Qure
Center for Complete Care
Blood Diseases. Cancer. Stem Cell Transplantation

Fanconi Anemia

What is Fanconi anemia?

Fanconi anemia (FA) is a rare and serious inherited blood disorder in which the bone marrow fails to make normal blood cells. This failure (which is an inherited aplastic anemia) means that the bone marrow does not make enough of any or all of three types of blood cells—red blood cells (to carry oxygen), white blood cells (to fight infection) and platelets (to help blood clot). FA affects children and is rare, affecting one in 100,000 births.

What are the signs and symptoms of FA?

FA is usually diagnosed before age 12 with symptoms due to anemia or bleeding. About two-third of children with FA have other physical defects such as short stature, small head, misshapen, missing or extra thumbs or fingers, curved spine, ear and eye defects, kidney or testes problems, skin patches, stomach and heart defects. FA can also cause the bone marrow to make faulty blood cells which leads to acute myeloid leukemia (AML), a blood cancer.

How is FA treated?

FA is a genetic disorder with no cure except for bone marrow transplantation (BMT). Treatment is based on how low or abnormal the blood counts are and the age of the patient.

Short-term—to monitor the disease and control symptoms.

- Regular blood-count checks
- Antibiotic treatment as needed (to fight infections)
- Blood transfusion as needed (to increase blood cell count)

Long-term—to improve quality of life and extend lifespan.

- Androgen therapy. This uses man-made male hormones (danazol) to help the body make more red blood cells and platelets for long periods. This therapy does not work as well for increasing the white blood cell count.
- Blood and bone marrow stem cell transplant (BMT). In BMT, blood stems cells are taken from a healthy donor (usually a family member) to replace the abnormal stem cells. The procedure is complicated and expensive and should be done by someone with experience in treating Fanconi Anemia. Stem cell transplant is the current standard treatment for Fanconi anemia that is causing major bone marrow failure.

What are the survival rates for FA?

The average lifespan for people with FA is around 20 years but there is much variation with some even living as long as 30-40 years, while others die before the age of 10 years. Bone marrow failure with anemia, infections or bleeding , leukemia and cancerous tumors are the most common causes of death related to FA. About 10 percent of people with FA develop AML. BMT offers the best chance of a cure, but carries its own risks (patients are still at risk for head and neck cancers), so discuss all treatment options carefully with your doctor.