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## (LI ~ NEWS N° 35 ~ FEBRUARY 2021

At the dawn of this New Year, we wished we'd gotten rid of this nasty virus that upset us so much in 2020. Alas ! this has not been the case and we still have to learn to live with it for a long time.

Our working habits have changed: no more travelling, no more taking part in events but more time spent in the office since now everything is organised online.

The lack of fundraising opportunities has become a problem. Companies are financially fragile and thus less keen to fund our projects.

We are reducing expenditure to a minimum, but we are still there and we hold on, for you all, patients and families who count on us, on our commitment and our support.

There a little more than one year left to make up the necessary funds to organise the 6th Cutis Laxa Days held in Ghent (Belgium in September 2022. We will make it, we believe in it. Yes We Can !!!

Marie-Claude Boiteux, Chair

### NEW CONTACTS, FAMILIES' NEWS

Madalynn, Liam, Héloïse, Amelia, Brittany and her mother, Roxanne, Richelle and 4 members of her family, James and Charles, Ehran, Lisa, Emam are new members of the big Cutis Laxa family.

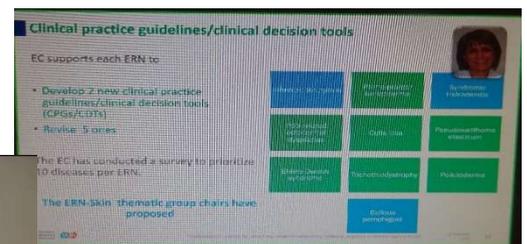
We are now 466 patients and their families, coming from all parts of the world. We are united and we get our voices heard. We support each other. And as the Rare Disease Day motto declares: « We are RARE, we are STRONG, we are PROUD ».

### MEETINGS, EVENTS AND EXHIBITIONS

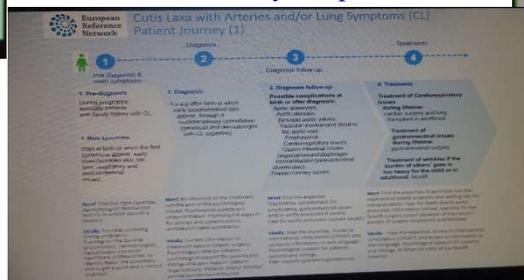
As previously mentioned, since september 2020, none of the events we should have taken part in were held physically, In Real Life. Nevertheless we took part in many important events with particularly rich lessons and contacts.



2nd October 2020 : ERN-Skin annual Board Meeting was a great success : 4 new affiliated partners joined, coming from 4 countries not yet represented in



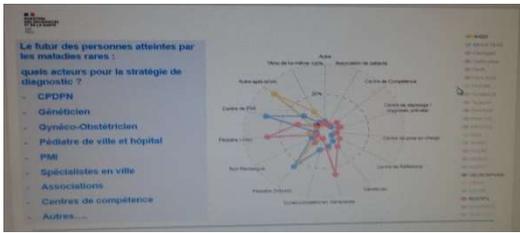
our ERN ; the results of the ongoing work, and especially the announcement that Cutis Laxa is one of the 10 disorders selected by the coordinators of the subthematic groups that will be prioritized for the development of



clinical practice guidelines/clinical decision tools- these show show the dynamism of ERN-Skin. The patient representatives (ePAGs) presented their work too, especially the « Patient Journey » for several of the disorders concerned by ERN-Skin.

3rd November 2020 : The first Congress organised by the French Alliance for Rare Disorders gathered together over 250 people. Elected representatives and Healthcare professionals presented the opportunities, improvements and perspectives for rare disorders for the coming 10 years.

The subjects of lively workshops were ; Access to diagnosis; Access to treatments and Improving the life journey. To improve patients' quality of life depends on improving the relationship between local and hospital professionals.



**Rational for 3 New ERNs**

- Incomplete coverage by some of the 24 networks: Actions
  - New diseases included in the 2019 update (ERPs)
  - Continuous extension of diseases by ERNs: premature request for recognition by national authority and approval by the DoH
- Large disease groups not addressed by any ERN
  - infectious diseases
  - gynecologic-obstetric, fertility patients and pregnancy
  - mental health diseases

5th and 6th November 2020 : Patient Representatives (ePAG) gathered

together, as they do every year, to share improvements, difficulties and successes of their work in the ERNs. Supported by the

Eurordis team, this coming together shines a light on the benefits, but also the gaps, of the organisation of the ERNs. As Enrique Terol, in charge of Rare Disorders issues at the Health Executive Board in the European Commission, explained, issues linked to gynecology, pregnancy and family planning are not covered by any of the actual ERNs. This annual meeting was also the opportunity to gain new knowledge/competencies to improve our work of patient advocacy.

**Closing Plenary How to organise a structured dialogue with your wider patient community?**

**Idea 1: Connecting the communicators**

- Virtual or hybrid: collect the best of both worlds (social media, and content) to create a community
- Key benefits: enables to keep awareness of project with better effects of the PANG, connecting language barriers, and/or languages
- Key challenges: time to create, build the links (not being able to meet in person)

**Idea 2: Intro ERN Webinar to Patients**

- Virtual or hybrid webinar presenting the ERN, what it does, the role of PANG, - indicators distributed by centers and patients
- Key benefits: patients get to know what is the ERN, Potential to give more info on PANG role (Structure & how it works)
- Key challenges: Content (ERN structure and content), time, language, including the support issues that patients of ERNs face

**Tableau de bord sur le partage des résultats de...**

- Waiting on best ideas (Choice multiple)
  - How 1: Connecting the Communicators: 96%
  - How 2: ERN webinar to Patients: 31%
  - How 3: Create a Local ERN directory for accepting patient community support: 30%
  - How 4: Handbook on how to make a comprehensive, short and clear document: 45%
  - How 5: Focus on the patient experience: 28%
  - How 6: Making good practice: creating tools to support centers in Africa, Central Asia: 31%
  - How 7: ERN webinar to Patients: 17%
  - How 8: ERN webinar to Patients: 73%

27th November 2020 : The Annual FIMARAD meeting (French Health Network for Rare Skin Disorders). Core discussions of the day were : Taking stock of the work done in 2020; Issues of the MDPH files (Local House for Disabled People); How to involve Patient representatives;

organising the workshops for 2021; and ePAGs' work.

**Mieux comprendre**

Dossier évalué par une commission des droits et de l'autonomie des personnes handicapées (CDAPH)

- Prescription de compensation du handicap (PCH)
- Cartes, RQTH, affilation assurance vieillesse

**3ème JOURNÉE NATIONALE FIMARAD**

**L'évaluation des situations et l'identification des besoins en situation de handicap**

MDPH 13-pole enfant : -22 Episemaine -1 à 2 CDA (mois)

Activité 2019 : 11 800 familles ont déposé un dossier à la MDPH 35 548 décisions rendues

**3ème JOURNÉE NATIONALE FIMARAD**

Webinar interface showing participants and speakers.

**Activités de FIMARAD en lien avec le PMARS**

Action 4.2 : Créer un observatoire des traitements placé au sein de comités consultatifs multidisciplinaires d'évaluation (observatoire bureau/COPE) dans chaque filière de santé maladies rares

- Groupe de travail de la filière « Médicaments/Recherche » : création de l'observatoire des traitements de la filière (action 4.2 du PMARS)
  - Site d'échange
  - de concertation
  - de centralisation de l'information concernant le repérage de nouvelles molécules en développement et un recensement des médicaments existants, avec ou hors AMM

L'objectif de ce groupe de travail est de mettre en place une stratégie spécifique en matière d'accès aux médicaments

- Recrutement d'un chef de projet scientifique/médical
- veille bibliographique avec une contribution d'information concernant les molécules d'intérêt à partir de données de la littérature et de celles de l'observatoire au sein des CHU et CCAM

Action 4.4 : Mieux encadrer les pratiques de prescriptions hors AMM

- GT DGS-DGOS : piloté avec 8 FAMP référentes dont FIMARAD

**GT Impasse diagnostique**

- Buts
  - Problématique de l'impasse diagnostique
  - Identifier patients en impasse (BNOMIR)
  - Mise en place du recueil complémentaire pour les patients en « impasse et errance diagnostique » choix des items (obligatoire/non obligatoire), choix unique ou plusieurs choix
  - Homogénéisation des règles de codage et de remplissage dans le Set de Données Minimum
  - Algorithme décisionnel (selon les pathologies)

1st December 2020 : The Annual Meeting and 2nd Conference of the French Federation for Skin (FFP) : Following the 2020 annual report and the projects for 2021, the FFP conference opened the floor to experts on the impact of Covid-19 sfor skin disorders, paediatric dermatology, users' participation on the evaluation of health products and dermatology research. Needless to say that it was an intense and riveting morning.

**Programme (1/2)**

10h15 - 10h40 : « Impact du Covid-19 sur les affections dermatologiques »

- Madame le Professeur Marie BEYLOT-BARRY, Présidente de la SFD, Chef du service dermatologie au CHU de Bordeaux

10h40-11h05 : « La Dermatologie pédiatrique : ses spécificités, ses avancées »

- Madame le Docteur Christine LABREZE, Dermatologue au CHU de Bordeaux

11h05 - 11h30 : « Contribuer à l'évaluation des produits de santé et aux rencontres précoces entre le HAS et les industriels dans le cadre du développement de recherche clinique »

- Monsieur Marc GUERRIER, Conseiller médical du Service Engagement des Usagers de la Haute Autorité de Santé

**Programme (2/2)**

11h30 - 11h55 : « ATU/RTU : état des lieux et perspectives »

- Professeur Bernard GUILLOT, Professeur Emérite, Faculté de Médecine de Montpellier

11h55 - 12h10 : « La recherche dermatologique aujourd'hui et demain »

- Professeur Olivier DENEIRE, chef de service dermatologie CHU de Montpellier, Président du Conseil scientifique de la SFD

12h10 - 12h45 : « La Fédération Française de la Peau : Une nouvelle organisation en 2021 pour soutenir et développer les missions des associations de patients en dermatologie »

- Georges MARTINHO, Délégué Général et Jean-Marie MEURANT, Président de la FFP

**Assemblée Générale Mardi 1er décembre 2020**

**La Fédération Française de la Peau**  
Une nouvelle organisation pour soutenir et développer les missions des associations de patients en dermatologie

Intervenants : JM. Meurant, Président / G. Martinho, Délégué Général

**LA CUTIS LAXA**

Association Française des Malades de la Cutis Laxa

Carte de France montrant les associations régionales.

**SYMPOSIUMS SATELLITES**

**PROGRAMME JDP 1-5 DECEMBRE**

**E-POSTERS**

**ACCUEIL**

**Village des associations**

Direct | Sessions enregistrées | Programme | E-Posters | Exposition | Espace MCO | Symposiums | Point Info

2nd to 4th December 2020 : Paris Dermatologic Days ,which we were able to attend online, so as to be visible despite everything. Nevertheless we sorely missed the opportunity for face to face meetings with representatives of laboratories to talk about our activities.

## WE NEED YOU TO MAKE PROGRESS

As part of the patient representatives' work in the ERN, a workgroup on « **Pregnancy and Family Planning** » has been set up. To feed into our work, we are looking for your testimonies about your journey, your difficulties, your successes, your pains and your victories. Thank you for sending them at your convenience : by post at CLI's address, by email at [mcjlboiteux@aol.com](mailto:mcjlboiteux@aol.com) or via facebook in a private message at <https://www.facebook.com/marieclaud.boiteux>

As for the workgroup on « **Diagnosis Roving and Impasse** » in the French Network of Dermatologic Rare Disorders, your testimonies are more than welcome. Thank you for sending them at your convenience : by post at CLI's address, by email at [mcjlboiteux@aol.com](mailto:mcjlboiteux@aol.com) or via facebook in a private message at <https://www.facebook.com/marieclaud.boiteux>

## THANK YOU FOR YOUR CONTRIBUTION

## LEGISLATION ~ SOCIETY



### Rare diseases challenges in the US, barriers to diagnosis, care and treatment

The National Organisation for Rare Diseases (NORD) has conducted a survey from October 2019 to March 2020 to assess how far RD patients living with a rare disease (RD) in the United States (USA) are accessing care today. 42% of the respondents experienced delays in diagnosis because of limited medical specialisms. Another challenge is the geographic barrier in accessing care. Also, more RD patients have no access to school today (26% compared to 5% in 1989), and 62% are unable to attend work compared to 23% in 1989. The report states that research in RD will also contribute to boost access to treatment. (© Orphanews 24.12.2020)



### Rare 2030: Backcasting the future of European Reference Networks

The project Rare 2030, a two-year policy EU Pilot Project, commissioned by the European Parliament and coordinated by EURORDIS, aims at guiding reflection on rare disease policy in Europe through the next ten years and beyond, based on prospective exercises. The project will deliver recommendations in early 2021 towards the future scenario deemed most desirable through a broad consultation with over 200 stakeholders. The Rare 2030 Prospective Study has paid particular attention to the status of European Reference Networks (ERNs):

- **Governance and Strategic positioning of ERNs**
- **Integrating ERNs to national systems and frameworks**
- **The role of ERNs in virtual care delivery and cross-border healthcare**
- **ERNs, research, and the data ecosystem of the future** (© Orphanews 11.12.2020)

## THE MEDIA

**November 2020** : « Practitioner's Magazine », French monthly magazine for healthcare professionals « Living with.....Cutis Laxa »

**11th November 2020** : Recordings from the FIMARAD annual meeting (in French):

[https://www.youtube.com/watch?v=71EwHwy5g\\_4](https://www.youtube.com/watch?v=71EwHwy5g_4)

**December 2020** : How we got in contact with Solhand (in French): [https://www.solhand.org/coups\\_de\\_coeur/cecile-un-10eme-cas-a-tout-prix](https://www.solhand.org/coups_de_coeur/cecile-un-10eme-cas-a-tout-prix)

**With the end of the Brexit transition**, British Centers ceased all participation in the ERNs. Nevertheless, Patient representatives (ePAGs) can still take part in them as their status is different from that of Healthcare Centers.

Rare Disease  
International Day  
28th February 2021



### The challenges of people living with a rare disorder :

300 Million people worldwide ask for their rights to be fully respected. That is the aim of the campaign « Universal Health Coverage for Rare Disorders » led by RDI and EURORDIS at the United Nations.

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

**SAVE THE DATES**



[www.cutislaxa.org](http://www.cutislaxa.org), by the end of 2020, Our Website had reached 50 000 visits since its opening in 2002.



« Cutis Laxa, Story of a Rare Disorder, Patients' testimonies »

This book is available in French, English and Spanish. We will send it to you on request, using the form below, at €25 per copy.

**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/>

*Take good 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.....
- 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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 BIC : AGRIFRPP881

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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