NF1 CAB Report

NF Patients United

March 2026

EXECUTIVE SUMMARY OF NF1-CAB TALK WITH SPRINGWORKS

The NF1 Community Advisory Board ("NF1-CAB") meeting held on 5 December 2025, with representatives of SpringWorks Therapeutics ("SpringWorks") focused on discussing NF1-related symptoms and shared decision-making processes by NF1-CAB participants. A total of 6 NF1-CAB members participated from the following countries to participate in the NF1 CAB: Austria, Greece, Poland, Spain, and the UK.

NF1-CAB members shared personal experiences and research findings on various NF1 symptoms, including high-grade gliomas, MPNSTs, cutaneous nerve fibromas, and neurodevelopmental effects, while highlighting the need for more research and effective treatments. The conversation ended with members sharing their perspectives on healthcare challenges across Europe, including access to care, multidisciplinary treatment approaches, and the importance of shared decision-making in managing this complex condition.

INTRODUCTION

NF1-CAB meeting began with introductions and updates. The meeting focused on discussing NF1-related symptoms and shared decision-making processes. The participants emphasized the importance of collaboration between industry, patient organisations, and academia.

The meeting began with introductions where both parties expressed their gratitude for the opportunity to have this talk and the hope for a collaborative future. The NF1-CAB team discussed the importance of patient input across the drug development cycle and expressed optimism about future face-to-face meetings. The agenda focused on NF1-related symptoms, with NF1-CAB members presenting on high-grade gliomas, followed by discussions on other symptoms such as MPNSTs, continuous neurofibromas, and neurodevelopmental issues. The meeting aimed to prioritise these symptoms in research and care, building on insights from the EU Pearl project.

TOPIC 1 – NF1-RELATED SYMPTOMS THAT SHOULD BE PRIORITISED FROM THE PATIENT'S PERSPECTIVE

NF1-CAB members shared their perspectives on NF-related symptoms and their impact on the daily lives of patients and caregivers. Among top ranked symptoms the following were discussed with the view of prioritising them in clinical trials: high-grade gliomas, MPNST, cutaneous neurofibromas, benign peripheral sheath tumours, bone manifestations.

NF1-CAB member from Spain discussed high-grade gliomas in neurofibromatosis type 1 that affect approximately 1-2% of individuals with NF1, noting their aggressive nature, poor prognosis, and the challenges in monitoring patients during transitions between pediatric and adult care. They are also a cause of high psychological and family distress.

NF1 CAB Chair continued the presentation on malignant peripheral nerve sheath tumors (MPNSTs) in NF1 patients, highlighting their prevalence, symptoms, and the limited effectiveness of current treatments, while emphasizing the importance of early detection and surgical resection. He mentioned that the typical age group is around 20 to 40 years old and this condition is less typical for children. In addition to extensive clinical burden, MPNSTs also reduce quality of life due to related anxiety, depression and physical limitations.

NF1 CAB member from Poland concluded by addressing cutaneous neurofibromas (CNs) – skin lesions very typical for NF1 that affect 60-80% of NF1 population. The burden of CNs increases with age and accelerates during puberty and pregnancy. Increased psychological burden and reduced quality of life are also associated with CNs. There is the need for non-surgical treatment options, and ongoing research, as there are no proven medical therapies yet.

NF1 CAB member from Greece discussed plexiform neurofibromas (PNs) that develop in more than 50% of patients with NF1 and present serious complications affecting tissues that cover and protect nerves. PNs have a severe impact on patients, including vital organs, pain, mobility issues, and risk of malignant transformation. The most usual treatments include surgery, systemic therapy and pain management. She emphasized the need for regular monitoring

NF1 CAB members from Spain addressed the neurodevelopmental effects of NF1, noting that these symptoms affect the entire NF1 population, with variable impacts on quality of life, employment, and cognitive development. They typically begin in childhood and persist throughout life. He emphasized the urgent need for screening tools to identify NF1-related neurocognitive issues as well as guidelines for holistic treatment, and called for more research into biomarkers, early identification tools, and interventions to mitigate these effects.

NF1 CAB Chair discussed optic pathway gliomas and other low-grade gliomas, which affect 20% of young children, with optic pathway gliomas more prevalent among female patients. Optic pathway gliomas may cause visual impairments and even blindness, while low-grade gliomas, depending on their location, can cause headache, seizure, weakness, and other neurological impairments. Annual ophthalmological assessments and MRIs are recommended for monitoring, with systemic chemotherapies used for treatment.

NF1 CAB member from Poland covered the topic of bone manifestations in NF1 which are numerous and include ligament disorders, bone dysplasia, impaired bone healing, osteopenia, osteoporosis, and some other abnormalities. These conditions may progress with age and become a dominant problem around school age and puberty. Specific rehabilitation and developmental support are needed including such procedures as surgery, external or internal stabilization of the spine.

He also presented the topic of juvenile myelomonocytic leukemia (JMML) – a rare disease which accounts for approximately 1-2% of all paediatric leukemias and is more aggressive in NF1 patients. It is usually diagnosed around the age of 2 to 4 but can sometimes be present in older children. HSCT (Hematopoietic Stem Cell Transplantation) is now the best available treatment for JMML.

The meeting then focused on the challenges faced by NF1 patients, highlighting the devastating impact of the condition and the lack of effective treatments for many symptoms. NF1 CAB Chair shared a personal experience of dealing with a child with NF1 who died from JMML despite receiving experimental treatment. The group discussed the need for more research, potential treatments for NF1-related symptoms.

He also called for more inclusion of their members in future research activities, planning of design of clinical trials as well as raising awareness in the patient community. The ultimate goal is to develop preventive therapies to alleviate the burden of NF1 on patients and their families.

The group discussed the challenges faced by individuals with NF1, including its unpredictable progression, stigma, and impact on mental health and quality of life. NF1 CAB members shared personal experiences of living with NF1, highlighting issues such as limited access to care, school absences, and social isolation. The discussion emphasized the importance of multidisciplinary care and case management for patients and families, with plans to continue the conversation after a break. It also focused on discussing NF-related symptoms and the importance of patient involvement in clinical trials.

SpringWorks representatives expressed their gratitude for the valuable insights shared. NF1 CAB Chair highlighted the need for a holistic approach to addressing NF, including medical and psychosocial aspects, and suggested future collaborations to prioritize and align goals between patient communities and industry. It was unanimously agreed on the necessity of quantifying the psychosocial burden of NF to develop appropriate support programs.

TOPIC 2 – SHARED DECISION-MAKING IN NF1 MANAGEMENT

The meeting focused on shared decision-making in neurofibromatosis type 1, emphasizing its importance in managing this complex, lifelong condition. Participants discussed the need to consider various aspects, including scientific evidence, clinician expertise, patient values, and life goals, when making medical decisions. They highlighted the challenges of NF1, such as its unpredictable nature, multiple manifestations, and the need for tailored treatment plans. The benefits of shared decision-making were outlined, including building trust, improving treatment satisfaction, and potentially increasing adherence to treatments. The lack of specific research on shared decision-making in NF1 was noted, along with the need for evidence-based tools to support patient involvement in decision-making processes.

Specific examples were shared illustrating how shared decision-making can improve various aspects of care, including treatment adherence. One example concerned decisions on the frequency of MRI monitoring. While clinicians may recommend less frequent MRIs in the belief that this is less emotionally burdensome for parents, in some cases parents may feel more reassured by more regular imaging, as it allows them to better understand whether the condition has progressed over time. In such situations, parents may therefore prefer more frequent MRIs than those initially recommended by clinicians.

NF1 CAB Chair discussed the importance of making healthcare tools available in multiple languages and highlighted a web-based service as a best practice for patient-centered care. He emphasized the need for a structured patient pathway for NF1 patients, advocating for multidisciplinary care and addressing various aspects such as learning difficulties, ADHD, ASD, and mental health issues. He also shared Austria's experience in developing a holistic care plan for NF1 patients, which has been successful in standardizing and improving the quality of care. He also stressed the significance of shared decision-making in NF1 treatment, involving patients, families, and healthcare providers in the process to ensure personalized care plans.

NF1 CAB member from Spain shared his personal experiences with bullying and job instability, highlighting the long-term effects of neurofibromatosis on her career and mental health. NF1 CAB member from Greece discussed her son’s struggles with school-related psychological burdens, including bullying and lack of teacher support, which led to emotional distress and psychosomatic symptoms. Despite these challenges, he managed to overcome his issues with psychotherapy and continues to pursue his studies with determination and curiosity, aiming to focus on informatics and computer science at university.

The group discussed the importance of care pathways and shared decision-making in NF1 treatment, highlighting the need for digital tools to support patient education and data collection. They emphasized the challenges of multidisciplinary care, particularly during the transition from pediatric to adult healthcare, with two NF1 CAB members from Spain sharing their experiences of navigating different healthcare systems and specialist knowledge gaps. The discussion concluded with a call for better access to information about NF1 experts and specialist centers across Europe, with a suggestion to develop a map identifying the appropriate centers for patients.

The discussion focused on challenges in NF1 care across Europe, particularly the lack of adult specialist clinics and the difficulties patients face accessing treatment and care. NF1 CAB member from Greece highlighted Greece's situation where adult NF1 patients must travel to major cities for care, while NF1 CAB member from Poland shared Poland's progress in establishing coordinated care centers for patients up to age 30.

The meeting focused on healthcare challenges faced by NF1 patients across Europe, with participants sharing experiences from different countries. NF1 CAB Chair highlighted the need for patient portals and data sharing, while NF1 CAB member from Spain discussed Spain's portal system and NF1 CAB member from Greece described financial burdens in Greece.

The group agreed to continue discussions on potential collaborations and to share meeting materials, including a non-confidential report of this talk.

CONTACT US:

Global Network of NF Patient Organisations
info@nf-patients.eu
www.nf-patients.eu