138 impasse de Champs Gervais - 74890 Bons en Chablais - France - 33 (0)4 56 30 74 43 - mcjlboiteux@aol.com - www.cutislaxa.org
IBAN : FR76 1810 6000 4296 7525 0578 892 BIC : AGRIFRPP881

CLI ~ NEWS N° 37 ~ JANUARY 2022

The 6th Cutis Laxa Days will be held at the University Hospital of Ghent(Belgium) on **14th, 15th and 16th September 2022. Registration is open on our website: https://www.cutislaxa.org**

2021 has been a tough year: many online meetings, online training courses, online conferences, etc.
As the pandemic « slowed » a little in the Automn, we were able to organise and/or take part in various events.
And the routine work and preparation the Cutis Laxa Days in Ghent took a lot of time and energy too.
I will not say more here, but see more details below about our great news.

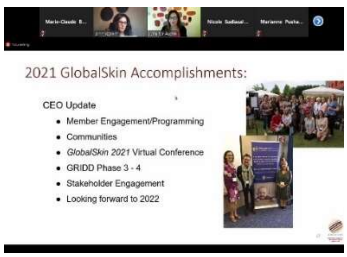
I wish you all a sweet New Year 2022.

Marie-Claude Boiteux, Chair

NEW CONTACTS, FAMILIES' NEWS

Cutis Laxa Internationale now gathers 499 patients coming from all parts of the world:
Kim, Amina, Nura Bay, Linda, Dawson, Riley, Cecelia, Rai, Sutton, Bodhi, Katia, Heather, Inter, Melany and her daughter Lilian, Husseini, Xenia as well as Samardashi and Arthur joined us since our last newsletter.
Our Big Cutis Laxa Family is glad to welcome them and give them all the help and support they need.

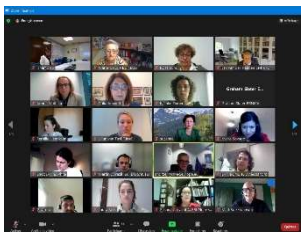
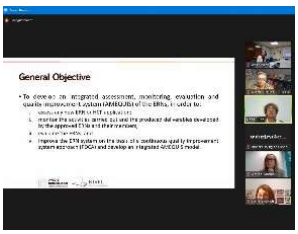
MEETINGS, EVENTS AND EXHIBITIONS



29th September : Globalskin Annual Meeting. The International Federation of Skin Patients' Organisations was set up in Canada. It has grown a lot and allows to share efforts worldwide for patients' improved quality of life.

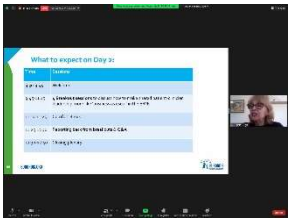
26th September , 3rd, 9th and 16th October As part of Pink October, dedicated to breast cancer and as part of the Blue Walk, dedicated to elderly people, Bons en Chablais' local Council organised several events that allowed CLI to collect over € 1,000. Thanks to all of you..

1st October Bersot Estate Agency gave us a € 300 chéque. They had proposed to their clients to make a donation to the organisation they had chosen for each transaction. A huge thank you to them.

26th-27th October AMEQUIS workshop. The global aim was to simplify and improve the revalidation steps for ERNs when applying for reassessment.

4th-5th November All ePAGS joint annual meeting with Eurordis to help each other and improve their patient representatives' work in the ERNs. These two days of hard work allowed us to establish the priorities that need to be improved.

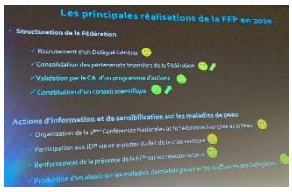


23rd November ERNskin Executive Committee

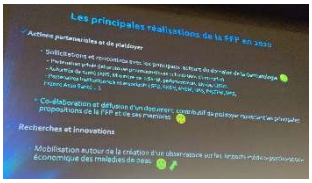
25th-26th November FIMARAD Annual meeting

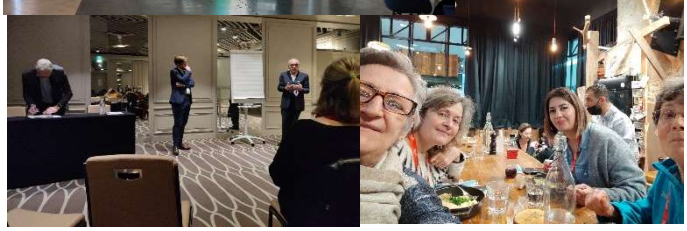
Those two events gave the opportunity to take stock of the actions and improvements achieved in 2021. Online training courses, using the CPMS consultation platform, setting up registries and specific activitiesfor each thematic group were the strong points for ERN-Skin. Regarding Fimarad, the results of the different workgroups showed the improvments still needed. These two institutions have similar objectives, one is at the French national level : Fimarad , the other is at the European level : ERN-Skin. They are both coordinated by Pr Christine Bodemer.


30th November The 3rd Conference of the French Federation for Skin was opened by Mr Olivier Véran, French Minister of Health and Solidarity. Convinced that democracy is necessary in health issues, he recognised the quality of the FFP advocacy and the real obstacle course its 21 member organisations can face. FFP represents more than 20 million patients. this third conference was an undeniable success in bringing together high quality speakers : health Professionnals and Institutions. The work of the FFP is recognised by all stakeholders in the field of dermatology.



After the Conference, the Annual Genarl Meeting was held and the annual report brought to light the many achievements in 2021. The FFP member organisations elected the new board of Directors and its new Chair : Marie-Claude Boiteux.



1st, 2nd & 3rd December For the first time this year we had a stall during the Dermatology Days of Paris. With its conferences, pharmaceutic and cosmetic laboratories stalls, Associations' village, and e-posters, there's no need to demonstrate the richness of those days.

It was the opportunity for many exchanges with health professionnals as well as with association leaders, all concerned by dermatology.


22nd December Christmas Market. Freezing cold and coming out of the pandemic meant that the this market was not as interesting as it should have been. But we stood firm all afternoon to welcome the hardy strollers. Despite the entertainment, takings were very low……. We will do better next year.


RESEARCH – MEDICINE – GENETICS

NEW MUTATIONS

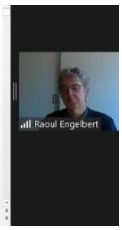
As I wrote in our previous newsletter, several new genetic mutations were recently found:

LOX : This gene of the 5-Lysyl oxidase family is involved in initiating of cross-linking of Elastin and Collagen. The mutation leads to cardiovascular, respiratory and bone symptoms, especially fractures. This is why it was initially considered to be a new type of osteogenesis imperfecta (glass bones disorder). But the discovery of fragmented elastic fibers allowed this mutation to be included in Cutis Laxa Syndroms. It is a recessive form.

EFEMP1 (Fibulin3): The consequences, besides lax skin, of this mutation are multiple hernias and joint hypermobility as well as mild intellectual/learning disability. It is a new recessive type of Cutis Laxa.

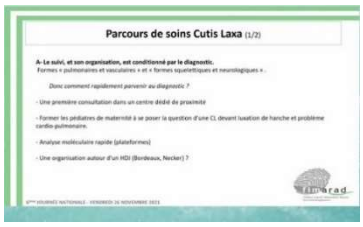
LTBP1 : This mutation is distinguished by lax skin, inguinal hernias, craniofacial dysmorphology, various heart defects and prominent skeletal features (short stature, brachydactyly, craniosynostosis,..). It is another new recessive type of Cutis Laxa.

PI4K2A : This 4th new mutation is characterised by the following clinical signs : lax skin, involuntary movements (neurological issue), dysmorphism and intellectual/learning disability. It is also a recessive type.



A 5th new mutation has recently been found and we are longing for it to be published so we can tell you about it.

Thanks to all the researchers for their amazing work in basic knowledge of Cutis Laxa. All these findings are essential to give patients better care and offer them a better quality of life.



Diagnosis Roving and Deadlock

This workgroup of FIMARAD (Rare Dermatologic Disorders French Network), of which Marie-Claude Boiteux is a member, is interested in these issues. An important piece of work for Cutis Laxa was presented during the FIMARAD meeting in November.

Due to the great diversity of types and symptoms, the quality of the healthcare journey for CL patients is intimately linked to diagnosis. So, it is not enough to diagnose a Cutis Laxa one needs to be really precise about which type of CL with the help of a molecular analysis (genetic testing).

The quality of the patient's healthcare as well as the quality of their life depend on the swiftness of the molecular diagnosis. In France, a number of systematic tests are carried out immediately after birth to evaluate the newborn's health (hernias, tone, mobility, hips,etc)..

A table has been created associating these systematic examinations with various CL symptoms to allow a faster clinical and molecular diagnosis. So, for instance, if the systematic examination of a newborn shows hip dysplasia, the next simple step is to see if the skin is lax and the newborn has hernias to suspect ARCL2A or Geroderma Osteodysplastica. The diagnosis can then be confirmed with a genetic test. Concordance of symptoms is the first step to a faster diagnosis.

LEGISLATION – SOCIETY

16th December2021 The United Nations General Assembly voted the First Relotion for People Living With a Rare Disorder

After a first adoption by consensus in November, the final text became an official United Nations Resolution with the vote during the General Assembly on 16th December.

Rare Disease International, together with Eurordis and the Non Governemental Organisations (NGO) Committee for Rare Disorders advocated relentlessly with United Nations members to elaborate the resolution "Addressing the Challenges of Persons Living with a Rare Disorder and their Families". This hard work has now been rewarded.

Australia : Government is funding the « RArEST » initiative for 3 years with an amount of $ (AUD) 1.9 million. This initiative is led by Universities and the Federation « Rare Voices Australia », to improve Education, Support and Training courses for Rare Diseases . (©Orphanews 16th November 2021)

The UN calls on France to completely change its idea of disability. They especially tackle too « medical » approach and « systematic » institutionalisation. (© Hospimédia)

Ratification of **the African Medicines Agency (AMA)** Treaty. One of its priorities will be to act against sub-standard and fake medicines.
And also, the **First African Summit on Rare Diseases** was held in Accra (Ghana) on 1-3 December. The main theme was to engage in a dialogue between all stakeholders in the national and regional rare disease eco-system in Africa. (©Orphanews 2nd December 2021)

In the USA : Establishment of 31 Centres of Excellence for Rare Disorders. They will cover several specialties and provide rare disease patient education, training courses for physicians, and will contribute to enhanced research on rare disorders by promoting collaboration in this field. (©Orphanews 02 Décembre 2021)

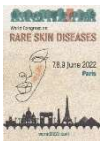
In Italy : Adoption of a National Law for Rare Diseases. This law provides a definition of rare diseases as one with a prevalence of less than five patients per 10 thousand. This definition fits in with the one used at the European level. The law also provides for the establishment of a National Committee for Rare Diseases within the Ministry of Health. (©Orphanews 2nd December 2021)

THE MEDIA

6th October : Le Dauphiné – Bons en Chablais : On the occasion of « Pink October » and in partnership with the local council of Bons en Chablais, Cutis Laxa Internationale held an information stall and sold various items.

11/10/21 Le dauphiné – Bons en Chablais : Pink and Blue Walk The health organisations of the town were associated with the Blue Walk dedicated to elderly people and with Pink October dedicated to Breast Cancer.

Autumn 2021 : La Gazette Bonsoise : Report of all the events organised on the occasion of « Pink October ».



SAVE THE DATE :
The First World Congress on Rare Skin Disorders will be held in Paris
on 7th to 9th June 2022

www.cutislaxa.org at the end of 2021, **our website** indicated **61 951 visitors** since its opening in 2002. Only 13 % of them visited more than once.

Leetchi Kitty :
€ 312 were collected, Thanks to all who participated.

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WE NEED YOU :
You have got ideas, you want to help us, you want to organise fundraising events for the benefit of CLI.
Feel free to contact us and we will work together to achieve your project.
We need you to be able to organise the next Cutis Laxa Days.

ASSOCIATION'S FINANCES: Think about renewing your fees and donations in **2022**. **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/

HAPPY NEW YEAR 2022 !

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

NAME……

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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Name of the Bank : Crédit Agricole Bons en Chablais
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

☐ **Do you want to receive CLI~News via Email ? yes** ☐ **no** ☐

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