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# CLI ~NEWS N° 36 ~ AUGUST 2021

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

In 14 months we will gather together in Ghent (Belgium) for the 6th Cutis Laxa Days.
All of us, patients, doctors and researchers are longing to attend this event as it is an exceptional time for sharing and acquiring knowledge.
It allows each patient attending to meet other patients and be informed on the latest findings on Cutis Laxa and the specific type they are suffering from.
It allows doctors and researchers to strengthen their network, meet patients and promote new research programs.

Organising these Days means a huge amount of work, applying for grants and planning logistics.

I will do all I can for those Days to be a success; it would follow the pattern that allowed for the unforgettable memories of the previous Days organised in France in Annecy (2016), Lyon (2011), Paris (2008), La Rochelle (2004) and Soyaux (2002).

Marie-Claude Boiteux, Chair

## NEW CONTACTS, FAMILIES' NEWS

Our « Big Cutis Laxa Family » grows day after day.
Its new members : Joanna, Ayse and her two brothers, Delphine, Elizabeth, Adar, Aakshay, Fabricio, Solange, Andrea, Zahid, Dilsa, Jannae & Julia, Fatiha and her sister, MaryAnn come from Poland, Turkey, Canada, Saoudy Arabia , USA, Israel, Brazil, Italy, Colombia, France.
All around the world, 479 patients are now part of our Cutis Laxa patients' community.

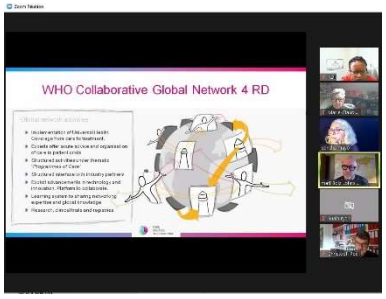
On 27th I heard with deep sorrow about Maddie's passing. She was 19 months old. I had talked a lot with her mother during her long stay in hospital. Rest in peace sweet angel, your pain has disappeared.

## MEETINGS, EVENTS AND EXHIBITIONS


*14th-16th January 2021 : "Hacking Health Lyon"* : During three days we took part in a Health Projects Contest. Even if we finish last, it was the occasion to meet young computer scientists willing to developp a diagnosis application. We are still working with them and sincerely hope that they will succeed.

***Rare Disease International (RDI) organised several events:***

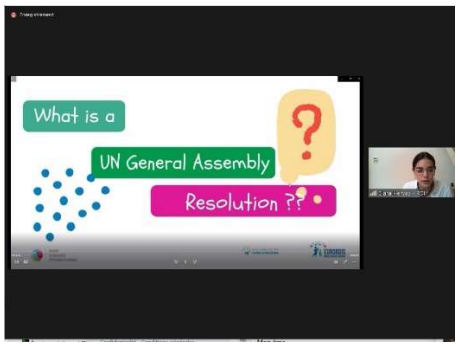

On 14th January , Webinar : Key Issues for a UN Resolution for Rare Diseases : Inclusion and Human Rights, Appropriate Care, National Stategies, Rare Diseases in the UN System, Monitor Progress and Implementation.




On 4th February, Webinar : « World Health Organisation Collaborative Global Network for Rare Diseases » (WHO CGN4RD) : Implementation of Universal Health Coverage from care to treatments; Expertise; Structured Activities under thematic « Programmes of care »; Exploit advancement in technology and innovation;

Collaboration Plateforms ; Sharing networking expertise and global knowledge ; Research, clinical trials and registries.

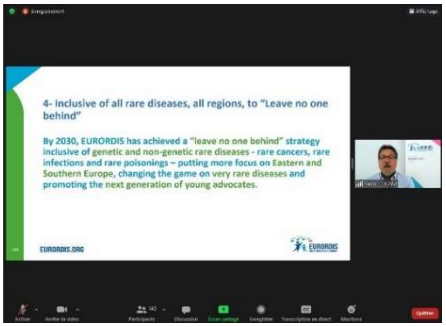
On 25th and 26th May : RDI members meeting and Annual Report : In 2020 RDI's activities were equally internal, with webinars for its members and external with outreach to Permanent Missions to the UN in New York. The aim is to promote Rare Diseases as a priority in International Policy and work towards a UN General Assembly Resolution for Rare Diseases with sustainable development goals.




***Globalskin (International Organisation for Skin Disorders) held two events early this year :***

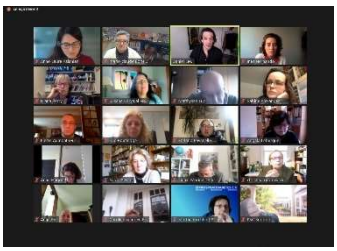
On 25th February : Webinar : « The Power of the Patient Community in the Rare Diseases Movement » with a great presentation from Dr Ségolène Aymé, first CEO of Orphanet.
On 3rd March : Structuring the European Community for Skin Disorders inside Globalskin.

***The first half of the year was also quite busy with Eurordis :***



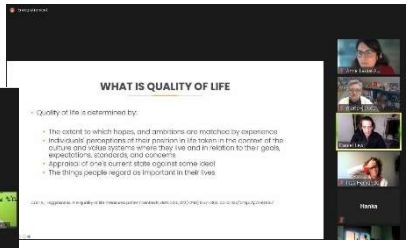

On 12th May : Annual Members meeting ; on 10th June : Annual Report - Setting up RDI (Rare Diseases International), editing surveys on a regular basis (Rare Barometer), helping patient representatives in the European Reference Networks (ePags), European Conference on Rare Diseases (ECRD), advocating at the European (EC) and international (UN, WHO) levels, etc… There is no lack of work. The actions led by Eurordis have changed, change and will continue to change the quality of life and care for people living with a rare disease in Europe and worldwide.


***And also workgroups, training and information for Patient Representatives (ePAGS)***



On 18th March : « Good Practice : How to measure results, What is Quality of Life, How to build up a survey, …»

On 9th June : ePAGS Steering Committee meeting




## RESEARCH – MEDICINE – GENETICS

**NEW MUTATIONS**

Even if I cannot tell you more since publications have not yet taken place, I am very happy and proud of our researchers who work on Cutis Laxa :

**5 NEW MUTATIONS have just been discovered.**
**For all of you whose precise type has not yet been identified, this is an extraordinary chance.**

You can be tested now with the new mutations. **Attending the 6th Cutis Laxa Days in Ghent in September 2022** can be the opportunity for you to know more about the precise type you are suffering from.
I hope I will be able to give you more information about those new mutations in our next CLI-News.

19th February : First ERN-Skin Scientific Day. 114 doctors and researchers attended. Each thematic group presented the work done on the disorders they are concerned with. For the Heritable Connective Tissue Disorders (HCTD), a study is taking place in the Netherlands to evaluate prevalence and severity of fatigue, pain, disability and global health for children and teenagers suffering from the most common HCTD.

During the last meeting of ERN-Skin Executive Committee, on 8th June, the agenda was very fully. After Marie-Claude Boiteux's presentation on the epags' work on « Patient Journeys » for each disorder concerned by ERN-Skin, the Patients Satisfaction Survey, Registries, e-learning, the SPOT application, results of 2020 and organising, in Paris in 2022, the next Rare Skin Diseases World Congres – these wide ranging questions showed ERN-Skin's dynamism and how it implements concrete projects.

## LEGISLATION ~ SOCIETY

On 23rd February : The European Commissioner for Health and Food Security, the French and Czech Republic Ministers of Health and Members of the European Commission opened the **RARE 2030 Conference,** gathering almost 590 attendees together. After two year of continuous work, Eurordis and the European Community annouced the 8 recommendations arising from this work. The future of rare diseases over the next 10 years starts now, comprising :

- ✓ A European policy framework ;
- ✓ Earlier, faster and more accurate diagnosis;
- ✓ A highly specialised healthcar ecosystem ;
- ✓ Guarantee the social and economic integration of people living with a rare disease;
- ✓ Encourage the meaningful participation, engagement and leadership of people living with a rare disease;
- ✓ Rare disease research is maintained as a priority;
- ✓ Data is used at its maximum ;
- ✓ Improve the availiability, accessibility and affordability of rare diseases treatments.

**French Federation for SKIN (FFP)**
Since the beginning of the year, Georges Martinho, FFP General Delegate, led several projects at the double all showing positive and very promissing results :

✓ The FFP plea was published in March. It gathers together the difficulties, needs and proposals of thousands of persons suffering from skin diseases in France, wether these diseases are rare or not. Delivered to all Health Authorities and elected Representatives acrossthe country, its aims are : long-term improvment of quality of life for skin patients ; advocacy against social and local inequalities ; improved support of care journeys ; getting patients' expertise recognised. The first reactions are very positive.

✓ National Office of Solidarity for Autonomy (CNSA) and Local Houses for Disabled People (MDPH) working towards the recognition of a « Skin Disability » and adapting the administrative steps to the reality and specificities of dermatologic diseases.

✓ Rallying Members of Parliament : After sending the petition, several meetings have taken place with Members of Parliament and Senators. The working areas fo the Members of Parliament and the FFP include meeting patients to increas knowledge of their needs, bringing the issues to the parliamentary debate and proposing amendments to legislation.

✓ Taking par in the inter-associative task-force of pharmaceutical industries : Following the recommendations published by the European Commission on how to improve the information on health products, a survey and a meeting allowed members of the FFP to share their point of view and take part in this work.

✓ Educational aids for children and teenagers : These aids have an informative and educational aim illustrated by concrete situations. We will soon start working with illustrators, various tools have been mentioned (books, videos,etc). The first two aids, for 6-11 year olds and 12-18 year olds, are taking shape and should be edited by the end of this year.

The FFP is doing great and has real concrete results.

**RARE DISEASES INTERNATIONAL**, Eurordis and the United Nations NGO Committee for Rare Diseases are leading a global campaign for the adoption of a UN General Assembly Resolution on Persons Living with a Rare Disease (PLWRD) and their families.

# THE MEDIA

On 16th March, Chiara's testimony (in French and in Italian) was posted on Facebook : https://www.facebook.com/photo?fbid=3509557279156318&set=pcb.3509561645822548

On 7th April, « Le Tribunal du net » edited an interview with Cécile (in French). Watch it again on our website https://www.cutislaxa.org/fr/quoi-de-neuf/ (videos) or on the "Tribunal du Net" page "https://www.facebook.com/letribunaldunet/videos/1782251148618795

In April and May, every week, Ewenlife published one of the 8 videos about Cutis Laxa. From diagnosis to medico-social care and treatments, all issues are tackled. You can watch them again (in French or with subtitles in your own language) on our website https://www.cutislaxa.org/fr/quoi-de-neuf/ (videos) or on Youtube : https://www.youtube.com/watch?v=XPI5X03FOEw&list=PLvUNl3idDQjTKEL_-tfQjZUE9ixRnH_aD

The drawing competititon "Story of my skin" organised by the René Touraine Fondation received high quality works : Please see below the 1st prize in the 14-18 age range.

**Patients' inequalities of treatment** (Opinion piece published in « Le Monde » on 15th May). Even if this piece concerned Bourneville Tuberous Sclerosis, another rare skin disorder, Cutis Laxa shares most of its arguments :

* The Diagnosis odyssey
* Various Symptoms that do not make the disease « visible » to the Authorities
* Only one support organisation, the only recourse to advocate for patients' rights, and the only source of funding of projects led by the organisation.
* Little/no support for research
* Depending on where they live, some patients do not have access to expertise for their disease.
* Refusal to cover travelling costs to hospitals
* It is necessary to break those inequalities in funding and favour patients instead of economics.

**Improving the healthcare experience**
**(survey Rare Barometer Eurordis-Rare Disesses Europe, 3 905 réponses)**

Issues to be worked on to improve the healthcare experience :

-Asking about patient's health after a consultation

-Encouraging patients and carers to take part in a group to help them manage their rare disease better.

-Helping patients or carers to manage emotions linked to patients' state of health.

Centers of Expertise have an essential role to play for Rare Diseases.


**www.cutislaxa.org**, Mid-June 2021, **Our Website** counted 55,454 visits since its opening in 2002.

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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 2019. **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 the flavours of Summer !

**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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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.

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