

# NF1 CAB Report

NF Patients United

March 2026

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## EXECUTIVE SUMMARY

The inaugural Neurofibromatosis Type 1 (NF1) Global Community Advisory Board (CAB) involved patients, caregivers, advocates and industry representatives in a structured, collaborative forum. Over two virtual half-day meetings, held on 28-29 November 2025, participants shared lived experiences and identified gaps in diagnosis, treatment, long-term support, access to reliable and easy to understand information and communication. The CAB aimed for open dialogue between the NF1 community and Alexion AstraZeneca, with the objective of ensuring that future strategies reflect the needs and priorities of those affected by the condition.

Day 1 focused on community-defined priorities. Participants described unmet needs across the spectrum of NF1 manifestations, including high-grade gliomas, malignant peripheral nerve sheath tumours, cutaneous and plexiform neurofibromas, bone manifestations and neurodevelopmental challenges. Many reported fragmented care and a lack of coordinated management for the multisystem nature of NF1. Mental health concerns, stigma, anxiety and the psychosocial consequences of visible and invisible symptoms were described as pervasive. Participants called for structured shared decision-making tools, improved screening and earlier access to therapies, and research on whether MEK inhibitors could address a wider range of NF1 symptoms beyond their current indications.

Day 2 explored the practical realities of accessing care across healthcare systems. Participants described variation in specialist availability, lengthy diagnostic pathways, and difficulties accessing medicines due to regional inequities or reimbursement barriers. Caregivers reported substantial emotional and administrative burdens, while young adults described navigating systems with limited support. The discussion also examined the often-unseen economic burden of NF1, including lost work, reduced educational opportunities, travel expenses, private assessments and the long-term financial impact of chronic care needs. A health-economics study undergoing and led by Alexion AstraZeneca in collaboration with medical and patient community would ideally quantify these impacts, with participants stressing the importance of interim findings and meaningful patient involvement in shaping methods and outcomes.

The participants called for continued collaboration through future CABs dedicated to shared decision-making, health economics and early community involvement in research planning. These next steps were identified as essential to improving care pathways, strengthening the evidence base, and ensuring that NF1 strategies align with the real-world priorities of the community.

## INTRODUCTION

The Neurofibromatosis Type 1 (NF1) Community Advisory Board (CAB) was established to bring together the NF1 patient community for the first time in a structured, collaborative setting. The primary motivation was to create a platform for open exchange, where the community could set its own agenda, share lived experiences and directly influence industry strategies.

This inaugural CAB meeting with Alexion AstraZeneca marked a significant milestone: it was the first time the NF1 community convened in this format, with a clear focus on patient-driven priorities. The meeting intended to foster transparent dialogue, with Day 1 dedicated to topics chosen by the patient community and Day 2 shaped by questions and objectives from Alexion AstraZeneca. This approach allowed both community and industry perspectives to be addressed.

This report tries to capture the discussion from the two 3-hour meetings held virtually on sequential days, on 28-29 November 2025. The discussion, facilitated by Patvocates and supported by Alexion AstraZeneca, brought together patients, parents or direct family of patients, patient advocates, and representatives from Alexion AstraZeneca to map the patient experience and identify critical gaps in care, communication, and treatment pathways. The insights presented here point to areas where systemic improvements are needed to better support those living with NF1.

The objectives of the NF1 CAB meeting were:

- To enable the NF1 patient community to set the agenda and discuss key priorities, challenges, and needs in an open forum.
- To provide a platform for industry partners, specifically Alexion AstraZeneca, to engage with the community, ask targeted questions, and receive direct feedback on topics of mutual interest.
- To maintain privacy, and at the request of the participants, the names of the participants are removed throughout this document.

The two CAB meetings were run on consecutive days, on an online conferencing program, recorded for transcripts, and chat comments were captured in a separate document.

## CAB PARTICIPANTS

A total of 12 patient advocates were present from the following 11 countries (4 continents) to participate in the NF1 CAB: Greece, Austria, Spain, the UK, Sweden, Italy, the Netherlands, Canada, China, Brazil, and Poland. 2 participants were living with NF1 themselves, while 9 participants were parents of NF1 children or other first-degree relatives of people living with NF1, and 1 NF1 patient advocate. An additional patient advocate from NFPU was an observer throughout the CABs. From the Alexion AstraZeneca side, 5 members of the team joined.

## DAY 1: COMMUNITY-DEFINED CHALLENGES IN NF1

### Symptoms that have unmet needs from the patient perspective

The first part of the meeting focused on symptoms that the NF1 community believed should be prioritised for research, clinical attention, and industry engagement. Participants pointed out that clinical priorities often fail to align with the lived burden of disease and that many manifestations remain insufficiently studied despite significant morbidity. Discussions centred on high-grade gliomas, malignant peripheral nerve sheath tumours (MPNST), cutaneous neurofibromas (cNF), plexiform and other benign peripheral nerve sheath tumours, neurodevelopmental difficulties, optic pathway gliomas and low-grade gliomas, bone manifestations, and juvenile myelomonocytic leukaemia (JMML). The community repeatedly reported that the evidence base for these manifestations is limited, fragmented, and lacking in long-term or patient-reported outcomes. One participant highlighted the limited subject-specific knowledge of their clinician, stating, “Our paediatrician... started googling in front of us.” Others explained that they often move between different specialists who treat isolated symptoms rather than the condition as a whole. One participant commented, “When I go to clinic, they treat whatever is in front of them, whether the tumour, the pain, the scan, but the rest of my life is invisible.”

High-grade gliomas and MPNST were repeatedly discussed as areas where mortality remains unacceptably high and therapeutic options are minimal.

Cutaneous neurofibromas (cNF) were described as a lifelong burden that affects not only physical comfort but also identity, employment, intimate relationships, and social participation. One person shared, “You stop wearing short sleeves. You stop going swimming. You make yourself small because the world does not know what it is looking at.” Itching, friction, bleeding, and unpredictable growth patterns were described as common but dismissed symptoms. One person described their cNF as “a thousand tiny reminders that you’re different.” People expressed their worries for their loved one, with one person saying,

***“I hope she will not be severely affected by cutaneous neurofibromas... it can devastate mental health.”***

Plexiform neurofibromas (pNF) were associated with pain, functional impairment and disfigurement, often requiring repeated surgeries with uncertain benefit. One caregiver recalled, “Each surgery bought time, but not much else. You prepare yourself for the next scan before you’ve healed from the last one.” Another person explained that their loved one

***“has a lot of pain... on a scale of one to ten, he has a four every day.”***

Participants described a need for better imaging protocols, earlier access to targeted therapies and more consistent follow-up. Many expressed interest in understand better how, when and the impacts of newer MEK inhibitor options but reported limited access and inconsistent prescribing practices across regions. A participant stated,

***“The drug exists, but not for me. Not in my country, not at my hospital. That’s the reality.”***

Neurodevelopmental challenges, including but not limited to ADHD, autism spectrum disorder (ASD) traits, depression, learning delays, motor difficulties and executive-function problems, were repeatedly mentioned as major yet undervalued contributors to disease burden. One parent explained, “The school battles are relentless.” Another caregiver added, “We pay privately for assessments because nobody in the system connects the dots.” Parents also highlighted the academic struggles their children face. 80% of children with NF1 have deficits in at least one area of their neuropsychological functioning and in many children with NF1 problems like ADHD, autism, learning difficulties, motor functioning or struggling to

control one's temper are the main issues that their families are dealing with. Of their daughter, one parent said, “she struggles with mathematics... that’s the only neurocognitive issue she’s facing.”

Adults reflected on lifelong educational and employment challenges that trace back to childhood, with one participant saying,

***“I wish someone had told my teachers that NF affects the brain too. My whole path could have been different.”***

Another adult patient shared that he had had six jobs in four years, with gaps in employment in between.

Optic pathway gliomas and low-grade gliomas generated concern about unpredictable progression and lifelong visual consequences. One participant commented, “You live scan to scan. Every MRI feels like a verdict.” Bone manifestations, including scoliosis, tibial dysplasia and long-term orthopaedic consequences, were described as a major gap in coordinated care, particularly by the participants present in the CAB who lived with that condition. Participants reported inconsistent assessment protocols and late referral for interventions that could have prevented disability. One participant explained that “we went from different specialists to different specialists and had very mixed experiences.” In contrast, one participant spoke about the role of luck in their diagnostic journey, “We had the lucky break that my son was diagnosed with a neuroblastoma... they did a full body MRI and found the gliomas.”

Participants reported distressing experiences related to clinician communication, particularly around discussions of prognosis. One participant described a profoundly distressing interaction with a clinician: “A young doctor told my wife she should enjoy the time she has left with our daughter... like she was going to die soon.”

Overall, there was a general experience within the CAB members’ experiences that symptom management in NF1 is fragmented, reactive and limited by a lack of specific guidelines. Participants called for multidisciplinary pathways, internationally harmonised practices, greater clinician awareness, and improved access to psychosocial and rehabilitation services. A summative comment from one participant captured the general sentiment: “NF1 is like twenty conditions happening at once. We need a system that sees the whole picture.”

Although MEK inhibition has demonstrated clinical benefit in some NF1 manifestations, the community expressed a strong desire for evidence on its potential to alleviate a wider spectrum of symptoms. Given the absence of effective treatments for many of the concerns raised, there is a clear imperative to investigate how MEK inhibitors, and future combination approaches, might modify disease trajectories, reduce cumulative symptom burden, and improve long-term functioning across the lifespan. Participants repeatedly voiced the hope that targeted therapies could address not only tumour growth but also the everyday realities of living with NF1. A coordinated research effort is therefore urgently needed to determine how MEK inhibition could meaningfully improve outcomes in the areas that matter most to the NF1 community.

## Shared decision-making tool for NF1

The discussion on shared decision-making (SDM) was in response to the need for a structured, patient-friendly resource to support individuals and families as they navigate complex and recurrent healthcare decisions across the lifespan. SDM is a priority for all age groups, noting that NF1 decisions often involve balancing benefits, risks, uncertainty and personal circumstances.

Participants broadly agreed that they often made decisions with insufficient information, limited context, or without understanding all available options. One participant described clinical discussions as “fast, technical, and overwhelming, like trying to negotiate in a language you don’t speak.” Another participant described the effects of insufficient information on their mental well-being, “I just need the answer. I don’t want to be in the air all the time, which is really damaging my mental health.” In one instance, a participant explained how their clinicians worried regarding their unwillingness to provide additional information or context,

***“Doctors sometimes say... don’t worry about it. Just forget about it... and that’s so dangerous.”***

Regarding coordination and treatment plan, participants expressed the burden placed on them to gather information regarding next steps, “It’s a lot of us chasing and calling... because we don’t really know what the plan is.” Another participant confirmed this by stating, “We don’t really know what the plan is... that coordination lands on us.”

The group discussed the need for a tool that would allow patients to prepare for appointments, clarify what matters most to them, and track choices over time. Many noted that NF1 decisions typically occur not as a single event, but as a series of decisions linked to tumour progression, school transitions, surgery timing, medication options and surveillance planning. One participant observed, “Every choice has a domino effect. An SDM tool would help you see the whole chain, not just the next step.”

Several participants described scenarios in which clinicians presented only one option, often reflecting institutional practice rather than patient preference. One parent recalled, “We were told ‘this is what we do here,’ not ‘what matters to you?’” Another participant commented, “Shared decision-making should feel shared. Right now, it often feels delegated, not shared.”

Transition from paediatric to adult care was repeatedly described as a vulnerable point where SDM could provide continuity. Adolescents often feel unprepared for the shift in responsibility, and parents struggle to adapt to a reduced decision-making role. Participants proposed that an effective SDM tool should incorporate symptom-specific modules, descriptions of common clinical pathways, explanations of uncertainty, and reflective questions about values, goals and lifestyle constraints. They suggested that it should include sections for school and workplace adaptations, fertility considerations, reproductive counselling, and psychological well-being. Several participants supported a digital format, with one saying, “We carry our phones everywhere. The tool should be right there with us.”

Multilingual versions were brought up as important. A participant from a non-European country noted, “Our healthcare system works differently. A tool that assumes Europe’s structure would not help me navigate mine.” Overall, the group saw SDM as a route to improving autonomy, reducing decisional regret, building trust, and ensuring that patient values shape care. One participant concluded, “Good decisions are personal. A shared decision-making tool would finally acknowledge that.”

Two examples of SDM tools were provided. The World Federation of Hemophilia is a global NGO and has generated an online tool supported by videos intended to support patients with this chronic lifelong condition: <https://sdm.wfh.org/>. Another example is the 7 workbooks for different situations, made by the International Kidney Cancer Coalition: <https://ikcc.org/kidney-cancer-decision-aid-my-treatment-my-choice/>. Both examples offer potential solutions for consideration when considering an SDM tool for NF1.

## Mental Health and Wellbeing

The final discussion focused on mental health and well-being, which participants described as central to the NF1 experience yet poorly integrated into routine care. One of the Spanish participants presented evidence of increased rates of depression, anxiety, insomnia, panic symptoms and social withdrawal among people living with NF1. Participants' testimonies strongly aligned with this evidence. Many explained that psychological burden accumulates across childhood, adolescence and adulthood, often intensifying during key life transitions.

Body image concerns and experiences of stigma were recurring themes. Participants spoke openly about how visible tumours affect self-esteem, identity and social interactions. Parents described the emotional toll on children who face bullying or exclusion at school. One parent said, "My daughter hated to change into sports clothes on PE days because when she was in the clinical trial, she had sores and red spots. That is a childhood being shaped." Participants described significant challenges for their children, particularly about peer relationships and acceptance. One participant expressed concern about their child, "His social environment... finding new friends... will be difficult for him."

A consistent concern was that clinicians rarely ask about mental health unless the patient raises it explicitly. One adult participant noted, "They ask about pain, vision, tumours. Nobody asks how you're coping." Several described feeling dismissed when reporting anxiety, as if psychological symptoms were secondary or unrelated to NF1. Anxiety is a prevalent topic for many participants. One person said, "They tell you it's normal to be anxious. But living in fear of the next scan is not normal. It's exhausting." Another noted that, "The biggest challenge... has been the feeling of anxiety... given the nature of this disease." The emotional burden and anxious state were addressed by another participant, "It's the emotional burden too... that constant anxiety and waiting for the other shoe to drop."

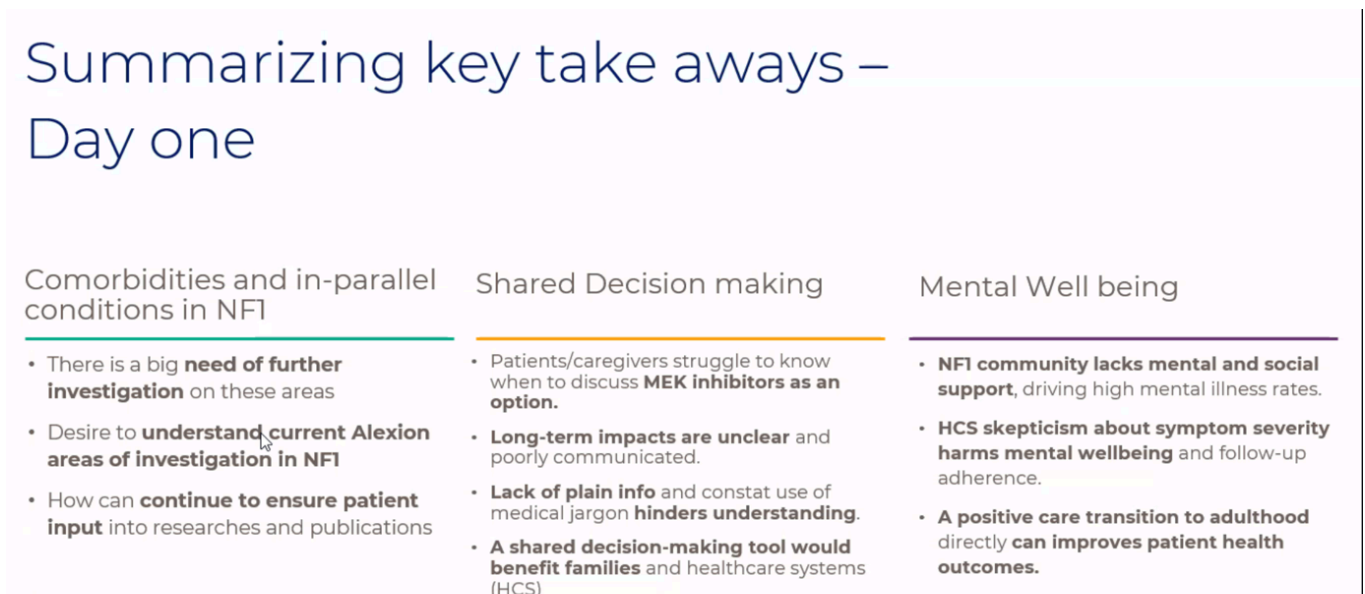
Access to mental health care varied widely across countries. Long waiting lists, limited insurance coverage and scarce NF1-informed specialists were recurring barriers. One caregiver shared, "We found a therapist who understood chronic illness, but we had to drive far and pay privately." Another participant remarked, "Support groups do more for my mental health than any formal service." Participants also described challenges related to communication and social interaction, "Interaction with other people was a big problem... he lacked a lot of language." Another participant highlighted how fear of being singled out acted as a barrier to support: "She doesn't want to be pointed out... she was refusing any type of strategy or care."

The group also discussed the economic dimensions of psychological burden. Difficulty maintaining stable employment, reduced educational attainment, and the cumulative financial impact of care were described as major stressors. One participant observed, “It’s not just the illness; it’s the cost of living with the illness.” Parents described challenges balancing employment with caregiving responsibilities, hospital visits and advocacy at school.

Participants suggested that mental health should be embedded in NF1 care pathways, with routine screening, referral to specialists, and access to psycho-oncology, family counselling and peer support. Many supported the idea of integrating mental health modules into any SDM tool developed for NF1. One participant summarised the collective experience: “NF1 affects the body, the brain, and the heart. A system that only treats the body is not treating the person.”

Another participant concluded, “The biggest misconception is that NF1 is only a tumour condition. For many of us, the emotional battles are the ones we fight every day.”

Figure from Alexion AstraZeneca summarising Day 1.



## DAY 2: BARRIERS AND REALITIES IN NF1 CARE

### Caring for someone with NF1

Participants were asked by Alexion AstraZeneca what they expect when trying to access high-quality care for NF1. Many described an expectation of coordinated multidisciplinary services, clear pathways and reliable specialist input, although this was not the reality in several countries. In Greece, one participant explained that “there is only one NF clinic, which is for paediatric patients and is in Athens,” although a private hospital team continues care into adulthood. Access to new medicines requires approval from the national regulatory authority, with an obligation for an MRI every three months to renew access. This requirement was described as burdensome and a barrier to continuity of care.

The Canadian participant described geographical inequity as a significant barrier. They explained that while large cities such as Toronto, Vancouver, or Montreal provide access to specialised NF clinics, remote and rural areas experience extremely limited availability. Several participants highlighted the burden of travel for care and associated travel costs. One participant noted, “Patients in rural areas might have to travel hours... which adds to costs,” while another explained, “This is a huge problem... if we move away from Athens, we have to journey to the capital for care.”

Even in major cities without NF clinics, such as Winnipeg, patients struggle to access coordinated care. This led to a discussion of a virtual NF clinic model that could link local providers with NF specialists to reduce the need for long-distance travel. Another Canadian participant described a lengthy process to secure reimbursement for selumetinib in Manitoba, noting that their organisation had to write multiple letters to the government before it was added to the formulary, even though the national rare disease strategy identified such drugs as a priority. They summarised the experience by stating that “it took longer than it should have”.

Participants from Spain and Italy described similar regional disparities. Those living near designated reference centres reported good access to specialists and structured multidisciplinary care, while those in other regions encountered long delays, limited diagnostic expertise and inconsistent access to treatment. A participant from Spain described “difficulty to access specialist care depending on the geographical area” and added that “patients with invisible symptoms, like pain, itching or cognitive problems, are often not taken seriously” when they attend non-specialist centres. Another noted that once within the specialist centre, “we had great access to our team and often they discussed with us the different options” for management.

Expectations for good care included clarity, responsiveness and a proactive approach from clinicians. A UK participant noted that young people in their organisation are often told to return only when symptoms worsen, despite already experiencing daily pain or anxiety. They described this as an approach that “doesn’t align with the fact that NF1 is a lifelong condition that requires ongoing monitoring, not reactive care”. Expectations for quality care included reliable diagnosis, continuity across the lifespan, consistent clinical information, and access to appropriate medicines without excessive administrative burden.

## Expectations for accessing the best care

Participants were asked what they expect when trying to access high-quality care for NF1. Many described an expectation of coordinated multidisciplinary services, clear pathways and reliable specialist input, although this was not the reality in several countries. In Greece, one participant explained that “there is only one NF clinic, which is for paediatric patients and is in Athens,” although a private hospital team continues care into adulthood. Access to new medicines requires approval from the national regulatory authority, with an obligation for an MRI every three months to renew access. This requirement was described as burdensome and a barrier to continuity of care.

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The Dutch participant, who supports a grown son with NF1, shared that even though there is an expertise centre easily accessible to them, and that their country-level care has been well organised, they have concerns about the next stage, as they grow too old to support their son. They still attend medical appointments with him, and he is not fully independent, despite assisted living arrangements, and with employment. There is little clarity about this next life junction. Expectations for good care included clarity, responsiveness and a proactive approach from clinicians.

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Across all countries, expectations for quality care included reliable diagnosis, continuity across the lifespan, consistent clinical information, and access to appropriate medicines without excessive administrative burden.

## Biggest concerns

Participants were invited to describe their most significant concerns in relation to NF1 care. Several referred to the psychological and social impact of the condition. One parent described their concern that their child’s isolation was worsening and felt strongly that schools need to play a more active role in promoting inclusion. Others referred to the stigma associated with visible tumours, learning difficulties or autism traits, and expressed frustration that public understanding of NF1 remains limited.

Participants also expressed ongoing anxiety about their child’s progression, particularly when moving to a new area. One participant shared, “My biggest concern... is his new life away from Athens... We hope that his problems will be minor ones,” reflecting worries about the future. Others described fears that aspects of the condition were being

overlooked, with one stating, “What I worry about is there’s just something that they’re missing.”

A frequent concern was inconsistency in clinical information, with one participant stating,

***“There’s not enough reliable information... we’re all desperate for it.”***

One participant from Austria noted that patient organisations often step in to “gather trusted information on NF and provide that to the patient community through all available channels,” and stressed the need for information to be co-developed with specialists so that it remains accurate and accessible. They described attending scientific meetings and reviewing literature to stay up to date and encouraged patient communities to identify their own knowledge gaps so that these can be addressed through structured education initiatives. Participants also addressed gaps in patient awareness regarding information sharing within the clinical setting, noting, “Patients don’t know they can self-report side effects... and few people are aware.”

Concerns regarding access to medicines were raised repeatedly. Participants from Brazil explained that even though the national regulatory authority had approved selumetinib, the health technology assessment process had not recommended reimbursement. One person described the resulting experience as “a stressful pathway to get access,” requiring significant advocacy and time. Chinese participants described disparities between regions and referred to a “doctor-centred system” that sometimes fails to recognise patient needs. They requested greater global coordination and more support for patient organisations, which they believed could help mitigate structural barriers in healthcare systems.

Participants also discussed the challenge of evaluating new therapies. Several asked how patient voices could be integrated into evidence packages and reimbursement discussions. One participant said, “Equipping us is not the same as influencing us,” in response to questions about industry engagement, and clarified that providing abstracts and scientific information is welcome if it builds capacity without directing advocacy outcomes. Another participant added that “patient experts and patient advocates are not lay patients” and therefore should be engaged respectfully and transparently in research discussions.

## Country realities

The final part of the discussion explored how NF1 care differs across countries. Participants described varied experiences shaped by national health systems, reimbursement policies, specialist availability and regional resources. Some of the most remarkable experiences were described as:

**Greece:** Greek participants described fragmented services and noted that NF events, conferences and specialist talks are rare and usually held only in Athens. Many families seek information online, including from patient organisations in other countries. They described the need for multilingual patient newsletters and materials that can reach people outside major cities. A participant noted that “patients and caregivers in Greece search for answers on websites, not only by asking other caregivers and patients from Greece but also from other countries,” due to a lack of regular local educational events.

**Spain:** Two Spanish participants described access to a national reference centre, but inadequate pathways for those living elsewhere. They also reported difficulties in early diagnosis, limited knowledge among general paediatricians, and substantial information gaps for families. The Italian participant described similar patterns, with well-regarded centres of excellence but notable territorial disparities. In both settings, the role of patient organisations was regarded as essential for bridging gaps.

**Canada:** The Canadian participant described a federal system where rare disease drug access varies by province. While some provinces provide rapid access, others require lengthy advocacy efforts for reimbursement. They described the need for virtual care solutions to overcome geographical barriers and allow remote patients to access NF specialists without extensive travel.

**China:** The participant from China referred to the rapid introduction of new medicines and supportive reimbursement policies in some regions but described disparities between provinces. They raised concerns about the doctor-centred model, saying that “China is a doctor-centered system” and they expressed the need for approaches that incorporate patient priorities. A participant described interest in more support from industry for patient organisations and registries, which could strengthen evidence generation and inform clinical practice.

**Brazil:** The Brazilian participant described challenges due to inconsistent reimbursement decisions and noted that access to NF medicines may require legal proceedings or individual petitions. They referred to a widely used NF textbook by a Brazilian specialist, which serves as a key informational resource for families seeking guidance.

Across countries, participants identified shared difficulties in access, continuity of care, public awareness and information quality. Several expressed the desire for international collaboration to harmonise standards and reduce regional disparities. One participant summarised the situation by stating that “it is all connected: social care, healthcare, research and education, public education” and argued that better coordination is necessary to improve outcomes for people living with NF1.

## DISCUSSION

The inaugural NF1 Community Advisory Board created a structured environment for patients, caregivers and stakeholders to examine the realities of living with NF1 and to identify priorities for improved care, communication and research. Discussions across both days revealed persistent gaps in symptom management, psychosocial support, and access to coordinated services. Participants described a wide range of unmet needs, including the burden of tumours, pain, visible disfigurement, neurodevelopmental challenges and the cumulative psychological effects of living with a lifelong condition. These experiences were often compounded by fragmented services, inconsistent information and limited specialist availability.

Shared decision-making emerged as a central requirement to improve the care experience. Participants described uncertainty when navigating options, inconsistent explanations from clinicians and difficulty balancing risks and benefits in the context of everyday life. A need for structured, accessible resources was expressed repeatedly. Integrating quality-of-life data into these discussions would support decision processes that reflect the lived experience of NF1 and ensure that outcomes valued by patients are given appropriate weight. A future CAB dedicated to decision-making could examine how quality-of-life measures are selected, communicated and used in clinical consultations, and how tools could support families and adults across different stages of the care pathway.

Mental health remained a pervasive concern. Participants described isolation, stigma, bullying and persistent anxiety related to disease progression. Psychological support was inconsistent across countries and often accessible only to those with personal resources. Integrating mental health services within NF1 care pathways would help address a major driver of reduced quality of life and could support adherence, coping and long-term wellbeing.

The discussions on access and country realities showed substantial variation in care structures, specialist availability and reimbursement decisions. Participants described geographical inequities, administrative barriers and slow access to medicines. Burden on

families includes travel, lost work, private assessments, assistive therapies and the long-term financial implications of educational disadvantage. A follow-up CAB focused on capturing these hidden burdens could support more systematic recognition and hone the modelling of health economics impact of NF1 with potential to inform planning, reimbursement decisions and policy development.

Participants were clear that future therapeutic research must involve the community at an early stage. They described interest in a broader evaluation of MEK inhibitors, including their impact on cutaneous tumours, pain, neurodevelopmental outcomes and functional abilities. Early engagement in research planning would allow community members to contribute their priorities and perspectives at the outset, shaping studies that address outcomes that matter to them. A dedicated research-focused CAB could support co-design of studies, develop approaches for meaningful patient evidence and ensure that NF1 research reflects the heterogeneity of the condition.

In addition to the medical aspects discussed, the CAB brought to light the need to capture the full economic burden of NF1, including indirect and often overlooked impacts on individuals, families, and society. The discussion made clear that existing assessments rarely account for the lived reality of people with NF1. One participant noted that traditional models “only consider the direct cost of a new treatment or a new intervention, and sometimes some of the related costs, but what is very often not taken into account is all the other indirect costs”.

These indirect elements were described as substantial, including lost income, lower education consumption, reduced career trajectories, time spent caring, and the high emotional load placed on households. One participant explained that “some of you had to give up jobs to reconsider your careers as caregivers,” adding that “many career pathways may be completely precluded to you” if you live with NF1. Another expanded on this by noting the societal cost of diminished participation in work and education when NF1 limits functional ability or when carers must reduce their availability: “the ability to contribute to society is significantly reduced”.

The meeting also identified the absence of data infrastructures necessary to quantify these hidden burdens. Participants cited “a lack of registries,” “lack of patient portals,” and limited real-world data, which together restrict the ability to conduct comprehensive evaluations of economic and social impact. The group agreed that patient involvement is critical in shaping such research, as patients are “in a best position to tell what those more indirect costs or the cost of lost opportunities could be,” and that evidence generated jointly “could be brought to the attention of decision makers” to develop a more accurate understanding of NF1’s total impact.

During the discussion, Alexion AstraZeneca representatives confirmed that a dedicated health-economics study is already underway. This project aims to “gather quality of

life of adolescents and adult patients living with NF1 and caregivers,” though it was noted that the study “will take time” and is expected to run for one to two years. Participants expressed support for producing interim milestones to keep the community informed and to ensure that findings remain relevant to advocacy efforts.

The session concluded with agreement that future CABs should be pursued, with three priority topics identified for further exploration.

1. Development of a Shared Decision-Making tool. A CAB dedicated to this topic could include defining the information needs of the community, identifying the key moments in the care pathway where such a tool would be used, and determining the type of evidence, language, and format (e.g. online vs printable) required to support meaningful patient-clinician discussions.
2. Understanding the impact of hidden burdens. Participants called for a structured examination of the often invisible personal, social, and economic pressures associated with NF1, with the intention of ensuring that these factors are incorporated into health economics modelling to better reflect the true societal burden of the condition.
3. Strengthening research collaboration. There was strong support for involving the community early in research planning so that priorities, lived experiences and meaningful outcomes shape the design and direction of future studies. Participants described the need for a value-based partnership that moves beyond transactional engagement towards sustained and collaborative involvement in research activities.

## **ACKNOWLEDGEMENTS**

This report was made possible through the contributions of patients and carers, who generously shared their experiences and insights. We extend our deepest gratitude to all participants for their candid discussions concerning both the challenges and opportunities for improving care in NF1.

We would like to acknowledge Patvocates for their role in facilitating the discussions, as well as Alexion AstraZeneca for supporting this initiative.

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