



# UNDERSTANDING Congenital Melanocytic Naevi



Written by

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in association with

**Caring Matters Now**



# What is a CMN?

*A Congenital Melanocytic Naevus (or CMN) is a type of birthmark. It is also a mole.*



## **Congenital**

= present at birth.



## **Melanocytic**

= to do with melanocytes.

*Melanocytes are cells in everyone's skin and hair that produce the pigment melanin. This is what gives us our hair and skin colour. The cells in a CMN look most like melanocytes and they produce pigment, so they are called melanocytic.*



## **Naevus**

= birthmark

*(the plural is naevi).*



Though CMN means birthmark or mole at birth, in fact, not all of them are present at birth.

Around 1% of CMN appear after birth, usually in the first year of life. They look exactly the same as the ones present at birth, but are called CMN "tardive", which means "late".

### **How often do CMN occur?**

Single small CMN are found in 1% of all newborn babies. However, large or numerous CMN are rare, occurring in around 1 in 20,000 births. The frequency seems to be similar around the world and amongst different populations.



# What does a CMN look like?

*Almost every CMN looks different to the next one.*

## Site

CMN can be on any part of the skin, including palms, soles and scalp, but also sometimes inside the mouth.

## Colour

The colour is usually some sort of shade of brown to black, but sometimes they can be more reddish. The colour is often very mixed inside the CMN, with smaller areas of different colour on a background colour. CMN at birth are often black, or even dark purple/red.



Large CMN with different colours within it. There are some open areas which can sometimes happen at birth but then heal.

## Hairiness

CMN usually have hair growing out of them, although this is often not visible at birth and can remain unnoticeable throughout life. The hair colour can be darker than the child's head hair, or the same, or very occasionally lighter. If a CMN is on the scalp there is usually more luxuriant hair growth over the lesion and the hair grows faster than on the rest of the scalp. Occasionally, CMN are completely hairless, even ones that occur in the scalp, or they can have patchy hair loss or hairs without pigment (like grey hair).

## Texture

The texture of large CMN tends to be different from that of normal skin, being softer, looser and more wrinkled. The CMN can be nearly flat or can be very folded or lumpy.



Flat texture on the back.

## Spontaneous lightening of CMN

In most children the CMN will lighten to some degree in the first few years of life. This is still being studied but it appears to be more prevalent in children with light or red hair and pale skin than in those with darker hair and skin colour. In some children the lightening is very dramatic and in very rare cases the CMN can disappear or stop producing pigment.



Particularly dramatic lightening in a CMN.

# Problems with CMN

## **Fragility**

Many CMN are more fragile than normal skin and can therefore tear more easily if they are knocked or scraped. However, they do not bleed any more than normal skin, and they tend to heal well with minimal scarring.

general skin surface and the limb, buttock or side of the face appearing obviously thinner than normal. The thinner area in practice functions just as normal – for example a thinner leg still works perfectly well.



Lumpiness in a CMN on the hand.

## **Dryness**

Some CMN are drier than the surrounding skin so needs to be washed without soap, and moisturised regularly (your doctor can advise you on this). Occasionally, CMN are very itchy which can be due to dryness or eczema in the CMN, or sometimes for no apparent reason. This itchiness can usually be treated using creams and ointments.

## **Underlying decrease in fat and muscle bulk**

This is usually only seen with larger CMN, particularly those on the buttocks or limbs. The presence of the CMN interferes with the development of the layer of fat and sometimes also the normal muscle bulk. This can result in the CMN appearing to be depressed below the



The right thigh is noticeably thinner than the left, due to less fat under the CMN in that area.



# The size and growth of CMN

CMN almost always grows in proportion to the child. In other words, CMN will usually continue to cover the same area of skin as at birth.

The eventual size of a CMN can therefore be estimated from the size at birth in proportion with the body covered. *The exception to this rule are "tardive" CMN, as these appear after birth and can therefore appear to grow and darken in colour in the first few months.*

The total number of naevi can increase after birth, although this does not always happen. It is more likely to happen in children with very large and multiple CMN, and new naevi are always relatively small.

## CMN classification

CMN are classified according to the size they will be in adulthood (known as "projected adult size"). This is a difficult definition to understand because different parts of the body grow at different rates, but it is used to give doctors a way of comparing different CMN. We no longer define CMN as small/medium/large/giant, but by the size in adulthood of the largest one and the total number of naevi. Usually, but not always, these two measures go together – in other words very large CMN are usually accompanied by lots of other naevi. These other naevi used to be called "satellites", but now we consider them to be smaller CMN. The classifications we use at the moment are projected adult size of <10cm, 10-20cm, 20-40cm, 40-60cm, >60cm.

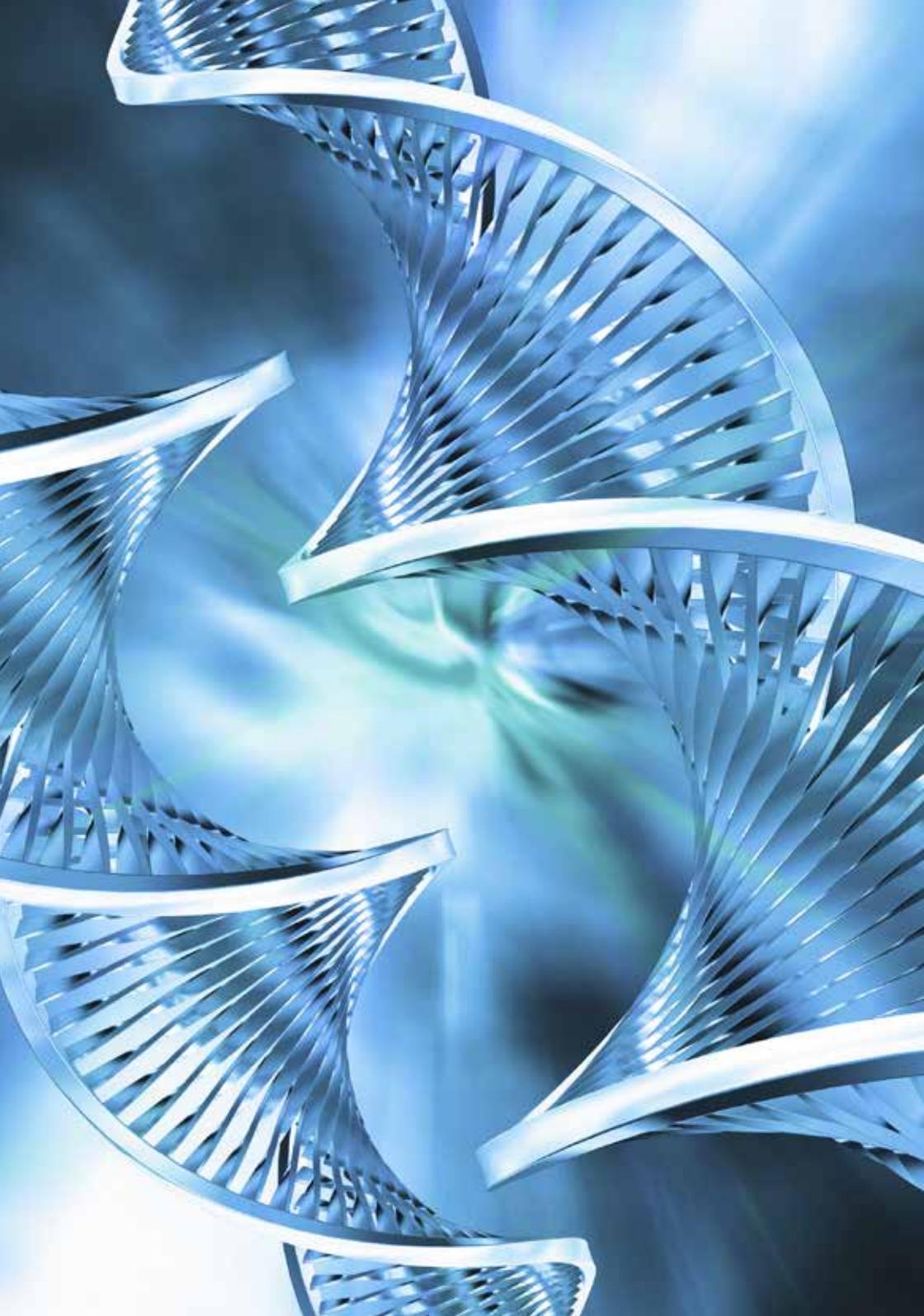
The total number of other naevi is classified as 0, 1-10, 10-20, 20-50, 50-100, 100-200, 200-400, >400.

Where the numbers are small we tend to count them, but for large numbers we estimate.



Multiple CMN lesions of different sizes, colours and textures.





# What causes CMN?

*CMN are caused by a change in a gene which happens to the baby while it is developing in the womb.*

A change in a gene is called a mutation.

A mutation means that there has been a spelling mistake in the gene, and a gene is just one of the instructions in our body. Mutations happen all the time when babies are being formed, and during our lifetimes, but many of them are completely unimportant. If, however, a mutation happens in an important gene, then it can have a big effect. The most common gene to cause CMN is called **NRAS**, which is the cause in about 70% of cases of CMN. NRAS is a very important gene and the mutation in CMN is a very important place in NRAS. The only other gene currently known to cause CMN is called BRAF, which is the cause in about 6% of cases. In the other cases the gene

is still not known. There doesn't seem to be any differences in problems for the patient caused by having different genes.

The mutation is not inherited from either of the parents but happens only to the baby during development. If the mutation happens very early in the baby's development then it will probably result in a very early miscarriage. If it happens a bit later then the baby can survive, but will have CMN, and may have the characteristic facial features and/or brain problems.

The mutation in the gene does not happen because of something the parents did or didn't do, it is just due to chance.





# CMN Syndrome

*Our recent research has led to a proposal of the term **CMN Syndrome**.*

## **CMN syndrome**

A syndrome is a collection of findings in one individual that can potentially be explained by the same genetic change. We believe the term **CMN syndrome** is useful to alert healthcare professionals to aspects other than the skin. Apart from the neurological association the new association is as follows:

### **Characteristic facial features**

Our research has found that most children with CMN have a similar looking face. This is a normal face, and in fact usually a very attractive face, but it is similar to other children with CMN. This is likely to be caused by the same mutation that causes the skin changes and the brain changes (when there are any).



## **Increased rate of weight gain in children with CMN**

This has been found in a large group of patients at Great Ormond Street Hospital (GOSH), but it is an average finding. In other words, we found that on average the children with CMN gained weight faster than those without, but this did not apply to them all, many have normal weight gain. It is not yet clear what causes this weight gain, but it seems to respond to the usual recommendations of eating more healthily and increasing the amount of exercise.

It is important for parents to ensure that their children do not become overweight in childhood, as it is a very difficult problem to address in teenage and adult years. It can be very difficult for an individual with CMN to cope with being overweight. In addition, being overweight in adulthood is bad for the individuals overall health.

## **Subtle hormonal problems with CMN**

Some children with CMN have early breast development. This has been found to be mild and harmless, and does not usually need any treatment. It also does not seem to lead to early puberty. It is however always sensible to inform the child's doctor. Undescended testes also seem to be an associated problem in boys with bathing trunk distribution CMN (in the shorts area). These need to be reviewed by a urologist to discuss whether they need to be surgically corrected.

# Complications of CMN

## **Neurological problems (problems with brain and/or spine)**

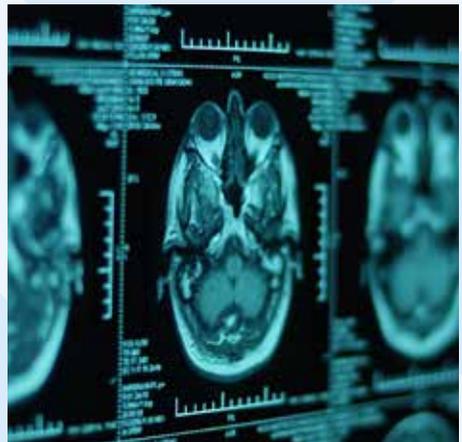
Neurological problems associated with CMN used to be called *Neurocutaneous melanosis*. This term is no longer used for two important reasons. The first reason is that some of the problems are not actually “*melanosis*”, in other words they are not a problem of pigment cells. The second reason is that in the medical literature *neurocutaneous melanosis* is often actually melanoma of the brain or spinal cord, and this has caused big problems with trying to advise families on diagnosis. We now prefer to call the neurological problems by their actual names – so if it is benign *intraparenchymal melanosis* (e.g. moles in the brain) we say so, and if it is melanoma of the brain or spine we say so. This allows us to give better information to parents and patients, and has allowed us to divide the causes into serious and less serious.

Overall, problems in the brain or spinal cord are the most common complication seen in children with CMN. The most common problem is pigment-containing cells (like a CMN) in the substance of the brain. This is called *intraparenchymal melanosis*. Other rarer problems include benign brain or spinal tumours, too much fluid in the brain, or abnormal brain structure. All of these neurological problems are more common with larger and more numerous CMN. Our current recommendations are that any child born with two or more CMN should have a routine MRI scan of the brain and spine, preferably by the age of 6 months.

The overall chance of finding an abnormality on an MRI scan in children with multiple CMN (two or more at birth) is around 20%, but only around

half of these children will have any actual problems. If they do have problems these can be fits (convulsions), developmental delay, or problems with their limbs. It is possible to have problems in development even when the scan is normal, but these tend to be milder. The reason for doing the scan is to pick up the rare cases of tumours and extra fluid on the brain that require an operation, and to allow us to monitor development more carefully in children with MRI findings. In addition, some of the findings alert us to monitor the children more carefully for the development of melanoma, by having more MRIs. Most children however will only have one scan.

Not only do we recommend this single screening scan, but anyone with multiple CMN who develops new problems with development or fits or limbs, or persistent headaches over a long period, or any problems which could be due to brain or spine disease should have a repeat MRI to look for the development of melanoma.





## Melanoma

Melanoma is a cancer of melanocytes, the skin cells which produce pigment and which make up CMN. It used to be thought that melanoma was very common in people with CMN, but we now know that overall it is rare, occurring in around 1-2% of all people with CMN over their lifetime. However, the risk is higher in people with brain or spine disease found on an MRI scan after birth (in these cases around 10%), and there is a peak of risk during childhood. When considering these risks, you need to bear in mind that every one of us has a risk of about 50% of developing some kind of malignant tumour at some time in our lives, and in the UK we estimate the risk of melanoma in the normal population at about 0.5%.

Melanoma in children with CMN can occur anywhere, not just within the CMN. It appears to be more common in the brain/spine rather than in the skin, at least in childhood. This may be different in later life. In the skin it can present as a lump in the CMN, in another area of skin, or in the lymph nodes (which are throughout the body). In the brain or spine, it can present with persistent

headaches with nausea/vomiting, or visual disturbances, balance problems or fits. Unfortunately, most often it seems to be the case that when melanoma occurs in a person with a CMN, it is aggressive and difficult to treat. Our recommendations are that any rapidly changing area in the skin should be reviewed by a Paediatric Dermatologist or Dermatologist, so that they can decide if a biopsy is required. Do remember however that lumps and nodules can appear in CMN quite frequently, and the majority are not melanoma. We also recommend that people who develop new neurological problems such as recurrent headaches, visual disturbance, fits, or developmental changes should be seen by a doctor and an MRI of the brain and spine carried out.

Knowledge of the NRAS and BRAF gene mutations has started to help in the treatment of melanoma arising in people with CMN. If melanoma is suspected these gene mutations can be tested for and that can help direct treatment.



# Complications of CMN

## Sun protection

Children with CMN can and should enjoy a normal life. They should be protected from the sun in the same way we would recommend for all children regardless of whether they have CMN or not. In particular, it is important that they should not become sun burnt.

As a general rule sun avoidance, coupled with good clothing protection, are much more important than sunscreen.

## Sun avoidance

Children should be kept out of direct sun as much as possible during the hottest hours of the day and in the hottest months of the year, for example, by staying in the shade. The hottest hours of the day are 10am-4pm, and the hottest months of the year are April to October. Outside these times there is usually no need for sun protection in the UK. This does not mean children should not go outside between 10am-4pm in the hot months, it just means they should stay in the shade where possible and wearing appropriate clothing.

## Good clothing protection

Sun hats with a brim that covers ears and the back of the neck as well as the face are ideal, as are longer sleeved tops and longer shorts/skirt.

Sunscreen is not a substitute for sun avoidance and good clothing protection. It should, however, be used as extra protection if the child has to be in direct sun during hot periods for areas that are not protected by clothes. The best sunscreens contain a reflectant barrier such as titanium dioxide; select a high protection factor (SPF), ideally 30 or more (or 50 or more for babies), and with high UVA protection (5 stars, usually on the back of the bottle). Sunscreen needs to be

refreshed every 2 hours or so, more often when swimming or sweating.

Other important points to remember:

- UV exposure is much higher when beside water or snow, so special care should be taken when swimming outside or skiing.
- The sun is more harmful at higher altitudes.
- The sun remains almost as harmful when it is cloudy, so even if the day is cloudy the child should wear appropriate clothing such as a sunhat during the hottest times of the year.
- Shade provides less protection when near water or snow or when the weather is cloudy.
- The increased risk of skin cancer in children with CMN probably applies to their whole body, not just on their CMN.



# Can CMN be treated with laser or surgery?

*We think that the decision to have surgery has to be made on an individual basis, and very much depends on whether the plastic surgeons think they can improve the appearance.*

*In cases of very large CMN, surgery is often not possible.*

*In other cases the following points should be considered:*

- 1** Many CMN will lighten spontaneously to at least some degree over a period of years. This can be monitored with repeat photographs.
- 2** Surgery has not been shown to reduce the risk of melanoma in the child.
- 3** Early surgery has not been shown to be advantageous. We do not do any routine surgery before 1 year of age.
- 4** The site of the CMN is very important – for example, the child may get more benefit if a CMN on the face is removed, compared to one hidden in the scalp.
- 5** The size of the CMN is very important – we have found that children with larger CMN were less pleased with the cosmetic result than those with small lesions which could be completely removed.
- 6** The number of naevi is important, in particular if the child has a tendency to develop lots of new ones, as this may reduce the benefit from removing some.
- 7** Whether you want your child to take part in the decision, in which case it is better to decide later.
- 8** What is involved in the type of surgery being offered – this will depend on the individual case.



If a CMN can be removed, for example by excision or serial excision (more than one operation but relatively straight-forward), the cosmetic benefits may easily outweigh the small risks associated with any operation. However, if a CMN is in a difficult place for removal, or if it is too large to ever be removed completely, then that balance changes. It is very important in these cases to take time to decide about surgery, particularly to see if the CMN is lightening over time.

Laser therapy cannot be used to treat CMN. It will often lighten the colour, but this is a temporary phenomenon, and the CMN will gradually (or sometimes rapidly) grow pigment again. Sometimes it appears that the colour after repigmentation is lighter than it was at birth, but we now know that this is because in that individual the CMN was going to lighten anyway, and the final colour is connected to the person's own hair and skin colour, not to the colour it was at birth (or to any longterm help from laser). The same thing applies to dermabrasion or curettage, which are other superficial removal techniques.

### What can we do about hairiness?

Hairiness only needs to be treated if it is causing a problem (e.g. making an area difficult to clean) or the child or family feel it looks better without hair. An electric shaver is best, and clipping the hair short rather than completely shaving often avoids problems of itchiness with regrowth. Hair removal creams etc should not be used as they can irritate the skin. Shaving does not affect the amount or thickness of hairs that grow. Hair regrowth after shaving is generally slow and the new hairs will have exactly the same appearance and feel as the original ones. Most parents find that they do not need to shave an area more often than once every couple of weeks in order to maintain a satisfactory appearance. Laser hair removal is possible but needs to be done regularly over a relatively long period and needs a general anaesthetic in children under teenage years. Electrolysis is slow and painful and is therefore not suitable for large areas.

### Psychological issues

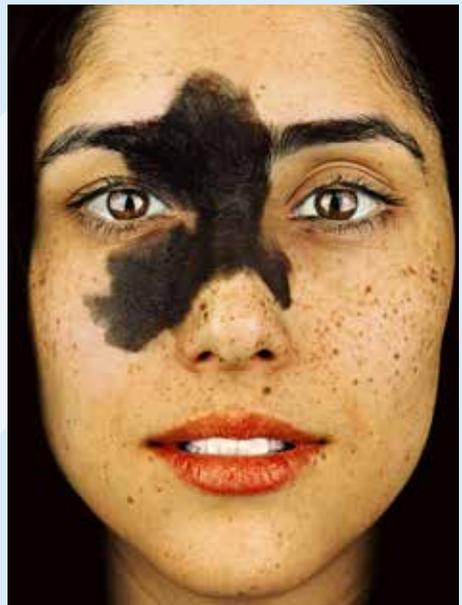
Children who grow up with a prominent CMN or many CMN may well have problems adjusting to their visible difference, particularly during teenage years. This, however, is a very individual thing, and varies depending on the child's personality, and on the support from family and friends.

It would be ideal if we were able to offer all children at risk some degree of automatic counselling during their childhood. At GOSH we currently have a psychology service available. However, not all families want psychology input, and not all hospitals have this facility.

We actively promote support group involvement for both parents and children, as well as recommending psychological support before transition to secondary school, and learning key techniques for coping with visible difference. For example see:

[www.changingfaces.org.uk](http://www.changingfaces.org.uk)

[www.ypfaceit.co.uk](http://www.ypfaceit.co.uk)



# Support and Research

## Support Group

Caring Matters Now is a registered charity and support group specifically for people with CMN and their families. Their aims are:

- To support those affected by CMN
- To raise awareness about CMN
- To raise funds for the CMN research

Support is provided in many different ways. The group has a dedicated telephone line, a network of trained support contacts, hold family support days and activity weekends, use social media and manage online forums to give the opportunity to chat, share experiences and seek advice. They also have a very informative website

[www.caringmattersnow.co.uk](http://www.caringmattersnow.co.uk)

They use these sources to spread news of the research findings and developments.

Caring Matters Now also coordinates a voluntary fund-raising programme to contribute significantly to the research into CMN at Great Ormond Street Hospital in London.

## CMN Research

Dr Kinsler is conducting long-term research into CMN at Great Ormond Street Hospital. If you would like to be involved and are not attending GOSH regularly, please ask your doctor (GP, Dermatologist or Paediatrician) to refer your child for a one-off appointment in order to participate in this research. The aims of this research are to understand what causes CMN, to identify those patients at highest risk of neurological and melanoma complications, and ultimately to design better treatments for CMN.

## Disability Living Allowance

Parents of children with extensive CMN or NCM may be eligible for Disability Allowance, but in general this is only for children with neurological problems or melanoma. If you would like further advice, contact your Citizen's Advice Bureau or the Social Services Department in any hospital your child visits. Alternatively, there is more information at [www.gov.uk](http://www.gov.uk).







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