

WHAT IS THE RISK OF MY CHILD INHERITING THE DISEASE?

It should be noted that FMF is an **autosomal** (affecting both girls and boys) **recessive** (presence of two mutations in the gene responsible for FMF, one genetic mutation inherited from each of the parents) **disease**.

There are several possible scenarios.

SCENARIO N° 1: TWO ASYMPTOMATIC CARRIER PARENTS

At each pregnancy, this couple:

- has a 25% chance of having a child carrying no mutations
- has a 50% chance of having a child carrying a mutated gene
- has a 25% chance of having a child with FMF, thus carrying two mutated genes

This could be explained by the fact that FMF may not be visible for several generations—two asymptomatic carrier parents have a 25% risk of having a child with FMF.

SCENARIO N° 2: ONE PARENT WITH FMF, ONE UNAFFECTED PARENT

At each pregnancy, this couple will have children carrying a mutated gene. They will be simple heterozygotes.

This couple will not have any children with FMF.

This situation could also be explained by the fact that FMF may not appear for several generations. However, since all of these children are carriers, they could be in scenario n°1 when they grow up and have children.

SCENARIO N° 3: ONE PARENT WITH FMF, ONE ASYMPTOMATIC CARRIER PARENT

At each pregnancy, this couple:

- has a 50% chance of having a child carrying a mutated gene
- can have a child with FMF, thus carrying two mutated genes

This couple has a 50% chance of having a child with FMF.

SCENARIO N° 4: BOTH PARENTS HAVE FMF

At each pregnancy, this couple will have children with FMF.

SCENARIO N° 5: ONE ASYMPTOMATIC CARRIER PARENT, ONE UNAFFECTED PARENT

At each pregnancy, this couple:

- has a 50% chance of having an unaffected child
- has a 50% chance of having a child carrying a mutated gene

None of the children will have FMF. Like scenarios n°1 and 2, this situation could also be explained by the fact that FMF may not appear for several generations.

Despite the above scenarios still there are cases where a patient with a single mutation will have full blown FMF.

"I WAS PREGNANT WITH MY SECOND CHILD WHEN THE GYNECOLOGIST ADVISED ME TO SEE THE GENETICIST at the hospital to find out if I should undergo an amniocentesis or not. She told me that I should because I was taking colchicine. She then proposed an appointment for the examination. I told her that couldn't make this decision without getting the internist's opinion. The internist was categorical—colchicine would not cause any malformations in the unborn child—so an amniocentesis was unnecessary."

Père de Jérémy, 13 ans

"UNFORTUNATELY, MY SITUATION WAS EXCEPTIONAL - being pregnant did not reduce the number of attacks. What should be done? What medication should I take to reduce the pain? The emergency doctor I called one evening was uncertain. I decided not to take any medication because I wasn't sure if there would be any effects on my unborn child. The next day, I got an appointment with my internist, who contacted my gynecologist to determine what medication I could be prescribed without risk of harming my child."

Aude, 35 ans

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**COLLECTION TO LIVE WITH
"I AM PREGNANT
AND SUFFER FROM FMF"**

WHAT IS FAMILIAL MEDITERRANEAN FEVER, WHICH IS MORE COMMONLY REFERRED TO AS PERIODIC DISEASE?

Familial Mediterranean fever (FMF) is a rare genetic disease in France. Attacks may last one to several days (3- 4) and generally expressed by fever and pain in the membranes of the abdomen (peritonitis), lungs (pleuritis) and/or joints (arthritis).

The symptoms may suggest virus infections, flu, or even—when the attacks are violent- may mimic «surgical abdomen».

In addition to the painfulness of these attacks, the main danger is that the disease will lead to amyloidosis with **kidney failure** (a very serious complication with the risk of having to undergo dialysis, kidney transplantation, or it may even result in death).

When the diagnosis is confirmed—often after years of diagnostic error—the person begins a long-term drug **treatment** (colchicine tablets) with regular follow-up by a specialist or the family doctor. This treatment **protects** the patient from renal complications (amyloidosis) and generally **reduces** the attacks frequency and intensity.

WILL I BE ABLE TO HAVE CHILDREN WITHOUT DIFFICULTY?

Yes. There is **no** risk of sterility in women with FMF under colchicine treatment. Cases of sterility have been noted in some patients who have suffered many inflammatory episodes and **were not on colchicine treatment**. Some untreated males sterility cases have been reported in patients suffering many inflammatory episodes, and some of them had also developed testicular amyloidosis.



CAN I TAKE COLCHICINE DURING PREGNANCY?

Definitely, since there is solid evidence that colchicine does not induce foetal malformations. It is thus **essential** to continue the treatment, at the usual dosage, throughout pregnancy. This is important for you and your baby, as the treatment will protect you from potential attacks and eventual premature delivery.

AND WHAT ABOUT COLCHIMAX?

It is **recommended** that this medication be **avoided** in favour of colchicine. Colchimax contains tincture *Opii* and therefore can induce foetal drowsiness, with a **risk** of addiction and withdrawal symptoms.

ARE THERE ANY RISKS ASSOCIATED WITH PREGNANCY?

Inflammatory conditions can increase the risk of miscarriage. Colchicine **reduces** or even suppresses this risk. In many women, periodic disease symptoms **disappear** during pregnancy. The mechanisms underlying this phenomenon are still unclear, but they seem to be linked with the hormonal metabolism induced by pregnancy, and with the effects of foetal inflammation regulators in the maternal blood.

It is still essential to have the **best possible follow-up** during pregnancy by planning a visit to an FMF internal specialist who will coordinate care with your obstetrician-gynecologist.

SHOULD I UNDERGO AN AMNIOCENTESIS?

No. Undergoing colchicine therapy during the **conception** and pregnancy period, in the **absence** of other factors, is not a sufficient basis for prescribing an amniocentesis. It is therefore important that the specialist contact the **obstetrician** in order to avoid mistakenly prescribing and amniocentesis for women patients simply because they have FMF and are under colchicine therapy at a regular dosage.

WHAT SHOULD I DO IN CASE OF AN ATTACK?

Make an appointment with your FMF internal specialist. This doctor will prescribe medication at dosages that are compatible with your pregnancy. It is **advised** that you make this appointment as soon as you know you are pregnant, do not wait until you experience an attack.



CAN I BREAST FEED MY INFANT?

There is no reason for a woman under colchicine therapy not to breast feed her baby, because very little of this medication passes into the maternal milk. It is recommended that mothers take their colchicine tablet just before the evening breast feeding, because the colchicine content in the milk peaks about two to three hours after the medication is taken, and it declines seven to eleven hours after. The baby will thus not absorb much colchicine, and the issue will be resolved when the mother stops the evening breast feeding sessions.

It should again be stressed that colchimax should not be taken during breast feeding—opt for colchicine instead.