Patient Journey Cutis Marmorata Teleangiectatica Congenita (CMTC)

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About CMTC

Cutis – skin
Marmorata – marbeled
Teleangiectatica – abnormal bloodvessels
Congenita: present at birth
Occurrence: unknown
CMTC is both a skin disease as well as a vascular malformation

Introduction

The global CMTC-OVM was established in 1997 in the Netherlands to support patients and families, bring people together for peer contacts, information provisioning and patient advocacy. CMTC-OVM arranges consultation between patients, clinicians, psychologists, and other stakeholders.

CMTC-OVM aims to stimulate a global collaboration in research and map the medical expertise in collaboration with the ERN-SKIN & ERN-VASCERN. These ERNs are the virtual expert centers networks with the aim to improve quality, safety and access to highly specialized healthcare for children and adults with rare skin and vascular diseases throughout Europe.

Purpose

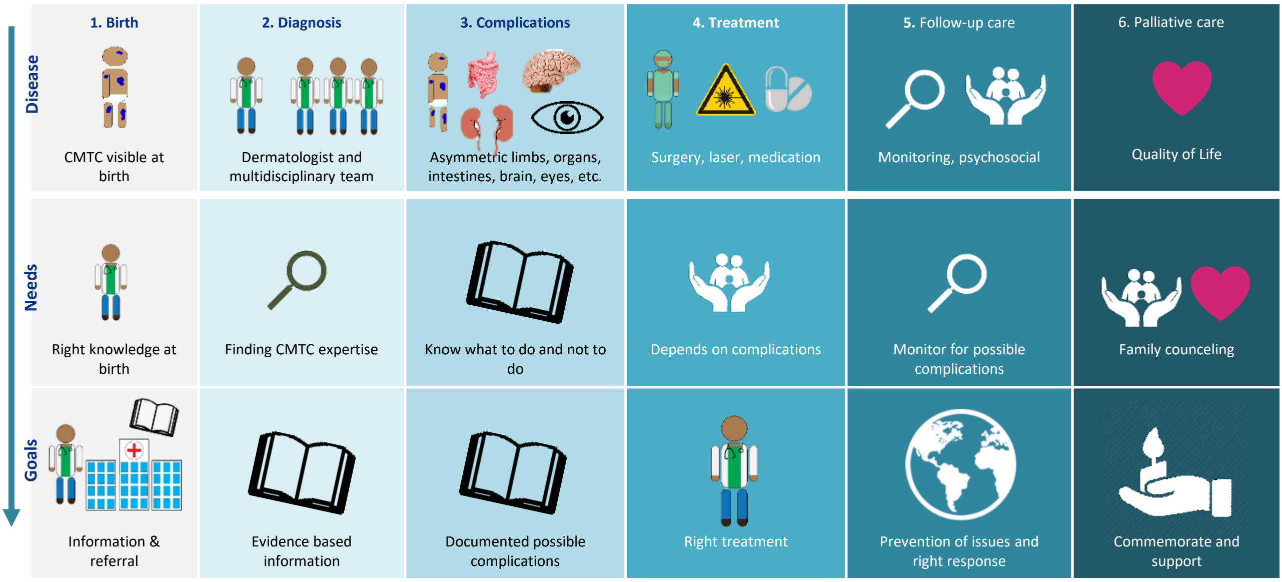
Patient Journeys represent the collective holistic perspective on the burden of the disease and the needs of patients and their families with first-hand experience of living with a rare disease. The Patient Journey for **CMTC** was developed from the perspective of patients and parents.

Methodology

A workshop was organized during the CMTC-OVM global conference in 2019. During the event a ERN-SKIN ePAG advocate collected feedback from the CMTC patient community and their families to draft their patient journey.

These outputs together with CMTC experience for over 20 years were useful to **map the needs and recommendations on ideal care across the different stages of the patient journey from the first symptoms at birth to the end-of-life care.**

The document was also shared via the CMTC social media channels to collect feedback from the wider patient community (about 10 people reacted globally).



	1. Birth	2. Diagnosis	3. Complications	4. Treatment	5. Follow-up care	6. Palliative care
Disease	CMTC visible at birth	Dermatologist and multidisciplinary team	Asymmetric limbs, organs, intestines, brain, eyes, etc.	Surgery, laser, medication	Monitoring, psychosocial	Quality of Life
Needs	Right knowledge at birth	Finding CMTC expertise	Know what to do and not to do	Depends on complications	Monitor for possible complications	Family counceling
Goals	Information & referral	Evidence based information	Documented possible complications	Right treatment	Prevention of issues and right response	Commemorate and support

Overall patients and families needs across the 6 stages

- ✓ Immediate access to diagnosis by a multidisciplinary team in the right medical expertise center.
- ✓ Psychological and psychosocial support.
- ✓ Peer support from patient organisations.
- ✓ Access to reliable and understandable information on the disease and treatment plan.
- ✓ Understand the treatment options (do' and don'ts) and the social impact on patients' future life.
- ✓ Information about the risks, the expected results and each step of the treatment.
- ✓ Financial support if the costs for treatment and surgery are not reimbursed.
- ✓ Smooth transition from care to home.
- ✓ Best holistic care approach.

Overall patients and families ideal care and recommendations across the 6 stages

- ✓ Timely diagnosis in a reference medical center.
- ✓ Access to information material on disease and treatment plan.
- ✓ Access to quick psychological and psychosocial support.
- ✓ Direct contact with patient organisations.
- ✓ Social inclusion programmes offered by the schools to inform and faciltate the integration of the children.
- ✓ Smooth transition from child care to adult care.
- ✓ Medical data record in one single platform and accessible for the patients and families.
- ✓ Access to other treatments (camouflage technique instead of laser).
- ✓ Access to the best medical and palliative care services.
- ✓ Medical and genetic data available for further research.
- ✓ Creation of a social media memoriam page.

Conclusion

The patient journey is a tool that enables patient advocates to share and gather the needs from the perspective of the wider patient community. This is a reference document to develop a common understanding of the disease and the burden of the disease on both the patient and family. It is a good instrument to engage with the clinicians and to address these unmet needs.

The CMTC patient community involved emphasized that there is an urgent need to access to a proper and quick diagnosis and that holistic care is required.

Design infographics by Quinten de Vries adapted from the original design by J. Meek of ERN GENTURIS (Eur J Hum Genet. 2020 Feb;28(2):141-143).