



Punch skin biopsy: information for families of children and young people with neuromuscular disorders

A punch skin biopsy is a short procedure to remove a small piece of skin tissue. This information sheet from Great Ormond Street Hospital (GOSH) explains what happens when your child comes to GOSH.

The sample of skin taken during the biopsy can be examined under a microscope or used to grow cells from the underneath surface (fibroblasts) of the skin. These cells can be tested for abnormalities of the DNA or to show whether a specific chemical reaction can or cannot take place.

Why does my child need a skin biopsy?

A punch skin biopsy is usually carried out to determine or confirm a diagnosis. There are also certain metabolic or chromosome disorders that can only be diagnosed by a skin biopsy. The doctors will discuss the reason for your child's biopsy with you.

Getting ready for the biopsy

Your child will usually have a skin biopsy as a day case on the ward. This means your child will arrive at the hospital, have the procedure and go home on the same day unless further investigations or treatment is needed. If your child is having another procedure under a general anaesthetic or sedation, the biopsy can be done at the same time.

What happens before the biopsy?

The nurse caring for your child will see you when you arrive on the ward to explain the procedure in more detail, discuss any questions or concerns you may have and ask you to give your permission for the procedure by signing a consent form. If your child has any medical problems, for instance, allergies, please tell the nurse about these.

The person bringing your child to the test should have 'Parental Responsibility' for them. Parental Responsibility refers to the individual who has legal rights, responsibilities, duties, power and authority to make decisions for a child. If the person bringing your child does not have Parental Responsibility, we may have to cancel the test.

What does the biopsy involve?

Skin biopsies are usually done while your child is fully awake, although the area of skin where the biopsy is taken will be numbed. Some children are worried by the procedure but you can help to distract your child by reading a story, playing a game or singing a song.

Depending on the area being biopsied, younger children can sit on your lap during the procedure. The biopsy takes about a minute. The biopsy site is numbed using a cream or spray before a local

anaesthetic is injected. Once the area is completely numb, the nurse or doctor will clean it with an antiseptic wipe.

The punch skin biopsy needle is gently inserted into your child's skin, rotated and a small circle of skin is carefully removed. The sample of skin will be sent to the laboratories for examination under a microscope and/or to grow cells from the underneath surface.

The biopsy site usually bleeds slightly straight after the procedure, but this will stop when pressure is applied to the site. The area will be held closed by Steri-Strips®, which are like strong sticky plasters, and then covered with a dressing.

Are there any risks?

There is a very small chance that the biopsy site may become infected afterwards. A child with known immune problems may be given an antibiotic cream to put on the area after the dressings and Steristrips® have been removed.

There is a chance that a scar, similar to a chicken pox scar, may form. This is more likely if your child has scarred easily in the past.

Going home

After the biopsy, you and your child will be able to go home unless any further treatment is needed.

The biopsy site may feel uncomfortable for a day or two afterwards. Your child may need paracetamol given according to the instructions

on the bottle, unless you have been advised otherwise.

After five days, you can remove the dressing. The Steri-Strips® may fall off of their own accord – otherwise they can be soaked off in the bath or shower when you remove the dressing. Your child can go back to nursery or school the day after the biopsy, but the area could be a bit sore if it is knocked.

You should call your family doctor (GP) or the ward:

- If the biopsy site is still painful more than three days after the biopsy was taken and normal pain relief does not work
- If the biopsy site is red or angry-looking
- If your child develops a high temperature and is not eating

How long will it take to get the test results?

Growing fibroblasts often takes four to six weeks, so results of biochemical testing may be available within six to eight weeks. However, some of the more specialised biochemical tests have to be sent away to a specialised laboratory, which only do tests infrequently, so can take up to six months to come back.

Analysis of the genes responsible for a disease (carried on the DNA) can also take several months. We will tell you when you can expect to have the results.

If you have any questions, please call the Neuromuscular Clinical Nurse Specialists on 020 7405 9200 ext 1195. You can also send an email to neuromuscular.cns@gosh.nhs.uk.

Having a punch skin biopsy



Your skin is the biggest part of your body. It stops bugs getting into your body and holds all your bones and organs in place. You are coming to hospital for a skin biopsy (said: bye-opp-see).



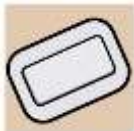
A biopsy is a tiny bit of body tissue. People in our laboratory (said: lab-or-ra-tree) look at it under a microscope (said: my-cro-scope). This helps the doctors work out how to look after you.



First we will put some cream on your skin where the biopsy will be taken. This makes your skin numb so you will not feel much.



Next we will give you an injection to make sure you do not feel any pain for a few hours. The doctor will quickly take a small bit of skin to send to our laboratory.



The area may bleed a bit afterwards but will soon stop. We will put some sticky paper strips over the area to hold it closed. We then put a plaster over the top to keep it clean while it gets better.



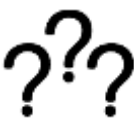
You might feel a bit sore where the biopsy was taken. You can take pain medicine to help.



You can go home a bit later the same day. Keep the dressing dry for 2 days. Tell someone if there is anything making the plaster wet.



Your doctor will get a report of the results of the biopsy. They will discuss it with you in clinic as soon as possible.



Please ask us if you have any questions.