138 impasse de Champs Gervais - 74890 Bons en Chablais - France - 33 (0)4 56 30 74 43 - mcjlboiteux@aol.com - www.cutislaxa.org
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CLI ~ NEWS N° 41 ~ AUGUST 2024

This first half of the year has been very busy.
Three major events allowed me to bring Cutis Laxa Patients' voice, further and higher, in Geneva, in Paris and in Philadelphia.
Being the only association worldwide advocating for the rights and needs of Cutis Laxa patients, we must attend these events as often as possible.
It takes a lot of time and energy, a lot of personal costs and a lot of patience for those who share my private life.
I want to thank them here, officially. Without their support, without their tolerance, without their demands, also, sometimes, I could not continue to work for Cutis Laxa patients.
THANK YOU !

Marie-Claude Boiteux, Chair

NEW CONTACTS, FAMILIES' NEWS

Daniella, Lucie, Sara, Jamila, Shelley, MDMBen, Mirxan, Maya and Andres have joined us. We are now in contact with 549 patients and families. Their isolation and loneliness facing Cutis Laxa has disappeared.

Welcome to you all, we are happy you joined our Big Cutis Laxa Family.

MEETINGS, EVENTS AND EXHIBITIONS

FACE-TO-FACE

In Geneva,taking the opportunity of the World Health Assembly, on **29th May,** two events were organised to bring the voice of rare and dermatologic disorders to the World Health Organisation (WHO).

Globalskin (IADPO) had choosen « Skin Diseases as a Global Public Health Priority – No Universal Health Coverage without Skin Health » as a subject to think about and work on.

Regarding **Rare Diseass International (RDI),** the importance of investment in rare diseasorders was at the heart of discussions with the final aim of The World Health Assembly drafting a resolution on rare diseases in 2025. A first step was made with RDI being admitted as an official partner of the WHO.

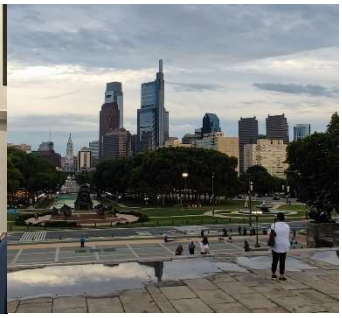
The most important event for rare skin disorders was held **in Paris from 12th to 14th June**. The **World Congress on Rare Skin Disorders (WCRSD)** gathered together doctors, researchers,patients and industries. The patient's point of view was at the core of common interests with a plenary session organised by the patients which attracted over 150 participants.

The session gave a voice to patient representatives coming from South America, Africa, Nepal and the Philippines.
Two other sessions provided a large space for patients' participation.

One was dedicated to Patient Education Programmes and the other touched on the future and how to increase our togetherness to improve diagnosis and treatments.

These were three intense days full of exchanges, discussions and future projects for patients' improved condition.

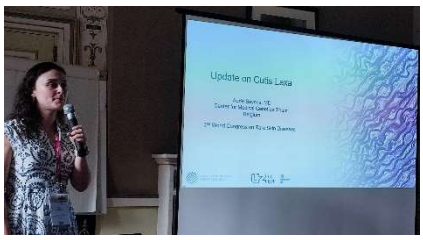
After two years of hard work, **the CONECT project** (Cardio-Ocular Network for Connective Tissue disorders) has come to the end of its funding.
It was important to meet to thing about the future of CONECT and the funding it requires, but also about common projects that may concern some of CONECT's members. This is why we gathered together in **Philadelphia 24th to 26th July**.
Besides the pleasure to see us without any screen and sharing special moments, we had a whole day of work and reflection about future projects. They are still in preparation, but I will certainly have the opportunity to let you know about them in future issues of CLI-News. It also was very important to exchange and see how our lives follow similar and parallel pathways.



ON-LINE ACTIVITIES

These are extremely numerous so it is difficult to give all details here. Whether it was Conferences (G5 Health, ECRD, etc), or Annual Meetings (RDI, Eurordis, etc), or workgroups (ERN-Skin, Globalskin, FFP, Fimarad, etc) or even Webinars or trainings sessions(ANSM, AMR, etc), being able to attend on line is more economical, more ecological, less tiring, even though face-to-face meetings allow or richer exchanges.

RESEARCH – MEDICINE – GENETICS



Cutis Laxa update During the World Congress in Paris in June, an update of the knowledge on Cutis Laxa was presented.
Pathophysiology of CL is represented as a defect of the elastic fibers. In the inherited forms, it is a defect of synthesis/assembly of elastic fibers, due to genetic mutations. When looking at the acquired form of Cutis Laxa, the process is different as it is instead a destruction of elastic fibers that were , at first, normal.
As of today, 18 sub-types have been identified in the inherited forms, together with related disorders such as Arterial Tortuosity Syndrome or MACS Syndrome. These identifications allow a new classification.

The forms with prominent connective tissue features (Tortuosity, Skin, Bones, Emphysema) and those with main neurological features with ou witout intra-uterin growth retardation are still there. The validation of this new classification is still ongoing. It will be a better tool for diagnosis, integrating clinical data, ultrastructural findings and pathophysiology. It will be the basis for novel guidelines..

The growing role of patient groups in healthcare research.

Patient groups contribute to research in three different ways :

- *In clinical trials and real world evidence :* The groups offer a unique perspective and act as research subjects, advisors, reviewers, and even researchers. They can be involved at any stage of clinical trials from the pre-approval and design to making sure the research results reach their patient community.
- *In Health Technology Assessment (HTA) :* This is a systematic process that evaluates technologies like medicines and medical devices. The assessment determines if they are clinically effective, cost effective or have any social/ethical impact. Patients and patient groups are increasingly involved in the process, providing insight into their condition and the impact of new technologies.
- *In Regulatory decisions :* Patient groups are involved in regulatory processes offering real-world evidence for better medical regulation. In Europe, their views are crucial for transparent communication on medicines and valuable input into the review of information on medicines like package leaflets and safety communications.

LEGISLATION ~ SOCIETY

Canada takes steps towards improving rare disease care. *(orphanews 2024.03.18)*
The network RareKids-CAN and the Canadian Network for Rare Disorders (RCMR) recently created by the Canadian government aim to improve quality and availability of healthcare and treatments available for Canadians living with a rare disorder.

Sweden launches its first national strategy on Rare Disorders *(orphanewzs 2024.03.18)*
The National Council for Health and Well-Being (CNSB) identifies four priorities to elaborate the strategy : Building a project based on patient groups' needs and reaching a consensus on aims and priorities ; relying on existing structures of care and taking into account the role of the different stakeholders ; focusing on the clarification of roles and spreading knowledge ; remaining in the field of responsibility of health and medical care. A public consultation is ongoing.

The World Skin Health Coalition (WSHC) ended its campaign « Not just my skin » on 19th April. It reached over 5 million people. Materials of the campaign were viewed over 17.8 million times. Over 6,800 people in 130 countries across 6 World Health Organisation regions signed the open letter urging health policy leaders to act.

In Belgium, the Federation RaDiOrg for rare disorders publishes propositions for a better future for people living with a rare disorder. *(orphanews 2024.05.23)*
The current Belgian Plan for Rare Disorders was established in 2013. This is why RaDiOrg together with the workgroup of the Belgian College for Human Genetics and Rare Disorders sent these propositions to all political parties. They were positively welcomed. The Minister of Health described them as a marker for the future project of the National Plan for Rare Disorders.

Members of the European Parliament approved the provisional agreement on the European Disability Card which aims to enhance mobility for people with disabilities across the European Union. (*orphanews 2024.05.23)*

Scotland publishes first Genomic Medicine Strategy *(orphanews 2024.05.23)*
The 2024-2029 plan intends to create a more coordinated genomic medicine service across the country to establish an equitable, person-centered and rights-based service.

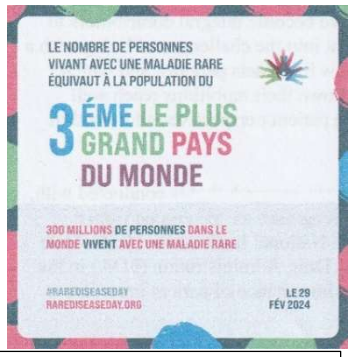
THE MEDIA

12th June 2023 Doctissimo : interview with Pr.Bert Callewaert : All you need to know about Cutis Laxa (in French) : https://www.doctissimo.fr/sante/maladies/maladies-de-la-peau/cutis-laxa-la-maladie-de-la-peau-relachee/a89563_ar.html
30th January Patient Stories: Dave Jacob : A late diagnosis of Cutis Laxa and creating ThinkGenetic (in English) : https://www.greygenetics.com/a-late-diagnosis-of-cutis-laxa-and-the-creation-of-thinkgenetics/
February 2024 Patient View : The growing role of patient groups in healthcare research. (in English) : https://www.patient-view.com/2024/the-growing-role-of-patient-groups-in-healthcare-research/
06 Mars Le Monde : Rare Disorders - the Challenge of Diagnosis
15 Mars Festival de la Communication Santé : Interview with Catherine Baissac, Patient relationship – Pierre Fabre Dermato-Cosmetics (in French)
28 Avril Charente Libre : The « Brin d'Aillet » run in Sireuil (France) to the benefit of Cutis Laxa Internationale (en Français)

5 % of the global population are suffering from a rare disorder

Equity for people living with a rare disorder means equitable access to diagnosis, treatments, care, social protection and opportunities

INFOGRAPHICS



300 million people worldwide live with a rare disorder. That is equivalent to the population of the 3rd biggest country.

Understanding mothers' experiences through narrative analysis *(orphanews international 2024.02.13)*

A study was recently published in « *Qualitative Health Research* ». Even if this study was made for Prader Willy Syndrome, the results and recommendations that result can apply to any rare disorder. Researchers identified themes focusing on the complexity and rarity of the disorder, including the desire to be normal, how ordinary becomes extraordinary, isolation, behavior and normative standards, and alternative stories of mothering. Based on these results, four key recommendations could be established:

- Recognition of the challenges of mothering a child with complexity;
- Moving beyond functionality and impairment to participation and quality of life;
- Considering anew how to tell the families stories and their need for support ;
- Engaging with mothers to determine care priorities.

The findings of this study highlight how more qualitative research methods such as narrative analysis can be used in rare disease research to identify care and policy priorities to improve the lives of people living with a rare disease and their families.

WE NEED HELP

For several years, our Spanish translations have been made by volunteers, friends or just acquaintances. For the past three years, these translations have been done with online tools. I am not sure about their quality ……. If you or someone you know wishes to help us, we would be very happy to entrust the Spanish translations to you. Feel free to contact me if you need more details : mcjlboiteux@aol.com

THANK YOU

Note from the person who currently supports the English translation: this is not a task that takes long and Marie-Claude usually gives me plenty of time. I find it extremely interesting to keep abreast of the work that CLI is doing.
In consideration that this is the ONLY organisation in the world engaged in supporting patients with Cutis Laxa and which, at the same time, is advancing the work of many other rare conditions, it is indeed a very small thing to do. I would encourage any Spanish speaker to get involved. Thank you.

Our Website :

Since January 1st, 2024, 2,532 people visited our website.

The pages mostly visited were genetic transmission, Cutis Laxa definition and photos.

ASSOCIATION'S FINANCES

Think about renewing your fees and donations **in 2024.**

Do Not Forget Us,

do not forget the patients who count on you, without you we would not be able to help them anymore. You will find a membership and/or donation form at the end of this newsletter.

MANY THANKS IN ADVANCE

FOLLOW US ON FACEBOOK

The Facebook Private Group : Is dedicated to patients, their parents, doctors and researchers. If you need to share with other sufferers, other parents, what Cutis Laxa means in your life, come and join our private group: http://www.facebook.com/groups/62977351521/

The Facebook Public Page : If you wish to follow us and get all the news from the rare diseases and disability world, in France and worldwide : https://www.facebook.com/CutisLaxaAssociation/

ENJOY SUMMER REFILL YOUR ENERGY

Thank you for filling and sending back this form with your membership fee and/or your donation

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FIRST NAME……..……..…….

ADDRESS……

ZIP CODE……..…….TOWN……..……

COUNTRY…….……

☐ **Wants to support Cutis Laxa Internationale** by sending his/her membership fee : **€ 30** (when paid in Euros) ; due to the very high cost of foreign currency exchange for small sums, if you cannot pay in Euros we require the equivalent of € 50 in your own currency.
☐ **Wants to send a donation**. Amount……..
☐ **Orders the book « Cutis Laxa, Story of a rare disorder, Patients' testimonies » and I add the amount of** ☐ **25 € to my membership fee and/or donation**

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