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FANCONI ANAEMIA: A REVIEW

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Abstract

Fanconi anaemia is a rare genetic disorder which leads to bone marrow failure and results in decreased production of all types of blood cells. The frequency of occurrence is greater in South-African Africaners,sub-saharan blacks, and spanish gypsies than in the overall world population. This rare genetic disease occurs when two people with the recessive gene have children. There is no cure available for falconi anaemia,the treatment for falconi anaemia is ***symptomatic.***

Keywords: *fanconi anemia,rare genetic disease,growth factors*

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INTRODUCTION:

Fanconi anaemia is a rare genetic disease .FA is the result of a genetic defect in a cluster of proteins responsible for DNA repair.FA is primarily an autosomal recessive genetic disorder. This means that two mutated alleles(one from each parent) are required to cause the disease. There is a 25% risk that each subsequent child will have FA. About 2% of FA cases are X-linked recessive, which means that if the mother carries one mutated Fanconi anemia allele on one X chromosome, there is a 50% chance that male offspring will present with Fanconi anemia. Scientists have identified 17 FA or FA-like genes: FANCA, FANCB, FANCC, FANCD1(BRCA2), FANCD2, FANCE, FANCF, FANCG, FANCI, FANCJ (BRIP1), FANCL, FANCM, FANCN (PALB2), FANCP (SLX4), FANCS (BRCA1), RAD51C, and XPF. FANCB is the one exception to FA being autosomal recessive, as this gene is on the X chromosome. Genetic counseling and genetic testing is recommended for families that may be carriers of Fanconi anemia.

Because of the failure of hematologic components to develop — white blood cells, red blood cells and platelets — the body's capabilities to fight infection, deliver oxygen, and form clots are all diminished.

As a result, the majority of FA patients develop cancer, most often acute myelogenous leukemia, and 90% develop bone marrow failure (the inability to produce blood cells) by age 40. About 60–75% of FA patients have congenital defects, commonly short stature, abnormalities of the skin, arms, head, eyes, kidneys, and ears, and developmental disabilities. Around 75% of FA patients have some form of endocrine problem, with varying degrees of severity. Median age of death was 30 years in 2000

EPIDEMIOLOGY:

The frequency of occurrence is greater in South-African Africaners, sub-saharan blacks, and spanish gypsies than in the overall world population, leading to expected birthrates in these subpopulations of around 1 cases per 40,000 births. Among Ashkenazi Jews in the United States, the carrier frequency is approximately 1 case per 90 people, with a projected birthrate of 1 case per 30,000 people.

The male-to-female ratio in the literature cases is 1.2:1, although equal numbers are expected in a disorder with over 99% autosomal recessive inheritance. Fanconi anemia has been diagnosed in patients from birth to age 49 years, with a median age of 7 years. Individuals with birth defects are diagnosed at younger ages than are persons without birth defects.

CAUSES:

This rare genetic disease occurs when two people with the recessive gene have children, means a person must get one copy of the abnormal gene from each parent.

Fanconi anemia is due to an abnormal gene that damages cells, which keeps them from repairing damaged DNA.

SYMPTOMS:

- People with Fanconi anemia have lower-than-normal numbers of white blood cells, red blood cells, and platelets (cells that help the blood clot).
- Not enough white blood cells can lead to infections. A lack of red blood cells may result in fatigue(anaemia).
- A lower-than-normal amount of platelets may lead to excess bleeding.
- Abnormal heart, lungs, and digestive tract
- Bone problems (especially the hips, spine or ribs, can cause a curved spine(scoliosis)
- Changes in the color of the skin, such as darkened areas of the skin, called café au lait spots, and vitiligo.
- Deafness due to abnormal ears
- Eye or eyelid problems
- Kidney(s) that did not form correctly
- Problems with the arms and hands, such as missing, extra or misshapen thumbs, problems of the hands and the bone in the lower arm, and small or missing bone in the forearm
- Short height
- Small head
- Small testicles and genital changes

Other possible symptoms:

- Failure to thrive
- Learning disability
- Low birth weight
- Intellectual disability

DIAGNOSIS:

- Pregnant women may have amniocentesis or chorionic villous sampling to diagnose the condition in their unborn child.
- Bone marrow biopsy
- Complete blood count (CBC)
- Developmental tests
- Drugs added to a blood sample to check for damage to chromosomes
- Hand X-ray and other imaging studies(CT scan ,MRI)

- Hearing test
- HLA tissue typing(to find matching bone-marrow donors)
- Ultrasound of the kidneys

TREATMENT:

There is no cure available for falconi anaemia,the treatment for falconi anaemia is **symptomatic**.

Medicines called growth factors (such as erythropoietin, G-CSF, and GM-CSF) can improve blood counts for a short while. A bone marrow transplant can cure the blood count problems of Fanconi anemia. (The best bone marrow donor is a brother or sister whose tissue type matches the person affected by Fanconi anemia). People who have had a successful bone marrow transplant still need regular check-ups because of the risk for additional cancers.

Hormone therapy combined with low doses of steroids (such as hydrocortisone or prednisone) is prescribed to those who do not have a bone marrow donor. Most people respond to hormone therapy. But everyone with the disorder will quickly get worse when the drugs are stopped. In most cases, these drugs eventually stop working. Additional treatments may include:

- Antibiotics (possibly given through a vein) to treat infections
- Blood transfusions to treat symptoms due to low blood counts

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