Videos en español

La ANEMIA de FANCONI

~ Divulgative Project ~

English videos

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UAB

Fanconi Anemia RESEARCH FUND, INC.

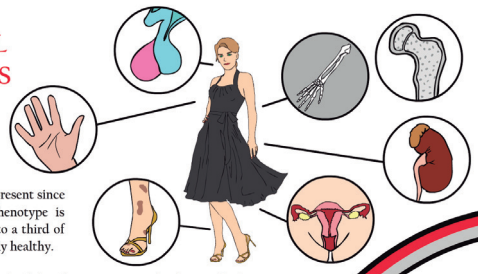
Divulgative interest

It is essential to make people aware of rare diseases, as its rareness should not be a reason to let it be unknown, but even more of a reason to spread the word of its nature. Particularly, Fanconi Anemia has had huge implications on many scientific fields, especially those concerning cancer. Therefore, Fanconi Anemia should be leading the way as a representative of this marginal group of diseases.

Objectives

This project is meant for arising Fanconi Anemia awareness to general population, as a representative of rare diseases. Its intention is to explain the disease by developing several videos that will be accessible to everyone. These videos will show animated drawings that wil complement the explanation given in both Spanish and English for it to reach to a greater extent of people. Additionally, a collaboration with a Fanoni Anemia foundation is meant to take place.

CLINICAL FEATURES

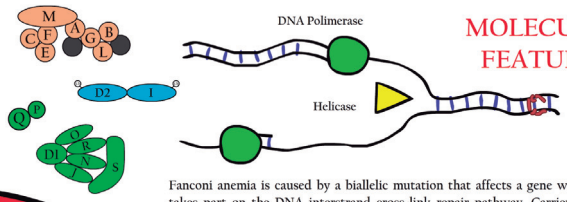


Fanconi Anemia is a genetic disease that is often diagnosed at the age of 5-15 years, even though it is present since birth. However, the phenotype is very heterogeneous: up to a third of the patients are physically healthy.

This disease is characterized by three aspects: aplastic anemia (a reduction in quantity of blood cells), acute myeloid leukemia (blood cancer derived from myeloblasts), and solid tumors (mainly those that affect the oral cavity).

Other traits of these patients include skeletal anomalies involving both the hand thumbs and the forearm, several endocrinopathies, café-au-lait skin spots, kidney anomalies, hypogonadism, and osteoporosis.

MOLECULAR FEATURES



Fanconi anemia is caused by a biallelic mutation that affects a gene whose product takes part on the DNA interstrand cross-link repair pathway. Carriers of a single mutation may also have consequences, such as FANCS (BRCA1) or FANCD1 (BRCA2) carriers, who have a higher risk of developing breast and/or ovarian cancer.

Eighteen Fanconi genes have been identified and classified by function in: the core complex, whose function is to detect the damage, the ID2 complex, whose function is to recruit the downstream effectors, and the downstream effectors themselves that work directly by repairing DNA.

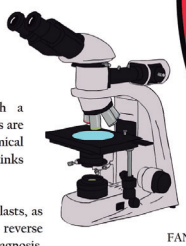
Therefore, when affected cells (mainly hematopoietic cells) face interstrand cross-links, DNA damage cannot be repaired, and they undergo apoptosis. However, some cells may evade the apoptosis pathway, accumulating mutations that will later lead into a cancer.

INTRODUCTION

Fanconi Anemia is a rare disease that affects only one per each half a million people.

It was first described by Guido Fanconi in 1927 and, since then, only 2.000 patients have been reported.

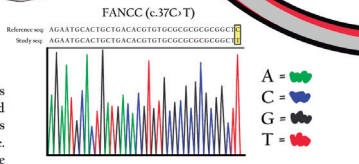
GENETIC DIAGNOSIS



Fanconi Anemia is diagnosed with a chromosome breakage assay: blood cells are tested for their hypersensitivity to chemical agents that cause interstrand cross-links (Diepoxybutane or Mitomycin C).

However, it is preferable tu use fibroblasts, as they cannot undergo a process called reverse mosaicism that makes difficult the diagnosis. Prenatal diagnosis is possible in this disease as this test can also be done on fetal cells.

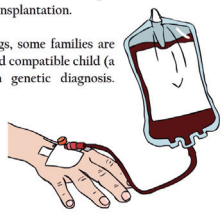
Additionally, a retroviral complementation test is useful when it comes to identifying the affected gene: cells are infected in vitro with a retrovirus containing a healthy sequence of a Fanconi gene. Only those cells infected with the healthy sequence that matches that of the affected gene can survive, uncovering this way the affected gene.



Finally, sequencing technologies allow for the identification of both the gene and the source mutation.

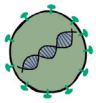
THERAPY

Fanconi Anemia is kept at bay with periodic blood transfusions and androgen administration. However, the only current cure that treats the hematopoietic problems of Fanconi Anemia patients is a hematopoietic stem cell transplantation.



As donors should preferably be siblings, some families are attempting to give birth to a healthy and compatible child (a saviour sibling) by pre-implantation genetic diagnosis. However, the success rate is very low.

In addition, hematopoietic stem cell transplantation cannot cure non hematopoietic difficulties such as solid tumors. Therefore, a follow-up of patients is required (bone marrow aspirations, and oral cavity and gynecologic examinations). Administration of the human papillomavirus vaccine is recommended.



Concerning the future, gene therapies are on the way, including both the replacement of the affected gene by a healthy gene and/or the use of designed nucleases.

Drawing and animation

Drawings and animations were done by using Adobe Flash Professional CC. This is the last installment of the Adobe Flash series, a multimedia program that is used for web designing and game development, among others.

Free website Apowersoft was required for video conversion.

Bibliography

1) A. R. Smith and J. E. Wagner (2012)
2) S. Longerich (2014)

Acknowledgments

Jordi Lesan - Jordi Surrallés - Elsa Velasco

Voice recording

Voice was recorded using Cool Edit Pro 2.0, a sound recording and editing application. Nowadays it is known as Adobe Audition, as Adobe systems acquired it in 2003.

In addition, a volunteer composed the soundtrack using the MuseScore application.