



# Gathering FA patients' experiences and perceptions for HTA processes - example of France

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**International Ataxia Patient Conference**

Hosted by Euro-ataxia

28 – 29 October 2025





1) *Feb 2023* => AFAF contribution  
via EURORDIS: letter about the  
impact of FA in daily life

HAS Questionnaires +  
Audition when possible

2) *Oct 2023* => Early Access Program (EAP)

3) *Oct 2024* => Assessment for reimbursement application

4) *Nov 2024+ Jan 2025* => Early Access Program 1<sup>st</sup> renewal

5) *Aug 2025* => Re-submission of the Assessment for reimbursement application

6) *Nov2025* => Early Access Program 2<sup>nd</sup> renewal

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



**The Voice of Patients**



**1<sup>st</sup> Survey**

- 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**



**Total : 209 responses**

All answers are  
anonymous  
and accessible  
only to the  
research team

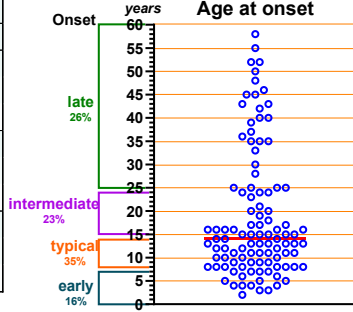
# Real life experiences

## Turning experiences into data

### Who are you?

#### Characteristics of FA individuals in France

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



### What's your daily living?

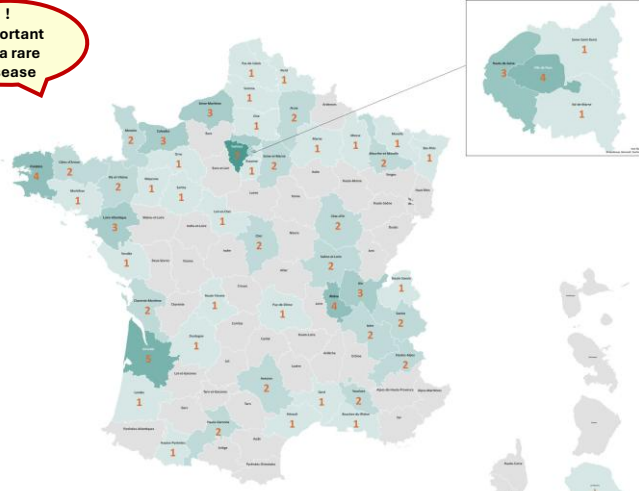
#### Symptoms with the heaviest impact on daily life

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

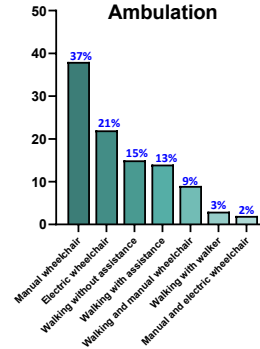
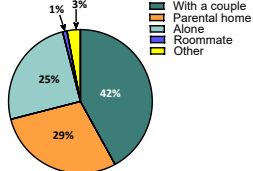
Main symptoms encountered	Symptoms with the greatest impact on quality of life	Symptoms that patients are most worried about as the FA progresses
Upper limb coordination, manual dexterity	83%	Balance / walking / falls 22%
Fatigue	77%	Upper limb coordination, manual dexterity 17%
Balance / walking / falls	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%
Diabetes	13%	Pain 4%
		Spasticity 4%
		Swallowing difficulties 4%
		Diabetes 2%

#### Geographical distribution and type of residency

! Important for a rare disease



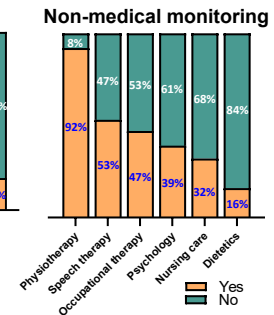
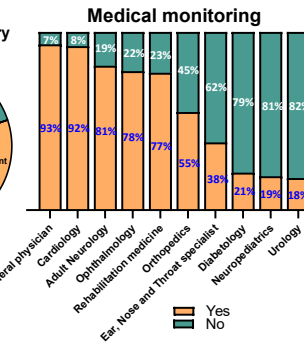
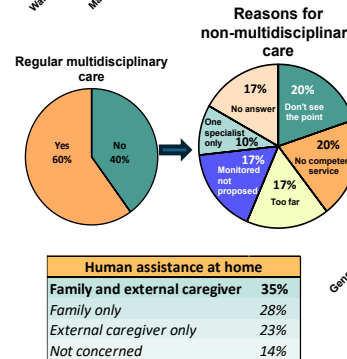
Living place



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



We used these results to **support the EARLY ACCES to Omaveloxolone (Skyclarys®)** (before EU approval)

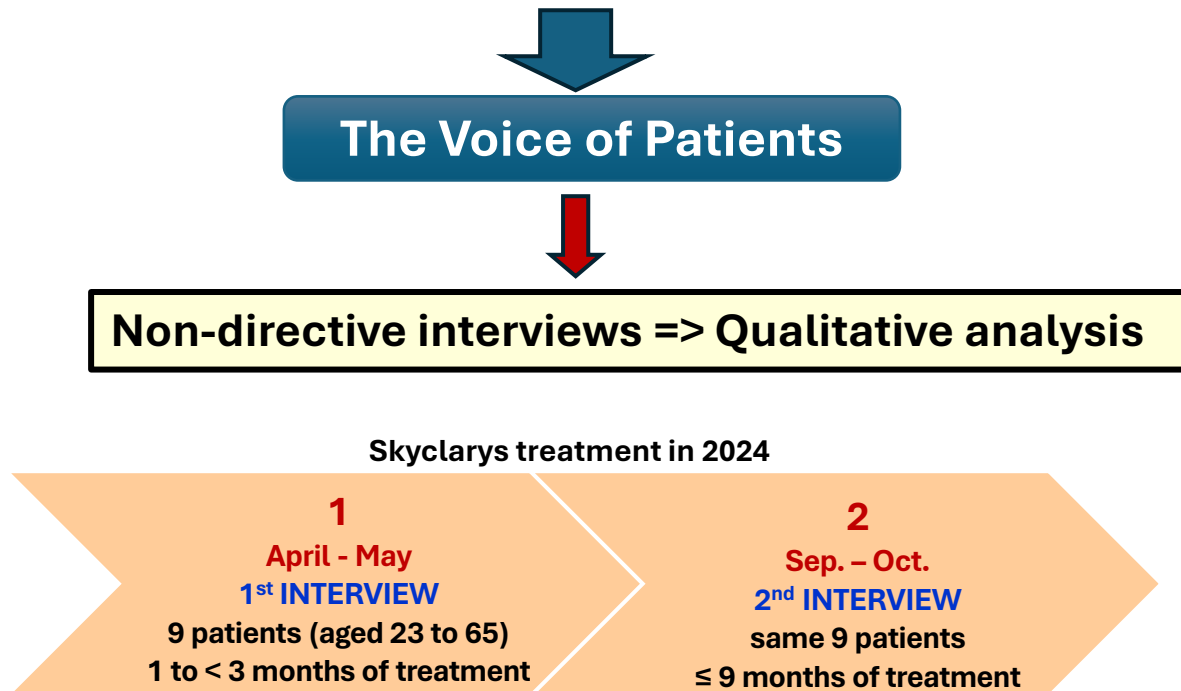


- ✓ Early Access to Skyclarys was granted by the French National Health Authority (HAS) by Dec. 2023.
- ✓ Patients started to be treated with Skyclarys under the Early access Program, from Jan. 2024.

# Real life experiences

## *Turning experiences into data*

Next: We wanted to understand the **impact of Skyclarys treatment** on patients' everyday life



- **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 <sup>st</sup> interview at	2 <sup>nd</sup> interview at	1 <sup>st</sup> interview at	2 <sup>nd</sup> interview at	1 <sup>st</sup> interview at	2 <sup>nd</sup> interview at	1 <sup>st</sup> interview at	2 <sup>nd</sup> interview at	1 <sup>st</sup> interview at	2 <sup>nd</sup> interview at	1 <sup>st</sup> interview at	2 <sup>nd</sup> interview at		
<b>Interview 1 (F/age 23)</b> 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"	"It's through the eyes of others that I see changes, for example my speech is better"		"Not many stunning effects, and no progression of the disease either"		
<b>Interview 2 (M/age 24)</b> 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"		
<b>Interview 3 (F/age 30)</b> 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		
												"My mum says that my legs don't go everywhere when she helps me to do things"	"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"
<b>Interview 4 (F/age 31)</b> 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"	
<b>Interview 5 (M/age 32)</b> 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"		
<b>Interview 6 (F/age 35)</b> 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"	
<b>Interview 7 (F/age 43)</b> 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"	
<b>Interview 8 (F/age 51)</b> 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.

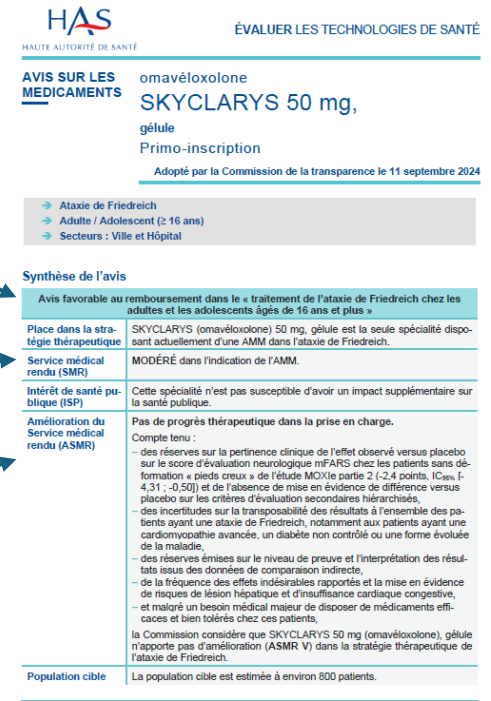


Bigger words represent a higher instance of reporting and smaller words represent a lower instance of reporting.

We used these results to support the **Assessment for Reimbursement application (Oct. 2024)**

✓ 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 (SMR) was **"moderate"**
- The Improvement of Medical Service Provided: **"No therapeutic progress in patient management"** (ASMR V)



**HAS** HAUTE AUTORITÉ DE SANTÉ ÉVALUER LES TECHNOLOGIES DE SANTÉ

**AVIS SUR LES MÉDICAMENTS** omavéloxolone  
**SKYCLARYS 50 mg,**  
gélule  
Primo-inscription  
Adopté par la Commission de la transparence le 11 septembre 2024

→ Ataxie de Friedreich  
→ Adulte / Adolescent (≥ 16 ans)  
→ Secteurs : Ville et Hôpital

**Synthèse de l'avis**

**Avis favorable au remboursement dans le « traitement de l'ataxie de Friedreich chez les adultes et les adolescents âgés de 16 ans et plus »**

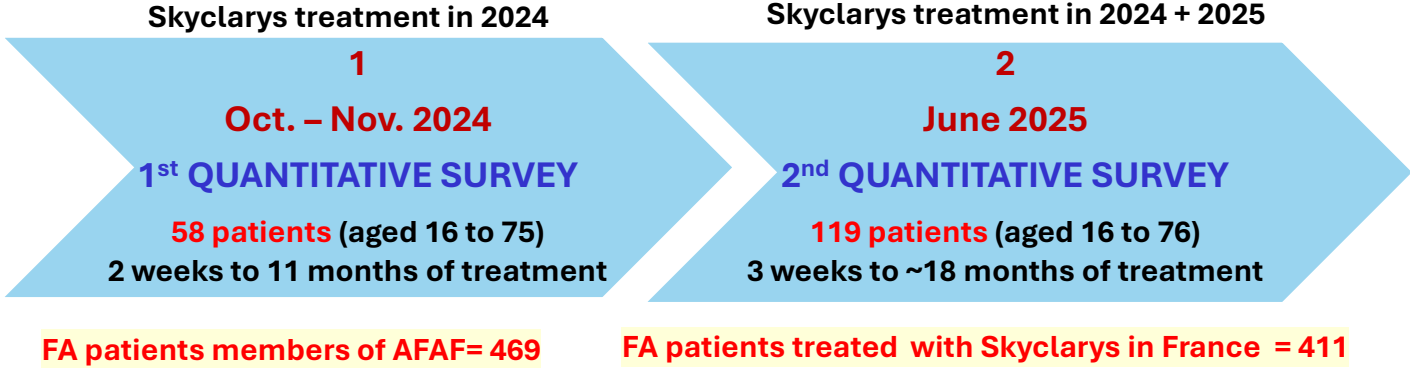
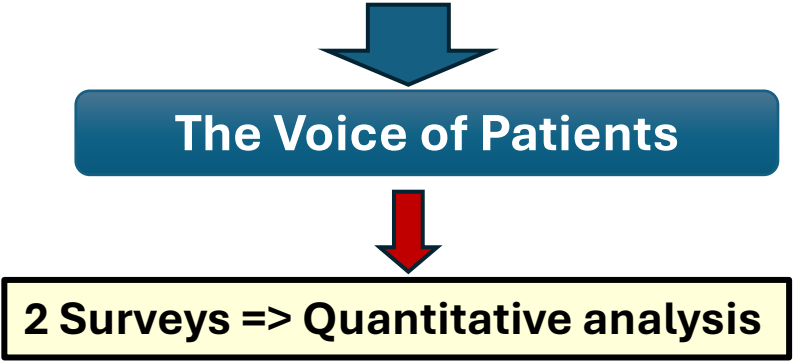
Place dans la stratégie thérapeutique	SKYCLARYS (omavéloxolone) 50 mg, gélule est la seule spécialité disponible actuellement d'une AMM dans l'ataxie de Friedreich.
Service médical rendu (SMR)	MODÉRÉ dans l'indication de l'AMM.
Intérêt de santé publique (ISP)	Cette spécialité n'est pas susceptible d'avoir un impact supplémentaire sur la santé publique.
Amélioration du Service médical rendu (ASMR)	<p>Pas de progrès thérapeutique dans la prise en charge.</p> <p>Compte tenu :</p> <ul style="list-style-type: none"> <li>- des réserves sur la pertinence clinique de l'effet observé versus placebo sur le score d'évaluation neurologique mFARS chez les patients sans déformation « pieds creux » de l'étude MOX/Le partie 2 (-2,4 points, IC<sub>95%</sub> [-4,31 ; -0,50]) et de l'absence de mise en évidence de différence versus placebo sur les critères d'évaluation secondaires hiérarchisés,</li> <li>- des incertitudes sur la transposabilité des résultats à l'ensemble des patients ayant une ataxie de Friedreich, notamment aux patients ayant une cardiomyopathie avancée, un diabète non contrôlé ou une forme évolutive de la maladie,</li> <li>- des réserves émises sur le niveau de preuve et l'interprétation des résultats issus des données de comparaison indirecte,</li> <li>- de la fréquence des effets indésirables rapportés et la mise en évidence de risques de lésion hépatique et d'insuffisance cardiaque congestive, et malgré un besoin médical majeur de disposer de médicaments efficaces et bien tolérés chez ces patients,</li> </ul> <p>la Commission considère que SKYCLARYS 50 mg (omavéloxolone), gélule n'apporte pas d'amélioration (ASMR V) dans la stratégie thérapeutique de l'ataxie de Friedreich.</p>
Population cible	La population cible est estimée à environ 800 patients.

HAS - SKYCLARYS 50 mg, gélule - septembre 2024

# Real life experiences

## Turning experiences into data

Finally: We wished to know the impact of **the long-term treatment** with Skyclarys in a **bigger population**

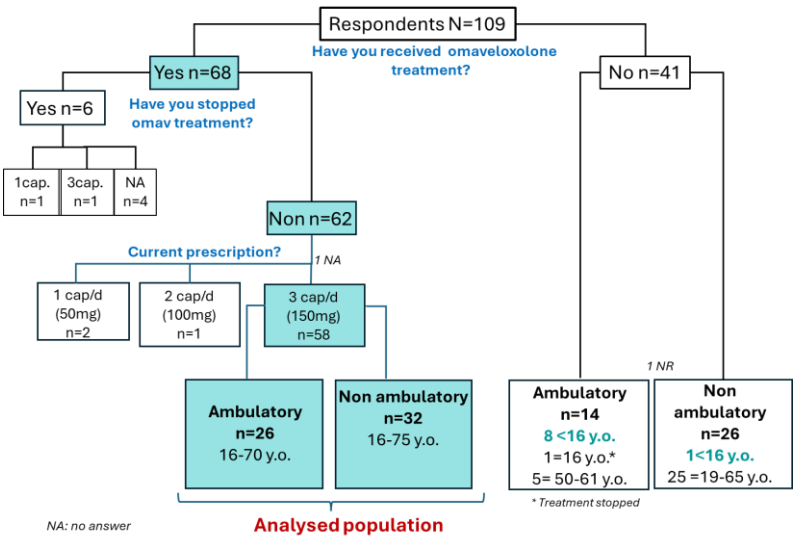


- A **questionnaire online** only to AFAF members (1) and to all French FA patients (2) on LimeSurvey platform.
- **Semi-open questions** on the five most frequently cited elements during the nine non-directive interviews: balance, coordination, motor skills, speech, and fatigue (feelings/positive and 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.

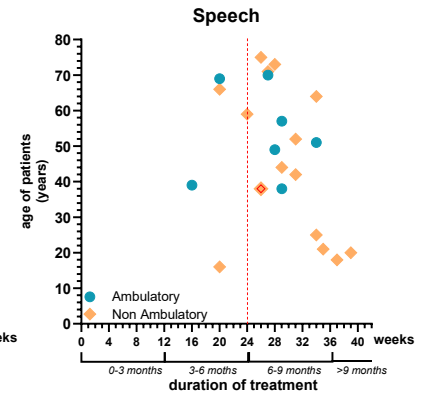
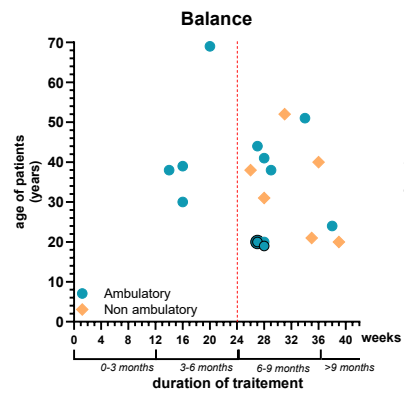
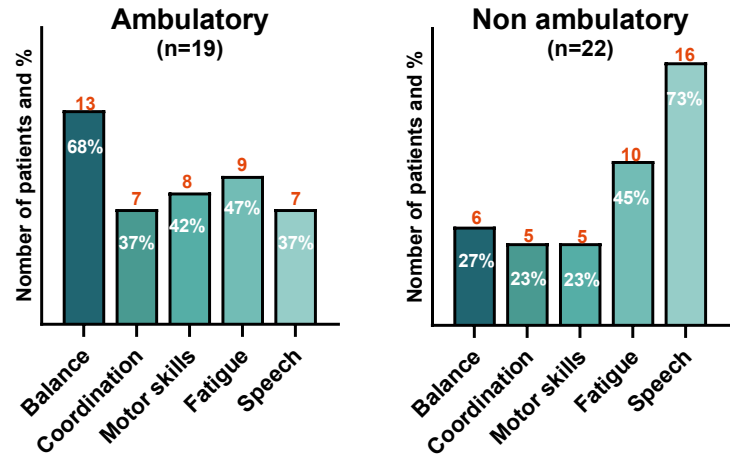
# Real life experiences

## Turning experiences into data

### 1<sup>st</sup> QUANTITATIVE SURVEY: 2 weeks - 11 months of treatment



### Positive effects



We used these results to support the application for the 1<sup>st</sup> renewal of the Early Acces (Dec.2024 and Jan.2025)

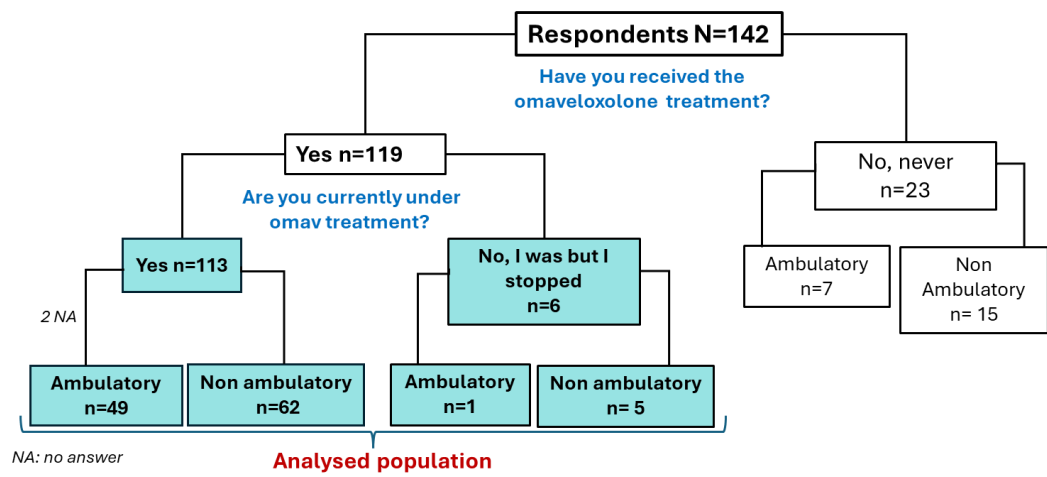


✓ Early Access to Skyclarys was granted for another year

# Real life experiences

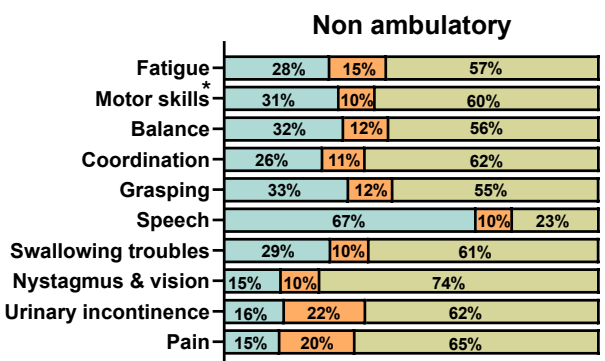
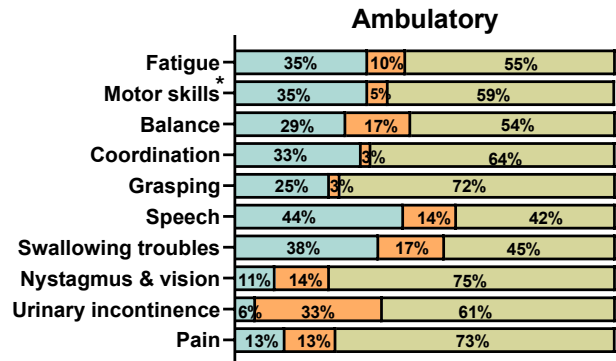
## Turning experiences into data

### 2<sup>nd</sup> QUANTITATIVE SURVEY: 3 weeks to ~18 months of treatment



Patients impacted by difficulties in:

Analysed population	Fatigue	Motor skills	Balance	Coordination	Grasping	Speech	Swallowing	Nystagmus & visual disturbances	Urinary incontinence	Pain
Ambulatory (n=50)	80% (n=40)	74% (n=37)	82% (n=41)	78% (n=39)	72% (n=36)	72% (n=36)	58% (n=29)	56% (n=28)	36% (n=18)	60% (n=30)
Non ambulatory (n=67)	79% (n=53)	78% (n=52)	75% (n=50)	79% (n=53)	76% (n=51)	78% (n=52)	73% (n=49)	58% (n=39)	55% (n=37)	60% (n=40)



The improvement appeared at	The improvement appeared at		
	0 - 6 months	6 - 12 months	> 1 year
Fatigue (n=29)	21 (72%)	7 (24%)	1 (3%)
Motor skills (n=26)	20 (77%)	6 (23%)	0 (0%)
Balance (n=28)	23 (82%)	4 (14%)	1 (4%)
Coordination (n=25)	17 (68%)	8 (32%)	0 (0%)
Grasping (n=25)	19 (76%)	6 (24%)	0 (0%)
Speech (n=50)	40 (80%)	9 (18%)	1 (2%)
Swallowing (n=23)	20 (87%)	3 (13%)	0 (0%)
Nystagmus & vision (n=9)	6 (67%)	2 (22%)	1 (11%)
Urinary incontinence (n=7)	3 (43%)	4 (57%)	0 (0%)
Pain (n=8)	4 (50%)	4 (50%)	0 (0%)

■ Improvement  
■ Deterioration  
■ Neither one nor the other

\* Motor skills: i.e. during physiotherapy sessions

# Real life experiences

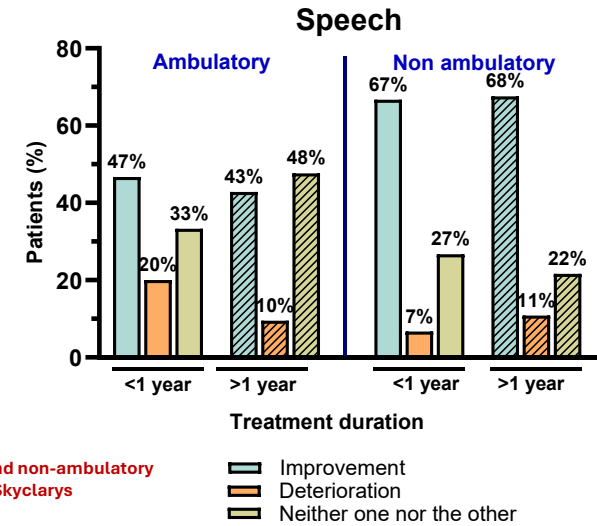
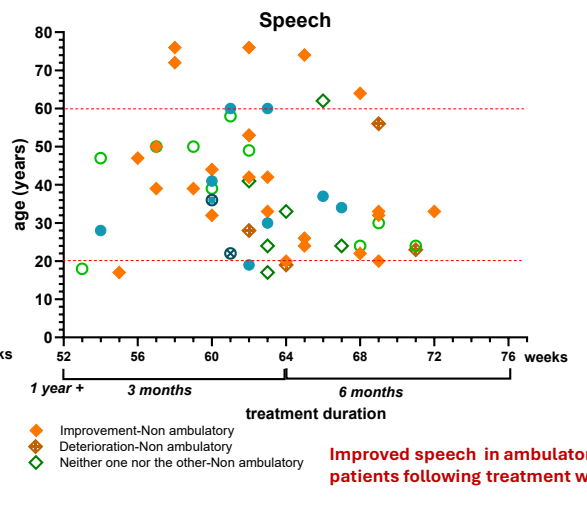
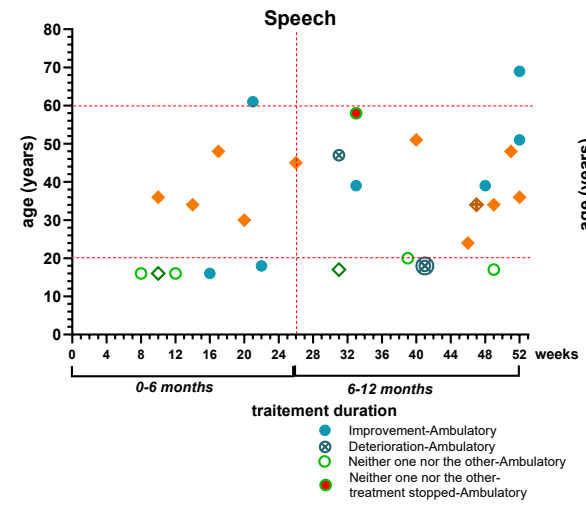
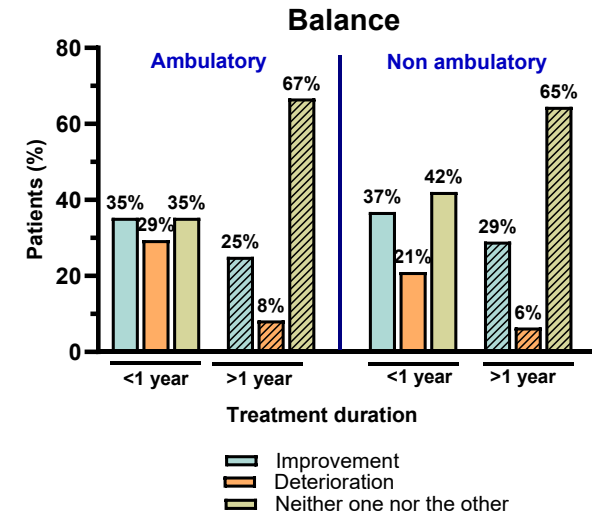
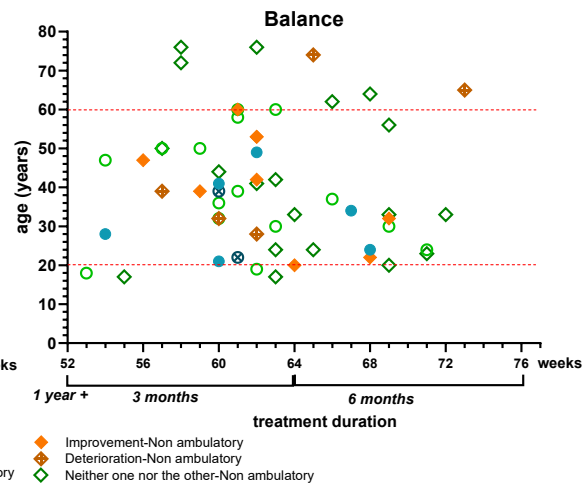
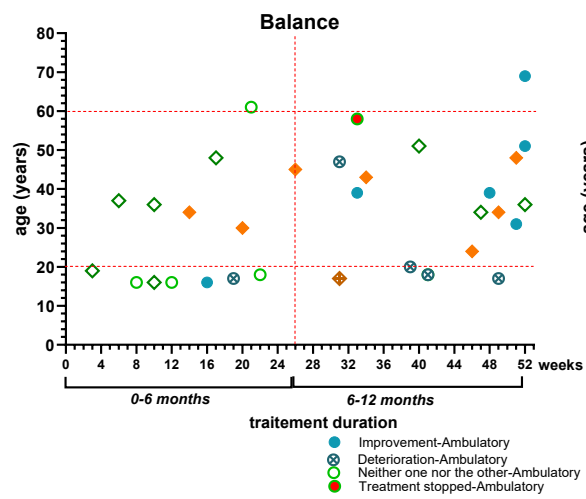
## Turning experiences into data

### 2<sup>nd</sup> QUANTITATIVE SURVEY

### 3 weeks to ~18 months of treatment

#### Patients of <1 year of treatment

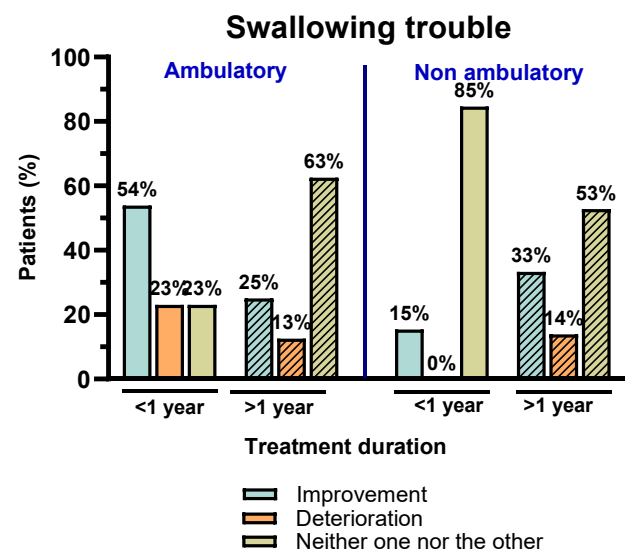
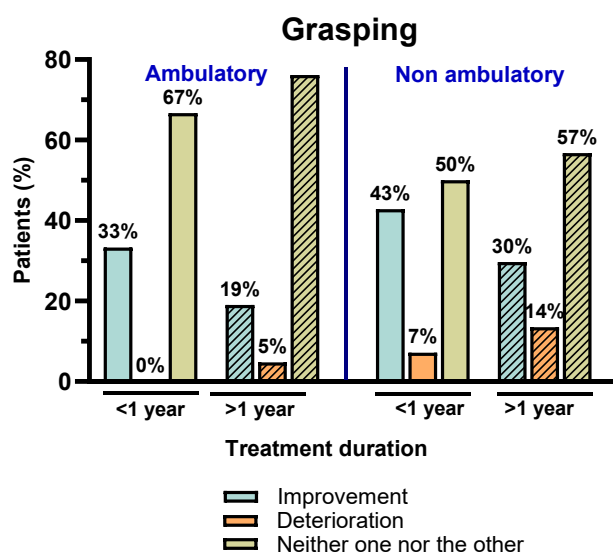
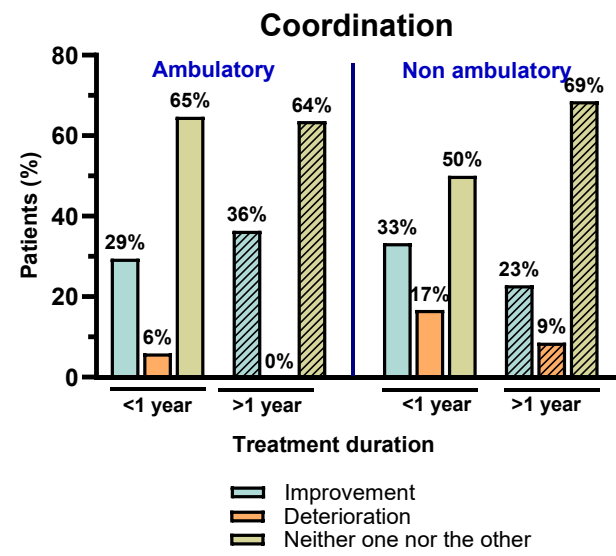
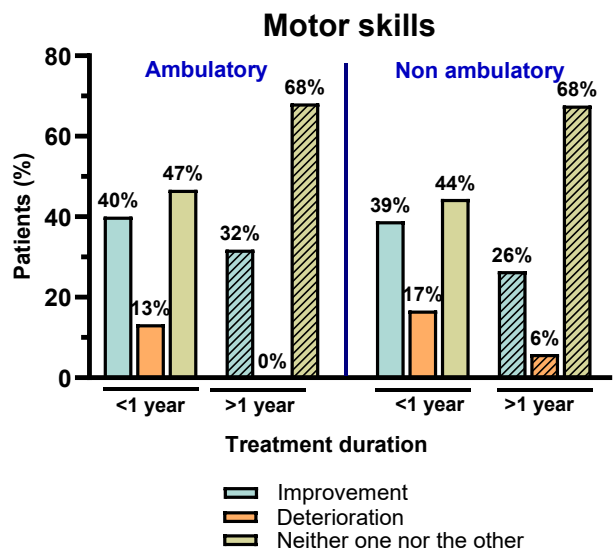
#### Patients of >1 year of treatment



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

### 2<sup>nd</sup> QUANTITATIVE SURVEY

3 weeks to ~18 months of treatment



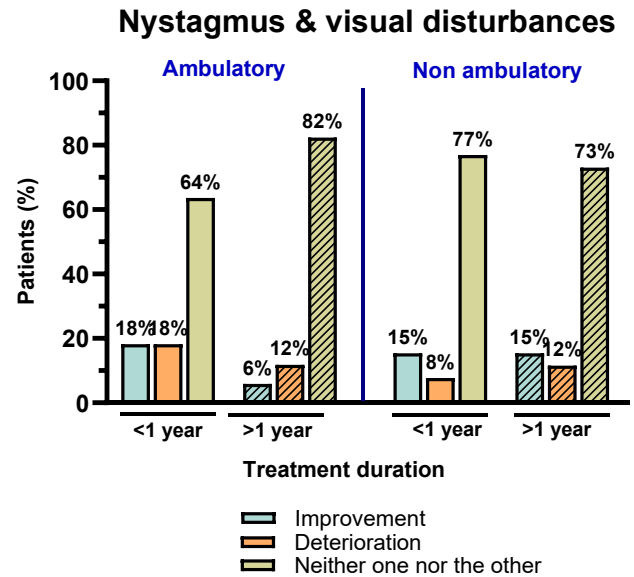
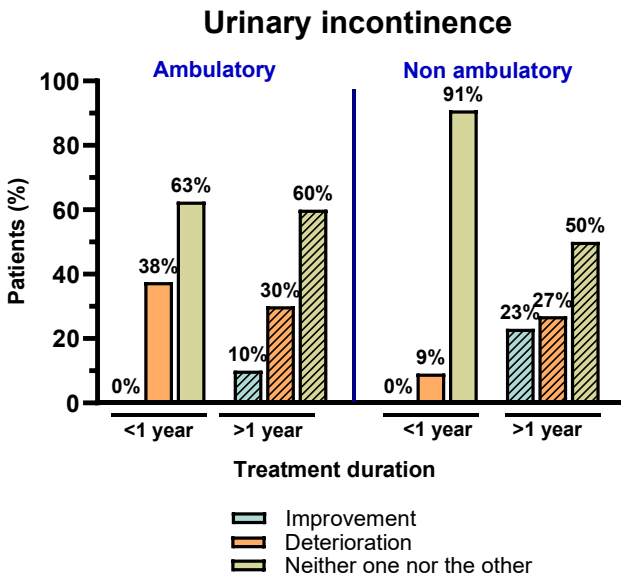
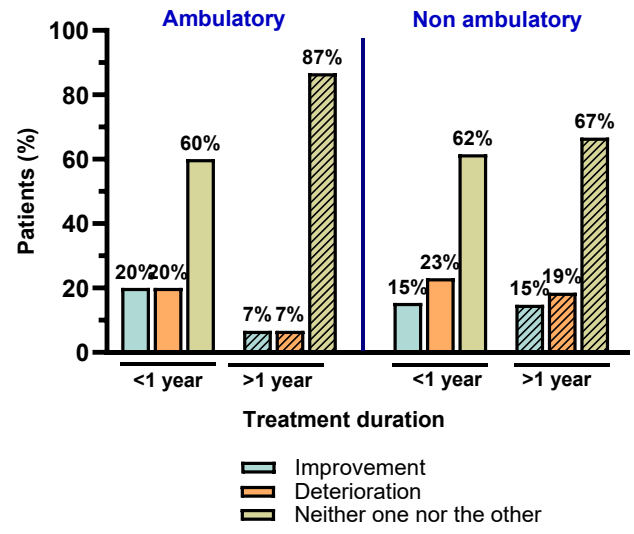
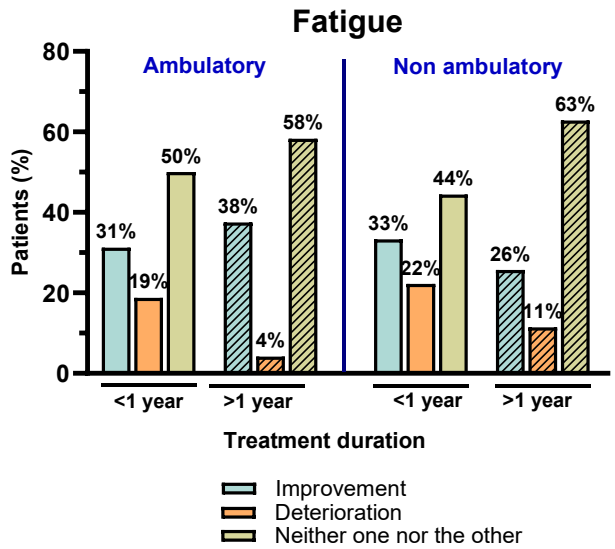
# Real life experiences

## Turning experiences into data

### 2<sup>nd</sup> QUANTITATIVE SURVEY

#### 3 weeks to ~18 months of treatment

#### Pain



*After more than a year of treatment, perceptions described as "neither improvement nor deterioration" of the disease, could be interpreted as a sense of stability, with the disease remaining unchanged.*

# Real life experiences

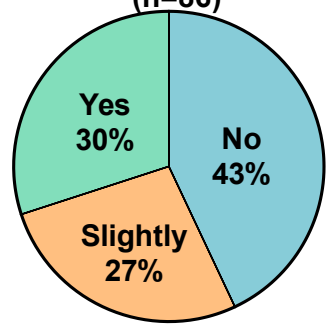
## Turning experiences into data

### 2<sup>nd</sup> QUANTITATIVE SURVEY

3 weeks to ~18 months of treatment

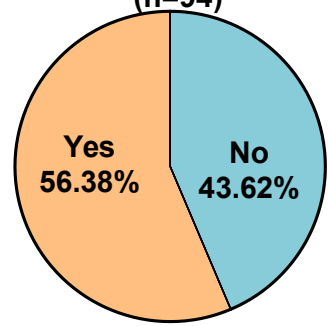
#### Stabilisation of the disease

Ambulatory & Non ambulatory (n=86)

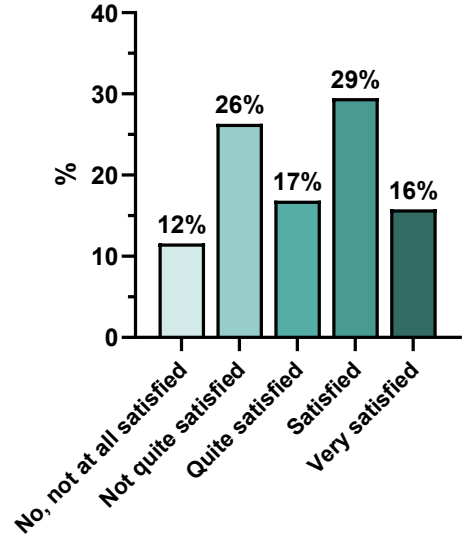


#### Improvement of quality of life

Ambulatory & Non ambulatory (n=94)

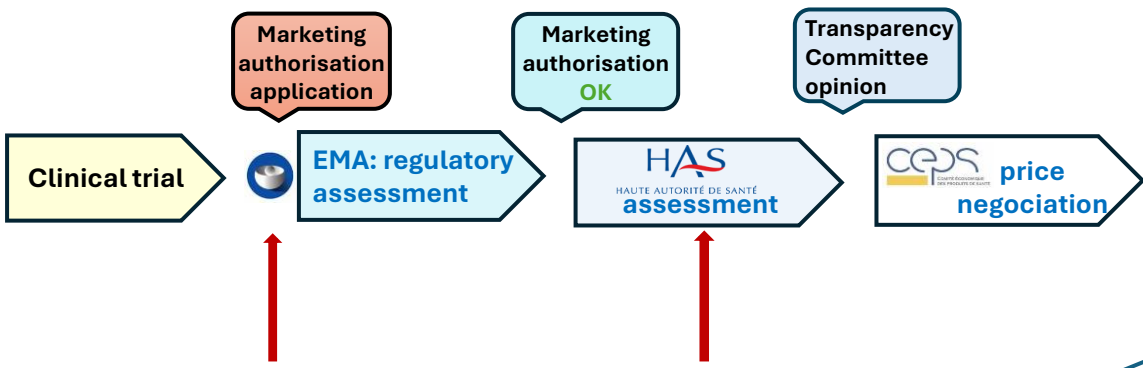


#### Level of satisfaction of Skyclarys



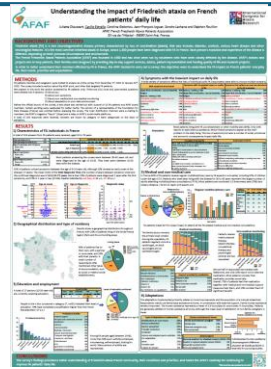
We used these results to **support the re-submission of the Assessment for Reimbursement application (Aug.2025)** and will be used support the **2<sup>nd</sup> renewal of the Early Acces (Nov.2025)**

# Conclusion

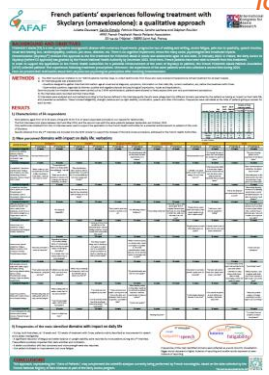


- 1) *Feb 2023* => **AFAP contribution via EURORDIS: letter about the impact of FA in daily life**
- 2) *Oct 2023* => **Early Access Program (EAP) Survey on FA patients in France**
- 3) *Oct 2024* => **Assessment for reimbursement application => Non-directive interviews on Skylarys effects at <9 months of treatment**
- 4) *Nov 2024+ Jan 2025* => **Early Access Program 1<sup>st</sup> renewal => Quantitative Survey Skylarys effects 2 weeks to 11 months of treatment**
- 5) *August 2025* => **Re-submission of the Assessment for reimbursement application => Preliminary results Quantitative Survey Skylarys effects 3 to 72 weeks of treatment**
- 6) *Nov 2025* => **Early Access Program 2<sup>nd</sup> renewal => Full results Quantitative Survey Skylarys effects 3 to 72 weeks of treatment**

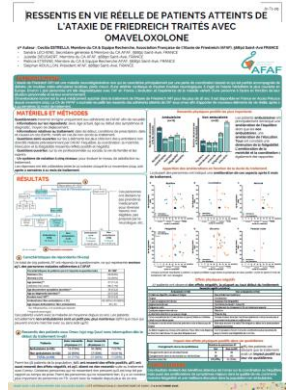
**HAS Questionnaires + Audition when possible**



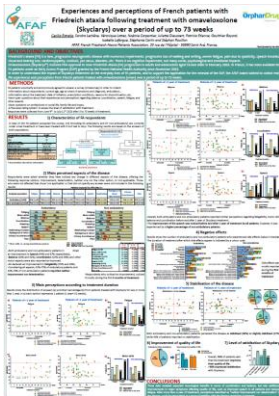
ICAR2024



ICAR2024



RARE 2025



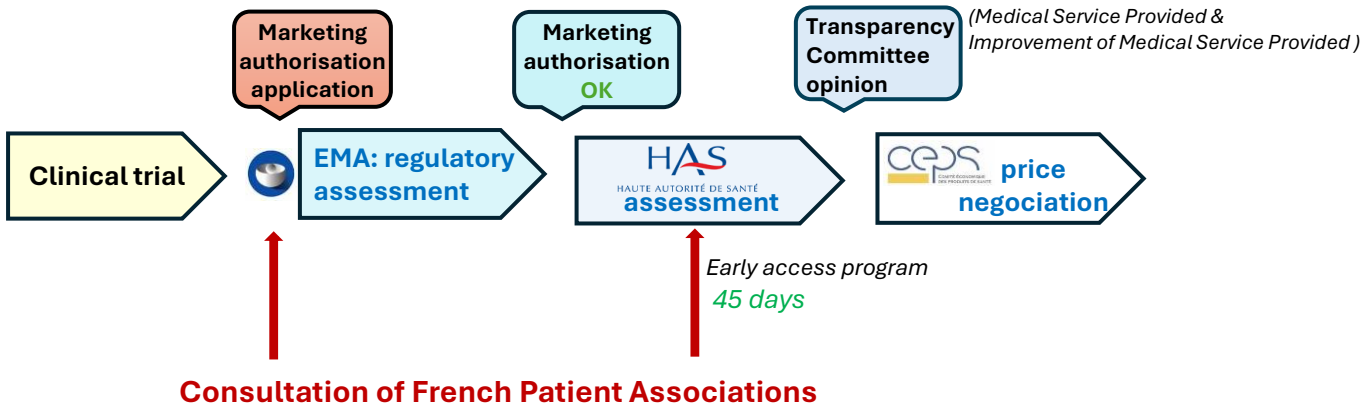
WODC2025

We are convinced that these interviews and surveys not only can inform health authorities about what it really means to live with FA, but also contribute to bring a real change for people living with FA.

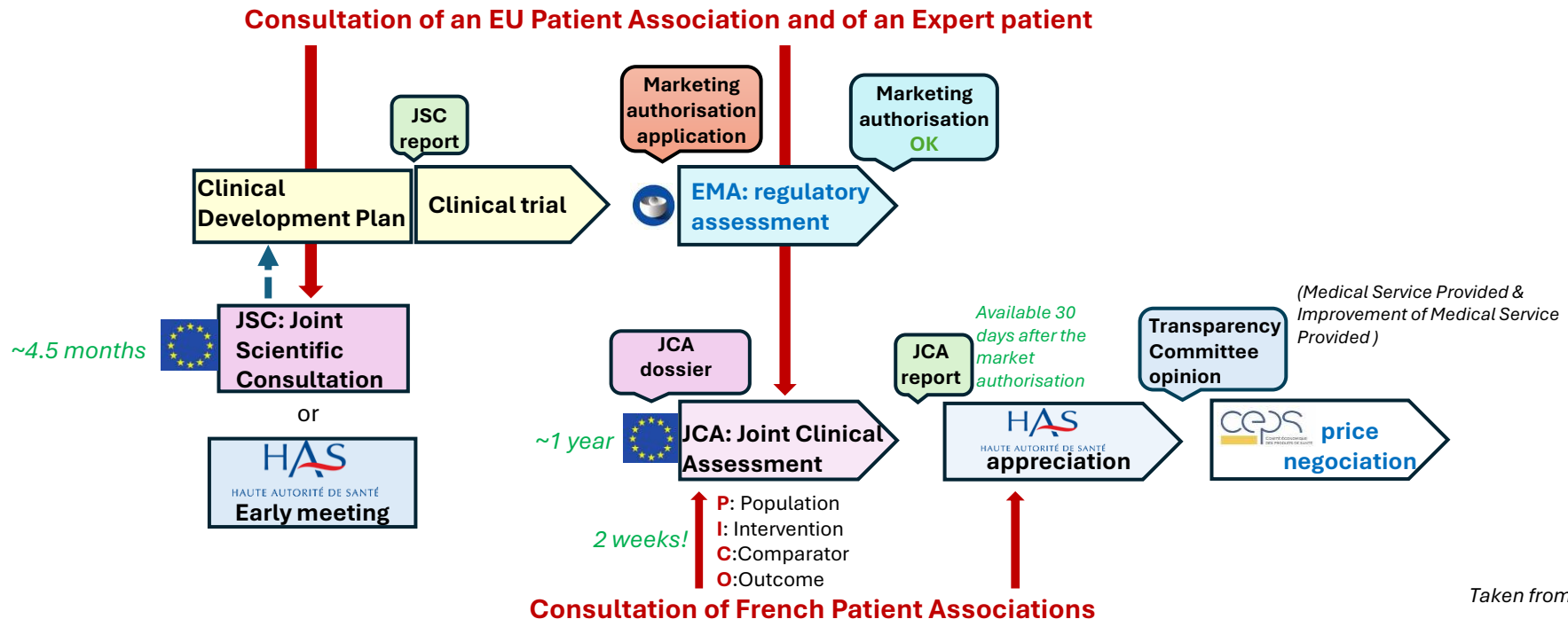


# Changes on Health Technology Assessment in France

## Before



## Now



**THANK YOU FOR YOUR ATTENTION!**