

Appendix C Information sheet for adolescents over 15 and adults



Deutsche Fanconi-
Anämie-Hilfe e.V.

Information for people with Fanconi Anemia (Aged 15 and Older)

Title of the study: "Reducing the burden of squamous cell Carcinoma in Fanconi anemia"

Hello,

We would like to invite you to be part of a research study. You are being asked to participate because you have been diagnosed with Fanconi anemia (FA).

Why are we doing this project?

People with FA have a higher than average risk of developing mouth cancer at young ages. Because of this, it's important that you have your mouth checked regularly by an experienced head and neck doctor (ENT). Mouth cancer can be treated relatively easily when detected early, but when discovered in later stages, treatment becomes more difficult. This project will help us learn if we can use simple, noninvasive techniques to collect and test samples for early signs of oral cancer and pre-cancer.

What will you be asked to do?

- **Health questionnaire:** We will ask you to answer questions about yourself, your medical history, and current health. You may choose to leave any questions you do not wish to answer blank.
- **Photograph and Oral map:** We will examine the inside of your mouth carefully and document any spot, mark or visible lesion on a diagram ("oral cavity map") so we can remember where they are.
- **Brush samples:** We will ask you to give us a small amount of cells from your mouth using a soft brush. First, we will ask you to gently brush six places in your mouth with a soft brush under our guidance for LOH analysis (LOH means Loss of Heterozygosity). You will not receive a report from this analysis. (Note for FA participants who have received a bone marrow transplant: At this time, this test does not work well for people who have had a bone marrow transplant. Researchers would still like to have brushes from transplanted patients to try to improve this test. These samples will be frozen and stored until a better method is found.)

We will also ask you to brush any visible lesions under our guidance for further cytological analysis. This test looks at the shape and morphology (changing) of the cells for any pre-cancerous and cancerous changes. You and your doctor will receive a report of this test on the visible lesions if you wish.

Extra brush samples: We would like to test for early signs of serious mouth problems in FA using other research procedures. If you agree to provide extra samples we will ask you to brush certain places in your mouth once more under our guidance, as well as any visible spots, for another study looking at specific bacteria present in the mouth. You will not receive a report from this analysis.

Saliva sample: If you agree to donate a saliva sample, you will be asked not to eat, drink, smoke, or use oral hygiene products (like toothpaste or mouthwash) for at least half an hour before. You will rinse your mouth with water, place a white pad (like a small sponge) in your mouth, and then, after about 2-5 minutes, put the pad into a cup we provide.

What do we hope to learn with the samples you donate?

We hope to use the samples you brush from your mouth to see whether it is possible to use a new method, called an "LOH" analysis, to find pre-cancerous cells very early in people with FA. When we find visible spots in the mouth, brush samples from these areas can be tested for suspicious cells using an existing test, called a "cytological analysis." Extra brush samples will be used to investigate whether there is a link between certain types of bacteria and the development of head and neck cancer in people with FA. Through collection of saliva, researchers hope to learn whether we can find molecules in saliva that would indicate the presence of early cancer.

You do not need to agree to participate in all of the above in order to be part of this project.

Possible risks and side effects of participating in this study

Brushing the mouth and collecting saliva are generally painless. The brushing might give you a tickling sensation and may cause irritation in places where the skin is already sensitive.

Analysis of brush samples may indicate an abnormality that is later found to be normal by a traditional tissue biopsy. This could happen because the brush test finds pre-cancer cells earlier than a pathologist can find them. However, it is also possible that the brush test was done too early or was inaccurate and a tissue biopsy was therefore not necessary.

Possible benefit of participating in this study

A possible advantage of participating is that if a suspicious spot is seen, you and your doctor will be alerted right away and you will be able to watch it carefully.

CAUTION: We could miss abnormal areas in our inspection. You should continue having regular checkups with your local doctor or dentist. If you find unusual spots in your mouth that do not vanish after 2-4 weeks, talk to your doctor right away.

Time to think it over

Please take all the time you need to think about whether you would like to participate in this project. Please do not hesitate to talk with family, friends and/or the project staff if you have any questions before you decide.

Confidentiality

The information that we collect about you during this project will remain confidential. Only the research team will have access to your name and personal information. For minors, please note that your parent(s) can request and receive your data including your responses to the questionnaire.

Participation is voluntary

You are completely free to decide whether you want to participate in this research project or not. If you decide to participate, you are free to change your mind and discontinue participation at any time.

Further information

We will ask to collect brush samples from FA patients who wish to participate at least every second year. If after reading this document you would like more information, or have new questions at any time before, during, or after the project, you can contact the Fanconi Anemia Research Fund by phone at 541-687-4658 or by email at study@fanconi.org.

Signing the consent form

If you decide to participate, we will ask you to confirm that you agree through signing an informed consent form. You may choose whether you want to participate in all parts of this project or only in some. If you change your mind about participation, you may contact us and indicate your desire to discontinue participation at any time. Parents or guardians of minors must also sign the consent form to let their child participate in the study.