PCHR insert for babies and children diagnosed with Neurofibromatosis Type 1

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Introduction

This is an additional insert for your child's Personal Child Health Record book (PCHR), which is issued to all new babies in the UK. These extra pages have been produced by The Childhood Tumour Trust, in collaboration with Dr Susan Huson (CMFT), Dr Carly Jim (MMU), Dr Shruti Garg (CMFT) and Dr Ellie Paul, for babies and children who have been diagnosed with Neurofibromatosis Type 1 (NF1). The pages give additional information that aim to help you and health professionals monitor the health of your child. Babies and young children with NF1 have just the same needs as any other child, but they may also have additional requirements. You should take your child for routine health checks and immunisations in the usual way (see main PCHR). It is not possible in this small booklet to cover all topics relevant to your child's health and well-being. Your local healthcare team, or one of the agencies listed at the end of this insert will be able to provide more information should you require it.

Neurofibromatosis Type 1 – Health problems

Babies and children with NF1 should have some extra health checks (see next section) so that if there is a problem it can be identified, examined and if possible treated as quickly as possible. There is no such thing as a typical child with NF1. Children with the condition are as different from each other as are all children, however NF1 brings with it certain medical complications. A child with NF1 is at increased risk and therefore requires further monitoring: these are listed in the following pages. It is not possible for this booklet to cover all the conditions which may occur more frequently in children with NF1. Those discussed above are some of the most common. Any concerns you may have about your child's health should be discussed with your local health professionals.

Neurofibromatosis Type 1 – Definition and diagnostic criteria

NF1 is a genetic condition which occurs in approximately 1 in 3000 people, half of these will be new mutations and half will have inherited it from their parents. NF1 is a condition which causes tumours to grow on nerve endings, the majority of children with NF1 will have multiple flat brown birth marks (Cafe au Laits). The diagnosis of NF1 is usually straight forward, and is based clinically on the NIH diagnostic criteria, and confirmed if necessary through blood tests. Further information about NF1 and the management guidelines can be downloaded from this website www.mangen.co.uk

Neurofibromatosis Type 1 – Extra child health checks

The next page describes the extra health checks which are needed for your child and the age these should be carried out. Details are given about the actual tests or procedures advised. Your GP or Health Visitor will be able to explain these to you. Different areas organise their child health services in different ways so the schedule will not always be followed precisely. However if you think that your child has missed out on one of these checks take this book along to your GP or Health Visitor or Paediatrician and ask if you can have the checks carried out.

Neurofibromatosis Type 1 – Suggested schedule of health checks

The following are suggested ages for health checks. Check at any other time if there are parental or other concerns.

What to check	How NF1 can affect this	Age when checks needed	Action
Development	Early milestones are usually normal in NF1, if delayed the child needs assessment for non-NF1 causes. NF1 often affects fine motor skills. Review development - noting coordination and speech difficulties. Children with NF1 often have mild short stature and relative macrocephaly which only need investigating if crossing centile lines. Precocious or late puberty occur slightly more commonly on NF1	Development - mainly 0-5 years Growth and head circumference 0-16 years	Children with significant delay should be referred to child development team. Children with early/delayed puberty should be referred to a paediatric endocrinologist
Education and behaviour	Most children with NF1 have a normal IQ but can find learning harder than their peers. Coordination can affect handwriting. Up to 50% of children with NF1 have ADHD, with more concentration problems than hyperactivity. They respond well to small doses of stimulant medication. Approximately 40% of children have ASD	Learning and behaviour problems usually obvious by age 8 and won't then develop if not present.	Consider REFERRAL for professional assessment of educational needs. Seek advice from SENCO, child development team or CAMHS

Hypermobility and flat feet	Children with NF1 often have hypermobility and flat feet. This may cause them to be easily tired and to complain of aches in limbs	Usually obvious by age 5 years	REFERRAL to physiotherapist advised if hypermobility is suspected and to podiatrist for flat feet
Skin	Skin neurofibromas – usually start to develop in teenage years but can be seen in younger children. Can present with itching. Plexiform neurofibromas - these develop in around 25% of NF1 children. Early signs may be very large areas of café au lait which then become thickened, the area may have excessive hair growth	Skin neurofibromas - are more commonly seen in adults with NF1. Plexiform neurofibromas - usually present by mid-teens and if not there then will NOT develop. Doctors with a lot of NF1 experience may pick up subtle signs earlier	REFER to an NF clinic, dermatologist or plastic surgeon for advice concerning the skin, especially if increased pain or growth
Eye checks	Most NF1 eye problems present by age 8. The minimum recommendation is for NF1 children to have their vision checked once a year by a Paediatric Ophthalmologist until age 8 years. Some centres offer younger children (who won't complain about visual loss) 6 monthly checks until 4-5 years. From 8 years onwards an annual check with a high street optician is recommended but the chance of a problem is very low indeed	Most NF1 gliomas cause visual loss by age 7 years	Once visual loss is detected the child should be referred to a regional centre for assessment by a Paediatric Oncologist

Back and limbs	Scoliosis – NF1 children have an increased risk of scoliosis. The child should be examined for signs of curvature of the spine at least once a year during entire growth period. Pseudarthrosis - This presents with curvature of the long bones, usually in the lower leg. The curved bone can fracture and then not heal without special treatment	Scoliosis - if not present by end of growth spurt unlikely to develop Pseudarthrosis - if not present by time child is walking it won't develop	Any concerns re scoliosis or long bone curvature mandates referral to a specialist orthopaedic surgeon
Neurology	Neurological symptoms should be reviewed annually, particularly ataxia, headaches, loss of consciousness and visual disturbance. BUT NF1-related neurological problems may present between checks and then parents should ask for urgent assessment with their GP or NF1 doctor		Significant symptoms need assessment by paediatric neurologist or neurosurgeon as appropriate

Neurofibromatosis (T1) – RECORD OF HEALTH CHECKS

Date	Age	Type of test	By whom and where	Result

Notes