

The panel discussion

ERN-Skin TRAINING SESSION

GHENT Monday 16th December 2019

On December 16 Marjolein van Kessel (Naevus Global) was invited to participate in a panel for a training session within the European Reference Network Skin in Ghent. The audiences were doctors and professors from various countries with both medical and genetic backgrounds.

Together with 3 other panel members from various countries we were asked to discuss 'patient burdens in rare skin disorders - what do physicians need to know?'. The training course was intended for the doctors and researchers from the ERN-skin. About 60 people were present, many of them were doctors in training. It was the first time patient representatives were asked to join a meeting like this for a panel discussion. We were honored to be given this opportunity. "NOTHING ABOUT US, WITHOUT US"

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Introduction round :

Marie-Claude BOITEUX, Chair and co-founder of "Cutis Laxa Internationale", Mother of Cécile, 29, diagnosed with ADCL, ELN mutation. (mcjlboiteux@aol.com)
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Ingrid JAGNEAU, founder of "Debra Belgium", Mother of xxxx , 34, diagnosed with EB xxxxx, (ingrid@debra-belgium.org)
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Marjolein VAN KESSEL, CMN organization, Mother of Dafnis, (1998-2012), diagnosed with CMN and NCM, (marjoleinvankessel@gmail.com)

Lex VAN DER HEIJDEN, Chair of CMTC Netherlands Organisation, Father of daughter, 26 years, diagnosed with CMTC (president@cmtc.nl)



Bert Callewaert (dermatologist UZ Ghent), Christine Bodemer (dermatologist Hôpital Necker Enfants Malades, Paris), Lex van der Heijden (CMTC), Marjolein van Kessel (CMN), Ingrid Jagenau (DEBRA), Marie-Claude Boiteux (Cutis Laxa)

Input in panel discussion based on CMN experience

Marjolein discussed the psychological impact information has on families. A rare disease also had a large impact on siblings. An ongoing problem is the lack of information to make good decisions on treatments. There's also little information available on research and clinical trials.

It would be helpful to have online consultation, for which we hoped CPMS* would work. In an ideal situation a patient with medical complications would like to have a consultation with a personal doctor and a medical expert on the disease join at the same time through an online consultation.

There's a problem for people from Eastern Europe (or further abroad) who don't have access to treatments like plastic surgeons. They can't find surgeons in Western Europe who have time and finances to treat people from Eastern Europe. Therefore people raise money to get an expensive treatment in the USA.

* CPMS, Clinical Patient Management System, is a secure web-based application to support European Reference Networks in the diagnosis and treatment of rare diseases across national borders.

Answers to other questions:

1. Personal experience concerning diagnostic pathways / expert advice / research?
 - a. **Diagnosis**
 - Need to fight to be taken seriously when discovering yourself what you are suffering from (internet, literature,.....)
 - Getting a diagnosis can be a stroke of luck or research for information from the patient
 - There is no silly question to ask every doctor you end up seeing
 - Impact of information given
 - Patients/Families need psychological support when receiving the diagnosis
 - Siblings also need to be supported
 - b. **Follow up**
 - Lack of multidisciplinary work, only dermatologists are in the ERN when patients also need other HCPs.
 - Patient's need to be "in control" of one's health
 - Need for a dialogue with clinicians
 - Where to find expertise for all symptoms, not only dermatology?
 - Patients/Families need to get accurate information based on test/medical results
 - Scattered medical information
 - From childhood to adulthood and to old age
 - Lack of reimbursements for treatments/ointments
 - c. **Research**
 - Lack of information on clinical trials/research/treatment
2. What are the main social burden and how could ERNs help tearing down these social burdens?
 - a. **Psychological**
 - Anxiety

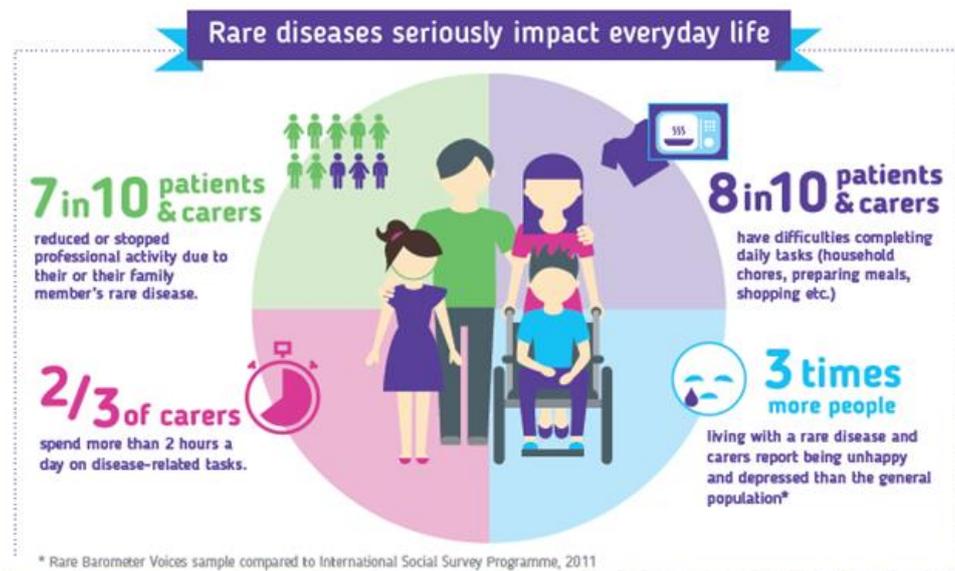
- Depression : People with RD are 3 times more depressed than the general population
- Self-esteem
- Facing gazes

b. Daily

- Juggling care and daily life
- Going out Shopping
- Choosing clothes
- Difficulties completing daily tasks
- Some decisions we take for the problems today may have a long term impact

c. Family

- Family impact
- Family carers : no rest, no salary, no retirement
- Private life / Love Life
- Having children?



d. School / Job

- Going to school
- Choosing studies
- Choosing a job
- Reduce or stop professional activity because of RD

e. Financial

- Care for patients getting old
- Financial burden when treatments are not reimbursed
- Need to evaluate quality of life to improve :
- Care
- Clinical Research
- Psychological Research
- Politics : "Skin diseases matter"

3. What is the patient view on achievements of ERN's: hopes/wishes?

a. Follow-up

- Treatments
- Setting up multidisciplinary networks including nurses, psychologists, etc

- More cross-ERN collaboration
- List of “disease-experts” available for patients across Europe (facilitate cross-border care)
- Lay versions of Clinical Practice Guidelines
- Develop guidelines for patients and doctors, creating a real network of dialogue

b. Research

- Facilitate access to clinical trials to all patients, when there is no trial in their own country
- Registries/data
- Data and registries : comprehensive data strategy including patients organisations
- We don’t want new data silos
- What are the ERN needs in term of data ?
- Need to have a say in what data is being collected, for what purpose and how it is collected

c. Improving on

- Raising awareness
- Integration in National Health Systems. At least one ERN-Skin center in each country to disseminate information and knowledge from ERNs (guidelines, training, research, etc)
- Sharing knowledge
- Sharing experiences regarding the use of existing clinical practice guidelines

d. Other

- State of the art of the Disability card, updates and usage
- Transversal ERN Workgroups to train school workers on RD management

4. What is patients’ view on how to organize research?

- Patients’ involvement: they are the data
- Educating patients in the basic concepts of research, clinical trials’ processes and terminology to get them involved in research activities.
- Patients need to remain patients – not epidemiologists or physicians – and contribute from their own experience
- Involving patients progressively in the research process  Doctors must be willing to spend time preparing and going through
- Education for researchers, regulators and all stakeholders on how to partner with patients on research-related activities
- Research is not only about genetics and treatments. It must also be on psychology, social issues, burden,...