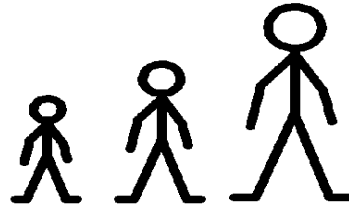


Personal Health Record



Neurofibromatosis Type 1

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Neurofibromatosis Type 1 Personal Health Record

You have been given this booklet because you or your child has a condition called **Neurofibromatosis Type 1 (NF1)**. People with NF1, may see a number of doctors and specialists, and this booklet will help you and your health care team ensure you/your child receive the correct care and appropriate check ups. Please bring it with you whenever you visit the genetics clinic or any of the following and ask them to complete the relevant pages:

- Your GP *(ask them to complete page 13-14)*
- Hospital Outpatients Clinic or Emergency Departments *(ask them to complete pages 5 or 6 and 13 or 14)*
- Your Dentist *(ask them to complete page 13 or 14)*
- Your Optician *(ask them to complete page 13 or 14)*
- Any other health care appointment *(ask them to complete page 13 or 14)*

You may also wish to use the plastic pockets in the front and back of this folder to store appointment letters, letters from your GP and hospital doctors, or anything else related to your NF1 that you want to keep safe.

Spare pages for this booklet are available from the Regional Genetics Service.

Important Note: This record does not replace your hospital, GP or other medical records which will continue to exist alongside this one.

This is the Personal Health Record of:

Name	<input type="text"/>	GM No.	<input type="text"/>
Address	<input type="text"/>	Date of Birth	<input type="text"/>
		Telephone	<input type="text"/>

Useful Contacts:

NF1 Doctor	<input type="text" value="NAME"/>	<input type="text"/>
NF Nurse/Genetic Counsellor	<input type="text" value="NAME"/>	<input type="text"/>
GP	<input type="text" value="NAME"/>	<input type="text"/>
Optician	<input type="text" value="NAME"/>	<input type="text"/>
Dentist	<input type="text" value="NAME"/>	<input type="text"/>
School (if applicable)	<input type="text" value="NAME"/>	<input type="text"/>
NF Association Spec. Advisor	<input type="text" value="NAME"/>	<input type="text"/>





NF1 CLINIC DETAILS

Genetics Service should attach a printed sticker here with clinic address and contact details.

Specialist Doctor Details

Sheet Number





People with NF1 are often looked after by more than one specialist, and this page is a record of all the doctors who are involved in caring for you/your child. If you visit a specialist doctor who isn't listed, they should add their details below.

Name	Title/Specialty	Address/Phone Number
		
		
		
		

Specialist Doctor Details cont...

Sheet Number

People with NF1 are often looked after by more than one specialist, and this page is a record of all the doctors who are involved in caring for you/your child. If you visit a specialist doctor who isn't listed, they should add their details below.

Name	Title/Specialty	Address/Phone Number
		
		
		
		

Information for You and Your Family

What is Neurofibromatosis Type 1?

Neurofibromatosis Type 1 (NF1) is a genetic disorder. The effects are variable from person to person and it is not possible to say how or if someone will be affected in the future.

Some people with NF1 will have skin features only.

One third may develop medical complications during their lifetime.

Because of this, people with NF1 need to take extra special care of themselves and have regular health checkups.

What Causes NF1?

It is caused by a change in one of your genes. Genes are responsible for individual characteristics such as eye and hair colour. People with NF1 have a change in the NF1 gene.

Half the people who have NF1 are the first person in their family to be affected. Others may have a parent with NF1 from whom they have inherited the faulty gene. If you have NF1, there is a 1 in 2 chance of passing this on to each child.

Whenever a person is diagnosed as having NF1, we check immediate family members thoroughly to see if anyone else in the family is affected.

What are the Features of NF1?

Almost all people with NF1 have one or all of the following:

- Café au Lait patches
- Freckles in unusual places
- Lumps and bumps on the skin

Some people may have additional health problems including:

- Learning difficulties
- Eye problems
- Problems with your back or bones

There are more serious complications (including uncommon cancers), which fortunately are rare.

Signs of a serious complication could be:

- INCREASING HEADACHES
- SIGNIFICANT CHANGES TO THE EYES OR VISION
- AN INCREASE IN THE SIZE OR HARDNESS OF AN EXISTING LUMP, OR PAIN IN A LUMP WHEN RESTING

If you/your child experiences any of these or any other worrying symptom, you should contact your GP or NF Doctor
IMMEDIATELY!

Information for Health Care Professionals

Diagnosis and Follow Up

Neurofibromatosis Type 1 (NF1) is an autosomal dominant condition occurring in approximately 1 in 3000. Half of affected individuals inherit NF 1 from their parents, In the other half it results from a spontaneous mutation. An affected parent has a 50% risk of passing on the gene to offspring.

The diagnosis of NF1 is usually straightforward. The gene responsible is known, but diagnosis is usually made on clinical grounds. Detailed diagnostic criteria are listed in our NF1 Clinical Management Guidelines. In the majority, the diagnosis can be confirmed by 5 years of age, or earlier if there is an affected parent. If there are no signs by 5 years, the diagnosis can usually be excluded.

The Role of the Regional Genetics Service

A clinical geneticist is usually involved in the diagnosis of NF1. They also have a role to play in ensuring patients receive appropriate follow up including genetic counselling, and education for patients and their families. Follow up may also be carried out by paediatricians, other hospital consultants or GPs. The clinical geneticist may also act as a co-ordinator where a number of different doctors and specialities are involved. These services may also be extended to other at risk family members.

Warning Signs in NF1

NF1 is variable and unpredictable, and the majority of people affected will have skin signs only. However, there are more serious (and rare) complications, that require investigation and referral to an appropriate specialist.

CONSIDER REFERRAL TO AN APPROPRIATE SPECIALIST
If there are neurological symptoms, painful or enlarging neurofibromas, hypertension, or any other unexplained or persistent symptoms including pain.

Advice and Further Information

Clinical Management guidelines for NF1 are available on request from the Regional Genetics Service or can be downloaded from our website www.mangen.co.uk

If you are still unsure about any aspect of caring for someone with NF1, you can telephone the Regional Genetics Service or visit our website for specialist advice.

NF1 CLINIC DETAILS

Genetics Service should attach a printed sticker here with clinic address and contact details.

Information for Education Professionals

How can NF1 affect learning ability?

Neurofibromatosis type 1 (NF1) is one of the more common genetic conditions that may have an impact on the way children and adults learn and perceive the world in which we live. The majority of children with NF1 have an intelligence that falls within the normal range, however a significant proportion (>60%) experience difficulties that fall within the Specific Learning Difficulty spectrum. Severe learning difficulty in NF1 is unusual (<8%)

A specific learning difficulty can be indicated when performance falls significantly below a child's general intellectual ability. For children with NF1, performance may fail to reflect their general intelligence. This can cause difficulties with acquisition of language, reading, writing, listening and mathematics.

What kind of problems could there be?

Children with NF1 can have problems with concentration, co-ordination affecting both fine and gross motor skills, memory, visuo-spatial skills, organisation and planning, sequencing and processing. Speech and language problems have also been identified. Day to day performance can be erratic and inconsistent, so progress made one day is lost the next making it difficult to build on fundamental concepts.

A proportion of children with NF1 demonstrate immature social skills and poor understanding of social cues. This can lead to peer friendship problems, social isolation, and over-familiarity with adults in authority.

There is an increased incidence of Attention Deficit Disorder (ADD)/ Attention Deficit Hyperactivity Disorder (ADHD) and Autistic Spectrum Disorder (ASD) associated with NF1.

The added health concerns imposed by NF1 together with the cosmetic implications the condition brings, compounded by absence from school to attend hospital appointments, can contribute to a sense of difference and uncertainty. Striving to enable a child with NF1 to achieve their full potential, in spite of these barriers to learning, is the challenge for their teachers.

What steps should be taken?

Early identification of any difficulties is critical to ensure a child does not fall significantly behind their peers. Early assessment therefore is important to enable appropriate resources from the Special Needs range of support to be made available. Strategies drawn from Specific Learning Difficulties (SpLD) techniques can be effective in helping children with NF1 to achieve their full potential.

Teachers should discuss concerns about performance and behaviour with parents and encourage appropriate referral via GP or paediatrician. Additional therapeutic services including speech therapy, physiotherapy and occupational therapy should be considered. In itself, NF1 is not a barrier to achievement and children with NF1 can make substantial progress in response to

Further information and practical teaching suggestions are available by contacting the NFA or visiting their website www.nfauk.org

NF1 Diagnostic Checklist

Your NF1 doctor will complete this page when you are first diagnosed with NF1 or at your first NF1 clinic appointment. They will update the details when necessary.

TWO OR MORE OF THE FOLLOWING FEATURES		Date	Dr's Initials
1	Six or more café au lait macules over 5mm diameter in prepubertal individuals (more than 15mm postpubertal)	<input type="checkbox"/>	<input type="text"/>
2	Two or more neurofibromas or one plexiform neurofibroma	<input type="checkbox"/>	<input type="text"/>
3	Freckling in the axillary or inguinal regions	<input type="checkbox"/>	<input type="text"/>
4	Optic glioma	<input type="checkbox"/>	<input type="text"/>
5	Two or more Lisch nodules (iris hamartomas)	<input type="checkbox"/>	<input type="text"/>
6	A distinctive osseous lesion such as sphenoid dysplasia or thinning of the long one cortex with or without pseudarthrosis	<input type="checkbox"/>	<input type="text"/>
7	A parent or child with NF1 by the above criteria	<input type="checkbox"/>	<input type="text"/>

Note to Doctors: If the patient's NF1 skin features are limited to one or more body segments they may have segmental/mosaic NF1. If an adult has no neurofibromas consider referral for genetic testing for the mild NF1 variants.

Genetic Testing Yes No Date of testing Result

A Summary of How NF1 Affects Me/My Child

Your NF1 doctor will complete this page. It is a summary of how NF1 affects you or your child. Your doctor will check that this page is up to date at your NF1 review appointments and fill in any positive findings. There is additional space on the next page for your doctor to make more detailed notes if necessary.

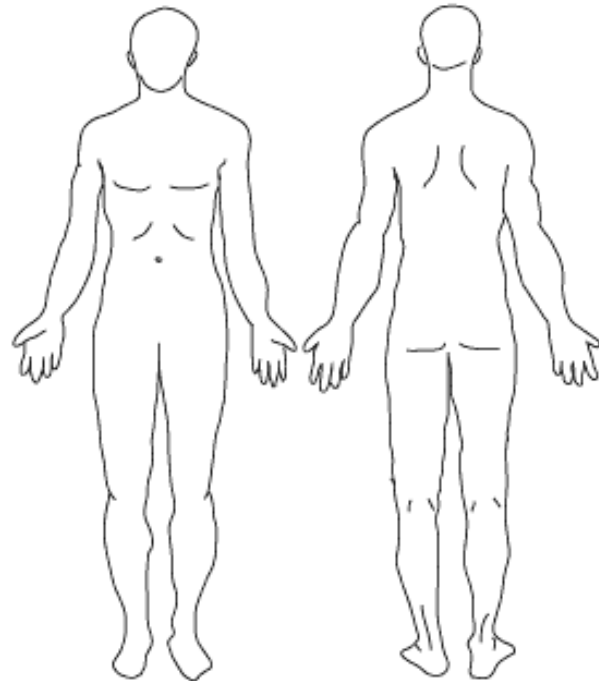
NF1 Related Features	Date & Initials	Comment	NF1 Related Features	Date & Initials	Comment
Cutaneous Neurofibromas			Nodular Neurofibromas (On major peripheral nerves)		
Plexiform Neurofibromas			Other Skin Features (E.G. Xanthogranuloma)		
			Scoliosis		
Learning/behavior problems			Other Orthopedic Issues		
			Visual Problems		
Other NF1 Related Issues			Neurological Problems		
			NF1 Related High BP		
Other Health Issues NOT NF1 Related					
Medication Details					

A Summary of How NF1 Affects Me/My Child... more details

Your doctor can use this space to make additional notes.

Please highlight any areas affected with complications and/or areas to be monitored. E.g. large areas of cafe au lait pigmentation.

Notes:



Please initial & date all entries

NF1 Review Appointments

Sheet Number

This page will be completed when you visit your NF1 Doctor for an appointment. Complications in NF1 are uncommon, but it is important for you and your doctor to be aware of the warning signs, and to carry out regular checks so that any potential problems are identified early. At this appointment, the doctor will carry out an examination and review any symptoms.

Date Doctor

Clinic Location

BP =	Ht = ()	Wt = ()	Hc = ()
------	----------	----------	----------

Doctor's Comments:

Next Review Due

Date Doctor

Clinic Location

BP =	Ht = ()	Wt = ()	Hc = ()
------	----------	----------	----------

Doctor's Comments:

Next Review Due

Details of Other Hospital Visits and Appointments

Sheet Number

People with NF1 may be under the care of several specialist doctors and health care professionals. This page provides space for them to record brief details about the appointment and will be a record of who you saw and why.

When (Date)	Where (Hospital/GP etc)	With Who (Name)	Doctor's Comments

Details of Other Hospital Visits and Appointments

Sheet Number

People with NF1 may be under the care of several specialist doctors and health care professionals. This page provides space for them to record brief details about the appointment and will be a record of who you saw and why.

When (Date)	Where (Hospital/GP etc)	With Who (Name)	Doctor's Comments

Sources of Information

Neurofibromatosis Association

The Neurofibromatosis Association

Quayside House, 38 High Street, Kingston on Thames, KT1 1HL

Helpline: 0845 602 4173

Website: www.nfauk.org

The NF Association is a registered charity that provides help, support and advice to those affected by either form of Neurofibromatosis, their families and the professionals working with them. To do this they employ a team of hospital based professional Neurofibromatosis Specialist Advisors around the UK. They provide relevant, up to date information on both forms of Neurofibromatosis for professionals, families and individuals and also fund research. The Association provides activity holidays and breaks for children and adults, and encourages the establishment of local groups and volunteers throughout the UK. They also seek to raise awareness of the NF1 amongst the general public through their fundraising activities and website.

Contact A Family

Helpline: 0808 808 3555

Website: www.cafamily.org.uk

The Contact a Family website is for families who have a disabled child and those who work with them or are interested to find out more about their needs. Contact a Family is the only UK charity providing support and advice to parents whatever the medical condition of their child. They have information on over 1,000 rare syndromes and rare disorders and can often put families in touch with each other.

Internet

The internet contains a wealth of information about medical conditions and treatments. Users need to be confident that the information they read on the internet is reliable and accurate. The Judge Health website **www.judgehealth.org.uk** has guidelines that aim to help you make informed decisions about Web sites and gives advice on how to search the internet for health information.

My Notes

You can use this page for any other information you wish to remember or record. For example, use it to write down any questions you want to ask at your next appointment. Ask your NF1 Doctor for more sheets if you need them.

A large, empty rectangular box with a thin black border, intended for the user to write their notes. It occupies the majority of the page's vertical space below the introductory text.

The development of this document has been a truly collaborative process. We would like to thank everyone who has contributed by sharing his or her experiences and knowledge especially:

Maggie Ponder

Diana Baralle

NF Association

North West Regional Genetics Service NF1 Consensus Group

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North West Regional Genetics Service: 0161 276 6506 www.mangen.co.uk

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