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Clinical and socio-economic comments on Faconi's anemia case: Venezuela

Abstract

This document deals with the clinical and socio-economic implications of Fanconi Anemia (AF). This pathology has been well documented since the Swiss pediatrician Guido Fanconi first detected it in 1927. The methodology used was the exploratory documentary review of a theoretical-explanatory nature, since the author made an analysis of scientific texts and/or publications most relevant on the subject. He also gives his opinions about this disease and its importance in the Venezuelan economic and political context.

Keywords: fanconi anemia, medullar aplasia, congenital, Venezuela

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Development

AF is a congenital pathology of recessive nature, this means that the disease has its origin during intrauterine development, whose carrier genes come from two abnormal copies (maternal and paternal). In this sense, it can be said that AF is a complex chromosomopathy from the genetic and symptomatological point of view, because despite the fact that the child is born with the disease, its manifestations vary from one patient to another, being the school age among the 7 and 10 years old, when the first clinical findings begin to appear.[1]

Among the signs and symptoms we can mention: fevers without apparent cause above 38°C, bruises on different parts of the body, brown skin spots, lack of appetite, jaundice (yellowish color on the skin), anemia (low concentration of hemoglobin) thrombocytopenia (progressive decrease of platelets), leukopenia (decrease in white blood cells) and in chronic cases a generalized hematological picture can appear, confirmed by the three previous symptoms called pancytopenia.[2]

The genes related to AF are: FANCA, FANCB, FANCC, FANCD1/BRCA2, FANCD2, FANCE, FANCF, FANCG/XRCC9, FANCI, FANCJ/BRIP1/BACH1, FANCL, FANCM, FANCN/PALB2, FANCO/RAD51C, FANCP/SLX4 / BTBD12, FANCQ/RCC4/XPF, FANCR/RAD51, FANCS/BRCA1, FANCT/UBE2T, FAN-CU/XRCC2, FANCV/REV7, FANCW/RFWD3. As can be noted, there are more than twenty genes related to the pathology, which are well identified. This provides valuable information for the early diagnosis of FA. However, the socio-economic context and the experience of health professionals is crucial to diagnose this disease in a safe and timely manner.[3]

In the Venezuelan case, it is usually pediatric hematologists and in some cases pediatric oncologists who have the greatest responsibility in their hands, since they are the specialists par excellence in the treatment of FA. Although there are protocols of standardized laboratories for the hematological and genetic diagnosis of FA such as genetic sequencing techniques (Sanger or NGS) and MLPA, public and private hospitals in the country face a serious health crisis, the consequence of which has been the migration of these professionals to other countries in Latin America, the United States and Europe, leaving our specialized health centers abandoned and destitute ok these doctors.[4]

For the diagnosis of FA, doctors in most cases must send blood samples from suspected patients abroad so that they can be analyzed. Italy is the reference country for the hematological and cytogenetic study of FA. In Venezuela there are some international NGOs and foundations that collaborate with this noble work, they have even managed to perform bone marrow transplants successfully in Milan-Italy. In this case, a histocompatible donor is necessary; generally a brother of the patient is the donor in most cases, since the risk of postoperative immunological rejection is reduced. However, a good technological platform with the support of doctors specialized in the area, would be the way for a more effective treatment of AF in Venezuela. We know that the crisis has almost completely wiped out the health system and the policies of the current regime do not worry about the installation of a humanitarian system, especially for the most vulnerable families.[5]

Also, the administrative bureaucracy for the granting of passports and visas is increasingly precarious, and this makes it difficult for patients to be transferred abroad to be attended to in a timely manner. I believe that it is necessary and urgent to establish humanitarian support networks with a conscience framed in the needs of patients and their families, because the situation in the country creates a psychological stressful situation for the patient, and as we know, it is proven that the survival of the affected person depends partly on the applied treatment and the state of mind. Stress proteins play a crucial role in the general deterioration of health and are important bio-indicators of the psychological picture of an individual.[6] For this reason, changes in the socio-political environment affect the family economy, decreasing the purchasing power to access medicines and other vital fluids such as plasma, blood, among others, used in the treatment of FA.[7] It is important to call for the inalienable and universal rights that an individual with any underlying congenital pathology possesses. The AF progressively deteriorates the health status of those affected and puts their potential risk of death. When the pathology enters its chronic phase the hematological values become unbalanced, causing an absence of important oxygen in the brain and generalized weakening. Even in the scientific literature, deaths of infants of school age have been reported. The severity of the AF is variant and the medullary aplasia causes in some cases self-destruction (apoptosis) of the hematopoietic stem cells and elevation of malignant and benign tumor promoter proteins, this must call for reflection to offer a better quality of life and hopes of survival through early diagnosis and bone

marrow transplantation in the short or medium term.[8] The task is not easy but not impossible, with assertive policies we can achieve agreements with foreign laboratories and prepare health professionals in the application of specific cytogenetic tests for AF in Venezuela and in this way, create the atmosphere conducive to save more lives and decrease deaths in vulnerable patients. One option may be to award fellowships to Venezuelan doctors, hematologists and oncologists, to study in Europe and the United States. To conclude, I believe that Venezuela is far from creating the necessary conditions for the care of these patients. However, everything is possible with the joint effort of the living forces of the communities and the creation of effective programs for the pre and post-operative monitoring of patients with FA, and of course, the habilitation and recovery of the existing hospital centers.

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Conflicts of interest

The author declares that no conflicts of interest exist in publishing this article.

References

1. Fanconi Anemia Foundation. *Help guide for the clinical diagnosis of hereditary diseases*. 2014. p. 1–5.
2. Sagaseta M, Molina J, Lezáun I, et al. Fanconi anemia. Current considerations. *ANAL Sis San Navarra*. 2003;26(1):63–78.
3. Lynn and Dave Frohnmayer. Fanconi anemia: A Handbook for Families and their Doctors. 3rd edition. 2000. p. 1–38.
4. Jesuit Refugee Service. *Report on Venezuelan mobility.* 2018. p. 4–16.
5. International Organization for Migration. Module II. *Human Mobility. Comprehensive border management in the Andean sub region.* Venezuela case: Perfil Migratorio del Ecuador publishers; 2012. 65 p.
6. MacMillan ML, Hughes MR, Agarwal S, et al. Cellular therapy for Fanconi Anemia: the past and future. *Biol blood Marrow Transplant.* 2011;17(1 suppl):S109–S114.
7. Bogliolo M. *Exome sequencing in Fanconi anemia: from diagnosis to the discovery of a new gene.* 2016. Spain: Autonomous University of Barcelona; 2015. p. 17–22.
8. Machín García S, Svarch E, Dorticós Balea E, et al. Medullary Aplasia. *Upgrade. Institute of Hematology and Immunology.* 1999;15(2):79–90.

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