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CLI ~ NEWS n° 14 ~ July 2009

Cutis Laxa Internationale was born on the Ile de Ré almost 8 years ago.

8 years of hard work, of fighting for the disorder to be known and recognised, of projects and actions to break sufferers' isolation worldwide. 8 years of tears and laughter shared with all those who joined us. 8 years of friendship and support for our action from people of the Ile de Ré. 8 very rich and enriching years.

I thank you all for those 8 years.

The fight is far from being ended. We still have many projects to achieve. Worldwide, sufferers count on us to break their isolation. We must continue to be the voice of Cutis Laxa sufferers so that it reaches every country in the world.

However, as voted at our Extraordinary General Meeting on 16th May, Cutis Laxa Internationale's Head Office will follow me as I leave the Ile de Ré and move to Haute-Savoie in September. Cecile will be studying in Paris next year. My other children, and my grand children, live in Haute-Savoie. I cannot envisage staying in Sainte Marie de Ré, alone. But it is not that simple to leave the Ile de Ré. Many memories, sad and happy, will remain here. I leave friends and a team who helped me achieve the daily tasks in the life of Cutis Laxa Internationale. They greatly supported me after Jean-Louis' death. Many thanks to them all. I hope I can count on all the volunteers who worked with Jean-Louis and I so that the Ile de Ré remains the symbol of our association's dynamism.

Marie-Claude Boiteux, Chair

NEW CONTACTS, NEW FAMILIES

The Cutis Laxa « Family » has grown again. There are now 156 members. On year old Paul lives in Germany. His mother is French and his father is German. Melanie, Susan C., Chris and Randi are American and all adults.

There are two new sufferers in France : Andrée and Philippe, both adults. Raffa is a little boy in the Philippines. Linah, a 16 month old little girl, lives in Saudi Arabia and her father is Syrian. Seher is Turkish.

And there is also an 18 year o young woman in Spain.

They all joined us since the issue N°13 of CLI~News. I am happy they found us and I welcome them among us.

In early 2009, Susan, Laura's mother, created a « Cutis Laxa Internationale » group on Facebook (internet communication tool). It is thanks to this group that Seher and Randi contacted. The increase in online communication tools is an opportunity for Cutis Laxa sufferers. Thanks to these tools we are more and more visible on the net, thus more and more accessible.

As Dr Zsolt Urban (St Louis, USA) received his grant agreement very late, the organisation of the 2nd Cutis Laxa American Day has been slightly modified. It was planned to be held in Dallas (Texas) in June and will finally be held in St Louis, but the date has not yet been decided.

Unfortunately sufferers' news are not all good news and I am very sad to announce the death of Philippe G. (France) and of Steven « Shorty » G. (USA). We will keep in touch with their families as long as they wish us to. In Memoriam Donations to CLI's totalled over € 1,600 at Philippe's funeral.

MEETINGS, EVENTS AND EXHIBITIONS



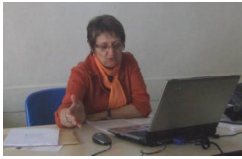
17th January : Health Meeting in La Rochelle. We welcomed many visitors to our stand. It was a very rich day of exchanges.

28th February : 2nd International Day for Rare Disorders.

21st March : Annual General Meeting of the CISS-PC in Angoulême (France): very interesting day where the results of the survey on Home Help in Poitou-Charentes were officially presented.

09th April : Presentation of the results of the survey « Inventory of Home Help » to the Health and Disability commission at the Local Council in Poitiers (France).

01st May : Brin d'Aillet run in Sireuil



16th May : CLI's Extraordinary and Annual General Meeting. We were not numerous but we shared a friendly drink in a convivial and hearty atmosphere after having proceeded to the reports and votes . You will find enclosed the minutes of these two important moments in the life of our association.

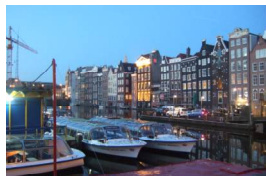


18th May : Cutis Laxa was presented at the International Film Festival in Cannes: Sébastien Maggiani, 13, directed a very short film on Cutis Laxa after sharing with us the Days in Paris on 9th and 10th May 2008. This 3mn long short film is entitled «Madame ». Cécile plays her own role in it. It is dedicated to Cutis Laxa sufferers and can be seen online at the following address :

http://www.dailymotion.com/video/x8ndh4_madame_shortfilms.

Here is what Sébastien said in conclusion to the scenario he wrote with Pascal Ange :

« Sufferers of the time, their faces already have the marks of time ! They are 5 , 13, 20 and they seem to be 2 or 3 times older.... They hear people calling them : « Hi Madame ! Hi Sir ! ». Words so hard to hear when you are so young. Time does not have the same value for them as for others because it speeds up on their faces at the rate of the disorder, Cutis Laxa disorder, premature aging of the skin. Time, this word does not means anything...Time you can read on faces, time of the years passing by. Times speeded up, and speeds up again, only leaving ignorance of a face without age, a wrinkled face with a child's smile...a face on which gazes and bitter words settle. Life is unfair to impose physical aging to a child. In our world a human being's beauty is too much linked to appearance and not to the person's sensitivity... Nasty gazes are our everyday life! criticism is our everyday life ! lack of understanding is our everyday life ! All of this builds the memories of a childhood broken by those remarks derived from the gap between the visible and the invisible. A childhood of enforced maturity..... »



20th May : I went with Nathalie to The Netherlands where she met Dr Eva Morava in Nijmegen. Dr Morava finalised a treatment for Cutis Laxa associated with osteoporosis, which is Nathalie's case. (see also « Research » below).

31st May : Triathlon of Sireuil

RESEARCH

In France

The research programme initiated by the AFM in 2002 having ended in 2007, the work lead by researchers and doctors as part of this programme was on hold in 2008. Today, initiated by Dr Pascal Sommer, new dynamics are being established with a view to creating a European network of doctors and researchers working on Cutis Laxa. In fact, in Europe, there are already several teams working on this very rare disorder. It is obvious that if we want to see the work move forward quickly and lead to therapeutic options, it is essential to exchange and share knowledge and findings while respecting individual copyright entitlements. Thus it is with this perspective that doctors/researchers will meet after the summer . They will establish and coordinate the basis of their future collaboration in order to (1) Improve information about the disorder, (2) Optimise the census, (3) Optimise the location of mutations and (4) Intensify therapeutic research.

In parallel to this project of a European network, Pascal Sommer continues working on the therapeutic properties of dill extract he has refined. This therapeutic option works well, in test tube, with the skin model carrying the mutation on Fibulin5. It is now necessary to move on to tests, in vivo, with an animal model. A lot of work, tests, and perseverance will be needed before we arrive at a “medication” that could ease the daily life for patients who suffer from Cutis Laxa with mutation on Fibulin5, even if it would not get rid of the disorder .

Worldwide



As you have just read, several teams work on Cutis Laxa in Europe. Among them, in the Netherlands and in Germany, two teams work especially on Cutis Laxa associated with low bone density, which is also called Geroderma Osteodysplasticum.

The Dutch team, with Dr Eva Morava of Nijmegen University Hospital, has found a treatment that allows a return to normal bone density after two years of therapeutic treatment.

Regarding the essentially German team, they have discovered the mutation causing this particular form of Cutis Laxa with low bone density. It is SCYL1BP1.

The appointment Nathalie had with Dr Morava will probably enable her to benefit from this treatment depending on the results of the tests she underwent, but also, finally, to receive the right diagnosis for her disorder,..... if the analysis of her DNA confirms it, after waiting for 37 years.....

LEGISLATION

Evaluation of the Plan for Rare Disorders in France (2005-2008)

The Plan for Rare Disorders comprised 10 strategic headings. The CODEV (Committee of Evaluation), in collaboration with Patients' associations, took these headings one by one to determine the success of the first French Plan for Rare Disorders. Explaining in detail the work of CODEV would need all this issue of CLI~News, but we should note the following positive points :

- ✓ Establishing the Centres of Reference
- ✓ The Local Centres for Disabled People (MDPH)
- ✓ Developing information on Rare Disorders with a specific collaborative effort with Orphanet .

The Plan was very ambitious and it was obvious that not all the aims would be achieved. Thus, the negative points are as follow :

- ✓ Funding research
- ✓ Social Care
- ✓ Training professionals

These negative points along with the continuation of funding for the Centres of Reference will have to be the aims of the 2nd Plan which will start in 2010 lead nationally by a cross departmental / ministerial group.

The Law « Hospital, Patients, Health, Location » (HPST)

This Law, on hospitals reform in relation to patients, health and location is being debated in view of a vote. The amendments brought to it are provoking great anxiety about health care in general, and not only for Rare Disorders.

The main points of concern are :

- ✓ There is a risk of abandonment from certain locations, mostly rural, as doctors' constraint for settlement is abolished
- ✓ Hospitals might change from being a Public Service to becoming « private enterprises » with « profitability » a higher priority than anything else .
- ✓ Therapeutic educational programmes run the risk of being offered to sufferers by doctors on their own , and not by a comprehensive health team. Programmes would also be evaluated by the Health Authority which, in this case, would become judge and jury, and risk major conflicts of interest. Moreover sufferer's close circles, even if directly concerned, could risk being excluded from these programmes.

Patients' associations and health professionals are very worried.

The economic crisis and health in Europe

In an open letter, the European Forum on Health called upon the EU decision makers to protect the statute of Health as a public priority. This letter was well timed as it was sent before the Spring European Summit dedicated to the worldwide economic crisis we are living through. EURORDIS and other members of the Forum wrote : « We are more and more anxious about official statements broadcast and with the impact of budget cuts announced in some Member States regarding health and social care expenditure ». And they ask for a « careful look at the possible investments in medical capacities, knowledge and infrastructure to contribute to economic recovery ». According to the members of the European Forum on Health, there is a « positive link between health and economics regarding long-lasting

development » and it is advisable to keep in mind that « countries who invested in health during the last century registered more lasting results than those who reduced their investments. » The letter ends with a strong call, urging governments to keep health at the top of their agendas. (Eurordis newsletter Mai 2009)

The Media

13th February 2009 : HSCNews *Bulletin* (International online bulletin of Health and Social Campaigners) dedicates an article to our financial worries after the Days of May 2008 and to the high cost of foreign currency exchange when donations are not made in Euros or when sums are too small.

01st February 2009 : Midi Libre : Report during the shooting of the short film « Madame »

05th March 2009 : Le Guide Santé « online » : Article in the heading « News »

23rd March 2009 : Charente Libre : AGM of the CISS and results of the survey on Home Help

23rd March 2009 : Sud Ouest : AGM of the CISS and results of the survey on Home Help

18th May 2009 : The short film « Madame » is in a report during the morning broadcast « Télé-matin » on France2 (French TV).

19th May 2009 : Sud-Ouest La Rochelle Ile de Ré: Cécile is on the front page with a long article about her role in « Madame »

26th May 2009 : France3 Atlantique (French TV) : Report during the news about Cécile and « Madame »

29th May 2009 : Radio Soleil de Ré : Interview with Cécile

June 2009 : « Maxi » Magazine (France) : Article about Cécile and « Madame »

THE ASSOCIATION'S FINANCES

Fees and Donations :

Many thanks to all those who already volunteered with their contribution. You all are, members and donors, our main source of funding. **To those who have not yet done it, Thank you for thinking about us. I know that our tax receipt cannot be used by those outside of France and I regret it a lot, but you can organise fundraising events in your area if you want to help us.**

Fundraising: Niamh's and Isabel's parents organised fundraising events (donations and selling items to the CLI's benefit). They collected over € 790. Many thanks to both of them.

Grants : The villages of **Saint Bonnet de Salers** and **Marthon** have just sent us their grants. Many thanks to both of them for remaining faithfully at our side.

Orphanet is finishing the first heading of a project integrating rare disorders in the international classification of diseases

The World Health Organisation (WHO) is reviewing the International Classification of Diseases (CIM-10). This system «aims to classify diseases, related health problems and external causes of diseases and traumas in order to compile useful sanitary information regarding deaths, diseases and traumas (mortality and morbidity)».

But, at present, only 240 rare disorders are included in CIM-10 with their own code.

In April 2007 WHO put together a group of experts on rare disorders, of which Dr Ségolène Aymé is the Chair, to examine the statute of these diseases in the system of classification. The research of information needed to establish the first issue of the new classification was entrusted to Orphanet. This classification will be a model for the overall review of CIM, as rare disorders cover all the fields of medicine.



Biostick : Your health on a key !

Your Patient's Medical File readable in 7 languages. It comprises the names and addresses of people to call in case of emergency, your medical background, your current treatments, etc ... our members are entitled to a reduced fee. Feel free to ask for it.

Le Guide Santé is a community portal, certified by HonCode, with whom we established a partnership. It comprises a lot of information about French hospitals. Feel free to visit it :

www.le-guide-sante.org

*fondation groupama
pour la santé*

Valre les maladies rares



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WARNING – TAKE NOTE !!! HERE IS OUR NEW ADDRESS FROM 15th SEPTEMBER 2009 ONWARDS

**CUTIS LAXA INTERNATIONALE
Lot . LES TERRASSES DE SAINT CERQUES N° 12
74140 SAINT CERQUES LES COTS
FRANCE**

WE DO NOT YET HAVE THE NEW PHONE NUMBER , BUT IF NECESSARY YOU CAN CALL THE CHAIR ON HER MOBILE / CELL PHONE ON 00 33 662 162 073. AND OF COURSE OUR EMAIL ADDRESS REMAINS THE SAME WE WILL GIVE YOU ADDITIONAL INFORMATION IN THE NEXT ISSUE OF CLI~News .

Thank you for sending back this form with your membership fee and/or your donation to the address mentioned below

CUTIS LAXA INTERNATIONALE ASSOCIATION
35, route des Chaignes - 17740 Sainte Marie de Ré - France

NAME..... FIRST NAME.....

ADDRESS.....

ZIP CODE.....TOWN.....COUNTRY.....

Wants to support Cutis Laxa Internationale by sending his/her membership fee : **€ 25** (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.....

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

Email address.....

Thanks to you it will reduce our mailing expenses.

As indicated in French law "Freedom and Computers" 06 July 1978, you have the right to have access and to rectify your personal mentions. If you want it, write to our Association.