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## LI ~ NEWS N° 34 ~ SEPTEMBER 2020

2020, What a difficult year !! The Covid-19 pandemic has turned everything upside down.

Many events have been cancelled or changed into online virtual meetings. We have had to learn how to work differently : teleworking, changing to part-time hours, ...

Patients with « rare » conditions have been directly hit: appointments, surgeries and care have been cancelled....

As a second wave is approaching, we must remain vigilant and follow the recommendations.

But life didn't stop during all those months. We worked to keep Cutis Laxa Internationale alive and transmit patients' voices.

Several projects are ongoing, even if some of them have just been postponed :

- The Cutis Laxa white book has just been issued ;
- The 6th Cutis Laxa Days, initially scheduled in June 2021, is postponed to September 2022

And many others...

New sufferers have joined us. Alas others passed away too soon. Jaiden, almost 16, and Zeinab, 21, have gone to join the stars. I am deeply sad and, once again, send their families all my thoughts and condolences in the name of Cutis Laxa Internationale.

Marie-Claude Boiteux, Chair

### NEW CONTACTS, FAMILIES' NEWS

Caetano, Tamapa, Defne, Tami, Yusuf, Tracey, William, Atay Robin, Sunusi, Hanna, Umar, Jo, Sarah, Talita, Randi, Carol, Eli, Gina, Sonia, Maria, James, Ayyaz, Mariya and Oluhle have joined us since the beginning of 2020.

Koweit, Dubai, Sierra Leone, Nigeria and South Africa are the additional colors on our map of world..

We now gather together 440 patients worldwide. Of them, 18,6 % have a Dominant type (ADCL) , 17,1 % a Recessive type (ARCL), 22 % an Acquired one and 40% do not know, or not yet, their exact type. The other types (MACS, OHS, GO,...) represent 2,3% of the total.



Since Cutis Laxa Internationale was set up, we have received many patients' and parents' testimonies. They are now collected in a newly published book : « **Cutis Laxa, Story of a Rare Disorder, Patients' testimonies** ». It is available in French, English and Spanish. We will send it to you on request, at €25 per copy.

### MEETINGS, EVENTS AND EXHIBITIONS



**09th February :** Shortly before lockdown, the Gospel choir had organized a masterclass and we were given the benefits. Cécile,



together with her brother and her sister took part in this singing and joyfull afternoon.



EUROPEAN COMMISSION HEALTH  
EUROPEAN COMMISSION

ORIGINATED ON 14-15 May 2020

# THE FUTURE OF DIAGNOSIS

NEW HOPES, FRUSTRATIONS AND CHALLENGES

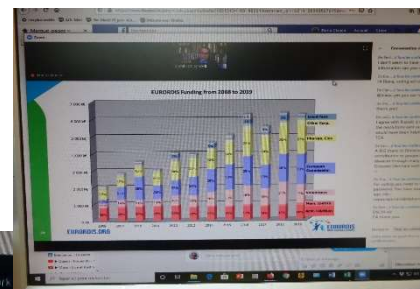
A EUROPEAN COMMISSION INITIATIVE

TOOK 20 MINUTES TO SUPPORT UNHAPPY COMPANY?

NEW BORN SCREENING

WHAT IS NEXT?

TIMING: FASTER, ACCURATE, DIAGNOSIS



**Patient Engagement in ERYs and ePAGs**

- Promote a meaningful patient engagement in ERY activities & provide regular support to ePAGs
- Define and pilot an ePAG model assessment framework to evaluate the impact of the ePAGs implemented in the ERYs. Roll out the evidence to all pilot patient organisations in the ERYs
- Promote and facilitate exchange in ePAGs good practices and to improve the management of knowledge needed to enable Europe to develop and implement relevant information
- Review the 24 ePAGs governance allowed to inform the process for the renewal of ePAGs advocates mandates in 2021 ahead of the next EN 5 years cycle. Review the ePAG Consultation contributes to the promotion of concrete methods to capture feedback on patient satisfaction & their experience across the ERYs (including Through Your Narrative)
- Deliver a comprehensive capacity-building programme Leadership School for ePAG patient advocates within EMERGEO Open Academy

Logos: European Patient Advocacy Group, European Endocrine Reference Centres, EMERGEO Open Academy.

# Advocate for Rare Diseases to be a Priority in the next Decade 2020-2030

- following the COVID-19 crisis, Promote a more integrated and ambitious EU Public Health Strategy in areas of action of high economic added value and high economic impact such as health threats, cancers, Alzheimer, mental illnesses, obesity and diabetes, and next diseases ...
- Prepare for the next decade of rare disease legislative & policy Framework to take the necessary steps to requalify rare diseases as a public health issue.

Execute last phase of Rare 2030 Foresight Study on Rare Disease Policy to create a new policy Framework for rare diseases. Consolidating all proposals and organizing them in the form of recommendations to the European Commission, EU Council and European Parliament in February 2021.

Based on rare 2030 recommendations, in line with the 2019 Court of Auditors special report on cross border healthcare and with recent Council Recommendations to review the rare disease policy, continue to consolidate political options with EU institutions to update, review or replace the overall EU rare disease strategy.

- Engage with upcoming EU Presidencies to support the EU policy strategic review
- Re-launch and expand the European Parliamentary Advocates for Rare Diseases network, in coordination with the National Alliances.
- Prepare for exceptionally strong advocacy efforts around the Rare Disease Day 2021.

EURODIS.ORG

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Slide content:

**RARE DISEASES INTERNATIONAL**  
Advocating for the integration of rare diseases in national policies within the Framework of UNC

**How to put rare diseases on the United Nations agenda Information & Brainstorming: ROI Webinar November 2017**

On site Meeting with Martín Remón - Permanent Commission of Spain to the WHO in 2018.

- ROI provided us with the template to encourage Permanent Missions to support the action of including DRs in the 'Sustainable' Development Goals when the 2030 agenda was being programmed.
- They immediately became interested in us!

On site Meeting with Foreign affairs of Spain  
Consejero de la Representación Permanente de España en Naciones Unidas

**RDI ACTION PLAN 2020-2021**

**Objective:** Inclusion of more in vitro diagnostics and therapies for RDI, contained essentially by relevant disease communities, into the WHO Expert List.

**Method:** In liaison with WHO ECL, EML, Qualification units and existing RDI members.

- Promote the opportunity to all potential applicants (in-house experts, device manufacturers, hospitals and company market holders).
- Present and explain EML and ECL programmes, opportunities and procedures to RDI members at large

**Activities 2020:**

- 2 webinars with WHO ECL and EML Secretariat
- 2 webinars with case studies / patient groups with experience
- Progressively involve relevant Expert Faculty members into analysis of current essential lists, guidance to RDI members, peer to peer learning, liaison with WHO
- Attendance by RDI members and Expert Faculty of ECL and EML Expert Committees open to stakeholders

**Coordination with INDIRG Rare Disease Treatment Advisory Working Group**

INDIRG  
International Network of  
Infectious Disease  
Rare Groups

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RARE DISEASES INTERNATIONAL

Advocating for the integration of rare diseases in national policies within the framework of UHC

## The international character makes consensus possible

- ❖ The European Recommendations helped us to position the issue of rare diseases locally.
- ❖ A global and common framework of action increase solidarity character.
- ❖ Working together Strengths us.
- ❖ ALWAYS take the Chance!

### Global Actions for RD



Rare Diseases International is the global alliance of people living with a rare disease of all nationalities across all rare diseases.

RD's mission is to be a strong common voice on behalf of rare disease patients around the world, to advocate for rare diseases at all levels, to ensure that rare diseases are not forgotten, to represent the community and ensure their participation in decision making.

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**7th August :**  
After several months in lockdown, it was a real pleasure to participate in the « Summer Music » evening in Fantasia Park in Annemasse (France).



Respecting all sanitary recommendations we were able to welcome people at our stall where they could find, besides our now usual sausages and french fries, information on Cutis Laxa as well as various jewellery and items we sell. A very friendly evening which allowed us to get some funds, more than welcomed with all our other scheduled events being cancelled.

## RESEARCH – MEDICINE – GENETICS

### ACQUIRED CUTIS LAXA :

➤ A scientific study on Acquired Cutis Laxa (ACL) will be led by Dr Bert Callewaert at University Hospital of Ghent (Belgium).

This type of Cutis Laxa has never been studied before.

It is a great opportunity to learn more about it : how it appears, what are its causes and maybe a first step to treatment. Worldwide patients with ACL can take part in it. The doors of hope are opening up for them.

➤ A possible improvement of the symptoms in Acquired Cutis Laxa associated with monoclonal gammopathy or multiple myeloma: A haematologist from Sao Paulo (Brasil) has had very satisfactory results in two cases with an autolog bone marrow transplant. For those two patients the results were more than encouraging though we certainly cannot talk about treatment or cure yet. Many studies and therapeutic trials are still necessary to get there. Nevertheless this is a huge leap for those patients.

### RARE SKIN DISORDERS AND COVID-19

An observational study was initiated by the European Reference Network ERN-Skin and the French Health Network for Rare Skin Disorders (FIMARAD) : « COVID-19 and rare skin diseases. European observational study (data research) during an epidemic ». It concerns paediatric and adult patients with rare skin diseases and suspected or confirmed COVID-19 infection who consulted a medical team that is part of the ERN-Skin or FIMARAD. Professor Christine BODEMER (Hôpital Necker-Enfants Malades – Paris, France) is the principal investigator..

The main objective of this European observational cohort study (research data) is to determine the impact of a COVID-19 virus infection in a cohort of patients with rare skin diseases and particularly whether these rare diseases and their treatments are risk factors of infection severity

### **SAVE THE DATES**

The 6th Cutis Laxa Days will be held at the University Hospital of Ghent( Belgium)  
on 14th, 15th and 16th September 2022

## LEGISLATION ~ SOCIETY

**The COVID crisis reveals how patients' and associations' role is poorly recognised** (Le Monde 2020.07.06)

In this newspaper article, three members of the French association Renaloo (kidney failure) make an unanimous statement : Associations of Health Users were forgotten in the heart of the sanitary crisis :

- There were no official recommendations for « at risk » people.

- Associations had to establish themselves « Recommendations for patients » based on international recommendations or those from other countries. ;
- Nothing was set up to allow « at risk » people to stop working as soon as the crisis started ;
- Cooperation with patients and their associations was considered incidental and with no priority ;
- Despite the collective stakes we all had to face, democracy in health suffered a lot ;
- Never quoted as being part of the various parties committed to protect vulnerable people, nevertheless patient associations were working on all fronts during this period ;
- In France, except a few cases, patients associations have not yet reached the place they deserve in the medical world;
- Government communication about vulnerable people focuses only on older aged people and forgets young and active people who are fragile too.

Nothing should be done without Us !

## THE MEDIA

**Avril 2020** : Rare Revolution Magazine : An article about David Ross «Meet David, a true RARE Revolutionary » and another one about Dawn Laney, Dave Jacob's daughter and co-founder of « Thinkgenetic », « RARE Heroe »

### WE NEED YOU :

**You have got ideas, you want to help us, you want to organise fundraising events for the benefit of Cutis Laxa Internationale. 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.**

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### Rare Diseases and Covid :

According to the survey lead by **Eurordis**, 9 out of 10 people living with a rare disease experienced interruption in care because of Covid. More than half of those who need surgery or transplant have seen these interventions cancelled or postponed.

And

According to a survey in the **UK**, 72% of family carers and siblings say they had to provide **a lot more** care. Overall parents talk about extreme exhaustion, stress and sleepless nights. For many of them, professional care just stopped.

### Our Website :

[www.cutislaxa.org](http://www.cutislaxa.org) has had 47 642 visits since its opening in 2002, that is almost 7,000 since 1<sup>st</sup> January 2020

### 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**

**A HUGE THANK YOU to two people - they will know who they are – for the € 6,000 they have donated. Thanks to you Ladies organising our coming Cutis Laxa Days seems more feasible.**

### 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/>

*Take care of yourselves !*

**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.....

☐ **Bank transfer :**

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Bank name: Crédit Agricole Bons en Chablais

Please don't forget to mention your name and complete address with the transfer so we can send you a receipt

☐ **Sending a cheque to**

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

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