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## (LI ~NEWS N° 42 ~ JANUARY 2025

With the uncertainty of today's world, where war, conflicts, violence, climate disasters seem to invade our daily life, I dare to hope that Cutis Laxa Internationale is a haven of peace. I dare to hope that glows of comfort and support brighten the life of the patients who joined us. This is what we are here for.

Receiving the diagnosis is violent and comes with a feeling of isolation and loneliness due to the rarity of Cutis Laxa. We do as much as we can for this feeling to dissipate quickly for those who find us. The answers we bring them and the fantastic work our rare diseases worldwide experts give hope to all.

23 years ago, when we founded CLI, we did not imagine the importance and the international impact it would have and there is still so much to do.

I hope that in 2025 all patients can have access to information on their disorder and are not isolated.

Marie-Claude Boiteux, Chair

### NEW CONTACTS, FAMILIES' NEWS

555: that's the number of patients who joined CLI, with the new members Kim, Tigui-Jamilah, Clare, Ali et Yassin, Isla, Marie-Astrid and her brother, Soumaya, Jonas et Goundoba. Welcome to all of them. We are happy you found and joined us.

Our Big Cutis Laxa Family is here to help you, support you and answer your questions.

Alas, we are very sad to let you know about two deaths: Lucille, who joined our private Facebook group in 2010 and was an active member and Safaa who was only 18. To their mourning families we give our support and thoughts in this difficult time.

Since we set up CLI in 2002, 27 patients have passed away. They remain in our hearts and thoughts.

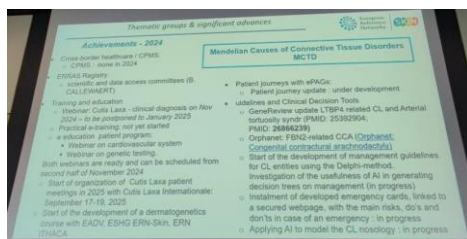
### MEETINGS, EVENTS AND EXHIBITIONS FACE-TO-FACE



16th-17th September 2024 : Event “Exp’Ose Ta Peau” (dare to show your skin) organised in Roubaix by LéoPharma Laboratory. Located in front of the town hall, we were able to raise awareness on skin disorders with passers-by. Then, in December, during the Dermatology Days of Paris, « Exp’Ose Ta Peau » had, again, a privileged place.



24th-25th October 2024 : ERN-Skin Board Meeting at “Institut Imagine” in Necker Hospital in Paris. This important annual gathering allow us not only to meet again and make time for face-to-face exchanges on future projects, but also to take stock of what we have achieved during the past year.





28th-31st October 2024 : European Elastin Meeting (EEM 2024) in Reims. All the scientists working on Elastin were gathered together on the initiative of the teams at Reims University. These days were fascinating, instructive and created therapeutic hope.



To top it all, it was a great joy to meet Pr Zsolt URBAN from Pittsburgh as well as researchers from Pr Romain DEBRET's team in Lyon.



All the exchanges, shared moments, the possibility to give patients' point of view, as well as relaxing times: these were the keys to the success for these 3 days. We also know that future collaborations will result from them. It was really important to be there.



### 08th November 2024 FIMARAD Annual Meeting

Patient organisation representatives attended the Annual Meeting of the French Network for Rare Skin Disorders.

On this occasion, they especially mentioned the progress of the working group « Diagnosis roving and impasse » with the analysis of the patient survey. The results of this survey are planned for publication.

26th November 2024 Pfizer Patient Forum, during which Marie-Claude Boiteux participated in a roundtable on « Minimising obstacles and maximizing impact: key learnings from pilot projects and advice for action ».



### 03rd-06th December 2024 : 6<sup>th</sup> Conference of the French Federation for Skin (FFP) et Dermatology Days of Paris (JDP)

This event is unmissable: the opportunity to gather together and evoke the achievements of the year, and explore future projects in roundtables and interactive workshops. It was also the last time I was leading the FFP Conference as my 3 years mandate chairing the FFP has come to its end.

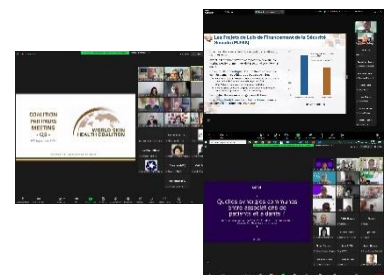


Thanks to our stand during the JDP, I have had the great joy, more than 30 years later, to see again Dr Karim CHERTI. At that time, he was a junior doctor in the genodermatosis unit at Hôpital Saint Louis in Paris. He was a member of the team who diagnosed my daughter, Cécile, and wrote his thesis on her case. It was a very moving moment for both of us.



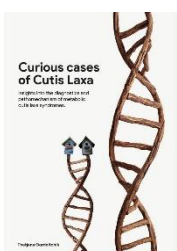
## ON-LINE ACTIVITIES

According to the rhythm established in 2024, attending numerous meetings and webinars online meant better progress with some projects and greater knowledge and competencies to carry on raising patients' voice.



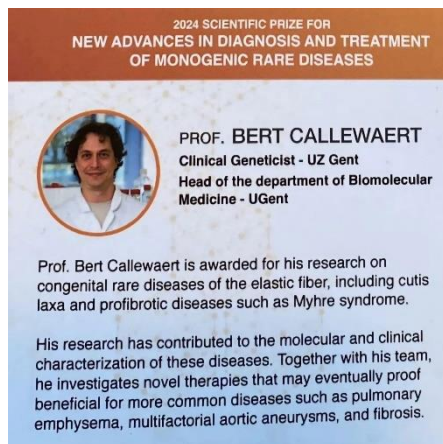
## RESEARCH – MEDICINE – GENETICS

Dr Thatjana Gardeitchik was a successful candidate with her thesis defence on 9th September 2024. The title is: « Curious cases of Cutis Laxa , Insights into the diagnostics and patho-mechanisms of metabolic cutis laxa syndromes ». You can find it online, in English only ([https://www.globalacademicpress.com/ebooks/thatjana\\_gardeitchik/](https://www.globalacademicpress.com/ebooks/thatjana_gardeitchik/))



**The 7<sup>th</sup> CUTIS LAXA DAYS will be held in GHENT – BELGIUM  
4<sup>th</sup> to 6<sup>th</sup> FEBRUARY 2026**





On 12th December 2024, Astrazeneca awarded its « Scientific Prize 2024 » to Pr Bert Callewaert.


We are very happy and proud that Pr Callewaert is one of the rare worldwide experts on Cutis Laxa. He is awarded the prize for his research on congenital rare diseases of elastic fibers, including Cutis Laxa and profibrotic diseases such as Myhre's syndrome. His research has contributed to the molecular and clinical characterization of these diseases.



Together with his team, he works on very rare forms of Cutis Laxa and identified several new genes and entities within the Cutis Laxa spectrum.

*« Elastic fibers play a crucial role in the elasticity and mechanical properties of tissues such as the skin, lungs and blood vessels.*

*We studied cutis laxa,, a group of conditions characterized by loose and excessive skin folds, and Myhre's syndrome, in which tissues harden or heal, to understand the structure and function of elastic fibers. This knowledge offers the possibility of finding treatments for age-related disorders. »* (Pr Bert Callewaert ©Fondation AstraZeneca)

 The AstraZeneca Award recognizes the innovative nature of his work and his contribution to the advancement of science.

## LEGISLATION ~ SOCIETY

On 30<sup>th</sup> August 2024, Rare Diseases International (RDI), the Arab Republic of Egypt, the State of Qatar and Spain co-hosted a public webinar on the campaign for a World Health Assembly (WHA) Resolution on Rare Diseases in 2025. The event featured discussions of why such a resolution is critical, what it can provide to people living with a rare disease (PLWRD) regionally and nationally and what needs to be done in order for the campaign to succeed. Despite recent global advances in recognizing the unmet needs of PLWRD, notably the 2021 UN Resolution on Persons Living with a Rare Disease and their Families, the community still faces a number of significant challenges. In particular, limited access to treatments and care, delayed diagnosis, and elevated financial burden. These struggles are often exacerbated in low- and middle-income countries.

A WHA Resolution on Rare Diseases would help address these challenges by building on the momentum generated by the UN Resolution and instituting a comprehensive, health-focused global framework for rare diseases with clear targets and deadlines for all WHO Member States. The campaign for such a Resolution was initially spearheaded by RDI, with Egypt, Qatar, Spain, Malaysia, and France agreeing to act as co-sponsors. (©Orphanews 2024.09.18)

## THE MEDIA

02nd September 2024 : Info-Chalon Official opening of the resource center for tele-expertise in dermatology in Chalon-sur-Saône.

10th September 2024 : Le Journal de Saone et Loire Official opening of the resource center for tele-expertise in dermatology in Chalon-sur-Saône.

15th November 2024 : Masterclass at La Sorbonne recording of an interview with Marie-Claude Boiteux for the videos used during the Masterclass at the University for Patients in La Sorbonne (in French) (<https://universitedespatients-sorbonne.fr/>)

29th November 2024 : Charente Libre Handing over the € 2,500 check to the benefit of Cutis Laxa Internationale by Triathlon de Sireuil

Autumn 2024 : la Gazette Bonsoise Cutis Laxa Internationale, beyond the frontiers of Bons en Chablais

**A huge THANK YOU to the leading team of the sport events organised in Sireuil, France (Trail du Brin d'Aillet and Triathlon). Thanks for faithfully supporting us, thanks to the energy you put in organising those events, thanks to the volunteers who work with you and support those sport events. Thanks also, of course, to all the sportsmen/women who participate: you are handing over € 2,500. That is enormous for us !!!**

**Thanks to you, we will be able to help patients come to Ghent, Belgium, to attend the Cutis Laxa Days and benefit from a consultation with Pr Bert Callewaert, worldwide expert for Cutis Laxa.**

**On behalf of all the patients, I am deeply grateful to you. Marie-Claude Boiteux, Chair**

### **Our Website :**

Since its opening in February 2002, our website allowed almost 75,000 visitors to get information on Cutis Laxa and for patients among them, to find the resources and reliable information they need and contact with other patients.

### **ASSOCIATION'S FINANCES**

Think about renewing your fees and donations **in 2025.**

### **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/>

Privacy Policy : You receive this newsletter because you are on our list. Send us a request to the address mentioned below if you wish to be taken off our mailing list.

*I wish you a Beautiful and Peaceful year 2025*

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

NAME.....

FIRST NAME.....

ADDRESS.....

ZIP CODE.....TOWN.....

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☐ **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**

☐ **Bank Transfer :**

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Please don't forget to mention your name and complete address with the transfer, as well as the details of the amount (membership fee, donation, book,...) so we can send you a receipt and the book if you ordered it.

☐ **Sending a cheque to**

**CUTIS LAXA INTERNATIONALE ASSOCIATION - 138 impasse de Champs Gervais - F-74890 Bons en Chablais - France**

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