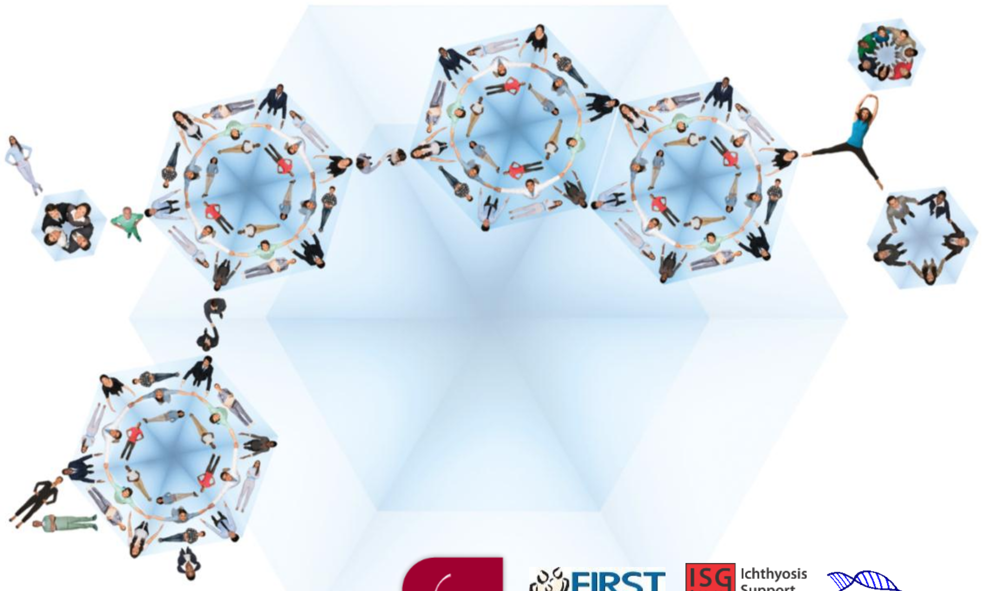
Lamellar ichthyosis

*A qualitative survey in the US and Europe**

November 2013



Background and objectives

A PROJECT CONDUCTED BY IFOP WITH THE SUPPORT OF FIRST, ISG AND ENI ORGANISATIONS

What is lamellar ichthyosis?**

It is a condition that mainly affects the skin. Infants with this condition are typically born with a tight, clear sheath covering their skin called a collodion membrane. This membrane is usually shed during the first few weeks of life, but remaining effects include scaly skin and outward turning eyelids and lips. People with lamellar ichthyosis typically have large, dark, plate-like scales covering their skin. Affected individuals may also have hair loss, a decreased ability to sweat and a thickening of the skin on the palms of the hands and soles of the feet. Less frequently, affected individuals have reddened skin and joint deformities.

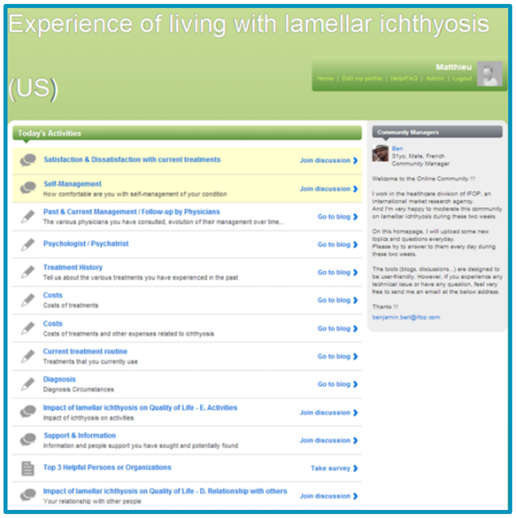
How common is lamellar ichthyosis?

Lamellar ichthyosis is estimated to affect 1 in 200,000 to 300,000 individuals in the United States.

Patient can describe their disease and needs

There is nothing more valuable than getting direct patient real life experience in any disease to improve awareness & understanding of the patient's life for any person. Such patient centric approach would help to share the LI psychological and practical impact sufferers and their families are facing on a daily basis. **Despite its orphan definition, LI clearly requires more innovative product / solution that would help patient and his/her family to have a better quality of life.**

Galderma is committed to deliver innovative medical dermatological solutions to meet the needs of people and serving the healthcare professionals to explore new and more effective treatment options.



Experience of living with lamellar ichthyosis (US)

Today's Activities

Satisfaction & Dissatisfaction with current treatments — Join discussion ›

Self-Management — How comfortable are you with self-management of your condition — Join discussion ›

Past & Current Management / Follow-up by Physicians — The various physicians you have consulted, evolution of their management over time... — Go to blog ›

Psychologist / Psychatrist — Go to blog ›

Treatment History — Tell us about the various treatments you have experienced in the past — Go to blog ›

Costs — Costs of treatments — Go to blog ›

Costs — Costs of treatments and other expenses related to ichthyosis — Go to blog ›

Current treatment routine — Treatments that you currently use — Go to blog ›

Diagnosis — Diagnosis Circumstances — Go to blog ›

Impact of lamellar ichthyosis on Quality of Life - E. Activities — Impact of ichthyosis on activities — Join discussion ›

Support & Information — Information and people support you have sought and potentially found — Join discussion ›

Top 3 Helpful Persons or Organizations — Take survey ›

Impact of lamellar ichthyosis on Quality of Life - D. Relationship with others — Your relationship with other people — Join discussion ›

Community Managers

Ben
31yo, Male, French
Community Manager

Welcome to the Online Community !!!

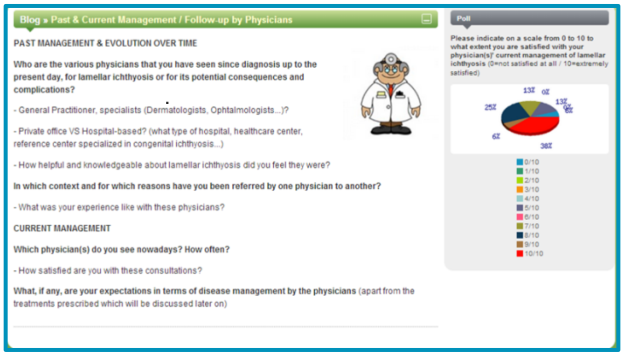
I work in the healthcare division of IFOP, an international market research agency.
And I'm very happy to moderate this community on lamellar ichthyosis during these two weeks.

On this homepage, I will upload some new topics and questions everyday.
Please try to answer to them every day during these two weeks.

The tools (blogs, discussions...) are designed to be user-friendly. However, if you experience any technical issue or have any question, feel very free to send me an email at the below address.

Thanks !!!

benjamin.berl@ifop.com



Blog » Past & Current Management / Follow-up by Physicians

PAST MANAGEMENT & EVOLUTION OVER TIME

Who are the various physicians that you have seen since diagnosis up to the present day, for lamellar ichthyosis or for its potential consequences and complications?

- General Practitioner, specialists (Dermatologists, Ophtalmologists...)?

- Private office VS Hospital-based? (what type of hospital, healthcare center, reference center specialized in congenital ichthyosis...)

- How helpful and knowledgeable about lamellar ichthyosis did you feel they were?

In which context and for which reasons have you been referred by one physician to another?

- What was your experience like with these physicians?

CURRENT MANAGEMENT

Which physician(s) do you see nowadays? How often?

- How satisfied are you with these consultations?

What, if any, are your expectations in terms of disease management by the physicians (apart from the treatments prescribed which will be discussed later on)

Poll

Please indicate on a scale from 0 to 10 to what extent you are satisfied with your physician(s)' current management of lamellar ichthyosis (0=not satisfied at all / 10=extremely satisfied)

** This is a qualitative study which highlighted trends and provided valuable information. But all quantitative results (satisfaction marks, ranking as well as mean price of treatments) have to be read carefully (No statistical value)*

***: source = ghr.nlm.nih.gov*

Key findings of this project

QUOTES AND PICTURES

LI shapes a strong character

"But the thing I've learned in life is to treat each day with positivity because one never knows what the next may bring" (Adult+, M, USA)

Still down moments

"I think these days everyone has to be perfect, perfect hair, perfect skin and wear the clothes that's 'acceptable' so I feel very pressured into trying to look like everyone else because of this stigma about not looking perfect". (Youngster, F, UK)

THE NEED OF COMMUNICATION

This rare condition often generates a feeling of loneliness / people not being able to truly understand what they are going through. Consequently, the exchange with other people with LI is extremely helpful.

- ***Physical resemblance is reassuring for most***
- ***Ability to discuss openly about any topic related to their condition and be understood***
- ***Possibility to learn good tips in terms of products, how to face difficult situations, and general self-management of LI***
- ***Helps feel supported / part of a community***

> *"[The online community]has been one of the best experiences ever for me. I am so grateful for being able to connect and share with the other members".*
> *(Mother of 11 yo boy, USA)*

Symptoms experienced & impact on quality of life

The symptoms mentioned by respondents include:

- **very dry skin which scales** / fissures (with a critical impact of climate or stress)
- **inability to sweat,**
- **itchiness,**
- ear complication, ectropion (lower eyelid turn outwards), hair loss and general predisposition to infection.

When asked about quality of life, most patients underlined the physical appearance (visible scales and flakes as well as red face) as it can lead to people stare, bad comments and can even go as far as bullying, which strongly impacts self-esteem.

Besides, **the inability to sweat implies a strong limitation in the daily life of patients** (sport practiced, type of vacation…) as well as a long preparation before any exercice. This was particularly mentioned by children and youngster who may feel excluded from peer groups, partly for this reason.

Attitude of patients

Most patients confirmed that Lamellar Ichthyosis shapes a strong character. It leads to a strong self-affirmation (particularly reinforced by religion in the US) it helps to be a better person (more empathetic, more altruistic) and a "fighter". **Despite the condition, a lot of patients explained that LI should not prevent them from doing anything they want**. That's why a lot of respondents highlited the importance of positive attiude and humor to develop self-mockery and be able to focus on the bright side of life.

However, some recognize the difficulty of always having this attitude. Feelings of sadness, anxiety and injustice are common in these patients. In addition,the difficulty to bear others' reactions as well as the difficulty to find a partner or to live in a world with pressure to look perfect was regurarly pointed out.

Overall this survey shows a typical evolution of attitude over the time. High school years are often the most difficult period (low self-esteem shyness, some bullying), but patients learn to accept that their skin is part of them and contibutes to making who they are, but does not define them.

> *"And the older I get, the more I realize there are many more people with much worse things to deal with. My skin has been a blessing in disguise, and I have truly learned a lot from it. If anything, I am thankful for it rather than angry about it." (Adult+, F, USA)*

Interactions & support

For a vast majority of patients, the family provides a very strong emotional support:

- ○ Understand condition and related constraints
- ○ Help face difficulties and frustration
- ○ Encourage
- ○ Protect against stares and bad comments.

The family also demonstrates a practical and financial commitment (finding the best doctors, major involvement in care routine)

Overall, most parents and patients indicate a particularly strong "parent-child" relationship.

Respondents also pointed out the role of partners who help boost self-esteem a lot **and friends** who are also essential, especially for the younger ones and singles.

Beyond these close relatives, **the role of support groups is critical**. They provide information & support (emotional, practical and financial though discount on products) and offer the ability to get in touch with other people with same condition.

Family is the #1 support for a vast majority of patients

The relevance of the online community

Given the strong need of communication, most participants showed extremely high satisfaction with this experience, especially mothers who are often even more in need of support.

Many patients / parents said they wish that they could have access to such an online community on a constant basis (exchange via a secure online platform with restricted access), ideally with a section dedicated to children

The impact of LI on activities

In addition to the care routine, the condition affects the daily life in several ways.
For instance, some patients mentioned few limitations resulting from dexterity and mobility issues.
A majority stressed that overheating becomes a concern when it comes to vacations (even going out requires some preparation).
Dressing is also fairly complicated and frustrating for people with LI (limited choice, clothes look worn quickly).
A lot of parents and patients also explained the **condition affects housework which becomes very time-consuming and tiring** (hoovering, sweeping, laundry…)
➔ **Overall, the management of LI and its consequences also generates extra expenses (water, laundry clothes, cleaning…)**

Respondents were generally happy with their professional environment but **appearance makes it more difficult to find a job**, especially in some specific fields (health, sales, outside works, restaurant…).

Finaly, **LI has also intimate consequences. Flirting/sexuality seems to be more complicated** (lack of self-confidence, embarrassment) but it is often said to get easier over time.

Main concerns for the future

In this study, patients stressed a major concern about the evolution of their health.

But, they were also concerned regarding their ability to find a partner / stay in couple and having children. The inability to afford treatments or cost of living was also mentioned, particularly in the US. **Parents, had strong general concerns regarding the future of their child** (personal and professional life)

Diagnosis, management and treatment routine

All parents confirmed that birth and diagnosis are extremely emotional situations generaly made up of contrasted feeling:
-Anxiety, fear, guilt
-but also relief when a name is put on the condition and when they are explained this disease is not life threatening.

This survey reveals that expectations regarding physician's management evolve over time.

Parents have greater needs in the 1st years (information, emotional support, appropriate recommendations, and need for an appropriate health insurance) and it often requires some time and several consultations before finding a dermatologist who meets their expectations.

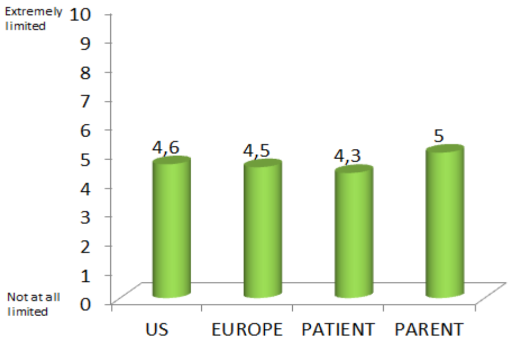
But, once parents and patients identified the most appropriate treatments, their expectations towards their physicians, decrease.

This survey confirms that daily routine is a real burden for most, especially because of the time it takes and repetition. (*See the figure on the next page)*.However, the level of satisfaction with current treatment remains fairly high.

The burden is also reinforced by the difficulties of self-management. Some body areas are more difficult to reach / treat (back, feet, hands and scalp). Besides, some symptoms are particularly difficult to relieve (overheating, itching).

> *"The most difficult parts for me are my feet, scalp, peeling my back and cleaning my ears, so my mother helps me, because I can hardly do that myself". (Youngster, F, GER)*

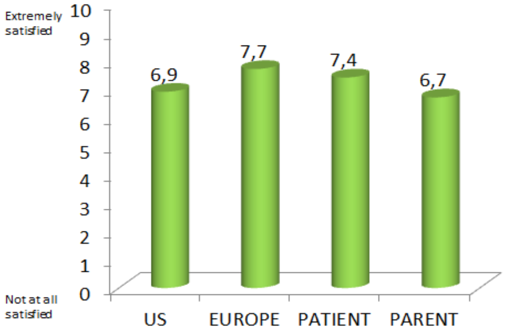
KEY FIGURES



Impact of LI on activities

In average, the impact could seem fairly limited. But, perceptions of the respondents vary a lot regarding to what extent they feel restricted because of LI in the activities they can do. Rates vary from 0 to 10 (it all depends on the severity of the condition and the support received)

Patients are aware of the crucial help brought by the products that they use, and show fairly high level of satisfaction



Satisfaction with current products

… is fairly high. There is a difference between the level of satisfaction in the US and Europe, which results mostly from a much more difficult access to all products in the US, finance-wise (retinoids and some expensive moisturizers+)

Description of the daily treatment routine

MORNING ROUTINE (~50 min)

- Shower generally for adults
- Bath generally for children → ~30min (up to more than an hour when exfoliating in-depth – which is a long and tedious process done ~2 times per week and once during the week-end)
- Applying lotions / creams → ~20min
- Wait some time for moisturizers to penetrate the skin before getting dressed → ~10min

DURING THE DAY (~10min)

- Number of moisturizer applications during the day varies a lot
- All of them carry at least one moisturizing product with them whenever they go out of home

EVENING ROUTINE (~40 min)

- Most take another shower or bath
- Then apply lotions / creams
- Some technics are also used by some to favor in-depth moisturizing (ex: wearing socks or saran wrap after application of moisturizer)

WEEK-ENDS

- Overall more intensive care (adults+)
- Conversely, the care routine is generally less intensive for children:

Focus on key treatment used and costs

Least expensive and most used

Most expensive Least used

OTC Moisturizers	Keratolytics & Steroids	Retinoids (oral or topical)
→ The main pillar for the treatment of LI → Applied every day (2-4/day) → A long process to find the most appropriate → Overall, a fairly good level of satisfaction	→ Very helpful to exfoliate and limit scales and flakes → Used 2-3/week on specific body areas → But with some negative effects (cost, odor, stings/burns)	→ Very efficacious in terms of skin appearance improvement → Rarely used (on some body part) → But issues in terms of tolerability, safety and costs (oral)
→ Generally fairly inexpensive but used a lot → Some products available with discount or for free	→ Fairly expensive → Keratolytics are used less quickly than OTC Moisturizers	→ Extremely expensive in the US, even with a health insurance + Blood tests and Doctor consultations required… → In UK and GER: all have free access to them on prescription

What would be an ideal treatment?

There is currently no cure for LI**. Thus, the ideal solution would obviously be a definitive treatment**. In that case, oral administration would be generally favored over topical

> *"I would like for genetic therapies to become the norm of new medications. Once scientists determine exactly the gene(s) that affect our skin, I would love to have genetic therapies that would be beneficial in fixing the skin so that way it would not look as it does (Adult+, F, USA)*

> *"Ideal treatments would be a cure so my beautiful daughter doesn't have to suffer. Giving her back the gene that she is missing would be ideal" (Mother of a 4yo girl, UK)*

> *"Ideal treatment would to be able to take an oral medication that helped my skin and not have to apply any topical besides a normal lotion". (Adult, +, F, US)*

Despite a fairly high level of satisfaction with current moisturizing products, there is a room for an improved product:

→ **High efficacy in making the skin look as 'normal' as possible**
→ **Long-lasting effect**
→ **Easy and quick to apply**
→ **Quick absorption and does not ruin clothes**
→ **With a cost effectiveness**

Study protocol / Methodology

2 English speaking online communities (qualitative methodology) had been set up by the IFOP agency from **June 3rd to June 30th 2013**

–20-25 participants per community (a mix of parents, young adults and adults was recruited).

–2 weeks of investigation to explore a large number of topics (treatment, costs, QoL, emotional impact, social and professional life, needs and expectations).

–A combination of tools (blogs, forum, mini polls).

–Everyday, new "activities" were uploaded on this secure Internet platform, and respondents were asked to reply to the questions, react to other respondents' posts, upload pictures…

Special thanks

Galderma would like to thank patients and their families as well as patient organizations for their support, involvement, time and valuable inputs to this research.

Patient organizations contact

Foundation for Ichthyosis & Related Skin Types, Inc. (FIRST)
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