

# Fanconi Anaemia (FA) Fact Sheet

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Fanconi Anaemia (FA), named after the Swiss paediatrician, Guido Fanconi, is one of the inherited anaemias that lead to bone marrow failure (aplastic anaemia). It is a recessive disorder: if both parents carry a defect (mutation) in the same FA gene, each of their children has a 25% chance of inheriting the defective gene from both parents. When this happens, the child will have FA.

Scientists have now discovered 15 FA or FA-like genes [A, B, C, D1 (BRCA2), D2, E, F, G, I, J, L, M, N, P and RAD51C]. These genes account for more than 95% of the cases of Fanconi Anaemia. Mutations in FA-A, FA-C and FA-G are the most common and account for approximately 85% of the FA patients worldwide. FA-D1, FA-D2, FA-E, FA-F and FA-L account for 10%. FA-B, FA-I, FA-J, FA-M, FA-N, FA-P and RAD51C represent less than 5% of FA patients. Some patients do not appear to have mutations in these 15 genes, so we anticipate that additional FA genes will be discovered in the future.

FA occurs equally in males and females. It is found in all ethnic groups. Though considered primarily a blood disease, it may affect all systems of the body. Most patients develop bone marrow failure, necessitating a bone marrow transplant. Many patients develop acute myelogenous leukaemia (AML) at a very early age. FA patients are extremely likely to develop a variety of cancers and at a much earlier age than the general population (20s, 30s and 40s). Patients who have had a successful bone marrow transplant and are therefore cured of the blood problem associated with FA still must have regular examinations to watch for signs of cancer. Many patients do not reach adulthood.

Fanconi Anaemia patients are usually smaller than average. FA usually reveals itself before children are 12 years old but in rare cases no symptoms are present until adulthood. Patients may feel extreme fatigue and have frequent infections. Nosebleeds or easy bruising may also be a first sign. Blood tests may reveal low white, red cells or platelet counts, or other abnormalities. Sometimes myelodysplasia or AML is the first sign of FA.

FA is sometimes evident at birth through a variety of physical defects. These may include any of the following:

- \* Varying degrees of hearing loss.
- \* Thumb and arm anomalies: an extra or misshapen or missing thumb(s) and finger(s) or an incompletely developed or missing radius (one of the forearm bones).
- \* Skeletal anomalies of the hips, spine or ribs.
- \* Kidney problems.
- \* Skin discoloration (café-au-lait spots); portions of the body may have a suntanned look.
- \* Small head or eyes.
- \* Mental retardation or learning disabilities.
- \* Low birth weight.
- \* Gastrointestinal difficulties.
- \* Small reproductive organs in males.
- \* Defects in tissues separating chambers of the heart.

The definitive test for FA at the present time is a chromosome breakage test: some of the patient's blood cells are treated, in a test tube, with a chemical that crosslinks DNA. Normal cells are able to correct most of the damage and are not severely affected whereas FA cells show marked chromosome breakage. There are two chemicals commonly used for this test: DEB (diepoxybutane) and MMC (mitomycin C). These tests can be performed prenatally on cells from chorionic villi or from the amniotic fluid.

Many cases of FA are not diagnosed at all or are not diagnosed in a timely manner. FA should be suspected and tested for in any infant born with the thumb and arm abnormalities described previously. Anyone developing aplastic anaemia at any age should be tested for FA, even if no other defects are present. Any patient who develops squamous cell carcinoma of the head and neck, gastrointestinal or gynaecological system at an early age and without a history of tobacco or alcohol use, should be tested for FA. Many FA patients show no other abnormalities. It is absolutely essential to test for FA before contemplating bone marrow transplantation for aplastic anaemia or treatment for cancer. FA patients respond extremely poorly to standard chemotherapy and radiation protocols.

While the total number of FA patients is not documented worldwide, scientists estimate that the carrier frequency (carriers are people carrying a defect in an FA gene, whose matching FA gene is normal) for FA is somewhere between 1 in 600 and 1 in 100.

## **IMPORTANT SOURCES OF SUPPORT AND INFORMATION**

### Fanconi Anaemia Family Support

A UK-registered Charity and Family support group set up by Families FOR Families

Web: [www.fanconisupport.info](http://www.fanconisupport.info)

Email: [fanconi@hotmail.co.uk](mailto:fanconi@hotmail.co.uk)

Tel: 07939 593993

### FARF (Fanconi Anemia Research Fund)

A US-registered Charity with 25 years Family support and research experience

Web: [www.fanconi.org](http://www.fanconi.org)

Email: [info@fanconi.org](mailto:info@fanconi.org)

### Fanconi Hope

A UK-registered Charity supporting the aims of the FA Clinical Network

Web: [www.fanconi.org.uk](http://www.fanconi.org.uk)

### Contact-A-Family

A UK-registered Charity, supporting families with disabled children (not FA-specific)

Web: [www.cafamily.org.uk](http://www.cafamily.org.uk)

Tel: 0808 808 3555

### The Family Fund

A UK-registered Charity, supporting families with disabled and seriously ill children (not FA-specific)

Web: [www.familyfund.org.uk](http://www.familyfund.org.uk)

Email: [info@familyfund.org.uk](mailto:info@familyfund.org.uk)

Tel: 0845 130 4542

### Camp Sunshine

A US-based Charity and Retreat for children with life-threatening illnesses and their families (FA-specific week every year)

Web: [www.campsunshine.org](http://www.campsunshine.org)

Email: [info@campsunshine.org](mailto:info@campsunshine.org)