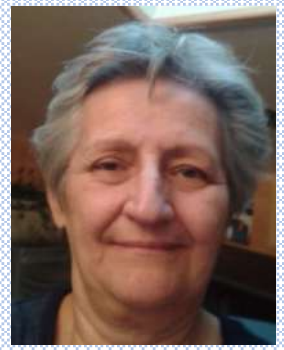
ERN-Skin ePAG

European Patient Advocacy Groups

ERN-Skin Board Meeting

2nd October 2020

Who are we ?



Marie-Claude Boiteux
Cutis Laxa Internationale
Mendelian Cause of Connective Tissue Disorders (MCTD)
France

Laurence Gallu
Association Pemphigus Pemphigoïde France (APPF)
Autoimmune bullous diseases
France

Giulia Volpato
p63 sindrome eec international
Ectodermal Dysplasias
Italy

Ingrid Jageneau
DEBRA- Belgium
Epidermolysis Bullosa
Belgium

Lex Van Der Heijden
CMTC and Other Vascular Malformations
Cutaneous Mosaic Disorders
Netherlands

Ulrike Holzer
Selbsthilfegruppe Ektodermale Dysplasie e.V.
Ectodermal Dysplasias
Austria

Marjolaine Van Kessel
Congenital Melanocytic Naevus
Cutaneous Mosaic Disorders
Netherlands

Jodi Whitehouse
Caring Matters Now
Cutaneous Mosaic Disorders
UK

Angélique Sauvestre
DEBRA - France
Epidermolysis Bullosa
France

Diana Perry
Ectodermal Dysplasia Society
Ectodermal Dysplasias
UK

Bente Villumsen
Hidrosadenitis- Denmark
Hidradenitis suppurativa
Denmark

Karin Veldman
Vereniging voor Ichthyosis Netwerken
Ichthyosis & Palmoplantar Keratoderma
Netherlands

2019-2020 Activities

1. Patient Satisfaction Survey
   - Aims
   - Accessibility

2. Patient Journeys
   - Gathering information from patients and the wider community
   - Common issues and ideal situations by disorder
   - Easy reading graphics
   - Collaboration with HCPs

Patient Satisfaction Survey

AIMS

- Measure the level of patient satisfaction after consultation in an ERN-Skin center
- Measure the impact of ERN-Skin centers to improve quality of care and performance
- Results will be crucial for the evaluation process of the ERN centers that takes place every 5 years

Patient Satisfaction Survey

- **The questionnaire has 26 items and it is divided in 4 main sections:**

1/ General information on the patient

2/ Consultation and follow-up (19 questions)

3/ Treatment prescription and therapeutic research (5 questions)

4/ Global satisfaction (2 questions)

A short blank section is available at the end of the questionnaire to add any comments.

Patient Satisfaction Survey

1-General Information

Who was the consultation for? ◻ Yourself ◻ Your Child ◻ Someone (child or adult) you are caring for

Which Centre was your consultation carried out in?

Hospital: Service: Town: Country:

What was the reason for consulting the Centre? ◻ 1st appointment ◻ Follow-up ◻ other, please specify.....

Who referred you to this service?

◻ Family doctor ◻ Local dermatologist

◻ Hospital dermatologist (or other medical service) ◻ Someone else, please specify: .....

2-Consultation and Follow-up Scale from 0 to 3, 0=no, not at all; 1=just a little; 2=yes, but incompletely; 3=yes and completely; N.A. = Not Applicable	0	1	2	3	N.A
Was it easy to find the contact information for the Centre and/or make an appointment?					
Was the consultation adequate from an emotional/psychological point of view?					
Were the location and space of consultation adapted to your specific needs and/or disability (dressing, solar filter, water points, etc.)?					
Was it a multidisciplinary consultation (seeing different specialists during the same consultation or the same day)?					
Did the consultation clearly propose psychological support?					
Did you feel the consultation considered all the necessary specialists to address your care?					
Did you understand the explanations and consequences of the rare disease?					
Do you feel well informed about the disease?					
In the case of a genetic disease, were you adequately informed regarding the inheritance risks?					
In the case of a genetic disease, were you referred to a specific genetics consultation?					
Are you satisfied with the follow-up within the Centre?					
Are you satisfied with the information on how to contact the Centre in case of emergency?					
Did you receive any information about the availability of peer support, such as disease specific national patient organisation and/or an international network and/or a national rare disease alliance?					
Has the Centre set up a local network for your follow-up?					
	0/No			3/Yes	N.A
In the case of a local network, it includes psychological follow-up					
In the case of a local network, it includes your local doctor					
In the case of a local network, it includes a nurse					
In the case of a local network, it includes social workers					
In the case of a local network, it includes other members (Please specify.....)					
3-Treatment prescription and Therapeutic research Scale from 0 to 3, 0=no, not at all ; 1=just a little ; 2=yes, but incompletely ; 3= yes and completely ; N.A. = Not Applicable	0	1	2	3	N.A
If a treatment already exists for the disease you consulted for, was the aim of the prescribed treatment discussed?					
Were alternative treatments discussed?					
Were side-effects/intensity/risks of treatment discussed?					
Were you given a specific contact in case of problems regarding at-home treatment?					
Do you know if therapeutic research protocols exist for the disease, or its symptomatic manifestations, in the centre where you consulted?					

4-Global satisfaction Scale from 0 to 10, 0=no, not at all ; 10= yes and totally ; N.A. = Not Applicable	0	1	2	3	4	5	6	7	8	9	10	N.A
Are you satisfied with how the multidisciplinary team took care of you?												
Are you satisfied with the hospital where the centre is located (premises, signage, reception, lift, etc.)?												

Please add any other comments you may feel useful or necessary:

Patient Satisfaction Survey
ACCESSIBILITY

- Translations are in progress and are already available in Danish, Dutch, English, French, German, Italian and Polish
  - Patient Representatives from other countries are asked to translate the missing languages

- Will be accessible online on the ERN-Skin website
  - There will also be a paper version for those who are not familiar with using the internet

Patient Journey

A “Patient Journey” is a testimony that reflects the natural history/needs of patients and their families for a specific rare disease. It represents the collective perspective on the burden of the disease and the needs of people with first-hand experience of living with a rare disease.

- Mapping the journey and identifying the needs and recommendations on ideal care for each specific skin disorder across the different stages from the first symptoms at birth to the end of life care
- Reviewing by both ePAG advocates and clinical experts (clinical presentation validated by some clinicians)
- Organising the information in easy reading graphics (table and illustration)
- Identifying the needs that are common for all ‘skin disorders’ (next steps)

Aiming to:

- Engage the wider patient community to consolidate common needs for a specific rare disease
- Engage in a discussion with the clinicians to highlight the different (unmet) needs of a specific rare disease, both medical and psychological, so the clinicians can respond to these needs
- Keep in mind both the professional and patient are experts

Patient Journeys

Stage of Journey (please adapt or modify stages if needed)	Timeline	Clinical Presentation / Symptoms	Identify Patient Needs	Ideal Outcome / Support
Pre-diagnosis	Pregnancy	• Family history with CL • IntraUterin Growth Delay	• Amniocentesis searching for known mutations	• Prenatal screening • Appropriated prenatal follow-up
Main symptoms	Birth	• No family History with CL • No IntraUterin Growth Delay • **Wrinkles and/or Lax skin** • **Wide anterior Fontanelle** • Ataxia • **Hip dislocation** • Dwarfism • **Micro or Macrocephaly** • Cerebral Cortex Malformation • Corpus Callosum Agenesis • Joint contractures	• Lack of knowledge at birth • incorrect diagnosis can generate fear and panic in the patient's family • **Where to find expertise? Dermatologists familiarized with CL in and/or outside my country** • Opinions given instead of information based on validated knowledge • **It takes time to get correct diagnosis with correct information**	• Knowledge at birth/fast referral (pediatrician, midwife, dermatologist) • Map of expertise • **Training for General Practitioners, Dermatologists, Pediatricians and other healthcare providers to identify the symptoms allowing to suspect the rare disorder**

Source: Cutis Laxa with Neuro and/or Skeleton Symptoms Patient Journey.

Cutis Laxa with Arteries and/or Lung Symptoms (CL) Patient Journey (1)

1 Pre-diagnosis & main symptoms... → 2 ...Diagnosis... → 3 ... Diagnosis follow-up... → 4 ... Treatments...

1. Pre-diagnosis

During pregnancy, specially patients with family history with CL.

1. Main symptoms

Start at birth or when the first symptoms appear: early loose/wrinkled skin, lax skin, respiratory and gastrointestinal issues.

Need: Find the right expertise. Dermatologists familiarized with CL in and/or outside a country.

Ideally: Prenatal screening during pregnancy. Training for the General practitioners, Dermatologists, Pediatricians and other healthcare professionals to identify faster the symptoms and to get a quick and a correct diagnosis.

2. Diagnosis

A.s.a.p after birth or when early loose/wrinkled skin appear, through a multidisciplinary consultation (geneticist and dermatologist with CL expertise)

Need: Be informed on the treatment options and on the psychological impact. Psychosocial aspects are underestimated. Psychological support for siblings and patients due to wrinkles/old aged appearance.

Ideally: Correct information on treatment options (plastic surgery). Psychological care for patients, psychological support for parents and siblings and peer support (patient organisations). Patients and/or families' advice/opinion heard/trusted.

3. Diagnosis follow-up

Possible complications at birth or after diagnosis:
Aortic aneurysm.
Aortic stenosis.
Bicuspid aortic valves.
Vascular involvement distal to the aortic root.
Emphysema.
Cardiorespiratory issues.
Gastro-Intestinal issues (inguinal/wound/diaphragm hernia/bladder/gastrointestinal diverticulae).
Bladder/urinary issues.

Need: Find the expertise. Treatments: symptomatic for emphysema, gastrointestinal issues and/or aortic aneurysm if severe. Fear for aortic aneurysm rupture (death)

Ideally: Map the expertise. Access to international consultation (CPMS?) and access to information in own language. Psychological support for patients, parents and siblings. Peer support (patient organisations).

4. Treaments

Treatment of Cardiorespiratory issues during lifetime:
cardiac surgery and lung transplant in adulthood

Treatment of gastrointestinal issues during lifetime:
gastrointestinal surgery

Treatment of wrinkles if the burden of others' gaze is too heavy for the child or in adulthood: facelift

Need: Find the expertise, in particular too few experienced plastic surgeons and waiting lists for transplantation. Fear for death due to aortic aneurysm. Information limited, specially for the facelift surgery (costs? duration of the results? burden of surgery on patients and families)

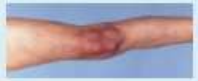
Ideally: : Map the expertise. Access to international consultation (CPMS?) and access to information in own language. Psychological support for parents and siblings. All financial costs of the facelift covered.

Patient Journey Cutis Marmorata Teleangiectatica Congenita (CMTC)

Lex van der Heijden president@cmtc.nl
President CMTC-OVM, the Netherlands

cmtcovm · Global Patient Advocacy Group · European Reference Networks · SKIN VASCERN

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

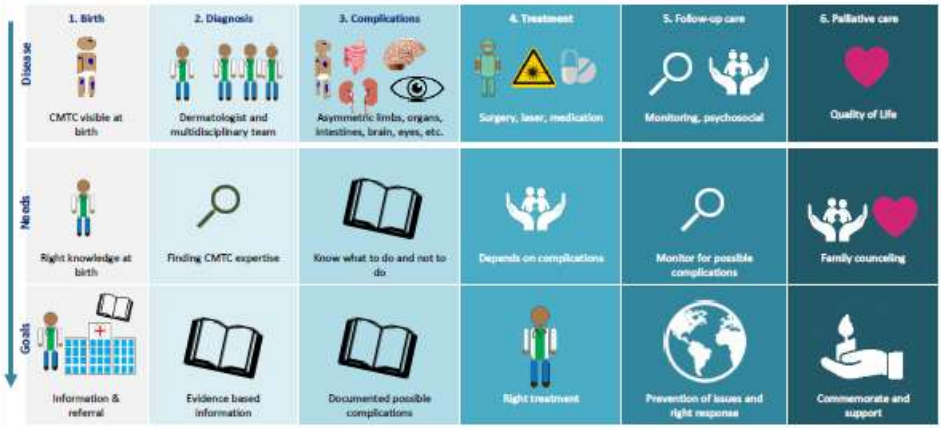
The global CMTC-OVM was established in 1997 in the Netherlands to support patients and families, bring people together for peer contacts, information provisioning and patient advocacy. CMTC-OVM arranges consultation between patients, clinicians, psychologists, and other stakeholders.
CMTC-OVM aims to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



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 facilitate 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).

ERN-Skin Patient Journey Posters presented during the ECRD online Conference 2020

Patient Journey Congenital Melanocytic Naevus Syndrome

Marjolein van Kessel, marjoleinvankessel@gmail.com
President Naevus Global, Utrecht, the Netherlands
Patient team, Naevus International, London, UK

Naevus Global · Naevus · European Reference Networks · SKIN

About Congenital Melanocytic Naevus (CMN)

Congenital – present at birth
Melanocytic- caused by a pigment producing cell
Nevus (plural, nevi) – birthmark
Occurrence:
Small CMN – 1 in 75 births
Largest CMN – 1 in 20,000 to 50,000 births
Congenital changes may appear in brain/spine, thus: **syndrome.**

Introduction

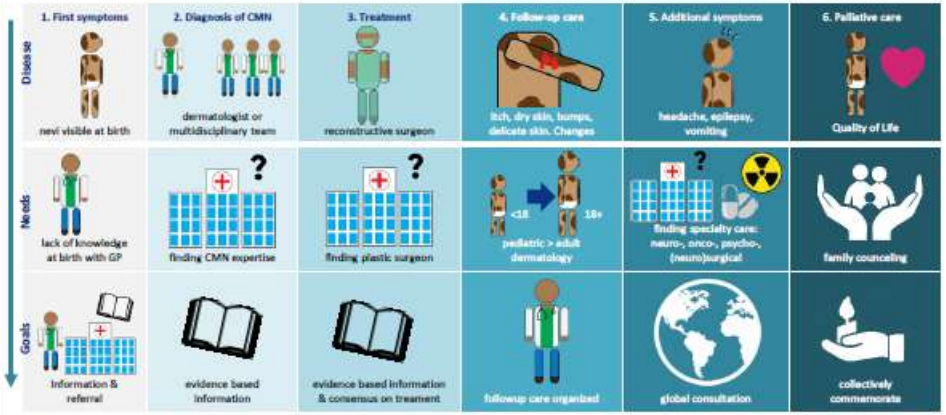
Naevus Global formalized in 2013 connects individuals and families around the world affected with rare forms of CMN. Together with Naevus International network, it provides mutual consultation between patients, scientists, clinicians, psychologists, and other stakeholders.
Naevus Global stimulates **cross-border collaboration in research, consensus guidelines** and international registries to integrate efforts and **map medical expertise,** in collaboration with the ERN-SKIN.
ERN-SKIN is a virtual expert centers network with the aim to improve quality, safety and access to highly specialized healthcare for children and adults with rare skin diseases throughout Europe.

Purpose

Patient Journeys represent the collective perspective on the burden of the disease and the needs of people with first-hand experience of living with a rare disease. The Patient Journey for **Congenital Melanocytic Naevus syndrome** was developed from the perspective of patients and parents, as a reference point for pathways and guidelines.

Methodology

SKIN ePAG advocates completed a mapping exercise of the needs of the CMN syndrome, across the different stages of the patient journey. These stages progress from **first symptoms, diagnosis, possible treatment (surgery), to follow-up care and palliative care.**
Patient needs at each stage of the journey are referenced under three levels: **clinical presentation; patient needs; recommendations on ideal care.**
A first version of the patient journey was presented at the Naevus International conference (2019) with 15 patients and ERN-SKIN patient advocates representing 10 countries in Europe, North & South America and Africa.



Challenge
Diagnosis possible neurological involvement.
Establishing prognosis and care strategy.
Care psychological impact of diagnosis.

Goal
Rapid referral to experienced care specialists
International mapping of centers and individuals of expertise.

Challenge
Psychological impact on families during the diagnosis.

Goal
Guidelines based on evidence based information.
Receive professional counselling to learn how to cope with the social impact. Systematic referral to patient organizations for peer support.

Challenge
Finding an experienced plastic surgeon.
Decision on whether to remove the nevus.

Goal
Further research to learn more about how to assess the risks of melanoma, possible treatments and their outcomes.
Psychological support for parents and siblings as well as patients.

Challenge
Transition to adult care. Patients and family members experience psychosocial challenges.

Goal
Well organized transition to adult care.
Professional counselling to learn how to cope with the social impact of the disease on the patients and their families' life.

Challenge
Finding the proper expertise when there are morbid or deadly symptoms. Experiences with clinical trials are scattered. Lack of statistics.

Goal
Access to highly specialized healthcare centers in Europe and beyond. International consultation.
Comparison of treatment efficacy with a standard and uniform outcome reporting.
Psychological support.
International peer support.

Challenge
Deep grief of families in case of loss.
Psychological support and guidance from professional councilors.

Goal
Retention of family expertise and experience.
Referral to grief therapists.
Patient organizations connect and collectively commemorate people who have passed away.

Conclusion

The patient journey is a tool that allows ERN-SKIN patient advocates to gather the needs and the perspective of the wider patient community.
This is a reference document to engage with clinicians to develop a common understanding of the disease and the burden of the disease it represents.
Clinicians can identify the gaps in care and treatment and develop Clinical Practical Guidelines to address these needs and map international expertise.
What's next? SKIN ePAG advocates who represent the CMN syndrome will organize a joint workshop with the clinicians to review this patient journey, identify the possible gaps and address patient needs.

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).
Photography: Anima Vera
May 2020

Patient Journeys

- Already Available:
  - CONGENITAL MELANOCYTIC NAEVUS
  - CUTIS LAXA / LUNGS – ARTERIES
  - CUTIS LAXA / NEUROLOGIC – SKELETON
  - CUTIS MARMORATA TELANGIECTATICA CONGENITA
  - NETHERTON SYNDROM /ICHTHYOSIS LINEARIS CIRCUMFLEXA
  - PEMPHIGUS / AUTO IMMUNE BLISTERING DISEASE
- Patient Journeys for other disorders are still on-going
- Next steps : When a majority of skin disorders covered by ERN-Skin have their Patient Journeys completed, ePAG advocates will present their journeys to the others to identify the common needs for a global Patient Journey for all ERN-Skin rare disorders

And now ? What projects ?

- Consolidation of the ePAG group in the Thematic working groups (filling the gap where there is no ePAG representation)
- Formalisation of the rules of participation of patient representatives in ERN (draft of ePAG Terms of Reference)
- Increase the engagement with the wider patient community
- Increase the collaboration with the clinicians in specific tasks such as Guidelines, Security cards, etc.

THANK YOU