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 the 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.

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<th>Disease</th>
<th>Needs</th>
<th>Goals</th>
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<td>nevi visible at birth</td>
<td>lack of knowledge at birth with GP</td>
<td>information &amp; referral</td>
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<td>evidence based information &amp; consensus on treatment</td>
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<td>finding CMN expertise</td>
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<td>finding plastic surgeon</td>
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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.

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.

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

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