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LI ~ NEWS N° 38 ~ SPECIAL ISSUE ~ 2022

**6th Cutis Laxa Days – Ghent – Belgium
 13th – 14th – 15th – 16th September 2022**



What a pleasure to meet again, especially after the Covid years that kept us away from each other!

The 6th Cutis Laxa Days were a huge success !!

It was the first time so many patients (23) gathered together;
 the first time so many countries (11) were represented; and
 the first time also that so many doctors and researchers interested in Cutis Laxa attended the Conference.

It is always very difficult to account for what happened during four days, in the most accurate way possible, especially when they were so intense : many moments of strong emotion, much information shared, much joy, much fun, but sadness too

Photos are witness to all this, what Cutis Laxa Days represent .

Marie-Claude Boiteux, Présidente

13th and 14th September 2022 – Clinics Days



Pr Bert Callewaert, Dr Karolien Aelbrecht, Mrs Deborah Wille and the full team of the medical genetics department in Ghent University Hospital welcomed us to allow patients to consult one-to-one in Genetics, Cardiology and Ophthalmology. They also had blood tests as well as a biopsy.

All those tests aimed at evaluating the consequences of Cutis Laxa on the organs examined, confirm the diagnosis initially given, allow molecular diagnosis when needed, inform patients on the medical follow up their case deserves, but also include all those willing to join the research programme led by Pr Callewaert.



Cheers and thank you to Bert's team. They were adorable and educationalist, together with a great medical expertise. We felt we were in good hands.



Activities for children were great, games and colour books and in the hospital and during the symposium



Several rooms had been mobilized, as much for the consultations as to offer a waiting and resting room for patients and for children, whether patients or not, and their siblings to have a place to play.

We want to deeply thank Pr Callewaert and his team for their attentiveness, their patience, their commitment and their great expertise in Cutis Laxa.

We also thank Ghent University Hospital for the catering.

15th September 2022 – Conference Symposium



In the beautiful historical buildings of Ghent University, participants were more numerous than ever before. We were able to organize this symposium thanks to funding from Ghent University, EADV, ERN-Skin, FWO and the European Programme EJPRD.



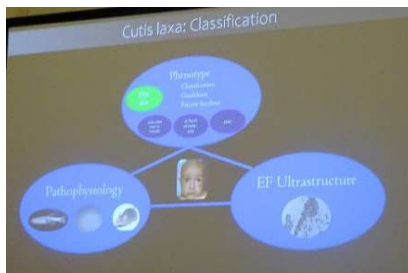
It was really moving to see such a crowded room ! Moderated by Paul De Brem, scientific journalist who has faithfully supported us since the Days in Annecy, this event allowed researchers and health professionals coming from Europe, Turkey and the USA to share with us the most recent information on Cutis Laxa.

Session I : Diagnosis and General Management in CL



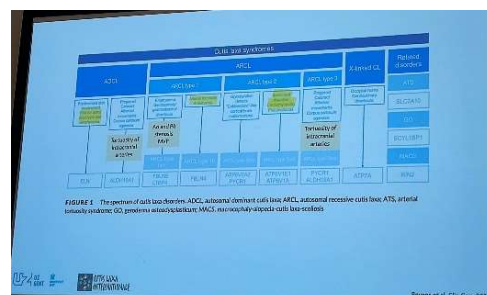
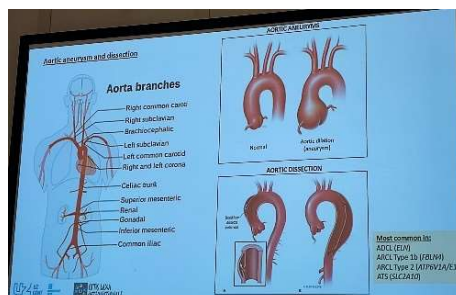
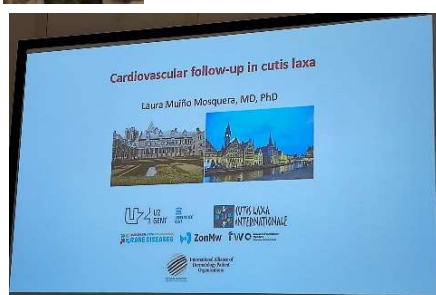
Prof. Dr. Bert Callewaert (Ghent – Belgium) : CL Classification and Guidelines for Global Management

This new classification will simplify the diagnostic process and identify relevant Cutis Laxa subgroups with regard to management and clinical research. In addition, identification of the underlying genetic defect may end the diagnostic odyssey, further stratify management, and may provide (im)partial answers to future health issues. Furthermore, it provides answers to potential recurrence risks in the offspring of patients and relatives as well as the use of diverse reproductive options. Finally, connecting with peers provides support and encourages participation into patient-centered research aiming at a better quality of life and-hopefully- more directed therapeutic options.



Dr. Laura Muño Mosquera, MD (Ghent, Belgium) : Cardiovascular Follow-up in CL

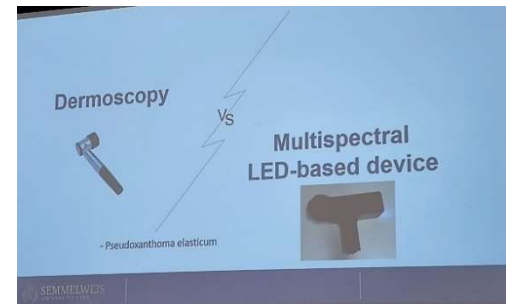
Cutis Laxa disorders usually present cardiovascular issues that need a specific follow up. Various devices can be used such as echocardiography, CT-Scan and MRI. Each one of those devices has its pros and cons and must be chosen according to what is examined.





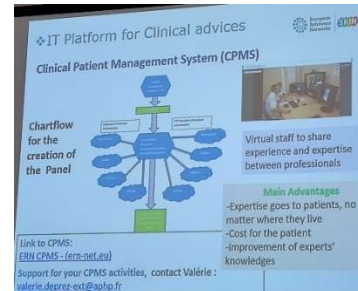
Mrs Klára Farkas, MD (Budapest, Hungary) : Potential applications and value of novel skin imaging techniques in connective tissue disorders

In this presentation we introduce novel imaging techniques including nonlinear optical (NLO) microscopy, multispectral imaging and high-frequency ultrasounds (HFUS) to visualize the characteristic changes in Connective Tissue Disorders (CTDs). The novel imaging techniques may prove useful in the early diagnosis of CTDs. They may be also used for the objective follow-up of the progression of CTDs, and the assessment of the efficacy of novel therapeutic approaches in the future.



Prof. Dr. Christine Bodemer, MD (Paris, France) : Embedding CL in ERN Skin & ERN-Ithaca

The European Reference Networks (ERNs) set up by the European Commission allow a collaboration between all health professionals concerned by a specific disorder throughout Europe. They also allow the discussion of more difficult cases, online, thanks to the Clinical Patient Management System (CPMS) platform.



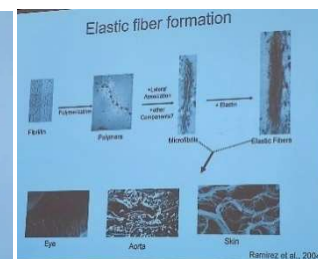
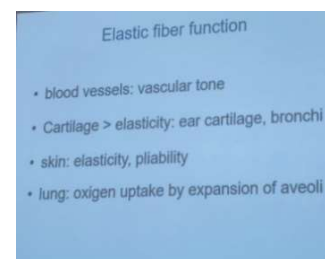
Session II : Pathophysiology and State-of-the-art in the different CL Subtypes



Prof. Gerhard Sengle (Köln, Germany): Matrix Biology – What can we learn from animal modelling ?



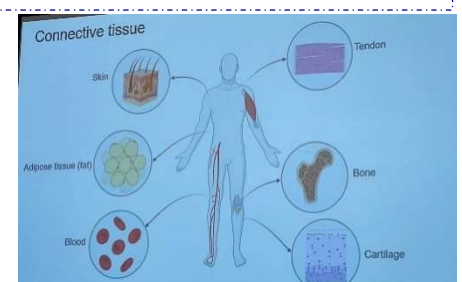
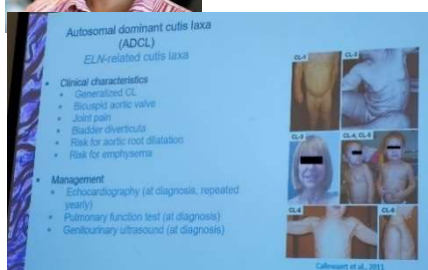
Understanding how the fine tune mechanisms of elastic fibers formation is perturbed in the different types of Cutis Laxa is crucial to design molecular therapies in preclinical trials using animal models.

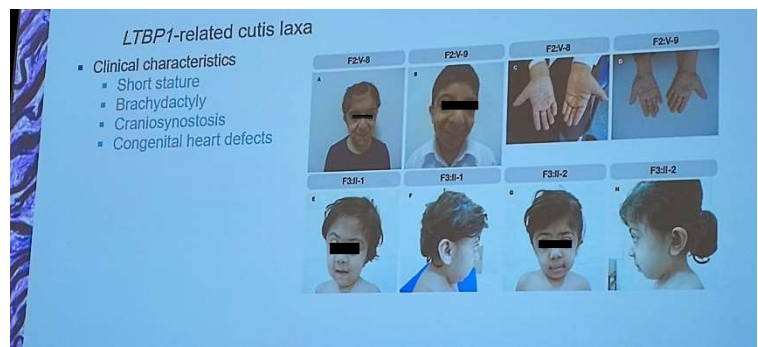
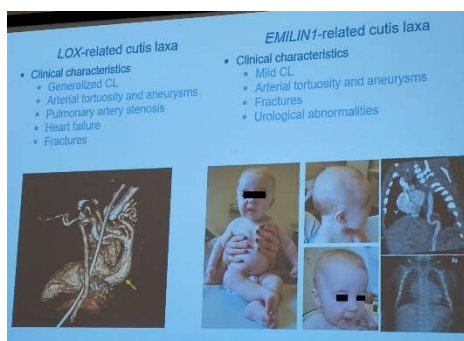


Mrs Aude Beyens, MD (Ghent, Belgium): Structural defects of Connective Tissue Proteins and CL

Elastic fiber assembly, or elastogenesis, is a complex process that is precisely regulated in a spatiotemporal manner and depends on proper growth factor signaling and mechanosensing.

The underlying molecular defect in cutis laxa syndromes affect the synthesis and/or association associated extracellular matrix proteins

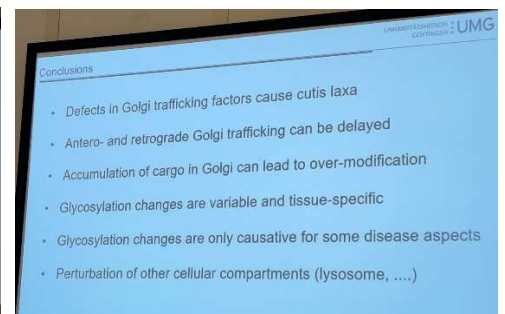
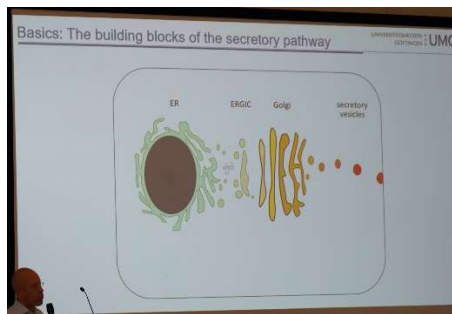
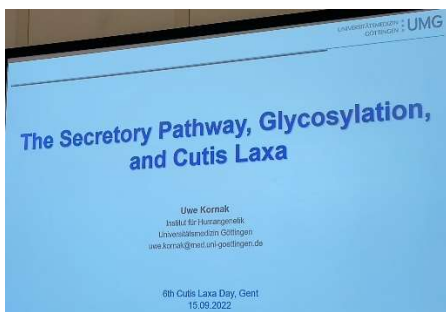




Prof. Dr. Uwe Kornak, MD (Göttingen, Germany): The Secretory Pathway, Glycosylation, and Cutis Laxa

Secreted proteins and membrane proteins have to pass through the secretory pathway, which is not only responsible for the transport to the correct cellular compartment, but also for the maturation and modification of these proteins. A central part of the secretory pathway is the Golgi compartment, which consists of several « pancake-like » membrane sacks (cisternae).

One important modification is glycosylation, which has been found altered in a whole group of Cutis Laxa disorders, that are also named congenital disorders of glycosylation (CDG).

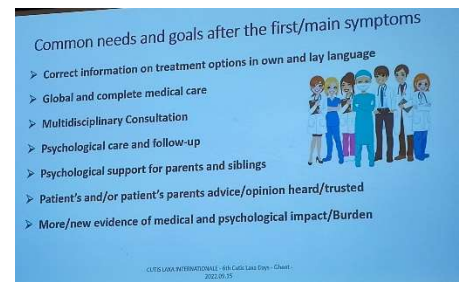
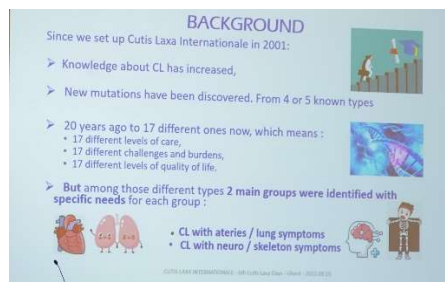
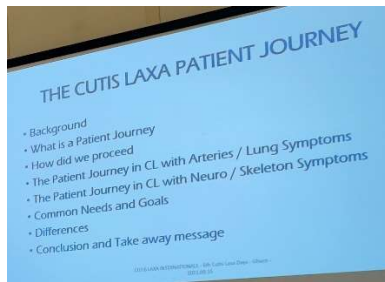


Lunch and Networking



It represents the collective perspective on the burden of the disease and the needs of people with first-hand experience of living with a rare disease. It is a baseline reference document for clinicians to develop a healthcare pathway and guidelines to address the identified needs.

Cutis Laxa is such an heterogeneous disease that we could not make only one « Patient Journey » but two to represent the specific needs of the patients with the different types of Cutis Laxa.



Mme Elif Yilmaz Gulec, MD (Istanbul, Turkey) : Cutis Laxa in Turkey : Challenges, Opportunities and Collaborations

Our Cutis Laxa patients are usually referred to us during neonatal period or early infancy.



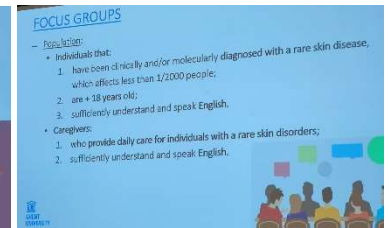
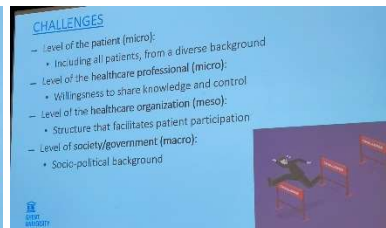
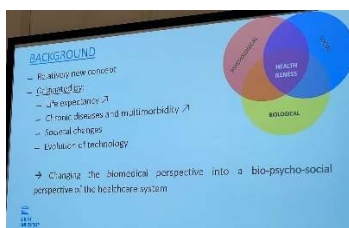
Many are confused with progeria, a rarer but more famous syndrome before admission.

Cutis Laxa needs a multidisciplinary approach, as soon as the diagnosis is made, the patient is referred to screening for possible accompanying organ disorders. Since the molecular defects determines organ pathologies and prognosis, molecular diagnosis is made as soon as possible.

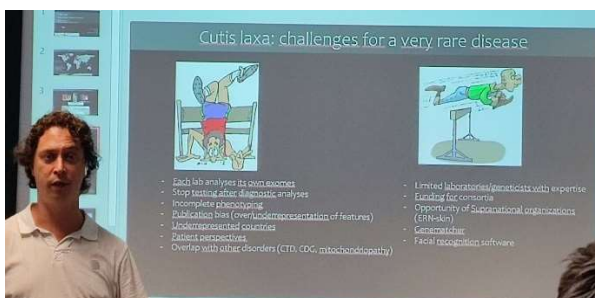


Dr. Karolien Aelbrecht (Ghent, Belgium) : Patient participation : a cornerstone !

Although the concept of patient participation still lacks of a clear definition, there is a well-supported consensus that it is the cornerstone of the bio-psycho-social perspective of our healthcare system and thus of a patient-centered approach in healthcare. In this presentation, we will give an overview of what patient participation entails, what the importance is, but also its challenges, and how our multidisciplinary team implements the concept of patient participation in practice and research.



16th September 2022 – Collaboration in Recherche



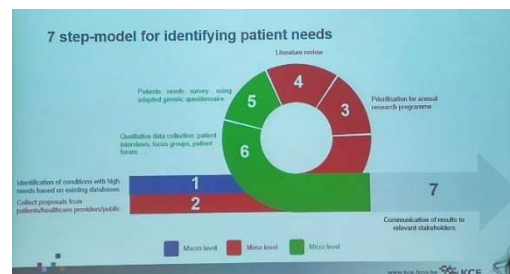
The aim of this event dedicated to research was to set up collaborative research among researchers



interested in Cutis Laxa, worldwide. It was also to ask patients to work together with researchers to let them know what main topics they would be interested in.

Cutis laxa: SWOT	
Strenghts Few labs, previous collaborations Large sample database Patient organizations	Weaknesses Extremely rare / small group Many phenotypes Data scattered in different labs Funding
Opportunities Consortia (e.g. ERN-skin): registries, biobanking, financing Financing of novel consortia Reaching out to other overlapping entities (CTD, CDG, Mitochondrial disease (MetabERN)).	Threats No referral to expert centres Underrepresented countries Group of diverse disease

Pr Callewaert presented the main challenges, strength and weaknesses Cutis Laxa represents. Then Dr Jens Detollenaere talked about how



patients' participation is organised as partnership in research in Belgium



After working in 2 groups (clinic and research, and patients) with those interested in taking part in the research collaboration, we reported our work in a general session. We ended this working day on research collaboration with the decision to set up an International Research Consortium on Elastic Fibers including a Patient Advisory Board. What a Success !!!!



The 6th Cutis Laxa Days is also

Gathering beyond continents.....



...Mums and Dads...



... Children's laughter and games



.... Shared moments ...



.... A Boat Tour in Ghent ...





It is such a beautiful town !



... Followed by a festive diner.





Time to go home..... It is so hard to leave each other.



Longing to meet again Roll on the 7th Days ...

Thanks to  **EUROPEAN JOINT PROGRAMME**  **RARE DISEASES**, to **bons**  en Châbles and to  **International Alliance of Dermatology Patient Organizations** who helped with funding