targets the three fundamental aspects of the disease: physical abnormalities, bone marrow failure, and malignant tumours. From the onset of the disease, patients should undergo frequent check-ups of their ears, urinary tract, psychomotor development, and eyes so that any abnormalities can be detected as soon as possible and the specialist can offer the most appropriate treatment option.[14]

The first-line treatment for the bone marrow failure that these patients develop is autologous haematopoietic stem cell or umbilical cord stem cell transplantation, with or without cytokines, androgens, and transfusion regimens. Transplantation is the only treatment capable of correcting the haematological defect, which determines success in more than 85 % of patients under 40. Furthermore, it eliminates the risk of developing myelodysplastic syndrome or leukaemia. Overall survival when the transplant is from an unrelated donor is 33 %, and there is an increased risk of graft-versus-host disease (GVHD), so the use of umbilical cord blood stem cells is recommended.[22]

The best results have been obtained in siblings with identical HLA, but this is not always possible. The main problem is that patients with Fanconi anaemia are susceptible to radiation and the chemotherapy required before transplantation due to their propensity for chromosome breakage, which initially posed a serious problem. Excellent results have been obtained with the chemotherapy drug fludarabine, which avoids the need for preoperative radiation. Androgens are used to correct haematological abnormalities caused by bone marrow failure, such as the depletion of red blood cells and platelets, and are effective in 50 % of individuals. However, when treatment is continued, the response diminishes. The androgen used would be oxybutazone. Cytokines such as granulocyte colony-stimulating factor (G-CSF), Neupogen®, or granulocyte-monocyte colony-stimulating factor (GM-CSF), Molgramostin®, can also be used to treat haematological problems.[23,24]

The research team considers stem cell transplantation to be the best treatment option for patients with this disease, but their life expectancy is short, so efforts to increase the effectiveness of this treatment and innovate in others should continue.

After transplantation, the physician who performed the procedure should decide how often bone marrow (BM) tests should be performed. In general, several BM aspirations and biopsies are performed during the first year and then at two years. Subsequent BMSCs are warranted if the patient has mixed chimerism, continues to depend on transfusions, or if there are concerns about low peripheral blood counts. A total body iron assessment should be performed one year after transplantation, as most patients received significant numbers of red blood cell transfusions. Repeated verification of serum ferritin levels may help monitor a trend, but ferritin is an inadequate measure of iron load. Liver biopsies or new non-invasive magnetic resonance imaging measurements offer more sensitive and specific results. Depending on the result, phlebotomy or iron chelation may be needed every month.

Endocrine issues are common in patients with AF and require lifelong endocrine evaluation and follow-up. Additional endocrinopathies, such as hypothyroidism, growth hormone deficiency, gonadal dysfunction, osteoporosis, and infertility, may arise after HSCT. After transplantation, all patients should undergo endocrine evaluation and continue to undergo these evaluations throughout their lives. Pay special attention to age, pubertal stage, and growth, as these are essential to determine the timing and extent of the individual's endocrine evaluation. HSCT can cause osteopenia, osteoporosis, and avascular necrosis of the bones, each of which can be accelerated by cumulative doses of glucocorticoids. A dual-energy X-ray absorptiometry (DXA) test should be performed one year after transplantation. No comparative normal values are available for children <5 years of age, but DXA testing can still be used to look for changes over time in these individuals. If the initial

DXA test is abnormal, the decision about treatment (vitamin D, calcium, bisphosphonates, or other agents) and when to perform follow-up DXA tests should be made in consultation with the patient's endocrinologist.

Growth and development should be assessed at least once a year. A formal neuropsychological evaluation should be performed in high-risk patients, especially those who received a transplant before the age of three. Early intervention is essential to help identify problems and optimise the patient's development. Although most AF patients are young and relatively selective in what they eat, endocrine and nutritional assessments should be performed to identify possible aetiologies if their growth is below optimal. Growth hormone therapy may be necessary for some patients. Metabolic syndrome is a constellation of central obesity, insulin resistance, glucose intolerance, dyslipidaemia, and hypertension, and is associated with an increased risk of type 2 diabetes mellitus and atherosclerotic cardiovascular disease. BMI survivors have an age- and bone mass-adjusted risk of diabetes and hypertension, which may place them at high risk for cardiovascular events as they grow older. Although there is no information to determine the exact risk of metabolic syndrome in patients with AF, this risk may be significant in those patients with AF who are particularly prone to diabetes. Therefore, all patients with AF should be monitored for early signs of metabolic syndrome and encouraged to follow a healthy diet and exercise regimen.[18]

The research team believes that hematopoietic stem cell or bone marrow transplantation is currently the only curative treatment for haematological complications resulting from bone marrow failure. However, it does not cure non-haematopoietic manifestations.

The need for close lifelong monitoring to detect the early onset of cancer has a considerable impact, making psychological support vital. When a bone marrow transplant is required, isolation for several weeks or even months after the transplant is a delicate situation that requires constant support. Sometimes, the stress and fatigue caused by the transplant are difficult for the patient and their family to cope with. The side effects of the transplant may compound these difficulties. Furthermore, avoiding post-transplant infections while waiting to develop functional immunity requires strict hygiene rules: long periods of absence from school or work, no use of public transport, no visiting public places, etc. It is sometimes difficult to establish social relationships, especially for young people, who may isolate themselves and remain on the sidelines. Support from the environment (family and carers) is essential to relieve the affected person and support them in the best possible way.[11]

The research team believes that early diagnosis in patients with FA (i.e., before the onset of haematological abnormalities) could provide more time to find an HLA-compatible donor suitable for bone marrow transplantation.

CONCLUSIONS

Fanconi anaemia (FA) is an autosomal recessive syndrome characterised by chromosomal fragility. For an individual to have the disease, both parents must be carriers, and the individual must receive the defective FA gene from both parents.

The chromosomal breakage test is the main method for confirming the diagnosis of Fanconi anaemia. Transplantation is the only treatment capable of correcting the haematological defect. Advances in the care of patients with FA have resulted in an increasing number of patients surviving for many years after transplantation.

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CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

AUTHORSHIP CONTRIBUTION

Conceptualization: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Data curation: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Formal analysis: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Research: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Methodology: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Project administration: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Resources: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Software: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Supervision: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Validation: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Visualization: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Writing - original draft: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.

Writing - revision and editing: Ileana Beatriz Quiroga López, Dionis Ruiz Reyes, Diana García Rodríguez, Madyaret Águila Carbelo, Adriel Herrero Díaz.