

ATAxia

Collecting information to support ataxia research and the HTA process

Emily Cutting

Research Manager

Ataxia UK



28 & 29 October 2025, Amsterdam

**International Patient
Organisation Conference**

Overview

Ataxia UK's work with industry

Surveys to understand the community

Assisting pharma with surveys

Ataxia UK's experience of the HTA process

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Ataxia UK's work with industry

- Regular meetings with Pharma companies to discuss how we can support their work.
- Proactive approach – reaching out to companies with ataxia programmes.
- Publication of a leaflet demonstrating our work with industry.
- Supported by 'Pharma Strategy Advisory Group'.

Ataxia UK's work with industry

- Connect companies with clinicians with interest in running clinical trials.
- Support study recruitment.
- Quarterly newsletter sent to researchers/healthcare professionals.
- Work as a network across Europe – with Euro-ataxia.
- Arrange input on patient information, trial design, consent forms
- Offer support with gathering information from the community (workshops, interviews, surveys...)



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<https://doi.org/10.1186/s13023-021-02091-x>

Orphanet Journal of
Rare Diseases

Health Open Research

RESEARCH

Open Access

The attitude of patients with progressive ataxias towards clinical trials



Gilbert Thomas-Black^{1,2}, Andrada Dumitrascu¹, Hector Garcia-Moreno^{1,2}, Julie Vallortigara¹, Julie Greenfield³, Barry Hunt³, Susan Walther⁴, Mackenzie Wells⁵, David R. Lynch⁵, Hugh Montgomery⁶ and Paola Giunti^{1,2*}

► AMRC Open Res. 2021 Nov 30;3:28. [Version 1] doi: [10.12688/amrcopenres.13036.1](https://doi.org/10.12688/amrcopenres.13036.1)

Symptom burden of people with progressive ataxia, and its wider impact on their friends and relatives: a cross-sectional study

[Anja Lowit](#)^{1,a}, [Julie Greenfield](#)², [Emily Cutting](#)², [Ruby Wallis](#)², [Marios Hadjivassiliou](#)³

Currently working on 'Gene therapy survey for people with genetic ataxias' as part of the TREAT-ARCA project

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Completed by 342 people (FA, inherited CA and idiopathic CA). Published in 2022.

Aim: To understand the attitude of people with progressive ataxia to clinical trials. To define the motivations for and barriers to trial participation.

Method: 29 question survey, distributed by Ataxia UK and FARA.

Questions related to:

Demographics

Personal motivation

Drug therapy

Trial design

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Just under 70% of participants would be prepared to undergo intrathecal drug administration.

The most important symptoms to be addressed by a trial were balance problems and ambulation, although these were superseded by speech problems in wheelchair users.

Reasons for non-participation included concerns about side effects, and the burden and cost of travel. Expense reimbursement was reported to be likely to increase trial engagement.

Drug repurposing trials proved popular. Placebo arms were considered a disincentive.

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Conclusion: This knowledge informs better, more patient focused trial design. This in turn may improve recruitment and retention of participants to future trials

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Surveys to understand the community

Aim: provide preliminary information on the presence and impact of medical symptoms and day-to-day challenges on people with ataxia and their friends and relatives.

Methods: Information was extracted from a survey by Ataxia UK. The views of 366 people with ataxia and 52 friends and relatives are reported.

Health Open Research

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Results: Questions looked at the symptoms of ataxia, but also the **impact** of the symptoms. This study begins to provide information that can be used in further research to explore the needs of people with ataxia and their carers, friends, and relatives.

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Published – can
be accessed
and referenced

We publicise to
researchers and
companies

Shared with the
community so
they see the
benefit of
surveys

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Surveys to understand the community

Currently working on 'Gene therapy survey for people with genetic ataxias' as part of the TREAT-ARCA project

Aiming to gather information on:

- Understanding of genetic therapies
- Concerns around genetic therapies
- Attitudes towards trials of genetic therapies

Future plans:

- Publish the analysed data, so conclusions are accessible and can be referenced
- Create educational tools to increase understanding of genetic therapies

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Assisting pharma with surveys

- In recent years, we have worked with PTC, Biohaven and Biogen on survey/interview-based research studies.
- We help with :
 - Providing patient perspective on the study design
 - Recruitment for the studies
 - Data analysis/publication

UNDERSTANDING THE SPECTRUM OF SCA1, SCA2, SCA3, AND SCA6 THROUGH THE EYES OF PATIENTS: BURDEN OF ILLNESS AND QUALITY OF LIFE

Lauren C Seeberger, MD¹; Melissa Wolfe Beiner, MD²; Michele Potashman, PhD³; Anne Neumann, RN, BSN⁴; Skyler Jackson, BA⁵; Austin R Letcher, MS⁵; Patti A Engel, BSN⁶; Lauren Moore, PhD⁷; Julie Greenfield, PhD⁸; Giovanni Ristori, MD^{9,10}; Laura Heller, PharmD¹¹

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PTC
THERAPEUTICS

PCR84

**The daily life and psychosocial impacts of Friedreich ataxia:
A qualitative study of patient lived experiences**

A Bever, MPH¹; SM Szabo, MSc¹; J Vallortigara, PhD²; P Giunti, MD PhD²; D Lynch, MD PhD³; G Vasco, MD PhD⁴; I Tomazos, PhD MBA⁵

¹Broadstreet HEOR, BC, Canada; ²Ataxia Centre, UCL Queen Square Institute of Neurology, London, UK; ³Division of Neurology, CHOP, PA, USA; ⁴Department of Neurorehabilitation and Robotics, Bambino Gesù Children's Hospital, IRCCS, Italy; ⁵PTC Therapeutics, NJ, USA

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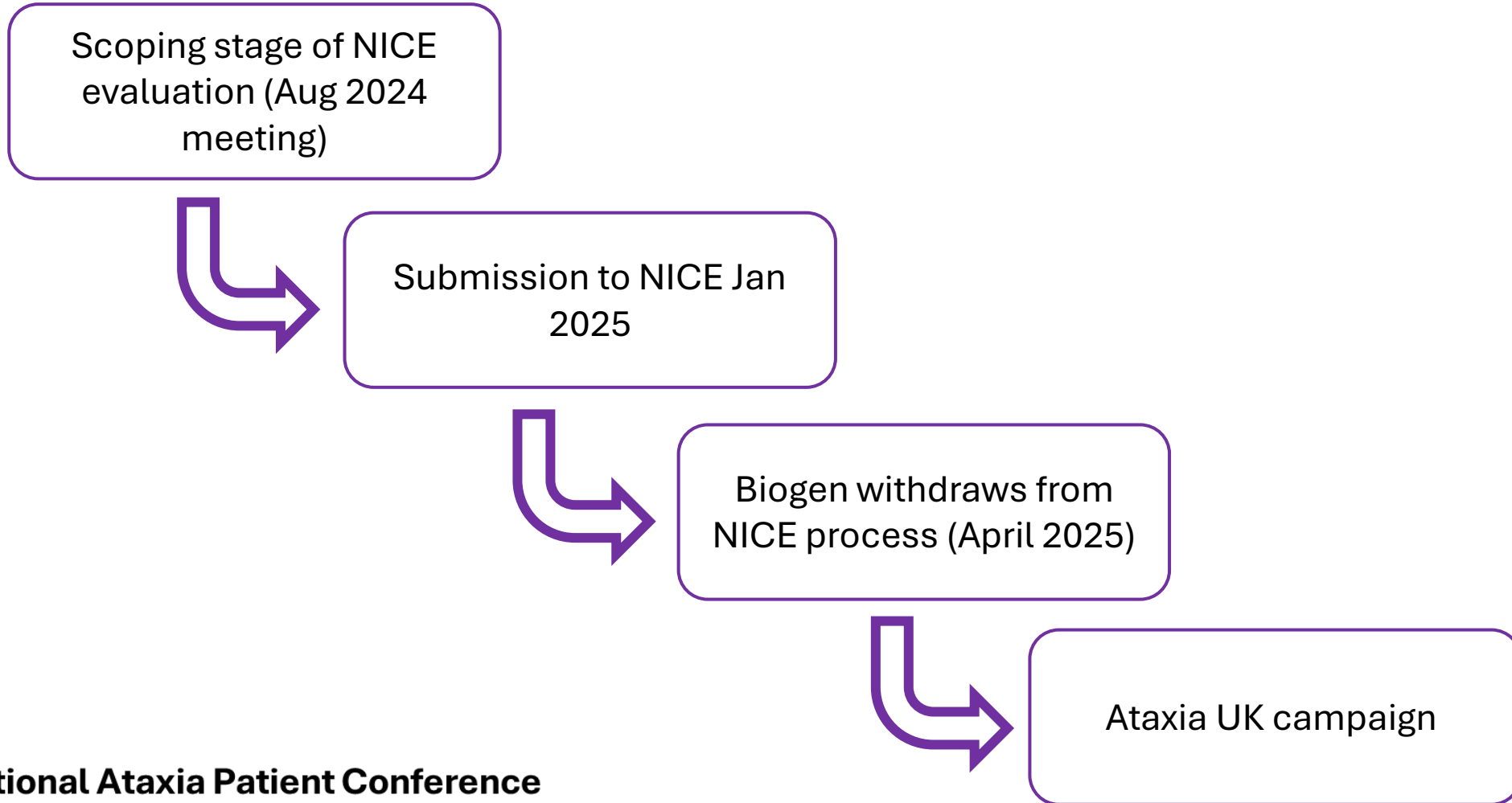
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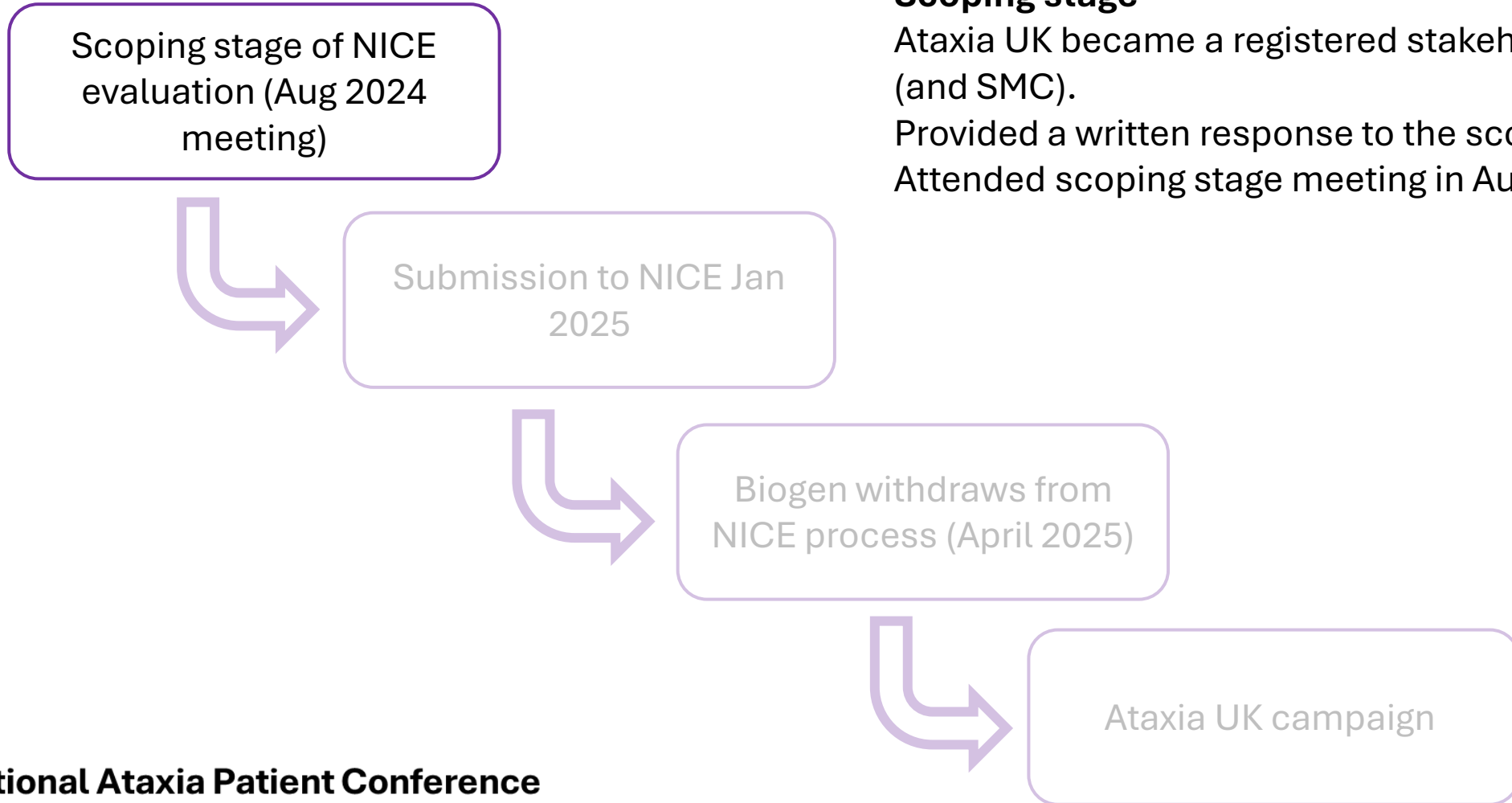
Our experience of the HTA process: **Omaveloxolone**

- Omaveloxolone is approved by the regulators in the UK (MHRA), but is not provided on the National Health Service (NHS).
- In England/Wales/NI, this decision is made by the National Institute for Health and Care Excellence (NICE).
- In Scotland, this decision is made by the Scottish Medicines Consortium (SMC).
- Ataxia UK supports people across the UK.

Omaveloxolone: England, Wales, NI



Omaveloxolone: England, Wales, NI



Scoping stage

Ataxia UK became a registered stakeholder with NICE (and SMC).

Provided a written response to the scoping stage.
Attended scoping stage meeting in Aug 2024.

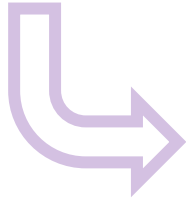
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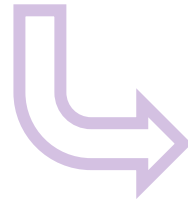


Omaveloxolone: England, Wales, NI

Scoping stage of NICE evaluation (Aug 2024 meeting)



Submission to NICE Jan 2025



Biogen withdraws from NICE process (April 2025)



Ataxia UK campaign

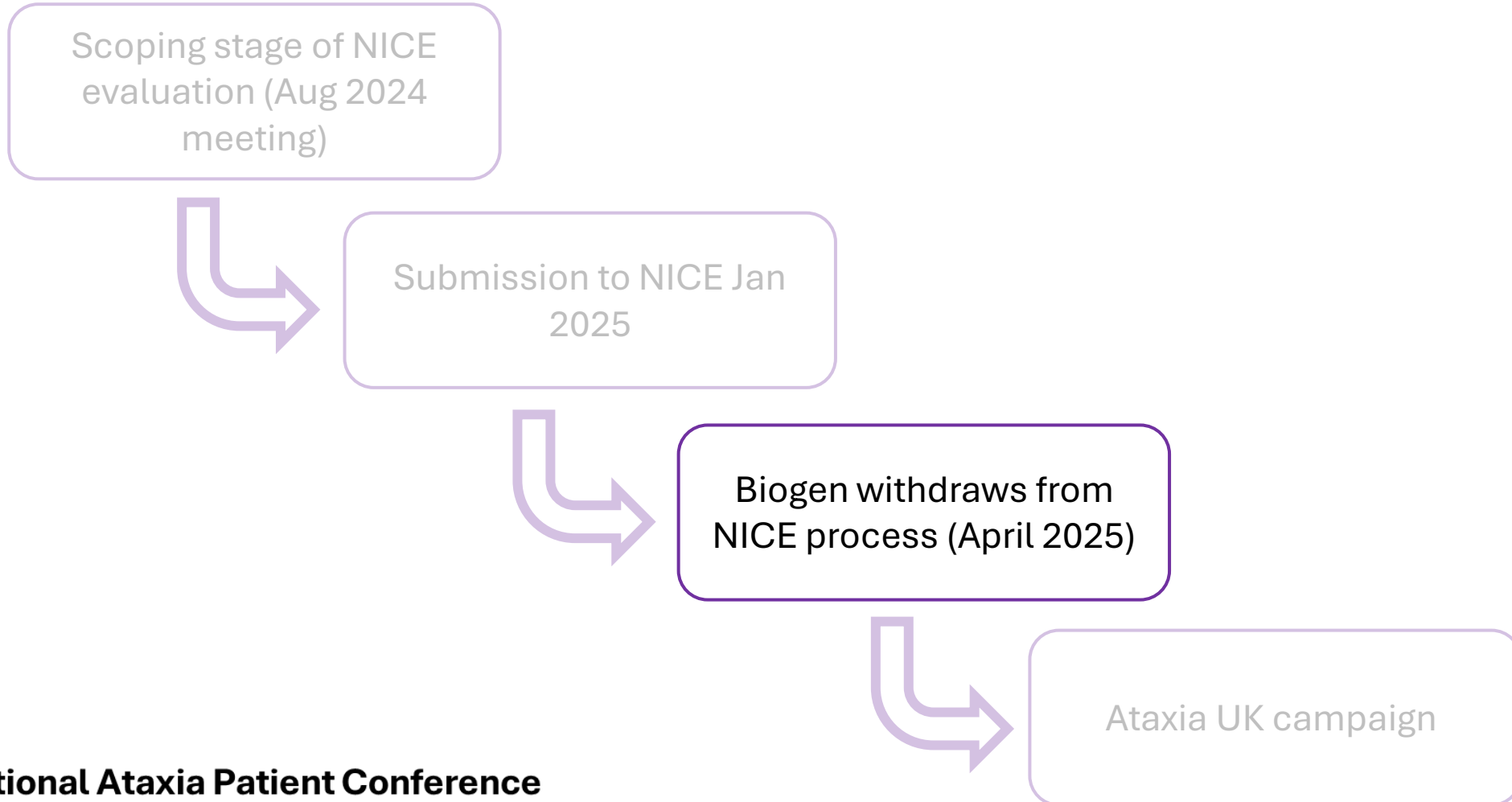
Ataxia UK submission to NICE

Included case studies from people living with FA, and those taking Omav (in the UK this is limited to those on the trial).

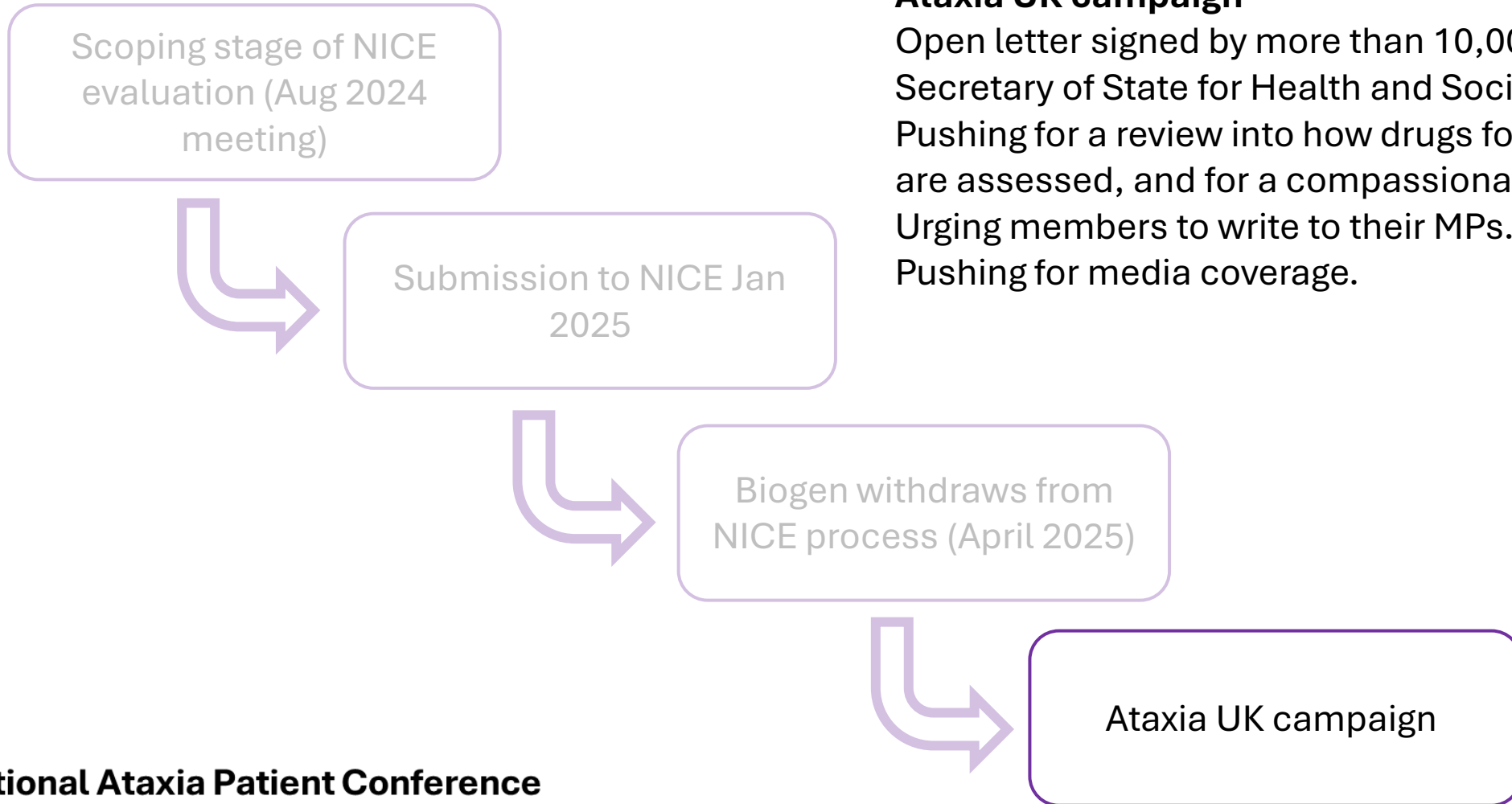
Information from surveys, including the AFAP French survey.

Began preparations for the evaluation stage (when the patient and clinical community would have had the opportunity to highlight the unmet need in FA and the potential of the treatment).

Omaveloxolone: England, Wales, NI



Omaveloxolone: England, Wales, NI



Ataxia UK campaign

Open letter signed by more than 10,000 people, to the Secretary of State for Health and Social Care.
Pushing for a review into how drugs for rare conditions are assessed, and for a compassionate use programme.
Urging members to write to their MPs.
Pushing for media coverage.

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Omaveloxolone: Scotland



Summary

- Not yet successful in the HTA process in the UK
- But we are prepared for future opportunities
- We are pushing for changes to the system in the UK to make access to future medicines easier
- Working with consultants as we learn about this process



Thank you for listening!

www.ataxia.org.uk

Please contact research@ataxia.org.uk if you have any further questions!

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