Experiences and perceptions of French patients with Friedreich ataxia following treatment with omaveloxolone (Skyclarys) over a period of up to 73 weeks

Cecilia Estrella, Sandra Lechêne, Véronique Lattes, Noémie Carpentier, Juliette Dieusaert, Patricia Etienne, Gaulthier Bayard, Isabelle Lafargue, Stéphanie Carlin and Stéphan Rouillon

AFAF, French Friedreich Ataxia Patients Association, 33 rue de l'Hôpital - 56890 Saint Avé, France.

*Work funded by the AFAF

BACKGROUND AND OBJECTIVES

Friedreich's ataxia (FA) is a rare, progressive neurogenetic disease with numerous impairments: progressive loss of walking and writing, severe fatigue, pain due to spasticity, speech troubles, visual and hearing loss, cardiomyopathy, scoliosis, pes cavus, diabetes, etc. There is no cognitive impairment, but many social, psychological and emotional impacts.

Omaveloxolone (Skyclarys®) received FDA approval to slow Friedreich Ataxia (FA) progression in adults and adolescents aged 16 and older in February 2023. In France, it has been available for FA patients under an Early Access Program (EAP) granted by the French National Health Authority since November 2023.

In order to understand the impact of Skyclarys treatment on the everyday lives of FA patients, and to support the application for the renewal of the EAP, the AFAF board wished to collect real-life experiences and perceptions from French patients treated with omaveloxolone (omav) over a period of up to 73 weeks.

METHODS

FA patients voluntarily and anonymously agreed to answer a survey (LimeSurvey) in order to collect:

-Information about respondents: current age, age at onset of symptoms and diagnosis, ambulation…

-Information about the treatment: date of initiation, prescription conditions, reasons for discontinuation, etc.

-Semi-open questions about their experiences and perceptions regarding balance, coordination, speech, fatigue, and other aspects.

-Open questions on professional or social life, family life and hopes.

-A five-level rating system: to assess the level of satisfaction with treatment.

Responses were collected from June 4th to July 11th 2025 after 3 to 73 weeks of treatment.

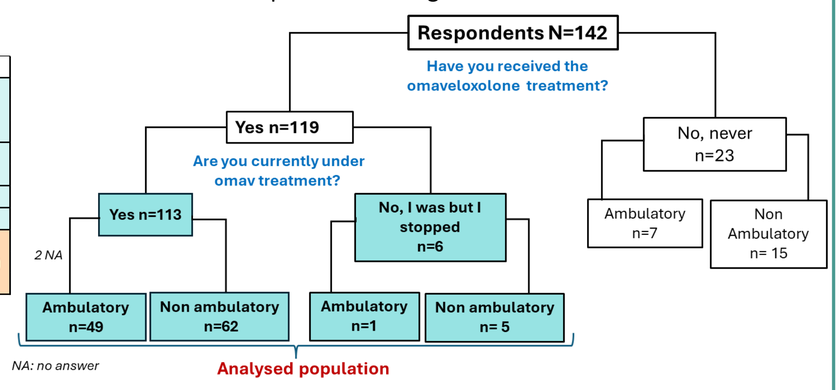
RESULTS

1) Characteristics of FA respondents

A total of 142 FA patients answered the survey, 119 (including 50 ambulatory and 67 non-ambulatory) are currently under omav treatment or have been treated with it but had to stop. The following results are based on the answers of both populations.

Characteristics of respondents	N=139*
Female	84
Male	56
Prefer not to answer	1
Age (years)(mean±SD)	37.09 (16.42)
Median (min-max)	36 (12-76)
Age at onset (years)(mean±SD)	17.34 (11.7)
Age at diagnosis (years)(mean±SD)	20.98 (13.47)
Ambulatory / Non ambulatory (n)	50/66
Age (years)(mean±SD)	33.6 (14.74)/ 38.6(15.75)
Median (min-max)	31.5(16-69)/36 (16-76)

** Characteristics informed by 139 patients*

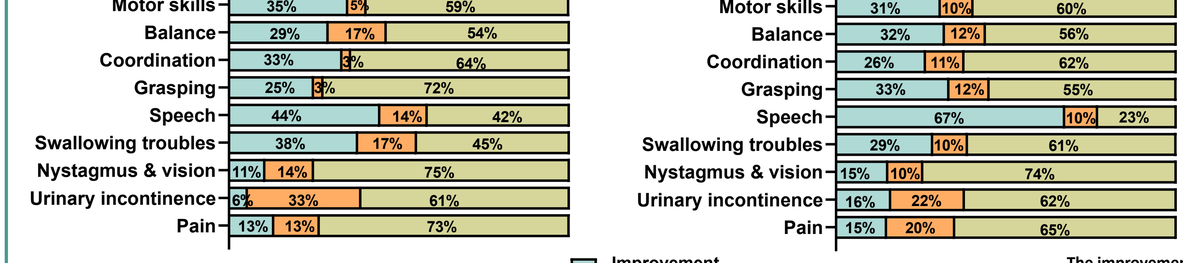


2) Main perceived aspects of the disease

Respondents were asked whether they have noticed any change in different aspects of the disease, offering the following response options: improvement, deterioration, neither one nor the other option, or not applicable. Those who were not affected that chose 'not applicable' or that did not specify any answer were not included in the following results:

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)



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

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%)

-Both ambulatory and non-ambulatory patients mainly noticed an improvement in **Speech** (44% and 67% respectively).

-**Balance** (29% and 32%), **Coordination** (33% and 36%) and other motor aspects were also reported as improved.

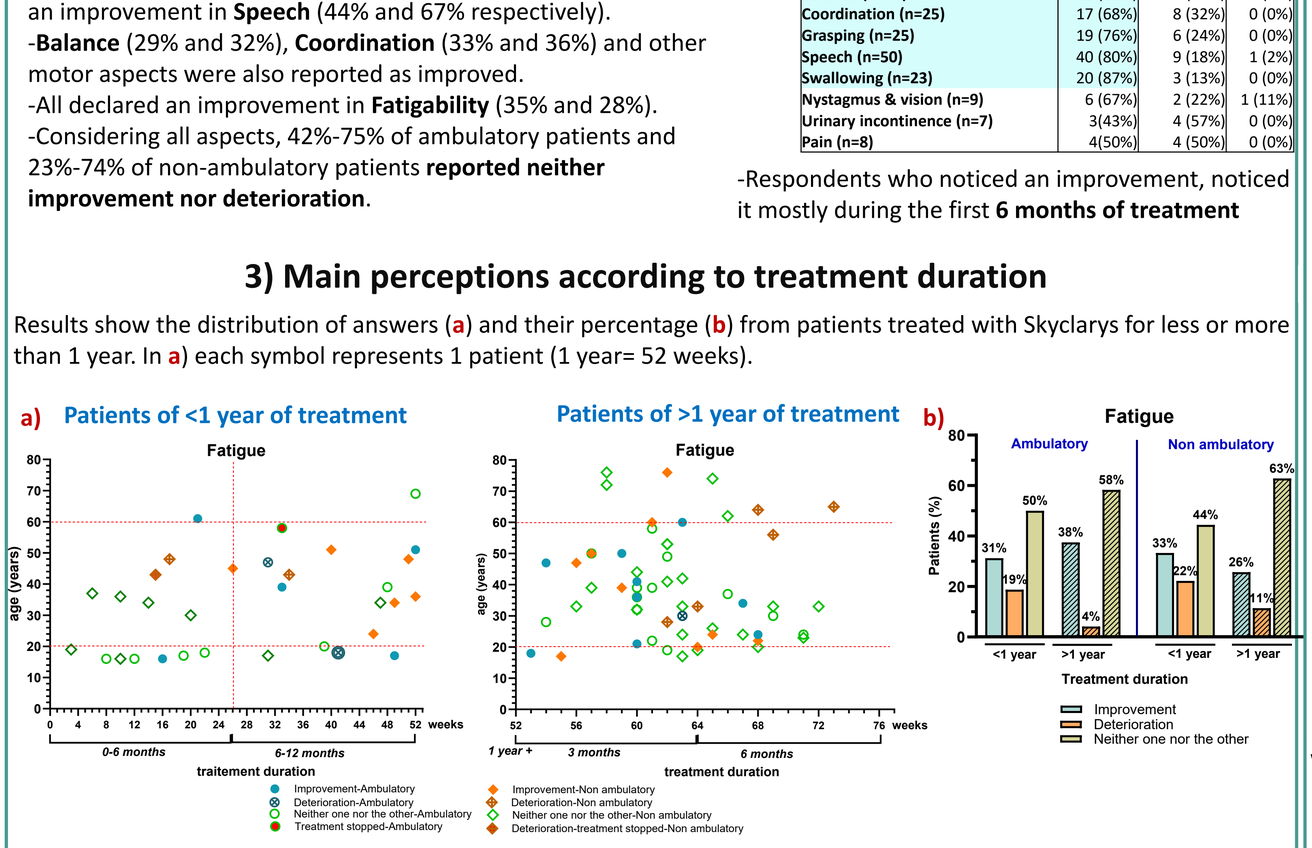
-All declared an improvement in **Fatigability** (35% and 28%).

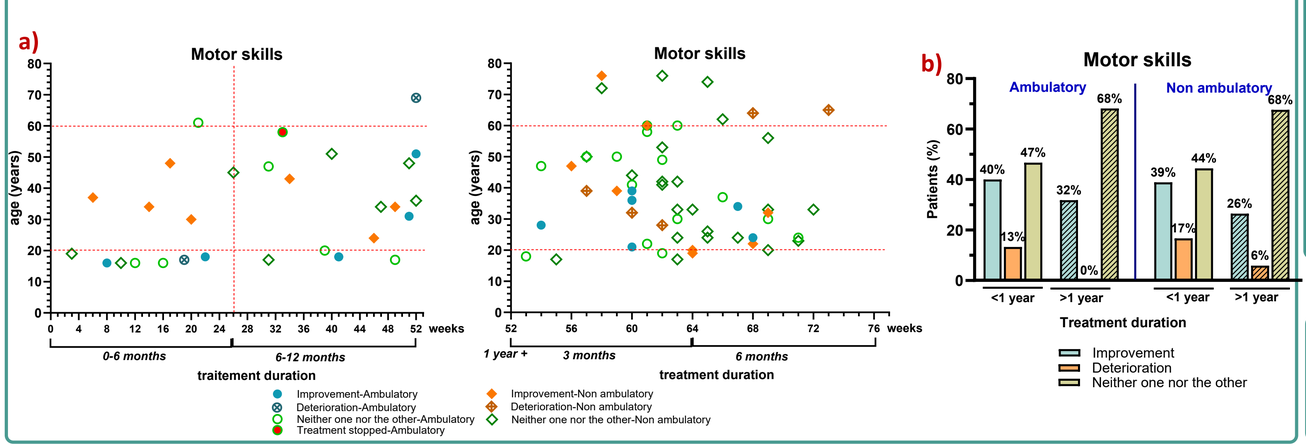
-Considering all aspects, 42%-75% of ambulatory patients and 23%-74% of non-ambulatory patients **reported neither improvement nor deterioration**.

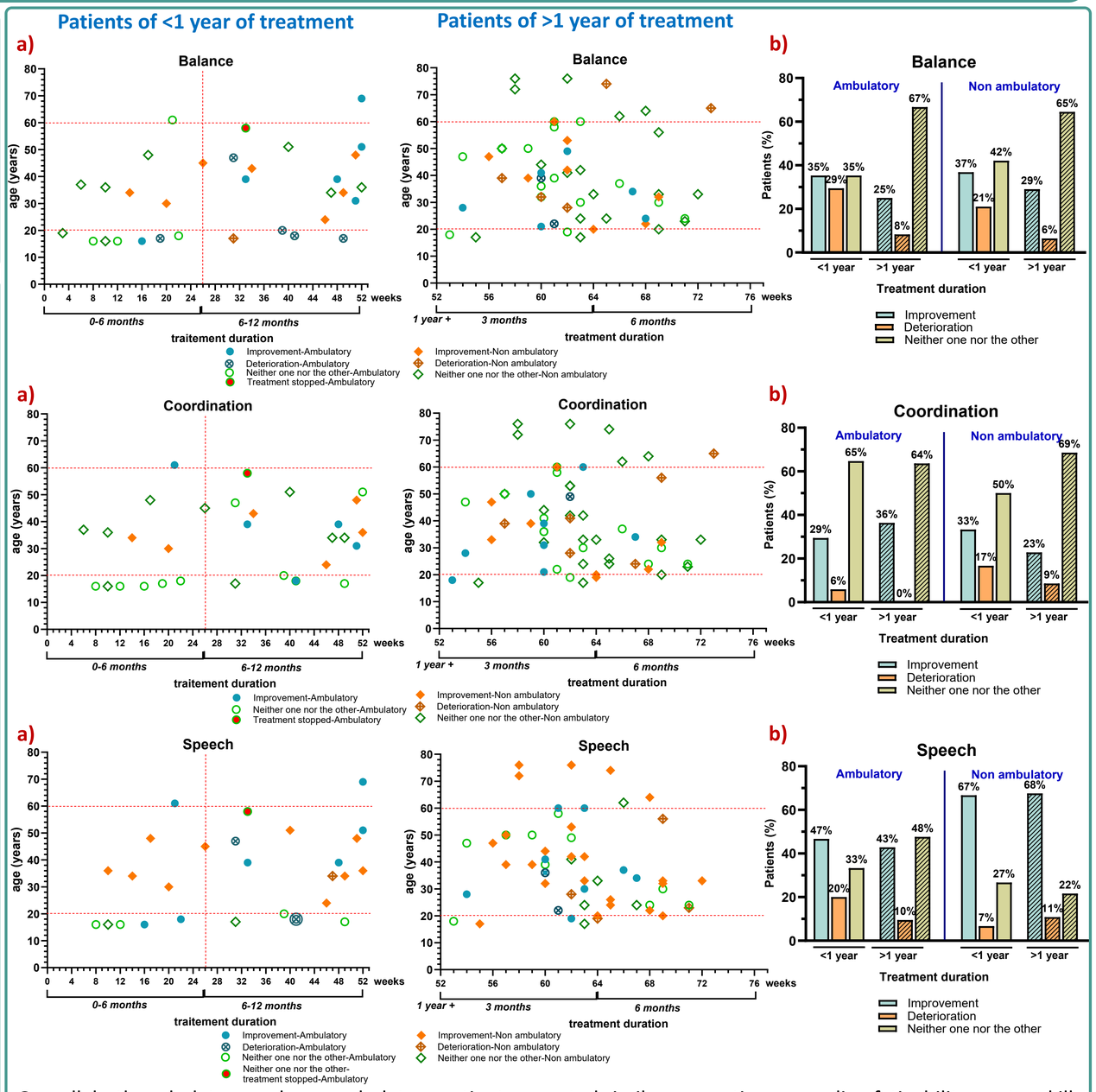
-Respondents who noticed an improvement, noticed it mostly during the first **6 months of treatment**

3) Main perceptions according to treatment duration

Results show the distribution of answers (**a**) and their percentage (**b**) from patients treated with Skyclarys for less or more than 1 year. In **a**) each symbol represents 1 patient (1 year= 52 weeks).





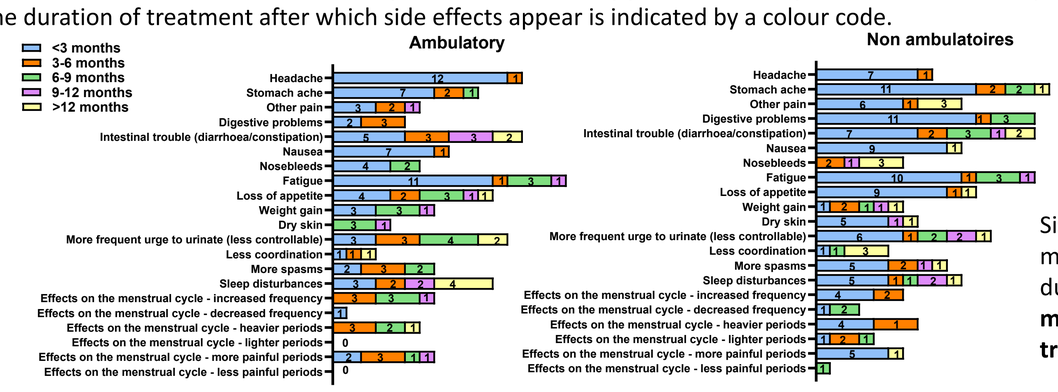


-Overall, both ambulatory and non ambulatory patients reported similar perceptions regarding fatigability, motor skills, balance and coordination before and after 1 year of Skyclarys treatment.

-The **improvement of the speech was noticed before and after 1 year of treatment by all patients**, however, it was experienced by a **higher percentage of non ambulatory patients**.

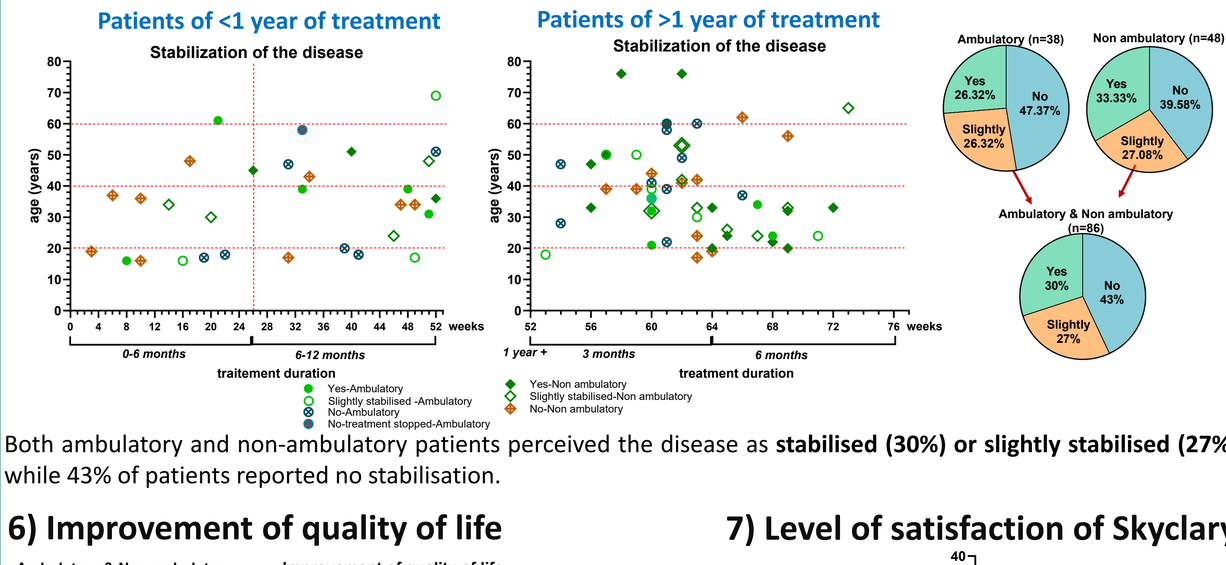
4) Negative effects

Results show the number of ambulatory and non-ambulatory patients who experienced side effects (value in brackets) The duration of treatment after which side effects appear is indicated by a colour code.



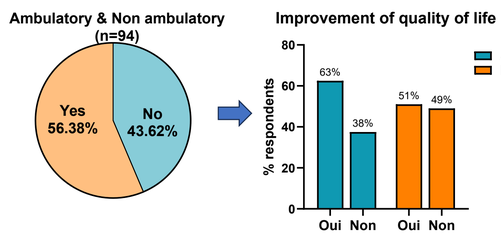
Side effects were mostly reported during the **first 3 months of treatment**

5) Stabilisation of the disease



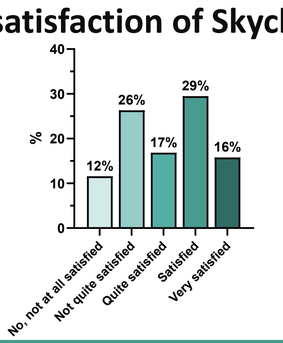
Both ambulatory and non-ambulatory patients perceived the disease as **stabilised (30%) or slightly stabilised (27%)**, while 43% of patients reported no stabilisation.

6) Improvement of quality of life



- Overall, **56%** of patients said that the treatment **improves their quality of life**.
- **>60% expressed satisfaction** with Skyclarys.

7) Level of satisfaction of Skyclarys



CONCLUSIONS

These data revealed expected neurological benefits in terms of coordination and balance, but also additional improvements in major symptoms affecting quality of life, such as improved speech in all patients and reduced fatigue. 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.