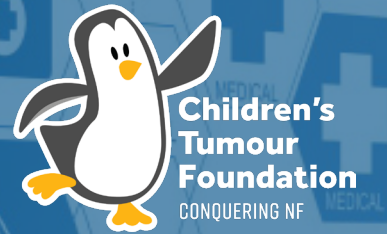


TREATING SKIN IN NF1



The information provided below is a summary of the presentation given by Assoc. Professor Mimi Berman on 7 March 2020 at the NF Information Day in Sydney.

Mimi Berman is the Head of the Genetics Department at the Royal North Shore Hospital (RNSH). She is also the Co-Chair of the ACI Clinical Genetics Network and the current President of the Australian Association of Clinical Geneticists (AACG) and is the Medical Advisor to The Children's Tumour Foundation.

TREATING ITCH IN NF1



WHAT ARE THE PSYCHOLOGICAL IMPACTS OF CUTANEOUS NEUROFIBROMAS?

Many international studies have shown that up to 70% of adults and up to 50% of children have reported that their neurofibromas were itchy and had a detrimental impact on their quality of life.

An Australian study of 60 adults conducted in 2015 showed that participants rated the cosmetic burden of their neurofibromas to be their main concern (even though they had other NF symptoms).

Patients in these studies commonly reported negative effects on their quality of life, particularly their self-esteem and self-confidence. They also reported shame, embarrassment and anger at the way their skin looked.

Royal North Shore Hospital (RNSH) recently conducted their own study and found similar results.



WHAT ARE THE USUAL TREATMENT OPTIONS FOR ITCH?

Many patients reported that they had tried various treatments, all with limited success. These included antihistamines, steroid creams and tablets, vinegar baths and other ointments.

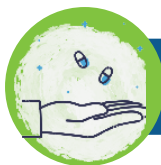
Most people choose to wear neutral fabrics and stay out of the sun in an attempt to alleviate the itch.



WHAT NEW DRUGS HAVE BEEN TESTED FOR ITCH?

RNSH trialed Doxepin in low doses. This is an antidepressant but also an effective treatment for nerve pain. Pregabalin (Lyrica)/ Gabapentin are nerve stabiliser drugs and were also trialed with success.

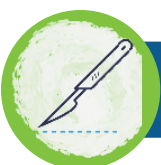
Both of these drugs were well tolerated by most participants and there was a significant reduction in the level of itch reported by patients. This resulted in significant improvements in overall quality of life.



HOW CAN I GET ACCESS TO THESE DRUGS?

The RNSH study is still yet to be published. Once this information is readily available, the CTF will distribute this on our website and social media pages (@ctfaustralia).

THE REMOVAL OF NEUROFIBROMAS



HOW CAN NEUROFIBROMAS BE TREATED?

Different drugs have been trialled in an attempt to shrink cutaneous (skin) neurofibromas. These include creams or tablets with a MEK Inhibitor (Rapamycin), chemotherapy and antihistamines (Ketotifen). Many of these have not reduced the size of the neurofibromas significantly.

The traditional method of removing neurofibromas is with a surgical blade. This is done by a specialist dermatologist or plastic surgeon. Other methods include electrodesiccation, which is where an electrode is placed very close to the neurofibroma and an electrical current destroys the tissue.

More recently, laser has been successfully used to reduce the appearance of neurofibromas. This can either be with a Carbon Dioxide (CO2) or a YAG laser (a form of crystal laser).



LASER TO REMOVE NEUROFIBROMAS

Laser is mostly applied to cutaneous neurofibromas that are less than one centimetre in diameter and are located superficially on the skin. The larger and more deeply set neurofibromas are still generally removed with a blade.

Whilst laser treatments are quite successful, prospective patients need to consider the fact that the procedure is painful and there will be a lot of scarring immediately afterwards. At least one week will be spent recuperating and patients will need to be very mindful of sun exposure.

Fractionated lasers can be used to improve texture of lumpy or pitting skin.

PROS OF LASER	CONS OF LASER
<ul style="list-style-type: none"> • No general anaesthetic required • Local anaesthetic • Can be done as an outpatient • Good results • Downtime is generally one week 	<ul style="list-style-type: none"> • Painful process despite local anaesthetic • Limited field (only face or back done in a single sitting) • Temporary scarring

If you have any questions regarding this information, our Support Team is available to help. You can reach them via phone on (02) 9713 6111 or by email at support@ctf.org.au