Patient Journey Congenital Melanocytic Naevus Syndrome

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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

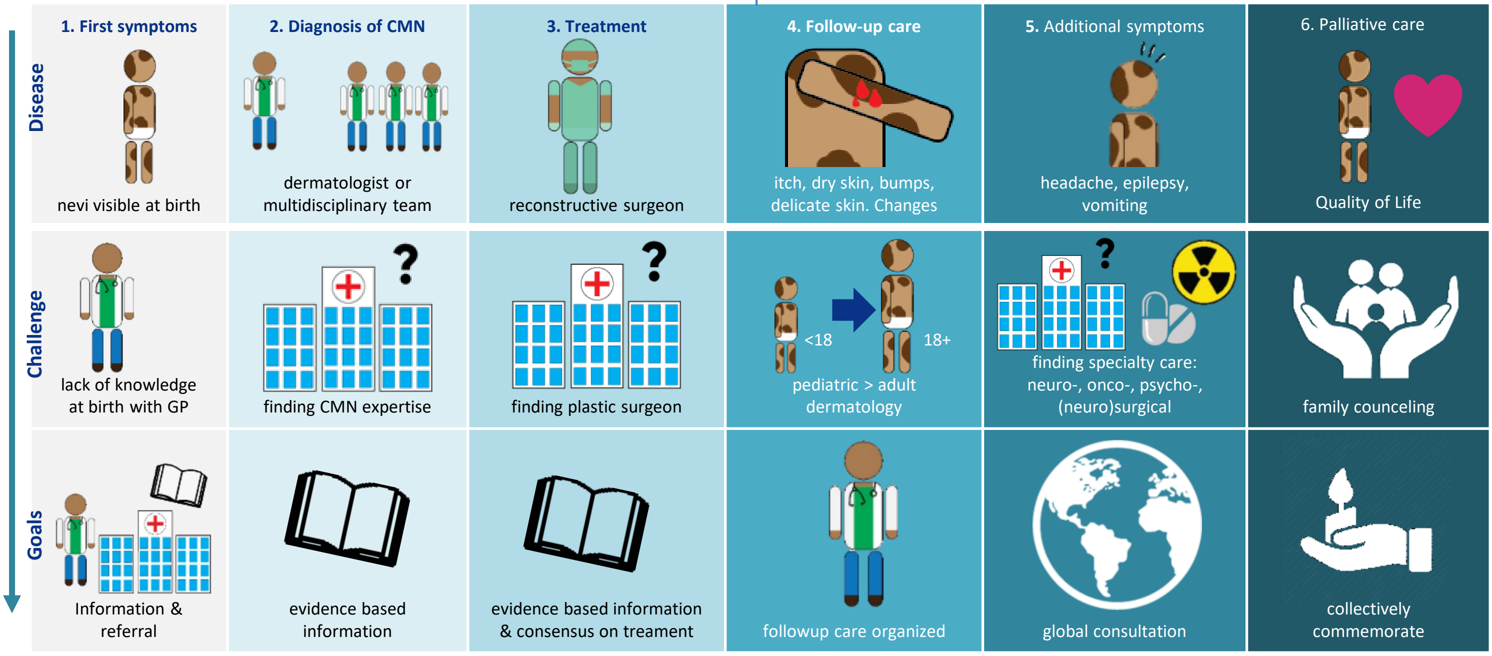
Patient Journeys represent the collective perspective on the burden of the disease and the needs of people with first-hand experience of living with a rare disease. The Patient Journey for **Congenital Melanocytic Naevus syndrome** was developed from the perspective of patients and parents, as a reference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



	1. First symptoms	2. Diagnosis of CMN	3. Treatment	4. Follow-up care	5. Additional symptoms	6. Palliative care
Disease	nevi visible at birth	dermatologist or multidisciplinary team	reconstructive surgeon	itch, dry skin, bumps, delicate skin. Changes	headache, epilepsy, vomiting	Quality of Life
Challenge	lack of knowledge at birth with GP	finding CMN expertise	finding plastic surgeon	<18 18+ pediatric > adult dermatology	finding specialty care: neuro-, onco-, psycho-, (neuro)surgical	family counceling
Goals	Information & referral	evidence based information	evidence based information & consensus on treament	followup care organized	global consultation	collectively commemorate

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.

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