Role of Health Visitors in identifying signs of Neurofibromatosis type 1 (NF1)?
• My name is Dr Carly Jim I am a Senior Lecturer at Manchester Metropolitan University (MMU) and one of the Trustees of the Childhood Tumour Trust (CTT).

• Together we have a joint campaign to improve earlier diagnosis of Neurofibromatosis type 1 (NF1).

• We believe health visitors have a crucial role to play in the earlier diagnosis of NF1.
What is NF1?

- NF1 is a relatively common genetic condition with an estimated birth incidence of 1/1700 for both sexes and all races (Evans et al., 2010); and prevalence of 1/2500 (Ferner et al, 2007).

- It is a tumour-prone condition that is associated with the risk of developing a number of different complications, these include:
- Tumours growing along nerves on the skin and inside the body
- Bone Deformities/Scoliosis (eg Curvature of the spine)
- Sight Problems/Blindness
- Hypermobility/Mobility Issues
- Cancer
- Learning Difficulties (eg Dyslexia)
- Social Skill Difficulties (eg Autistic Spectrum Disorder)
- Attention and Impulsivity (eg ADD/ADHD)
- Coordination Difficulties (eg Dyspraxia)
- Disfigurements
Despite how common NF1 is, many GPs, Midwives, Health Visitors and Paediatricians, have little awareness of NF1 as a consequence.

missed or delayed diagnosis is common
As people with NF1 could potentially develop varied complications, we at the Childhood Tumour Trust believe that it is essential to be diagnosed as early as possible and hope that with appropriate surveillance prognosis is likely to be improved.

Additionally identification of NF1 prior to producing offspring allows for informed reproductive decisions something which is crucial given the 50:50 transition of NF1 and the chance off-spring may be more severely affected (Huson, 2008).
So How Can Health Visitors Help?

The most frustrating thing about this missed diagnosis is that the hallmark feature of NF1 is a somewhat obvious flat brown birthmark called a Café au lait (CAL)

because birthmarks are common their significance is missed, but the key issue is the quantity, 95% of children with > 6 CAL greater than 0.5cm will have NF1 (Korf, 1992).
Café au Lait Marks cont.
But what if it's not NF1?

- Obviously not all birth marks are CALs and not all CALs mean that a person has NF1, but with a 95% chance that 6 or more CALs does indicate NF1 coupled with the fact that even if it's not NF1 multiple birthmarks might indicate a different genetic condition.

  95% chance that 6 or more CALs does indicate NF1

we believe that health visitors have a responsibility to ensure a referral to a genetics specialist.
Count the CALs

- At CTT our message is very simple:
  - if you can count to more that 6 and
  - if you can distinguish brown from other colours than you are capable of recognising one of the key diagnostic features of NF1.
  (its so simple even a child can do it, and no need for you to do invasive or expensive tests)
- As a further check if you can also ascertain if either of the parents has the CAL marks then that’s two diagnosis criteria and NF1 is almost certainly going to be confirmed by the specialist.
Count the CALs

- As a health visitor you often see the child fully undressed (e.g. at a baby weigh clinic) and this allows for a visual assessment, additionally you can discuss with the parent the child’s skin on a routine visit for something else.

- And if you want to help even more please share the poster with anyone you think could help identify the CALs and support our campaign for the logging of CALs in the Child Health Record (Red book)
COUNT THE CALs

- CALs (Café au lait patches) are flat brown birth marks. They can appear anywhere on the body.
- 10% of people have a couple of CALs but 95% of children with 6 or more have the genetic condition Neurofibromatosis (NF1).

- NF1 is extremely variable and some people may need specialist care.
- The incidence of NF1 is the same for both sexes and all races.
- Count the CALs and refer to genetics if there are 6 or more (> 0.5cm).

NOTICE
NEUROFIBROMATOSIS
More Information

- For more information on the Childhood Tumour Trust and our campaigns please visit our website and Facebook page.

- [http://www.childhoodtumourtrust.org.uk/](http://www.childhoodtumourtrust.org.uk/)

- [https://www.facebook.com/childhoodtumourtrust.org.uk](https://www.facebook.com/childhoodtumourtrust.org.uk)
References


