**Neurofibromatosis type 1 – Annual Review**

**Information for Parents**

**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 your child and to offer treatment and support. Exactly what the annual review involves will depend on your child’s age and how NF1 affects them.

This leaflet aims to provide some information about what the doctor will be looking for. However, it is important to remember that NF1 affects different children in different ways so not all the information that follows will apply to your child and just because one child with NF1 has problems in a particular area does not mean your child necessarily will.

**Skin**

What are we looking for?

* Neurofibromas - a type of benign tumour
  + Benign tumours do not spread to other parts of the body or grow in an uncontrolled way, though they may get bigger
    - A tumour which grows in an uncontrolled way is known as malignant
  + Cutaneous neurofibromas are on the skin
    - Do not become malignant
    - May be removed if they are catching on clothes and bleeding, or if the child is particularly concerned about how the neurofibroma looks
  + Subcutaneous neurofibromas are just under the surface of the skin
    - In childhood it is extremely rare for these to become malignant
      * Signs of this include pain, tingling or other unusual sensations or rapid growth of the neurofibroma

Possible next steps

* If your child is concerned about the appearance of their neurofibromas, they may be referred to a dermatologist (skin specialist) or a plastic surgeon, to discuss possible treatment or removal
  + If a neurofibroma is removed, there is a risk it may grow back

**Eyes**

Current guidelines are that all children with NF1 should be assessed yearly by an ophthalmologist (eye specialist doctor) until they are 7 years old. This is because children under the age of 7 are less likely to complain if their vision changes, and also because most problems with vision in NF1 develop before the age of 7.

The guidelines are not so clear after this, but we advise that regular eye assessments should continue every 2 years until adulthood, with additional reviews if the child’s vision changes in any way.

What are we looking for?

* Possible signs of optic pathway glioma (OPG)
  + An OPG is a tumour which grows on the nerves of the eye
  + Signs of OPG include:
    - Changes in the clearness of vision (acuity)
    - 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 your child has visual symptoms, they may have an MRI scan to rule out OPG

**Brain and nervous system**

What are we looking for?

* Tumours
  + Your child’s head circumference will be measured. Children with NF1 often have a larger head circumference compared to the average, which is not in itself a problem. However, it is important to monitor as rapidly increasing head size can be a sign of a tumour.
  + Tumours 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 children with NF1 can develop epilepsy, which can cause them to have symptoms such as epileptic fits (epileptic seizures)
  + If this affects your child, the annual review is a good opportunity to review how well controlled the seizures are and discuss any medication they might be taking

Possible next steps

* If there is a possibility that there may be problem with the brain or spinal cord, your child 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, particularly in the legs of young children
* Scoliosis – curving of the spine into an S-shape

Possible next steps

* Your child may be referred to an orthopaedic (bone) surgeon to assess them further

**Blood pressure**

What are we looking for?

* Some children with NF1 may have high blood pressure

Possible next steps

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

**Development**

What are we looking for?

* Height and weight
* Stage of development
* Co-ordination
* Speech and social communication

Possible next steps

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

**Education and behaviour**

What are we looking for?

* How your child is performing and behaving in school and at home
  + It is very helpful if you can bring their school report
* Sleep patterns
  + Any difficulties with getting to sleep or waking during the night
* Attention, concentration and distractibility
* Social interaction
  + Some children with NF1 might have difficulties forming friendships with peers
* Motor skills

Possible next steps

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

**Psychological**

Particularly in older children and young adults, the annual review is a good opportunity to check how the young person is coping with their NF1.

* If the young person might benefit, they may be referred for counselling
* If the young person is particularly troubled by their skin appearance, they may be referred to a dermatologist (skin specialist) or plastic surgeon to discuss treatment or removal of neurofibromas

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

* It is recommended that the transition from child to adult services happens gradually between the ages of 16 and 18 years old. Planning for this can start when the child has a learning age of 13 years old and/or when they are aged over 15 years.
* After the transition to adult services when your child is 18 years old, it is to continue annual reviews until the age of 25. After this, the frequency of appointments depends on the severity of the patient’s NF1 and the patient’s personal wishes.
  + As a minimum, it is recommended that blood pressure is measured at least once per year.
  + Additionally, it is important that patients and their families know what symptoms to look out for and who they should contact if they have any concerns.

**Will my child have routine brain scans?**

* MRI scans can show signs of NF1, such as commonly seen benign lesions, which do not grow and spread
* MRI scans can be used to find optic pathway gliomas (OPG)
  + There is evidence that there is no advantage to detecting OPG before they have caused symptoms, and early diagnosis will not change how the child is treated
  + So routine brain scans are not needed if the child is having regular eye reviews and the child and family know to report any changes in vision or in the appearance of the child’s eye
  + Children with communication difficulties may not be able to tell us if their vision changes, so may need more frequent scanning

**Between appointments, you should contact a doctor if your child experiences:**

* Symptoms associated with a neurofibroma
  + Rapid growth
  + Constant pain
  + A part of the body not moving or functioning 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
  + Signs of poor vision in younger children, like bumping into things
* 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