



UNDERSTANDING Congenital Melanocytic Naevus



Written in collaboration with Professor Veronica Kinsler
Great Ormond Street Hospital for Children, London

Introduction

Caring Matters Now is the only dedicated charity in the UK & Ireland supporting children and adults affected by Congenital Melanocytic Naevus (CMN).

Our 3 main aims are:

To support those affected by CMN

To raise awareness about CMN

To raise funds for the CMN research

Foreword by Jodi Whitehouse, Caring Matters Now Chief Executive Officer



This CMN medical booklet aims to provide healthcare professionals with accurate information about Congenital Melanocytic Naevus, helping to ensure everyone affected by CMN receives the correct diagnosis and ongoing medical care.

The medical information provided has been written in collaboration with Professor Veronica Kinsler, lead CMN consultant at Great Ormond Street Hospital for Children and Head of the CMN research lab at the Francis Crick Institute, London.

Caring Matters Now has been working in partnership with Professor Veronica Kinsler for over 20 years with one unified aim: developing effective treatments for children and adults affected by CMN. As a charity, we have donated over 1.8 million pounds to the CMN research led by Prof. Kinsler. The cause of CMN was found and more recently, the research team has developed an effective treatment for CMN. Subject to raising sufficient funds, we hope the treatment will then enter clinical trials in the near future. More details can be found on page 21.

To compliment the medical information, we have also included in this booklet details of the Support Pathway Caring Matters Now provides to children and adults affected by CMN. Our aim is to equip healthcare professionals with an understanding of the psychosocial support available to their patients affected by CMN, including how to signpost patients to the most appropriate support.

If you would like to reach out to Caring Matters Now for any additional information, please email info@caringmattersnow.co.uk

We hope you find this information booklet a helpful resource.

Jodi Whitehouse
Caring Matters Now CEO

What is CMN?

Understanding the terminology

Congenital means present at birth. **Melanocytic** is 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 simply means birthmark (the plural is naevi).

Type of birthmark

CMN is a brown birthmark, but 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 they occur?

How often CMN occur depends on the severity of the CMN. Single small CMN are very common, found in 1% of all newborn babies. At the other end of the spectrum, extensive or numerous CMN are much rarer, occurring in around 1 in 10,000-20,000 births. We estimate in the UK there are around 6000 babies born per year with a small single CMN, and around 50 babies born with the most extensive/numerous CMN.

What does a CMN look like?

Almost every CMN looks different.

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 a shade of brown to black, but sometimes they can be more reddish. It 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.



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 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.

Spontaneous lightening of CMN

In most children, the CMN will lighten to some degree in the first few years of life. This is now known to happen more in children with blonde 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 extremely rare cases the CMN can disappear or stop producing pigment altogether. Scalp CMN tend to lose their colour the most.





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.



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).

Itchiness

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, and it seems to get better with age.

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 muscle bulk that is normally present. This can result in the CMN appearing to be depressed below the general skin surface, or the limb, buttock, or sometimes the 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 well.



Register with Caring Matters Now as a medical professional to receive an annual email with the latest research findings, which will help you to provide the most appropriate care for patients affected by CMN.

Scan the QR code to register today.





Size and Growth of CMN

CMN almost always grow in proportion to the child. In other words, they will usually continue to cover the same area of skin as at birth. The exception to this are “tardive” CMN, as these appear later and can therefore appear to grow, or occasionally where part of a CMN is very pale, it can darken up in the first few months and appear to grow. The size that a CMN will end up can therefore be estimated from the size at birth, by looking at its size and where it is positioned. 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 have traditionally been 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, and it is currently being revised. Usually, but not always, the size of the largest naevus and the total number of naevi 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 that have traditionally been used are projected adult size of <10cm, 10-20cm, 20-40cm, 40-60cm and >60cm. The total number of other naevi is classified as 0, 1-10, 10-20, 20-50, 50-100, 100-200, 200-400 and >400. Where the numbers are small we tend to count them, but for large numbers we estimate. There are also different classifications which sometime may be available.

Caring Matters Now has a
Support Helpline for anyone
requiring information about CMN.

0300 373 3422

Sun Protection

People with CMN can and should enjoy a normal life. However sun protection is important for everyone, and possibly more important for people with CMN. As a general rule sun avoidance and good clothing protection are much more important than just relying on sun cream.

Sun avoidance

Sun avoidance is the most important way to protect against the sun. In the UK the hottest months of the year are April to October and the hottest hours of the day are 10am - 4pm. Outside these hours there is usually no need for sun protection. If people do go outside within these hours, they should stay in the shade where possible, wear appropriate clothing and use sun cream (See page 19 for details on key sun cream qualities).

Although the UK doesn't get a lot of hot weather, the UV can be high; for those times of the day avoid the sun as much as possible. The shade does not prevent you from UV exposure but it decreases it, so sitting under a tree or under a parasol and walking on the shady side of the street are really good ways to minimise sun exposure.

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.
- Shade provides less protection when near water or snow or when the weather is cloudy.
- The sun is more harmful at higher altitudes.
- The increased risk of skin cancer in children with CMN probably applies to their whole body and not just their CMN.





CMN Syndrome

CMN Syndrome

Recent research has led to the proposal of the term CMN Syndrome. A syndrome is a collection of findings in one individual that can potentially be explained by the same genetic change. The term CMN Syndrome is useful to alert other healthcare professionals to aspects other than the skin. Apart from the neurological associations these new associations are as follows:

Characteristic facial features

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 they are present).



Increased rate of weight gain in children with CMN

This has been found in a large group of patients at GOSH, but it is an average finding. In other words we found that on average the children with CMN gained weight faster than the normal population, but this did not mean that all the children did so. 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 years, and very difficult for an individual with CMN to cope with being overweight as well as an adult.

Subtle hormonal problems with CMN

Some girls 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 show this to the doctor looking after the child. For boys with bathing trunk CMN (in the shorts area), undescended testes also seem to be an associated problem. These need to be reviewed by a urologist for an opinion about whether they need to be surgically corrected.

Neurological Problems

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 what exactly their child has got. We now prefer to call the neurological problems by their actual names – so if it is benign Intraparenchymal Melanosis (likes 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 commonest 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. 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 i.e. 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. An MRI scan is recommended for all children born with multiple CMN to determine the presence of any pigment in the brain or nervous system. An earlier MRI allows for a clearer scan. MRI results can be used as a framework for the child’s development, as it will determine whether the CMN is internal. 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.

Children with medical complexities linked to CMN may be eligible for Disability Living Allowance, generally this is only for children with neurological problems or melanoma. Contact your Citizen’s Advice Bureau, Social Services in your local hospital or visit www.gov.uk for more information.

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.

Melanoma in children with CMN can occur anywhere, not just within the CMN. It appears to be commonest 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 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.



Meet Anna

Anna was born with CMN covering 50% of her body and has CMN internally, causing medical complexities. Scan the QR code to hear Anna's parents share their journey and how Caring Matters Now has been able to provide support over the years.



Register as a Medical Professional



Pictured: Dr David Atherton who established the CMN clinic and research, Jodi Whitehouse founder and CEO of Caring Matters Now, Professor Veronica Kinsler lead CMN research clinician.

We would love to send you an email once a year to update you on the latest research findings, which will help you to provide the most appropriate care for patients affected by CMN.

You can register with us by scanning this QR code.



What causes CMN

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

In recent years, the causes of CMN have started to be understood by the CMN research team studying the genes in CMN cells through skin biopsies, donated by people with CMN. These studies have found that changes (known as mutations) in a gene called NRAS are the most common cause of CMN of any size and any number. Overall, in one big cohort it causes 67% of cases. The BRAF gene has been found as a cause for 7% of CMN, which is much less common than the NRAS mutation. In some patients, there are no spelling mistakes, but two genes get stuck together, which shouldn't happen. This phenomenon stops the gene from doing its normal function causing the skin problems associated with CMN; this is known as a BRAF Fusion. These fusions are responsible for the CMN being very itchy and forming lumps or ridges. Research has shown that adverse outcomes did not differ between genotypes of people with CMN. It did not matter if the person with CMN had BRAF or NRAS 'spelling mistake'.

Specialised CMN Clinics

The specialised CMN clinic for children is at Great Ormond Street Hospital for Children, London and for adults is at Guys & St Thomas' Hospital London.

Scan the QR code for more details .



CMN is now understood as a Mosaic Disorder. Mosaic Disorders are defined as a group by a common disease mechanism - they are caused by a genetic mutation (change, sometimes also called a genetic variant) in a single cell in the developing baby during pregnancy. This leads to the baby being born with a mixture of normal cells (not carrying the mutation) and disease cells (carrying the mutation). It is this mixture which is referred to as mosaicism. This is a random mutation and is not inherited from either parent.



The effects of the mutation depend on lots of different things. The most important ones are when it occurs and whereabouts it occurs in the body. In general, the earlier the mutation happens, the more serious the disease will be. As a result of the variability, each child with a mosaic disorder is unique in their disease and needs to be assessed in an individualised unique way. Mosaic disorders affecting the skin are currently the best known because it is possible to see the effects on the skin as birthmarks. As a result, although they often affect many body systems as well as the skin, mosaic disorders affecting the skin are looked after by Paediatric Dermatologists and Dermatologists.

Are there any effective treatments?

Currently, there is no definitive treatment for CMN. As a charity we partner with Professor Kinsler and her research team at Great Ormond Street Institute of Child Health and The Francis Crick Institute, to fund the CMN research taking place. In recent years, Professor Kinsler's team have found the cause of CMN and now the team are working towards developing effective treatments to provide choice for those affected.



Surgery

The decision to have surgery has to be made on an individual basis. In cases of very large CMN, surgery is often not possible. We recommend that a medical opinion is always sought from Professor Kinsler and her team at Great Ormond Street Hospital on whether surgery is appropriate. When considering surgery, careful thought should be given to the following points:

1. Many CMN will lighten spontaneously over a period of years, which is related to the skin and hair colour of the child. This can be monitored with repeat photographs as it is often difficult to remember otherwise. It is very important in these cases to take time to decide about surgery, particularly to see if the CMN is lightening even more over time.

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.

4. The site of the CMN is very important - in one study the improvement was considered greater if a CMN on the face is removed, compared to one hidden in the scalp or on the body.

5. The size of the CMN is very important - in one study children with larger CMN were less pleased with the cosmetic result than those with small CMN 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.

9. Nodule removal depends on both the person and the nodule so, nodule removal should be considered on an individual basis by your doctor.

Caring Matters Now has a library of free Support Literature for all ages affected by CMN. Scan the QR code to access both digital and hard copy resources from our website.





Laser Therapy

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) develop pigment again. Sometimes it appears that the colour after repigmentation is lighter than it was at birth, but it is now known 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 the CMN was at birth (or to any long-term 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 the CMN would look better without hair. An electric shaver is best to use, and hair-removal creams should not be used because they can irritate the skin. Shaving does not affect the number or thickness of hairs that grow. Hair regrowth after shaving is generally relatively slow and the new hairs will have exactly the same appearance and feel as the original ones, if they are allowed to grow long again. 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.



Good clothing

Good clothing is the second most important way to protect against the sun. Appropriate clothes during hot weather really make a difference:

- a wide-brimmed hat which provides protection to the ears, back of the neck and over the face is probably the best thing you can do for sun protection for children especially under 10, as exposure to the sun then brings the greatest risk of skin cancer in later life.
- longer sleeve T shirts should be worn instead of strappy T shirts, so that the shoulders are covered.
- if shorts are worn, choose longer legged shorts to provide more coverage to the legs.
- UV suits which provide more extensive coverage and protection are really worth while.



Sun cream

Sun cream is not a substitute for sun avoidance and good clothing protection. However, it should be used for areas that are not protected by clothing, if someone has to be in the direct sun during the hottest times of the day.

The best sunscreens contain a reflectant barrier such as titanium dioxide. Select a high protection factor - for babies SPF 50 or more,; for children SPF 30 or more - and with high UVA 5 star protection. usually on the back of the bottle). Sunscreen needs to be refreshed every 2 hours or so and more often when swimming or sweating.

Meet Ada

Ada was born with CMN covering 70% of her body. Scan the QR code to meet Ada's family and to hear how Caring Matters Now has been able to provide support throughout Ada's life.





CMN Research Advancements

In June 2024, Professor Veronica Kinsler and her CMN research team at the Francis Crick Institute and Great Ormond Street Institute of Child Health published an article in the Journal of Investigative Dermatology detailing a huge breakthrough in their CMN research work.

It is hoped this approach will form the basis for clinical trials, subject to raising enough funds. This means that in the future the treatment could potentially be used to reverse CMN, therefore reducing the risk of affected children and adults developing melanoma. It could also potentially reverse more common types of at-risk moles as an alternative to surgery.

These results are very exciting, as not only does the genetic therapy trigger self-destruction of the CMN cells in the lab, but the researchers have managed to deliver it into the skin in mice. These results suggest that the treatment in future could potentially reverse CMN in people, however more testing will be needed before it can be given to patients.



The research team have designed a new genetic therapy that has silenced the NRAS gene in cells in a dish and in mice. NRAS is one of several genetic mutations causing Congenital Melanocytic Naevus (CMN). The research team used a genetic therapy called silencing RNA, which silences the mutated NRAS in CMN skin cells. The therapy was delivered through special particles directly to CMN cells. The team gave injections containing the therapy to mice with CMN; silencing the NRAS gene after just 48 hours. They also tested it in cells and whole skin sections from children with CMN. Importantly, silencing the gene triggered the CMN cells to self-destruct.



“We are very grateful to our patients at Great Ormond Street Hospital, who have been actively participating over many years to help us produce this new potential therapy. After more studies, we hope the therapy can soon enter clinical trials in people.” Professor Veronica Kinsler

You can read a plain language summary of the research findings by scanning this QR code.



Support

Caring Matters Now provides support in many different ways.

Helpline

Support Consultations with our Head of Support

Support Literature

Age Specific Support Guides

Red Book Insert

School Support Guide

Self Advocacy Pack

Adult Support Toolkit

Support Network

Regional Support Contacts

Regional Gatherings

Online Communities

Online Workshops

Online forums to share experiences and seek advice

Age Specific Events

Teen Support Initiatives

Young Ambassador Programme

Online Teens Resource

Adult Support

Tailored Support

For members experiencing medical complexities

For members exploring treatment

Special Events

BIG Weekend

Annual Research Presentations

Website: www.caringmattersnow.co.uk

Facebook: @CaringMattersNow

Instagram: @caringmattersnow

LinkedIn: Caring Matters Now

Caring Matters Now Support Pathway

0-4 years

We provide specialised early-years support and information for parents caring for babies born with CMN. Parents are offered a one-hour initial support consultation with our Head of Support.

From October to March, we host a monthly online baby and toddler group. We are committed to educating medical professionals to aid early diagnosis, with a particular focus on partnering with midwives and dermatologists across the UK and Ireland. Our Red Book Insert was created to support early diagnosis and provide clear medical signposting.



5-11 years

We support families throughout the primary years by offering age-specific support literature, Schools Guide, connections with our Peer Support Team, and access to family-friendly regional events.

We host bi-monthly Online Workshops exploring topics related to CMN, each stage of our Support Pathway, and the questions that matter most to our members at each life stage.



12-16 years

When a child member turns 12, they begin receiving direct support from our Teens Contact Team, which includes both a male and female volunteer. Teenage members are invited to participate in Level One of our Young Ambassadors Programme and to attend teen support events. We also provide support guides for those caring for teenagers living with CMN. Teenage members have access to an online self-help resource designed to improve self-esteem and confidence, with the option to work through the resource alongside our Head of Support.



**Any member experiencing a new problem related to their CMN is welcome to book a one-hour Support Consultation with our Head of Support.*

17-24 years

At age 17, members transition to receiving direct support from our Young Adults Contact, who helps guide them from child membership into adult membership.

Young adult members are invited to participate in Level Two of our Young Ambassadors Programme and to attend young adult support events. We have created a Self-Advocacy Pack to equip young adults with the knowledge and tools needed to manage their healthcare, communicate their needs, and navigate adult healthcare services effectively.



18+ years

Young adults are encouraged to register as adult members rather than continuing under a parent's registration. Adult members have access to our Adult Contact Team, are invited to adult support events, and may participate in adult-focused research studies. Adults experiencing new concerns related to their CMN, or those wishing to identify the genetic mutation responsible for their CMN, can be referred to the Adult Clinic at St Thomas' Hospital, London.



Over the past 4 years Caring Matters Now has grown at a phenomenal rate, with the number of families we support increasing by 100%. In 2021 we were supporting 389 families affected by CMN; in 2025 we are supporting 825 families affected by CMN. We hold dear our promise of providing support through FREE membership, without constraint, and to do this we rely on the generosity of giving. **For children and adults affected by CMN to access our Support Pathway, please register for free with Caring Matters Now by scanning this QR code.**





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