

## Frequently Asked Questions and other practical matters Cutis Marmorata Telangiectatica Congenita (CMTC)

Reviewers: Prof. Dr. A.P. Oranje (Paediatric Dermatologist, University Hospital Rotterdam, The Netherlands)  
 Dr. M.A. van Steensel (Dermatologist, University Hospital Maastricht, The Netherlands)  
 Dr. W.W. van der Schaar (Medical Psychologist, The Netherlands)

**Disclaimer: Despite all care having been taken in the preparation of this document we are unable to accept any responsibility should the information below prove to be incorrect. CMTC is a rare skin disease of which much remains unknown. This document will be updated regularly in order to remain as current as possible. If you do have any questions or comments then please contact the chairman (president@cmtc.nl)**

Category	Question	Answer
<b>What is CMTC</b>	What is CMTC actually?	<p>CMTC is the acronym for the Latin name Cutis Marmorata Telangiectatica Congenita. Translated into English this means a birth defect affecting the blood vessels that is visible as areas of marbled skin. The alternative name for this condition is Van Lohuizen syndrome. The Dutch paediatrician Van Lohuizen first described the syndrome in 1922. Internationally, both the names CMTC and Van Lohuizen Syndrome are in use.</p> <p>People affected by CMTC have in common that one or more areas of their skin are marbled. Generally this affects the limbs, sometimes the torso and less frequently the face. The condition is not limited to the skin however as this can also occur with defects in other tissues: too thin, too thick or too short bones; thin muscles and also some level of intellectual disability can be present. CMTC can also occur in the context of other syndromes, in which case it is itself a symptom.</p>
	Is CMTC life threatening?	CMTC is not life threatening. In fact, as far as is known to date 5 children have died from complications which can occur with certain syndromes in combination with CMTC. These then concerned the presence of syndromal CMTC.
<b>Causes</b>	What is the cause of CMTC?	It is thought that CMTC comes about through a fault during cell division in the unborn embryo. Each new life starts as a ball of cells that grows into an embryo. In an embryo a number of distinct layers can be identified and the

## Frequently Asked Questions and other practical matters Cutis Marmorata Telangiectatica Congenita (CMTc)

		cells of each of these layers later develop into the varied multiplicity of organs and tissues from which we exist. When a fault occurs in the layer known as the mesoderm then there is a chance that all tissues and organs that develop from this layer are affected. So aside from the skin, bones and muscles can also display abnormalities. When the genetic fault develops relatively late in pregnancy then only the skin will be affected. The fact that abnormalities frequently only occur on one side of the body supports this theory as to the cause.
	As a mother, have I done anything wrong?	No, whether a child has CMTc is down to chance.
	As a father, have I done anything wrong?	No.
<b>Laser Treatments</b>	Should laser techniques be applied in the treatment of CMTc patches?	Laser treatment of the very youngest children would not be advised. Any remaining wound/damage can be treated from an age of 7 years.  Possible complications: <u>EARLY</u> <ul style="list-style-type: none"> <li>• Infections.</li> <li>• Bleedings.</li> <li>• Crust formation.</li> </ul> <u>LATE</u> <ul style="list-style-type: none"> <li>• Pigment changes. In which the skin can appear either lighter or darker.</li> <li>• Scar formation.</li> </ul>
	When can laser treatment be started?	The use of lasers for children can mean that the child will need to be put under general anaesthetic, primarily to prevent panic and to ensure that the child remains still. Going through this process several times a year is not advisable for children from both a physical and mental wellbeing point of view.
<b>Asymmetry</b>	Is it possible for the body to develop asymmetrically, especially in head to toe dimensions (so where one side of the body develops at a different	Yes, this is a possibility although not common and is known as Hemihypertrophy. In certain cases this can be connected with the complication known as primary lymphedema. This is a disorder of the lymph vessels which regulate the fluid balance in the body. With primary

## Frequently Asked Questions and other practical matters Cutis Marmorata Telangiectatica Congenita (CMTC)

	rate than the other)? What can be done about this?	lymphedema there are too few lymph vessels to properly drain the body of fluids. As a result, one side of the body can become thicker than the other side.
	When should any intervention be considered if there is a difference in leg lengths?	In The Netherlands the guideline is that physical intervention is only appropriate when the difference in leg length is larger than 3cm. In addition, it should be noted that the age of the patient also plays an important role in deciding when to intervene.
	Can asymmetry also develop at a later age?	Only during the growth years generally speaking.
<b>Patch Locations</b>	What can be done if the patches are around the eyes and/or on the forehead?	In this case there is a chance of glaucoma (raised pressure in the eyeball that can lead to blindness) and brain abnormalities. In the latter case, an MRI scan is recommended.
	What can be done if the patches are on or around the nose and jaw area	Generally speaking no action needs to be taken.
	What can be done if the patches are in the ears?	Let an ENT specialist inspect the ears regularly and carry out a hearing test when needed.
	What can be done if the patches are in the mouth?	Let a dentist check development in the area regularly.
	What can be done if the patches are on the head?	Generally speaking no action needs to be taken.
	What can be done if the patches are on the neck?	Generally speaking no action needs to be taken.
	What can be done if the patches are on the legs?	In this case the legs may develop asymmetrically in length and girth. The most significant variation occurs mainly at a young age. During the physical development stage the patches are mostly less visible in terms of intensity.
	What can be done if the patches are on the buttocks and/or torso?	If there are likely to be other syndromes in combination with CMTC then the advice would be to determine with ecography (ultrasound) whether internal organs such as liver, kidneys, heart and lungs display abnormalities. Should this be the case, then an MRI scan can be used to get a better view of the precise nature of the variations.

## Frequently Asked Questions and other practical matters Cutis Marmorata Telangiectatica Congenita (CMTC)

<b>Genetic Condition</b>	Is CMTC genetically inherited?	As far as is known at present CMTC is not inherited. We do know of parents with CMTC that have children without CMTC.
<b>Rare Condition</b>	How rare a condition is CMTC?	CMTC is quite a rare condition. At the moment there are no figures available as to how frequently CMTC occurs.
<b>Complications</b>	Which complications could be found in combination with CMTC?	<p>To begin with, it should be noted that it is extremely difficult to determine whether any given complication is related to CMTC.</p> <ol style="list-style-type: none"> <li>1. Overgrown legs and arms (in length and girth).</li> <li>2. Undergrown legs and arms (in length and girth).</li> <li>3. Intellectual disability.</li> <li>4. Asymmetric development on one side of the body.</li> <li>5. Glaucoma.</li> <li>6. Syndromal symptoms.</li> </ol>
	Up to which age could associated disorders appear?	Abnormalities could in fact manifest themselves at any age. For example in the case of glaucoma which can result in blindness when this condition is not diagnosed over an extended period of time.
<b>M-CMTC</b>	What is M-CMTC?	M-CMTC stands for “macrocephaly” a special form of CMTC. The most significant characteristics of this type are a relatively large head size and a lower rate of intellectual development.
	M-CMTC or Cowden syndrome?	Certain cases concern not M-CMTC, but Cowden Syndrome. One of the differences between these is inheritance. M-CMTC is not inherited whilst Cowden Syndrome is inherited. Good clinical assessment can determine the difference after which molecular biological diagnostics can confirm the diagnosis.
	My child has M-CMTC and regular epileptic attacks. What is my best course of action?	Consult a pediatric neurosurgeon.

## Frequently Asked Questions and other practical matters Cutis Marmorata Telangiectatica Congenita (CMTC)

<b>Comparable Conditions</b>	CMTC is similar to which other conditions?	CMTC can appear similar to conditions such as Klippel-Trenaunay Syndrome or Sturge-Weber syndrome.
	Is it possible for a patient not only to suffer from CMTC but also KTS or SW?	This is indeed possible. As a result, a correct diagnosis is made more complex.
<b>Psychological Aspects</b>	My child with CMTC is being bullied. How can I best deal with this? As a parent, how can I prevent such problems?	<ol style="list-style-type: none"> <li>1. Involve their teacher(s) and ask if you can provide information to all the pupils in the form of a talk either by the child or by the parents. An important point to bring over is that CMTC is not infectious.</li> <li>2. Allow the class to ask questions so as to create a dialogue on the subject.</li> <li>3. Ask pupils to help when other people are bullying.</li> <li>4. Investigate whether pupils also have or have had similar problems.</li> <li>5. A “patch school” (this facility is offered at the University Hospital Rotterdam) from a psychologist can help.</li> </ol>
	The grandparents give the child with CMTC extra presents and attention in comparison with the other children. How should I respond?	Raise this issue with the grandparents and explain that they are in fact strengthening the “patient” role of the child and their disadvantaged position in comparison to the other children. In this way the child will not be able to build self-confidence and the other children will learn to display similar behaviour in order to gain equal attention.
	The child with CMTC does not appear to have any issues but as parent I do. I consider as a parent that the CMTC patches are unattractive on his/her body. How can I deal with this in practice?	The parents can best adopt a “WE” approach. In other words, both parents need to agree with each other how to deal with this. Ask for help if needed from a paediatric psychologist that also has children.
	When is the best time to discuss CMTC with the child? Should I as the parent take the initiative or can I better wait until the child asks questions?	Do not start the discussion yourself. Let the child do so first if possible, as otherwise it may appear that the child is the one solving the issue that the parents have with their view of the child’s disorder. Above all, the child may also ask questions that the parents have not yet thought about because the child has other priorities.
	What is the influence of stress on	There has been no scientifically proven link found.

## Frequently Asked Questions and other practical matters Cutis Marmorata Telangiectatica Congenita (CMTC)

	CMTC?	
<b>Other</b>	Can an infusion be applied or an injection be given in for example an arm or a leg that is affected by CMTC?	This is not recommended due to the chance of thrombosis (clotting in the bloodstream which could cause a stroke or heart attack for example).
	We have a child with CMTC and would like another child. How great is the chance that we will have another child with CMTC?	The chance of having a second child with CMTC is low. The same chance as for parents that have had children already without CMTC.
	My child with CMTC has regular nosebleeds. Does this have anything to do with CMTC?	As far as is known, there is no relationship between CMTC and nosebleeds.
	My child has a CMTC patch on her knee. If she falls on this patch and the wound bleeds then it appears to take a long time before the knee recovers. Is this normal?	This is normal.
	My child with CMTC has marked red veins in her eye. Does this have any connection with CMTC? What is my best course of action?	Probably also a symptom or has nothing to do with it.
	Should someone with CMTC patches be exposed to the sun?	This is generally not an issue if a good sun cream is used of at least factor 15.
	My child with CMTC has very flexible joints. Does this have anything to do with CMTC?	There is no known connection between CMTC and flexible joints (hyper mobility). Normally speaking, this hypermobility will reduce around 5 years after the end of the body's growth stage.
	My child with CMTC has two toes that have grown together. Does this have anything to do with CMTC?	This can occur when the child has CMTC patches on the legs and/or feet. We have several members who have two toes that have grown together.
	As a woman with CMTC, should I use an anti-conception pill?	If you have varicose veins then there is a risk of thrombosis. Discuss this with your doctor.



**Frequently Asked Questions  
and other practical matters  
Cutis Marmorata Telangiectatica Congenita (CMTC)**

	I have CMTC and would like to become pregnant. What should I do?	There are no restrictions in the case of non-syndromal CMTC.
	I have CMTC and an asymmetric body in the height dimension. I also have scoliosis. Are these two conditions related?	This is possible but would mostly not be the case.
	My child with CMTC has fingers that are growing bent. Does this have anything to do with CMTC?	No.