**Neurofibromatosis type 1 – Annual Review**

**Information for teenagers and young people**

**What is neurofibromatosis type 1?**

Neurofibromatosis type 1 (NF1) is a genetic condition which affects about 1 in every 3000 children. It may affect different parts of the body at varying ages, hence the need for an annual review.

**Why do we need an annual review?**

NF1 can affect several different parts of the body. Some of these effects might be present from birth and others might develop at different ages. The annual review is a yearly appointment to keep track of how NF1 is affecting you and to offer treatment and support.

This leaflet aims to give you information about what the doctors will be looking for at your annual review. It is important to remember that NF1 affects everyone in different ways, so just because some people with NF1 have difficulties in a particular area does not necessarily mean that you will.

**Skin - neurofibromas**

What are neurofibromas?

* Lumps on the skin
* Neurofibromas are **benign**, which means they do not spread to other parts of the body or grow in an uncontrolled way, though they may get bigger
* A lump which grows in an uncontrolled way is known as malignant
* It is **extremely rare** for a neurofibroma to become malignant when you are aged under 20 years old. After this, the risk increases but it is still **very rare**
* Signs to look out for include:
	+ Pain
	+ Tingling or other unusual sensations
	+ A neurofibroma rapidly growing bigger

Possible next steps

* If you are particularly concerned by how the neurofibromas look, you might be referred to a dermatologist (skin specialist doctor) or a plastic surgeon who will talk about possible treatment options
* This might include having the neurofibroma surgically removed. However, there is a risk it will grow back

**Eyes**

We recommend you should have regular eye reviews with an ophthalmologist (eye specialist doctor) every 2 years until adulthood, with extra appointments if you notice any changes between these appointments.

What are we looking for?

* Possible signs of optic pathway glioma (OPG)
	+ An OPG is a growth on the nerves of the eye
	+ Signs of OPG include:
		- Changes in the clearness of vision (acuity)
			* For example finding it difficult to read writing on the board at school or struggling to read small writing on your phone
		- Changes in colour vision
		- Squint – when one eye points in a different direction to the other
		- Bulging of the eye
		- Changes at the back of the eye – seen by a doctor on examination

Possible next steps

* If you have any visual symptoms, you may need an MRI scan to rule out OPG

**Brain and nervous system**

What are we looking for?

* Head size
	+ People with NF1 often have a larger than average head size, which is not a problem in itself. However, it is important we monitor it as rapidly increasing head size can be a sign of a growth on the brain.
	+ Growths on the brain can cause a rise in the pressure inside the skull. Signs of this include waking up with a headache and vomiting in the morning.
* Epilepsy review
	+ Some people with NF1 can develop epilepsy, which can cause them to have symptoms such as epileptic fits (epileptic seizures)
	+ If this affects you, the annual review is a good opportunity to check how well controlled your seizures are and to check how you are finding your medication

Possible next steps

* If there is a possibility that there may be problem with the brain or spine, you may be referred to a neurologist (specialist doctor) and we may arrange scans of the brain

**Bones**

What are we looking for?

* Bowing of the bones
* Scoliosis – curving of the spine into an S-shape

Possible next steps

* You may be referred to an orthopaedic (bone) surgeon for a further assessment

**Blood pressure**

What are we looking for?

* Some people with NF1 may have high blood pressure

Possible next steps

* If your blood pressure is high, we may do some other tests to look for causes of high blood pressure

**Growth and development**

The doctor will measure your height and weight and ask about your development.

Possible next steps

* If there are concerns about your growth or development, you may be referred to a hormone specialist (endocrinologist)

**Education and behaviour**

What are we looking for?

* How you are getting on at school or college
	+ It is very helpful if you can bring your school report
* Some people with NF1 might have difficulties in areas such as:
	+ Attention and concentration
	+ Being easily distracted
	+ Social interaction – forming friendships and getting along with others
	+ Getting to sleep at night

Possible next steps

* If there are concerns about any of these areas, you may be referred for a more specialist assessment by an educational or clinical psychologist, a community paediatrician or a neurologist

**Psychological**

The annual review is a good opportunity to check how you are coping with your NF1. Some young people might have difficulties coping with the appearance of their skin, low mood or problems interacting with other people. These are all things the doctors can help you with, so it is good to mention anything that might be worrying you.

* If you feel it might be helpful to talk to someone, the doctor can arrange counselling
* If you are particularly troubled by your skin appearance, you may be referred to a dermatologist (skin specialist) or plastic surgeon to discuss treatment or removal of neurofibromas

**Learning about your condition**

As you get older and become more independent it is important that you feel educated about NF1. The annual review is a good opportunity for you to ask any questions you might have about your condition.

**Planning for the transition to adult services**

* When you are 18 years old, your care will move to the adult services team.
* It is sensible to start planning for the transition to adult services early, so you can meet the team and feel comfortable that you know who to go to for advice and support. Planning usually starts when you are aged 13-15 years old.

**How long will the annual review continue?**

We recommend annual review appointments continue until you are 25 years old.

* After this, you may be seen less often, depending on how NF1 affects you and your personal wishes.
* As a minimum, your blood pressure should be measured at least once per year

It is particularly important that you know:

* What changes to look out for and when to contact a doctor for advice
* Who to contact if you have any questions or concerns

**Between appointments, you should contact a doctor if you experience:**

* Symptoms associated with a neurofibroma
	+ Rapid growth of the neurofibroma
	+ Constant pain
	+ A part of the body not moving or not working normally
* Changes in vision
	+ Change in the clearness of vision (acuity)
	+ Change in colour vision
	+ Development of a squint – one eye pointing in a different direction to the other
	+ Bulging of the eyes
* Symptoms which could indicate a problem with the brain or spinal cord
	+ Pain
	+ Numbness, tingling or pins and needles
	+ Headaches
	+ Loss of bowel or bladder control
	+ A body part not moving properly
* Signs that the pressure inside the skull might be high
	+ Waking up with a headache in the morning
	+ Vomiting in the morning