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CLI ~ NEWS N° 33 ~ JANVARY 2020

On 20th September 2004 I received a letter from Sweden. This was my first contact with Jonny. He was 55 at the time. He had been diagnosed with CL a few months after his birth. Since early childhood, lung issues linked to Cutis Laxa were a constant worry. But it did not prevent him from getting married and having a son.
Working as psychiatric nurse, and also musician, he was a very caring man.
In 2016 he attended the 5th Cutis Laxa Days in Annecy (France). His presence was a real gift to us all. The testimony of his life journey is a memory we cherish in our hearts.

Exhausted by respiratory and cardio-vascular difficulties, Jonny left this world on 11 December 2019.
We lost a friend but also one of the oldest members of Cutis Laxa Internationale.
We send our loving thoughts to his wife and son.

What a sad year 2019 was !!!! We were longing for its ending. Two other demises made the end of the year so dark : River et Esteban , little boys who only knew hospitals during their too short lives.

Marie-Claude Boiteux, Chair

NEW CONTACTS, FAMILIES' NEWS

Moctar's son, 7, is the first patient we know of in Senegal.
Akim, Felicity, Kofi, Kollin, River, Grace, Ashley & Janet, Tina, Rhian, Meagan & her mother, Zeynep, and Anaaaya also joined us in late 2019.

Today, Cutis Laxa Internationale gathers together 409 patients : 171 in North and Center America, 34 South America, 14 in Asia, 13 in Oceania, 152 in Europe, 18 in Middle-East and 7 in Africa.

MEETINGS, EVENTS AND EXHIBITIONS

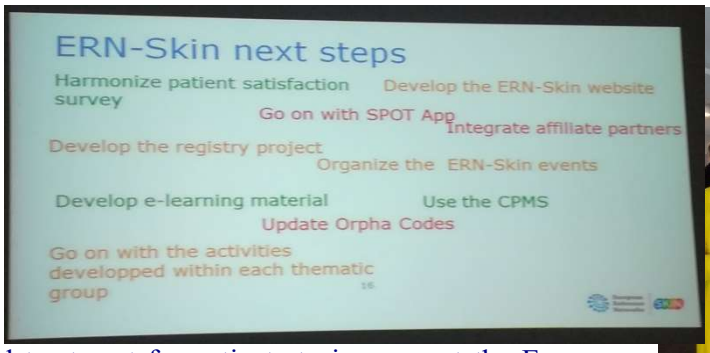
30th August Second evening With a stall in Parc Fantasia

29th November We received the cheque from the Triathlon of Sireuil

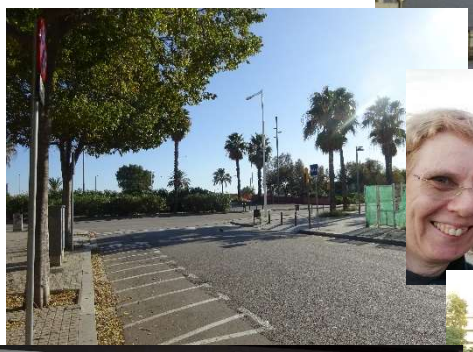
09th December We met the Gospel Choir… some projects for 2020…

3 - 4th October **:** ERN-Skin Board Meeting in Necker Hospital in Paris. Two days of intense work, whether in plenary session or in thematic groups.

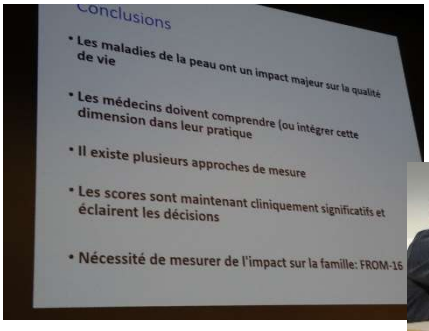
Patient representatives could express their wishes and dialogue with doctors and researchers to establish together the roadmap for the coming years. There is a huge amount of work to do but our will and synergy will allow diagnosis, care and treatment for patients to improve at the European Union level.



26 - 27th November Leadership School organised by Eurordis. After several online courses throughout the year, patient representatives in the ERNs met in Barcelone for the last two training days in Leadership. We will be issued with a Diploma at the end of the course. Even if the Catalan setting tasted like summer vacation, it was two days of real and hard work during which we increased our natural capacities and skills to become more efficient when interacting with health professionals in the ERNs. The aim of this Leadership school has been reached as we gained the competences required to improve the way we bring patients' voice forward where it matters most.



03rd December First Conference on Skin and Annual Meeting of the French Federation of Skin during the Dermatology Days of Paris. The presentation of Pr Andrew Y Finlay from Cardiff University highlighted the questionnaire on Quality of Life for patients with skin disorders, may they be rare or not. This questionnaire should be proposed to patients by all dermatologists. After this Patient Organisations took stock of their activities and difficulties.



16th December : ERN-Skin, training session in Ghent (Belgium). Organised by Pr Bert Callewaert, this session included a whole chapter dedicated to Cutis Laxa and all the scientific progress about it (see below the article in « Research-Medicine-Genetics »). Other pathologies were also presented and patients representatives took part in a panel discussion « Patients' burden : What healthcare professionals need to know ? ». Ghent University welcomed this session in its historic buildings. Pure marvel, as is all the city of Ghent..…

RESEARCH – MEDICINE – GENETICS

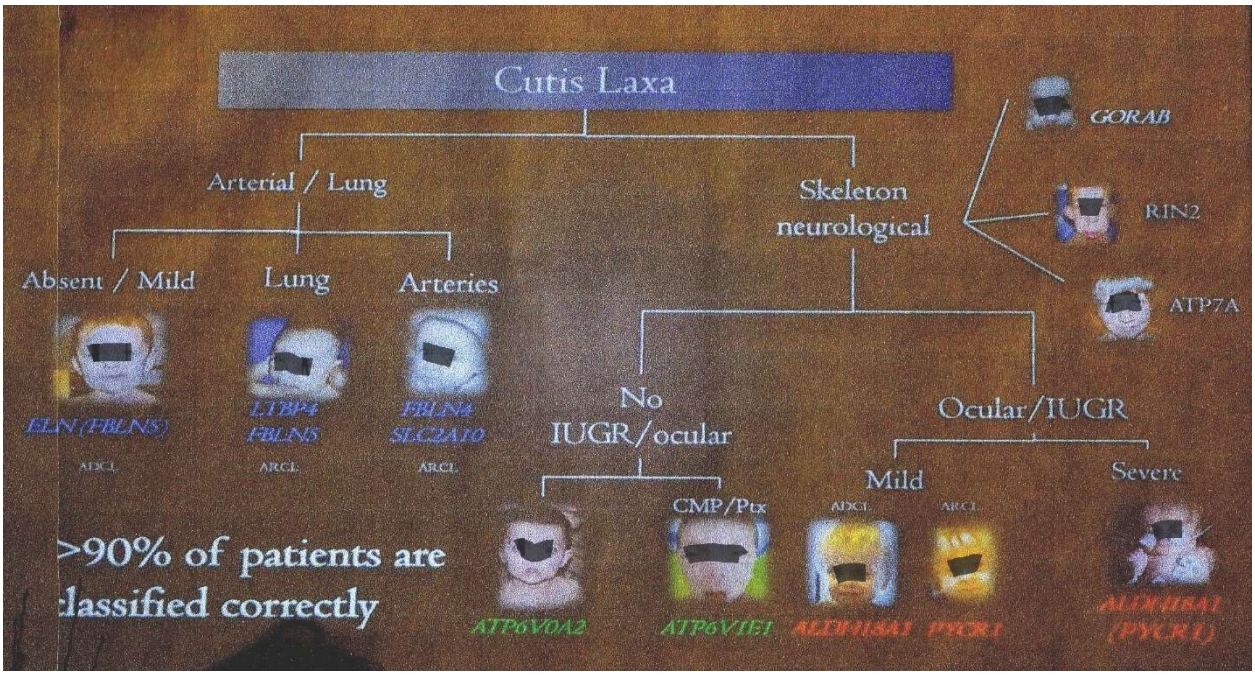
What's new about Cutis Laxa

During the training session in Ghent, the first part of the morning was almost totally dedicated to Cutis Laxa.
Several cases were presented by young doctors.

An overview talk on Cutis Laxa presented the clinical classification of Cutis Laxa disorders. Cutis laxa has an extensive clinical and molecular heterogeneity.

Based on a flowchart addressing the presence or absence of the main clinical symptoms, over 90 % of the people suffering from Cutis Laxa are correctly classified.

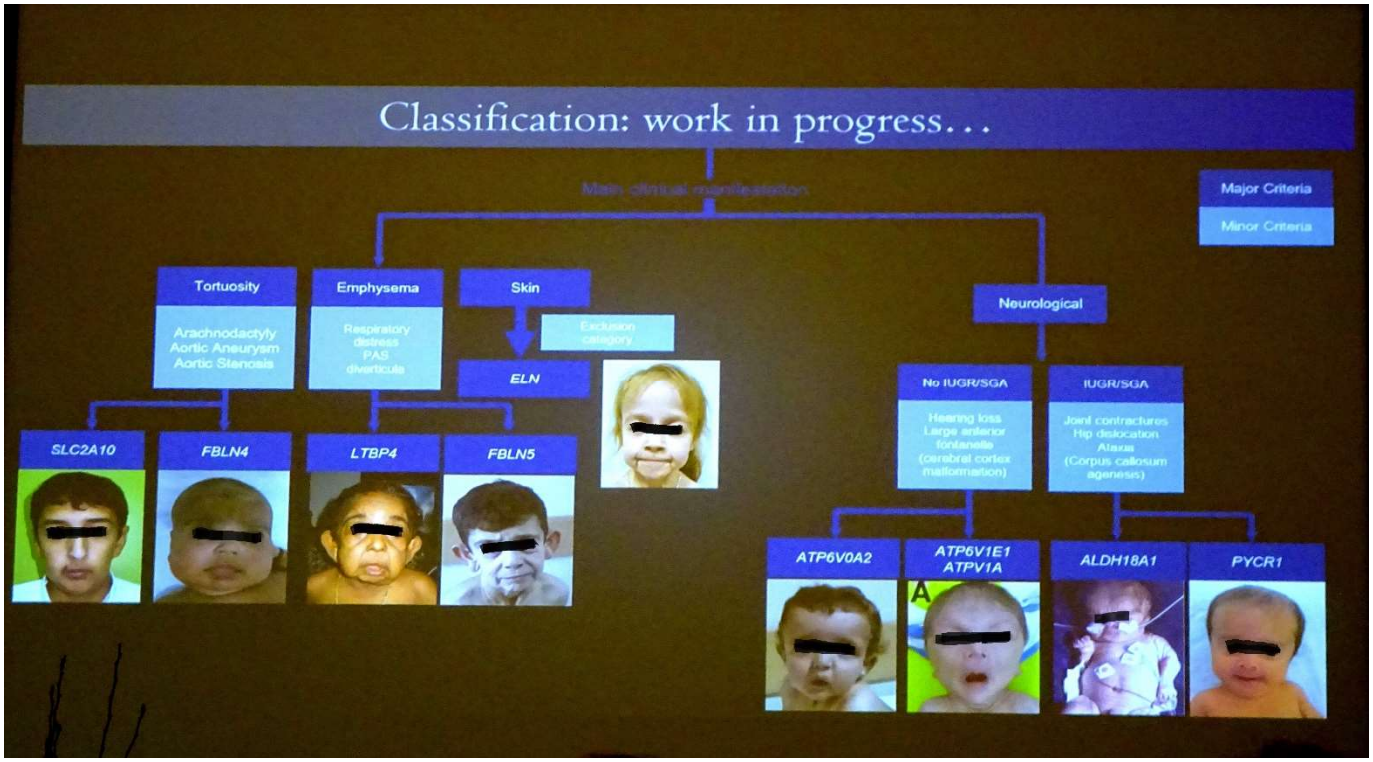
The main symptoms are: arterial tortuosity, emphysema, wrinkled/lax skin, neurological issues with or without intrauterine growth retardation (IUGR). In addition to those main symptoms, minor criteria are suggested for each type: arachnodactyly, aortic aneurysm, aortic stenosis, respiratory distress, diverticula, hearing loss, large anterior fontanelle, joint contractures, hip dislocation or ataxia.



The classification was based on a 'learning' cohort of patients known to the Ghent University Hospital. Refinement of the classification is ongoing based on a confirmation cohort from literature (650 patients). Patients are classified based upon clinical

examination and the main symptoms into major groups. Further refinement based on the minor criteria will eventually be able to predict the causal gen in over 95% of patients.

As an example, a patient with a lax skin, neurologic issues, intrauterine growth retardation as well as hip dislocation and ataxia, associated with ALDH18A1 mutation, will be diagnosed with Autosomal Recessive Cutis Laxa Type 3 or Syndrome De Barsy (ARCL3).



This new classification will help to a quicker diagnosis, interpretation of next-generation sequencing data and provide an opportunity of specific management and care for each type of Cutis Laxa.

THE PATIENT JOURNEY

Rare Skin Disorders are gathered together in the European Reference Network-Skin. Those disorders have multiple common points: late diagnosis, none or few treatments, difficulties facing other people's gaze, etc. The burden those difficulties represent is not always known by healthcare professionals. The Patient Journey is too often an Assault Course.

In order to help healthcare professionals better understand that burden, better manage it and thus give patients a better quality of life, patients must, themselves, evaluate their journey and make it known.

This is why patient representatives in all ERNs will circulate an evaluation grid to all patients. This « Patient Journey » grid gathers together the different stages that are inherent to each disease. Then each stage is described in 3 levels : Clinical presentation, challenges and needs identified by the patient, goals to improve care. The grids are then reviewed by both patients and professional experts.

Finally, this grid of the "Patient Journey" will allow patients to discuss their individual needs with only one aim : to improve care and quality of life. The "Patient Journey" will also be an important element in sharing knowledge from both patient's and healthcare professional's points of view. (European Journal of Human Genetics : " Patient Journeys" : improving care by patient involvement https://www.nature.com/articles/s41431-019-0555-6)

LEGISLATION ~ SOCIETY

WHO (World Health Organisation) signs a memorandum of understanding with Rare Disease International (RDI). According to this memorandum, the activities carried out as part of this agreement to collaborate will include goals and actions timed around WHO 13th Programme of Work that runs until 2023. In the first year, they will focus on harmonising the way rare diseases are defined internationally and on laying the ground for the development of a global network of centres of excellence for rare diseases (© RDI newsletter 2019.12.19)

At last Rare Diseases are included in the United Nations political declaration on Universal Health Coverage (UHC) : On 23rd September, two years of active advocacy by Eurordis and Rare Disease International were crowned with succes as the 193 member states of the United Nations adopted a historical political declaration on UHC including rare diseases. The principles of UHC state that all individuals and all communities must have access to the health services they need without any financial hardship. (©eurordis newsletter 2019.12.11)

How ERNs are included in the national policy regarding Rare Diseases : The French experience. The field of rare diseases is a rmarkable one in which care, knowledge improvement and the positive consequences for patients look like a virtuous circle. To reduce diagnostic delays and the number of cases of undiagnosed diseases, the French system of Reference Centers for Rare Diseases (CRMR) has been restructured in 2017. The process for approval 2017-2022 has been finalised for the 23 National Clinical Networks (Filières Santé Maladies Rares : FSMR) : 109 CRMR were approved, of which 387 are Centers of Reference (CR), 1 757 Centers of Competences (CC) and 83 Centers of Resources and Competencies (CRC). This is how the National Clinical Networks (FSMR) created in 2014 play a very important role regarding the European Reference Networks, a fact acknowledged by the European Commission in March 2017. Consequently FSMRs should be the organizational foundation of the 3rd French National Plan for Rare Diseases and were identified as mainsprings for research and innovation. (©orphanews 2019.07.31)

AUSTRALIA : Report on "Disability and rare disease: towards person centered care for Australians with rare diseases"
The recommendations of this report, commissioned by Rare Voices Australia, on behalf of partner organisations and people living with rare disease, were launched at the Parliament. They are focused on reforms which would deliver integrated care for those with health and disability care needs, improve the understanding of rare diseases by health and disability care workers and ensure that the systems can respond to the changing needs of people with rare diseases that are complex and sometimes difficult to predict.. (© orphanews 2019.12.06)

CHINA : An article published in *Bioscience Trends* acknowledges the great recent advances in the area of the needs of patients affected by rare diseases, but also highlights the recurrent difficulty in diagnosis and misclassifications. As a whole they emphasise the need for a more comprehensive and complete healthcare and social care system and advise the implementation of regional plans. (© orphanews 2019.09.18)

INDIA : IndiGen is the project announced by the country's Council of Scientific and Industrial Research (CSIR). It aims to the whole-genome sequencing of a thousand individuals from different populations, and to better understand genetic diversity around the South Asian country, while leading to clinical applications. (© orphanews 2019.12.20)

THE MEDIA

27th September : « Free Channel », Europe 1, French radio, Isabelle's testimony https://www.europe1.fr/emissions/La-libre-antenne/isabelle-est-atteinte-de-cutis-laxa-une-maladie-orpheline-qui-accelere-le-vieillissement-3922012?fbclid=IwAR3jUjNjHLCprKN2nR51F6hBSssOPTXzAhYtXgPr-HKyMduWNXGReMlmimk

01st December : Newspapers « De Morgen » (Belgium) and « De Volkskrant » (Austria) article in Dutch about Mohammed « Who helps Mohammed ? How charity stifles a Syrian family in Austria »
https://www.demorgen.be/nieuws/wie-helpt-mohamad-hoe-liefdadigheid-een-syrisch-gezin-in-oostenrijk-verstikte~b0765328/

02nd Décembre : Newspaper, Charente Libre (France) article on receiving the cheque from the Triathlon of Sireuil

10th December : TV Show, Turkey, report with Zeynep, a little girl recently diagnosed with CL
https://www.facebook.com/showtv/videos/2554386797988359/UzpfSTEwMDA0MDQ0OTc0MjIyMjpWSzoxMDE1ODA3OTkwOTQ4MTUyMg/

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.

Our Website : **www.cutislaxa.org** has had 40 721 visits since its opening in 2002, and almost 9,000 in 2019. Visitors come from 164 different countries. The top three countries in regards to the total number of visits are the USA, The United Kingdom and France. The times of the year that sum up the biggest number of visits are spring and summer.

New evaluation of the world population affected by a Rare Disease :

In an article published by Orphanet, Orphanet Ireland and Eurordis, the prevalence of rare diseases is of 3,5 to 5,9 % of the worldwide population. Rare cancers, infectious diseases and poisoning are excluded from this prevalence. Thus there is an approximate average number of people affected by a rare disease of 300 million worldwide (©orphanews 2019.10.08)

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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/

May 2020 be a gentle year for you !

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 :**
IBAN Number : FR76 1240 6001 2400 1806 3980 297 BIC Number : AGRIFRPP824
Bank Name : Crédit Agricole Angoulême Champs de Mars
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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