



Real life experiences

Understanding the impact of a rare disease on patients' everyday life

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AFAF - French Friedreich's Ataxia Patients Association

- **Friedreich's Ataxia:** loss of coordination (**ataxia**), progressive loss of walking and writing, severe fatigue, pain due to spasticity, speech troubles, visual and hearing loss, cardiomyopathy, diabetes, scoliosis, serious heart disease, etc. There is **no cognitive impairment**, hence the many social, psychological and emotional impacts.
- It is the **most common inherited ataxia in Europe** (~1,500 FA in France).
- **Each person's evolution and experience of the disease is different**
- **AFAF:** Founded in 1980
- Run by **volunteers**
- **Aim to help patients, their families and caregivers by providing day-to-day support services, advice, patient representation and funding quality of life and research projects.**



- **Psychological and Social Supports phone services** are **co-funded** by three associations: ASL-HSP France, AFAF and CSC (Cerebellar Syndromes).
- **Scientific Council** and **Medical and non-medical care Councils** shared

Real life experiences

How can your voice make a difference?

First: We need to **understand the impact of the disease** on patients' everyday life, their **needs, priorities** and **expectations**



The Voice of Patients



1st Survey

**Team of AFAF administrators (4 persons)*

Patients, families and caregivers, were invited to answer an online survey (during 1 month)
Close and open-ended questions divided into 4 sections:

- A) About you** (*age, age at onset, age at diagnosis, zip code etc*)
- B) About your symptoms**
- C) About your medical and non-medical monitoring**
- D) About adaptations to your daily environment**

- Before the official launch of the survey, a test phase was carried-out with a panel of 10 FA patients and AFAF board members.
- Distribution channels: e-mailing to members, the AFAF's magazine "Espoir" (*Hope*), social media platforms.



A total of 209 responses were received

Answers were shown by category or were categorized on the basis of verbatims

All answers are anonymous and accessible only to the research team

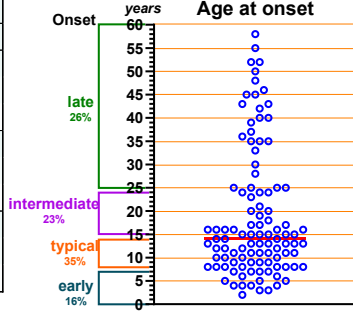
Real life experiences

Turning experiences into data

Who are you?

Characteristics of FA individuals in France

Total participants (n)	209
Total participants with FA (n)	104
Age of individuals with FA (years)	
Mean (SD)	41.3 (17.6)
Range	10-76
Age at onset (years)	
Mean (SD)	19.1 (13.9)
Range	2-58
Age at diagnosis (years)	
Mean (SD)	23.0 (14.9)
Range	4-59



What's your daily living?

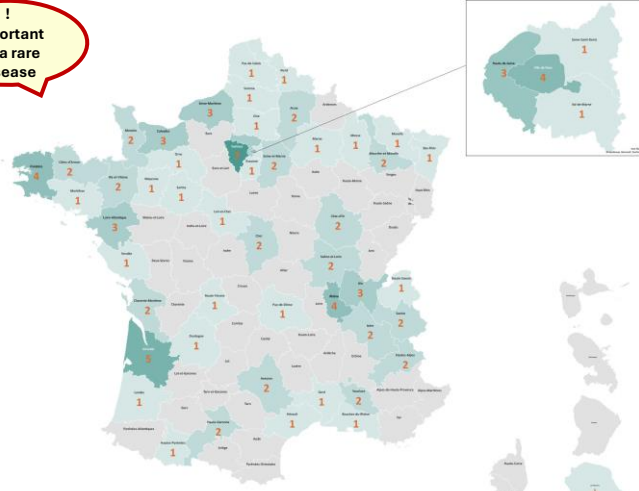
Symptoms with the heaviest impact on daily life

! Important for quality of life: how we can help?

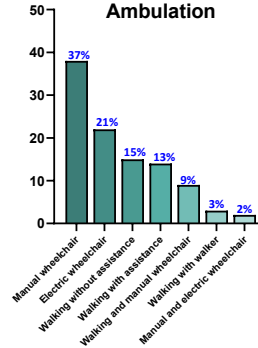
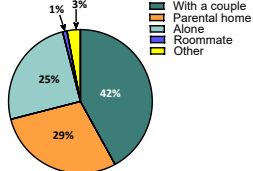
Main symptoms encountered	Symptoms with the greatest impact on quality of life	Symptoms that patients are most worried about as the FA progresses
Upper limb coordination, manual dexterity	83%	Balance / walking / falls 22%
Fatigue	77%	Upper limb coordination, manual dexterity 17%
Balance / walking / falls	73%	Fatigue 15%
Orthopedic problems (scoliosis, feet, etc)	63%	Speech problems 12%
Speech problems	59%	Incontinence / urinary / intestinal problems 9%
Spasticity	58%	Hearing problems 8%
Pain	50%	Speech problems 7%
Swallowing difficulties	50%	Heart problems 8%
Incontinence / urinary / intestinal problems	39%	Vision problems 7%
Heart problems	32%	Hearing problems 7%
Vision problems	30%	Orthopedic problems (scoliosis, feet, etc) 6%
Hearing problems	30%	Fatigue 5%
Hearing problems	30%	Pain 4%
Diabetes	13%	Spasticity 4%
		Swallowing difficulties 4%
		Diabetes 2%

Geographical distribution and type of residency

! Important for a rare disease



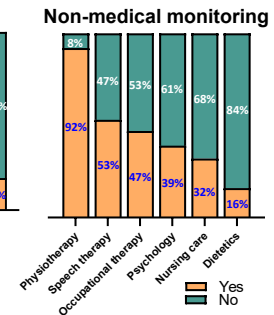
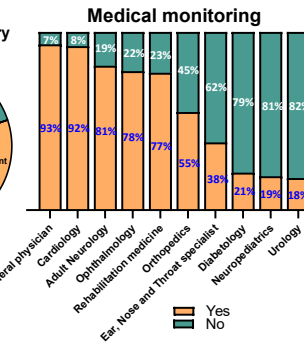
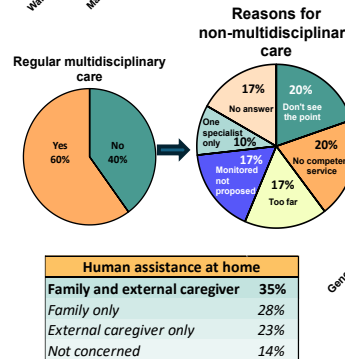
Living place



Complexity of everyday life	Social impact of the FA		
Stand, walk and move around independently and safely	21%	Loss of autonomy	20%
Handle small objects or pick up objects	17%	Social isolation	13%
Writing	16%	Loss of recreational activities	12%
Understand a conversation in a noisy environment	11%	Frustration	10%
Transfer independently (e.g. from wheelchair to bed)	8%	Loss of hope for the future	10%
Take a shower/bath	7%	Depression and/or anxiety	9%
Talking with others and being understood	6%	Difficulty maintaining relationships or creating new ones	7%
Eat without assistance	3%	Communication problems	6%
Drive	3%	Need to adjust work/school schedules	5%
Use digital tools (computer, smartphone, etc.)	3%	Financial difficulties	4%
Read a printed document (book, newspaper, etc.)	2%	Job loss or difficulty finding a job	4%
Sit without assistance	2%		

Medical and non-medical care

! Important for health needs



- ✓ **The survey's findings provided a better understanding of FA French community, their conditions and priorities, and fueled the AFAF's roadmap for continuing to improve FA patients' daily life.**



We used this solid data on the disease and its impact, based on the experiences of patients and their families, to **support the EARLY ACCES** (before EU approval) to **Omaveloxolone (Skyclarys®) ***



- ✓ Early Access to Skyclarys was **granted by the French National Health Authority by December 2023.**
- ✓ Since then, French patients have been able to benefit from this treatment.

Real life experiences

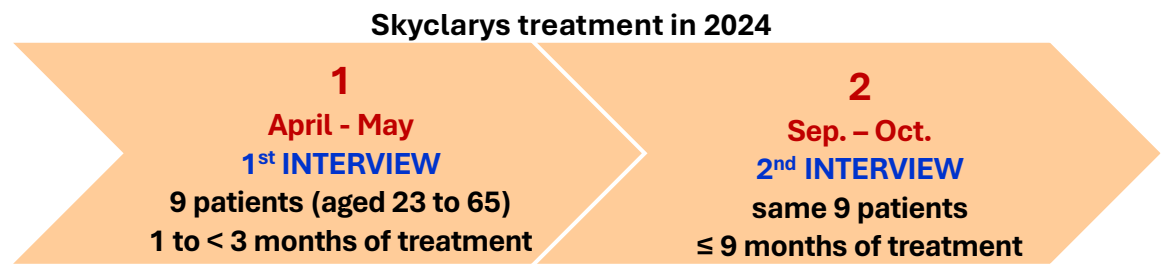
How can your voice make a difference?

Next: To support the application to the French Health Authorities for a potential reimbursement of the costs of Skyclarys, the AFAF collected patients' first experiences following treatment prescriptions



Non-directive interviews => Qualitative analysis

**questions are not pre-arranged*



- **Semi-structured non-invasive interviews** were carried out by 3 AFAF administrators: **patients were allowed to freely express their pre- and post-treatment perceptions**
- The **interviews** were **recorded** and then **transcribed**
- **Interviews** were **analyzed** by a different person from the AFAF, and **summarized according to the themes** defined in the **interview guide** (Perceptions before and since the treatment, hopes, perspectives, additional information).
- **Results were categorized into different domains** perceived by the patients as having an impact on their daily life, and **presented as verbatims**
- **Frequencies** were calculated as the total of patients giving an answer for each domain.

Real life experiences

Turning experiences into data

Main perceived domains with impact on daily life: verbatims

BEFORE SKYLARYS	FATIGABILITY		STRENGTH		BALANCE/ UPRIGHT STABILITY		COORDINATION		SPEECH		OTHER		
	1 st interview at	2 nd interview at	1 st interview at	2 nd interview at	1 st interview at	2 nd interview at	1 st interview at	2 nd interview at	1 st interview at	2 nd interview at	1 st interview at	2 nd interview at	
Autonomous, walking, balance problems, fatigability. Physiotherapy once/week. Yoga. Idebenone.	11 weeks	35 weeks	11 weeks	35 weeks	11 weeks	35 weeks	11 weeks	35 weeks	11 weeks	35 weeks	11 weeks	35 weeks	
	"No improvement so far"					"My physiotherapist tells me that there has been improvement in my balance"			"No improvement so far"			"Not many stunning effects, and no progression of the disease either"	
In wheelchair. Lives alone, transfers alone. Fatigue, loss of balance, pes cavus, handwriting problems, dyspraxia. Goes to the gym twice a week.	9 weeks	35 weeks	9 weeks	35 weeks	9 weeks	35 weeks	9 weeks	35 weeks	9 weeks	35 weeks	9 weeks	35 weeks	
	"When I wake up, I get out of bed and I'm more mobile, everything is much easier, I wake up and I want to move"	"I'm less tired so it's easier to do things better, more efficiently. I also fall less" "When you do a lot of transfers, it requires a lot of energy, but with the medication, it's better"	"Since the Skylarys, when I stand up with my parents help, I can walk a bit, it's not incredible but I can move my legs well enough"						"I feel like I can speak better, in fact I have more energy, it's as if I have more air"	"I find the greatest effect is in the voice. People around me have told me, even the doctor said, I really do have the impression that you speak better"		"Since I started taking the medication, the disease hasn't moved" "I feel really good. In fact I think the fact that I feel less tired plays a huge part in my mental state"	
In wheelchair. Not been able to stand up for 2 years. Physiotherapy 2 times/week, sports 2 times/week. Idebenone and oral contraception.	4 weeks	27 weeks	4 weeks	27 weeks	4 weeks	27 weeks	4 weeks	27 weeks	4 weeks	27 weeks	4 weeks	27 weeks	
												"Pain in the abdomen and menstruation twice in a month"	"Since taking Skylarys, I've had a lot of bleeding, several times a month, and abdominal pain. I've been very tired, so I quit taking it"
Difficulty standing up in the wheelchair stander, pes cavus, needs human assistance, cannot sit up in bed, severe nystagmus, no longer able to read. No physiotherapy.	10 weeks	33 weeks	10 weeks	33 weeks	10 weeks	33 weeks	10 weeks	33 weeks	10 weeks	33 weeks	10 weeks	33 weeks	
		"No change in fatigability"			"I don't need to grab onto my bed or mattress to sit up"	"I'm still sitting without holding on".	"I drop things a little less"		"I was able to play a video game again that I'd stopped, because there were just too many things I couldn't do, well, now I was able to do a bit more"	"People tell me I speak better than before, when I speak, people ask me less to repeat myself"	"I talk more, I don't restrain myself from talking"		"I don't know if it's due to Skylarys, but I was able to move my feet without help. I could do it just a few years ago, I was sure I couldn't do it anymore but I did it"
Autonomous, in a wheelchair. Fatigue (physical and psychological impact). Idebenone.	7 weeks	31 weeks	7 weeks	31 weeks	7 weeks	31 weeks	7 weeks	31 weeks	7 weeks	31 weeks	7 weeks	31 weeks	
			"I stand up in front of the parallel bars at the physiotherapist's, without holding myself up. I found myself more comfortable than usual"								"People around me notice little things about my speech"	"A friend I haven't seen in a year told me I was much better understood"	
Autonomous, manual wheelchair. Fatigue. Bladder problems: Ceris (Trospium chloride). Physiotherapy and gym.	7 weeks	29 weeks	7 weeks	29 weeks	7 weeks	29 weeks	7 weeks	29 weeks	7 weeks	29 weeks	7 weeks	29 weeks	
	"Much less tired after 1 month of treatment"	"Regarding the fatigue, it's not about feeling better, it's about going back to the way you were before, it's like better than before" "I'm no longer tired, I can talk for half an hour in front of people"	"One thing I stopped doing, and I'm starting to do it again, is to bend over in my wheelchair, and get up without my hands or anything, just with my back muscles"		"When it comes to sports, I try harder when I stand up straight, it's possible, so I do it! My physiotherapist is happy because I can do exercises better, for example, with my back"							"I have much less nystagmus"	
Self-education at home, walks with walker, yoga 2 times/day, writing, swimming, adapted pilates, dance, rowing. Physiotherapy, energy therapy, Toviaz and dietary supplements.	11 weeks	36 weeks	11 weeks	36 weeks	11 weeks	36 weeks	11 weeks	36 weeks	11 weeks	36 weeks	11 weeks	36 weeks	
	"Three weeks later after the treatment, I really felt less tired"	"I still feel much less tired, really, and much more energized"	"When I drop something on the ground, I pick it up, but I feel like I get up faster, yes, that's it"			"My physiotherapist sees that I'm more upright"			"And when I'm not tired, I feel like I speak more, it's more fluid"	"I've been told several times that I speak much faster, more fluid"		"I've been taking Toviaz only in the evening for the past three weeks" "I feel like I'm doing things a little faster, which is really encouraging, and for the mood, it's great"	"I quit taking Toviaz" "Before skylarys, I was coughing really badly, now I'd say I just have a light cough, so it's pretty good"
Upper limb problems. Walks with 2 canes. Does a lot of physical therapy.	6 weeks	30 weeks	6 weeks	30 weeks	6 weeks	30 weeks	6 weeks	30 weeks	6 weeks	30 weeks	6 weeks	30 weeks	
	"The first thing I felt, and really quickly within 15 days, was less fatigue"	"When walking with my walker I notice that I'm less short of breath" "I saw the neurologist again. I did the tests"	"I can bend over and straighten up my back" "I can do exercises if they have little amplitude, I"		"I have one knee more stable. I can hop from one foot to the other, and yesterday I tried to run in my garden, I succeeded a little!" "I stand more upright"				"I make longer sentences because I'm less tired"	"I can answer the phone at work without any problem, I feel better about this"		"Many very positive aspects, it changes everything in my life, I begin to do more things"	

Real life experiences

Turning experiences into data

Frequencies of the main identified domains were reflected as a word cloud for visualization.



Bigger words represent a higher instance of reporting and smaller words represent a lower instance of reporting.

- ✓ This approach **complemented the scientific analyses** performed by French neurologists, based on the data collected by the French National Registry of Rare Diseases as part of the Early Access program
- ✓ Skyclarys **received a favorable opinion** for reimbursement in the "treatment of Friedreich's ataxia in adults and adolescents aged 16 years and older"*

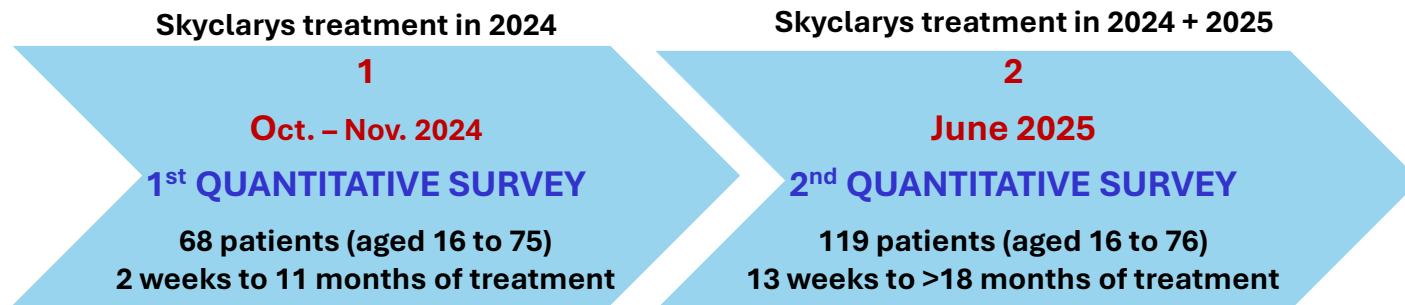
**The Medical Service Provided was "moderate"*

The Improvement of Medical Services Provided: "no therapeutic progress in patient management"

Finally: To support the application for the renewal of the Early Acces and for the resubmission of the reimbursement of the costs of Skyclarys, the AFAF collected patients' **experiences following treatment prescriptions**

The Voice of Patients

2 Surveys => Quantitative analysis

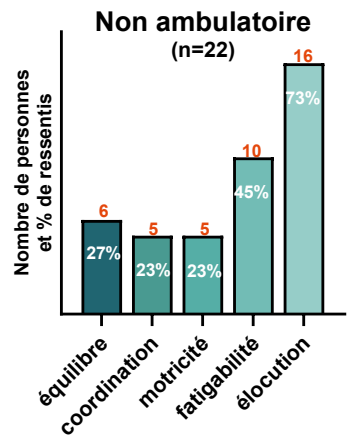
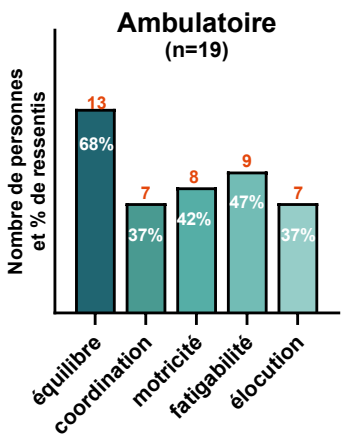
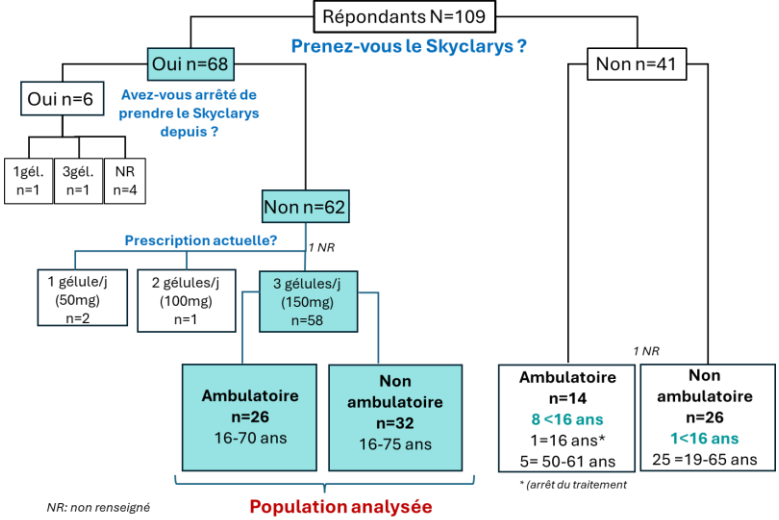


- A **questionnaire online** only to AFAF members (1) and to all french FA patients (2) on LimeSurvey platform.
- **Semi-open questions** on the five most frequently cited elements during the nine non-directive interviews: balance, coordination, motor skills, speech, and fatigue (feelings/positive and negative effects).
- **A choice of answers: Improvement / Deterioration / Neither / Not applicable.**
- **Open questions:** on professional or social life, family life, and hopes for the future.

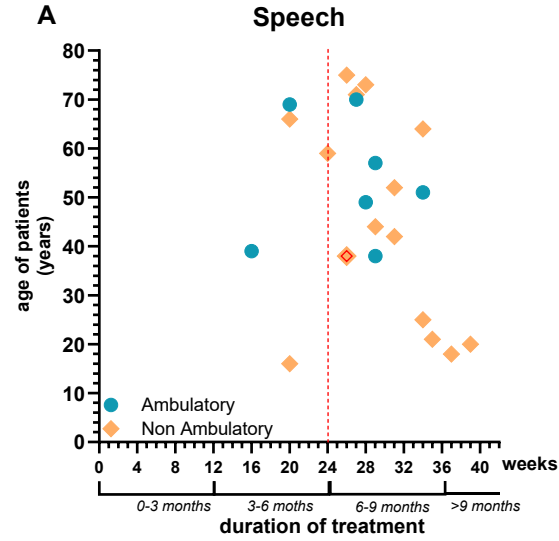
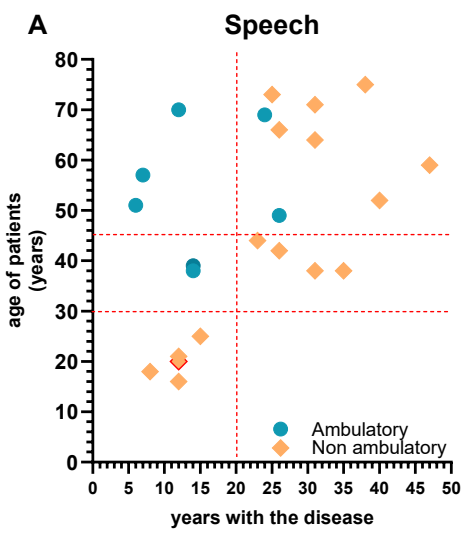
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Turning experiences into data

Characteristics of FA patients



Improved speech in ambulatory and non-ambulatory patients following treatment with Skyclarys



- The interviews and surveys may transform **patient opinions and experiences on topics that directly affect them**, into **facts and figures**
- These facts and figures
 - Can inform health authorities, politicians and decision-makers about what it really means to live with a rare disease.**
 - Can be shared with patient associations and the general public to bring a real change for people living with a rare disease.**
 - Contribute to actively involve patients in research on the topics that matter most to them.**

**For the patient association, this approach
fuel the roadmap for continuing to improve
patients' daily life**

The more of you
who respond
and share your
experiences, the
stronger our
voice will be!

THANK YOU FOR YOUR ATTENTION!