Fanconi Anaemia (FA)

Patient Information Series – What you should know, what you should ask.

What is Fanconi anaemia?

Fanconi anaemia (FA) is a disorder that causes bone marrow failure (sponge-like material inside the bones that produces bloods cells). This will manifest with low blood cells that carry the oxygen (erythrocytes or red blood cells - anaemia), cells that fight infection (leucocytes or white blood cells), and cells that help the blood clot (platelets).

This condition can also cause birth defects in various organs and body parts.
There is also an increased risk of developing blood cancer (leukemia, myelodysplastic syndrome) and solid tumors (most commonly of the skin, liver, brain, and kidney).

How does an FA happen?

FA is a rare genetic condition that affects 3 in 1,000,000 people. It is mostly inherited from parents as an autosomal recessive disorder (faulty gene needs to be inherited from both parents), but X-linked recessive inheritance has also been reported (faulty gene comes from the mother and male children are affected by the disease, whilst female children are not). There are many different genes linked to FA.

Should I have more tests done?

When FA is suspected before the baby is born, there are three tests that can be offered to you to confirm the condition:

- **Chorionic villus sampling** – taking a sample of the placenta by passing a thin needle through your abdomen and the womb into the placenta
- **Amniocentesis** – taking a small amount of amniotic fluid by passing a thin needle through the abdomen into the womb and the fluid surrounding the baby. The amniotic fluid contains the baby's skin cells that are then used for testing,
- **Cordocentesis** – taking the baby's blood sample from the cord by inserting a thin needle through the abdomen and the womb into the umbilical cord.

Any of the samples that we get from the above procedures can be used to do the chromosome breakage test, flow cytometry cell cycle testing, or genetic testing. The chromosome breakage test and flow cytometry cell cycle testing are the most used tests to diagnose FA. Genetic testing will look for genes linked to FA.

What are the things to watch for during the pregnancy?

Regular scans should be performed to exclude any associated abnormalities and to monitor the baby's growth and well-being. There are currently no treatments that can be offered during the pregnancy.

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What does it mean for my baby after it is born?

Due to the bone marrow failure, the child will be prone to infections, fatigue caused by anaemia, bruising, and bleeding gums. This can be treated with blood transfusions and medication to improve the symptoms. However, the final treatment for bone marrow failure is stem cell transplant, which has significantly improved the survival of the affected individuals.

Birth defects can include many different regions of the body. Skin discoloration is common (café au lait spots) as well as limb abnormalities (mainly of arms and hands). Children can also have short stature, small head or eyes, abnormal face and ears, and defects of reproductive organs, kidneys, and heart. Some children might have developmental delays.

Surgery might be required after birth or later on in life depending on the birth defects present. The main concern in the long-term prognosis is the increased risk of developing cancer. Because of this, the life span of people affected by FA is significantly shortened.

Will it happen again?

In cases of autosomal recessive inheritance where both parents carry a faulty gene, each of their children has a 1 in 4 chance of having FA. In cases of X-linked inheritance where the mother is the carrier, there is a 1 in 2 chance of having a son with the disease. Female children will not have the disease but might be carriers like their mother. Genetic counselling is advised following diagnosis of Fanconi anaemia in the family.

What other questions should I ask?

- How often will I have ultrasound examinations done?
- What options do I have before the baby is born?
- Can I meet the team of doctors that will be assisting my baby when it is born in advance?
- Is there anything that can be done for the baby before birth?
- How and where should I give birth?
- Will my baby need surgery after birth?
- Where can I get additional support during the pregnancy?
- Where can I get more information about Fanconi anaemia?

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