Leveraging patient voice and AI tools in cross-country mapping of quality of life and unmet needs in NF1

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OBJECTIVES

The primary objective of this research was to highlight common trends, and identify the distinctive needs of patients with NF1 and their caregivers across multiple countries. Specifically, we aimed to:

- Understand the impact of NF1 on the QoL of patients and caregivers
- Identify the fears and unmet needs of patients with NF1 and their caregivers
- Investigate barriers to treatment and access to healthcare
- Utilize dSML to capture authentic patient voices and insights, providing a more accurate and comprehensive understanding of their experiences
- Conduct a comparative cross-country analysis to explore differences and similarities in NF1 experiences and needs globally

CONCLUSIONS

- The use of AI technologies, specifically dSML, has provided valuable insights into the problems, fears and challenges faced by patients with NF1 and their caregivers worldwide
- Our findings indicate substantial differences in the impact of NF1 on the QoL across various countries
  - For example, physical pain is a major concern in Mexico, while psycho-emotional issues are more prevalent in Argentina
  - Informational unmet needs and barriers to treatment also vary
  - In Sweden and Malaysia, patients tend to ask more questions about symptoms, while in South Korea, financial issues bother patients the most
  - The analysis of barriers to treatment and diagnostics has shown similar patterns in Sweden, South Korea and Mexico: top barriers relate to access to treatment and lack of awareness about neurofibromatosis and treatment options
- Fear of disease progression is a common concern, except for in Romania in which specific fears such as bullying and academic performance were notable among caregivers
- The disparities in concerns and unmet needs highlight the importance of tailored approaches to support and care for patients with NF1 in different regions
- These insights can help healthcare providers, policymakers, and support organizations improve patient management, ensuring better and more timely care for patients with NF1 and their caregivers. By addressing the unique needs and challenges identified through this research, we can enhance the QoL and support for the NF1 community globally

PLAIN LANGUAGE SUMMARY

Why did we perform this research?
We wanted to understand the specific challenges and needs of people with neurofibromatosis type 1 (NF1) and their caregivers. This condition's impact on their daily lives, fears, and treatment barriers hasn't been thoroughly studied, so we aimed to identify common trends and unique needs in different countries.

How did we perform this research?
We used an AI-powered tool to analyze anonymous discussions from social media platforms like Facebook, Instagram and Naver. By examining posts from 2010 to 2023 in online groups, blogs, and health-related Q&A sites, we were able to listen to the real experiences and concerns of patients and caregivers.

What is deep social media listening?
Deep social media listening is a technique for collecting and analyzing anonymous data from social networks to understand what people are saying about their experiences. Using advanced technology, we can process large amounts of unstructured text from these platforms to identify the true needs and feelings of patients and caregivers.

What were the findings of this research and what are the implications
We discovered significant differences in how NF1 affects patients' and caregivers' quality of life in various countries. For instance, physical pain is a major concern in Mexico, while emotional problems are more significant in Argentina. Information needs and barriers to treatment also varied, with patients in some countries seeking advice on symptoms, and others looking for recommendations for healthcare providers. The fear of disease progression was common, with specific concerns like fear of bullying or poor academic performance in children mentioned in Romania. These findings suggest that understanding the specific needs and concerns of patients with NF1 in different countries can help improve their care and support globally.

Poster
Plain language summary
Supplementary material

Please scan this quick response (QR) code with your smartphone camera or app to obtain a copy of these materials. Alternatively, please use the link below.
https://rarediseaseresource.com/2024/CTF-06-20/NF1_QoL_aspects_social_listening_poster
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BACKGROUND

- The disease burden for patients with neurofibromatosis type 1 (NF1) and their caregivers is still largely unexplored
- Traditional research methods often fail to fully capture their experiences, needs, and concerns, which are crucial for improving care and support

METHODOLOGY

- The research utilized deep social media listening (dSML) with a proprietary tool designed to extract anonymized big data from open social media platforms
- Analyzed sources included online groups, blogs, and forums on Facebook, Instagram, Naver, Twitter, and other platforms spanning from 2010 to 2023
- dSML involves the collection and analysis of anonymized data from unstructured text in social networks
- This method allows researchers to capture the authentic patient voice, providing insights into patients' and caregivers' experiences, needs, and concerns that are often missed by traditional research methods
- dSML technology is particularly suitable for capturing the quality of life (QoL) aspects, unmet needs, barriers, and fears of the community of patients with NF1 because it leverages advanced natural language processing (NLP) algorithms to analyze large volumes of real-time, authentic patient and caregiver discussions
- By processing this data, dSML can identify emerging trends, common concerns, and specific unmet needs across different demographics and geographic regions
- This technology ensures that the collected data reflects the true experiences and challenges faced by the NF1 community
- The comparative cross-country analysis was based on data obtained using the same technology, allowing for consistent and reliable comparisons across different regions (scan the QR code for detailed methodology in the **Supplementary Material**)

RESULTS AND INTERPRETATION

The analysis of social media activity among patients with NF1 across regions unveiled distinct patterns of digital engagement (**Table 1**):

- South Korea had the greatest share of digitally active patients (31.6%), which may have been indicative of a robust online support network within the NF1 community
- Malaysia had an unusually high average number of posts per patient (49.08) despite a low share of digitally active patients (0.6%), suggesting a small but highly engaged subgroup
- Moderate levels of digital participation were reported in Sweden and Romania, with patients posting at similar rates (13.03 and 13.34 posts per patient, respectively), and shares of digitally active patients ranging from 5.6% to 12.4%
- A higher proportion of patients were digitally active in Argentina (7.6%) compared with Mexico (1.9%), yet both countries exhibited lower average posts per patient, suggesting less frequent but more widespread engagement

Table 1. Descriptive statistics for six countries

Region	Europe		Asia		Latin America	
	Sweden	Romania	South Korea	Malaysia	Argentina	Mexico
Number of posts	5667	4762	65,741	3092	6394	5168
Number of patients with NF1*	435	357	5474	63	1196	813
Share of digitally active patients (%)†	12.4	5.6	31.6	0.6	7.6	1.9
Average number of posts per patient	13.03	13.34	12.01	49.08	5.35	6.36

*Number of patients/caregivers who posted about their NF1 journey; †Percentage of patients who go online to discuss their problems and seek alternate opinions (calculated based on the reported NF1 prevalence of 1/3000).
Data collection occurred between 2017–2022 for Sweden, 2016–2022 for Romania and Argentina, 2016–2021 for South Korea and Mexico, and 2010–2023 for Malaysia.
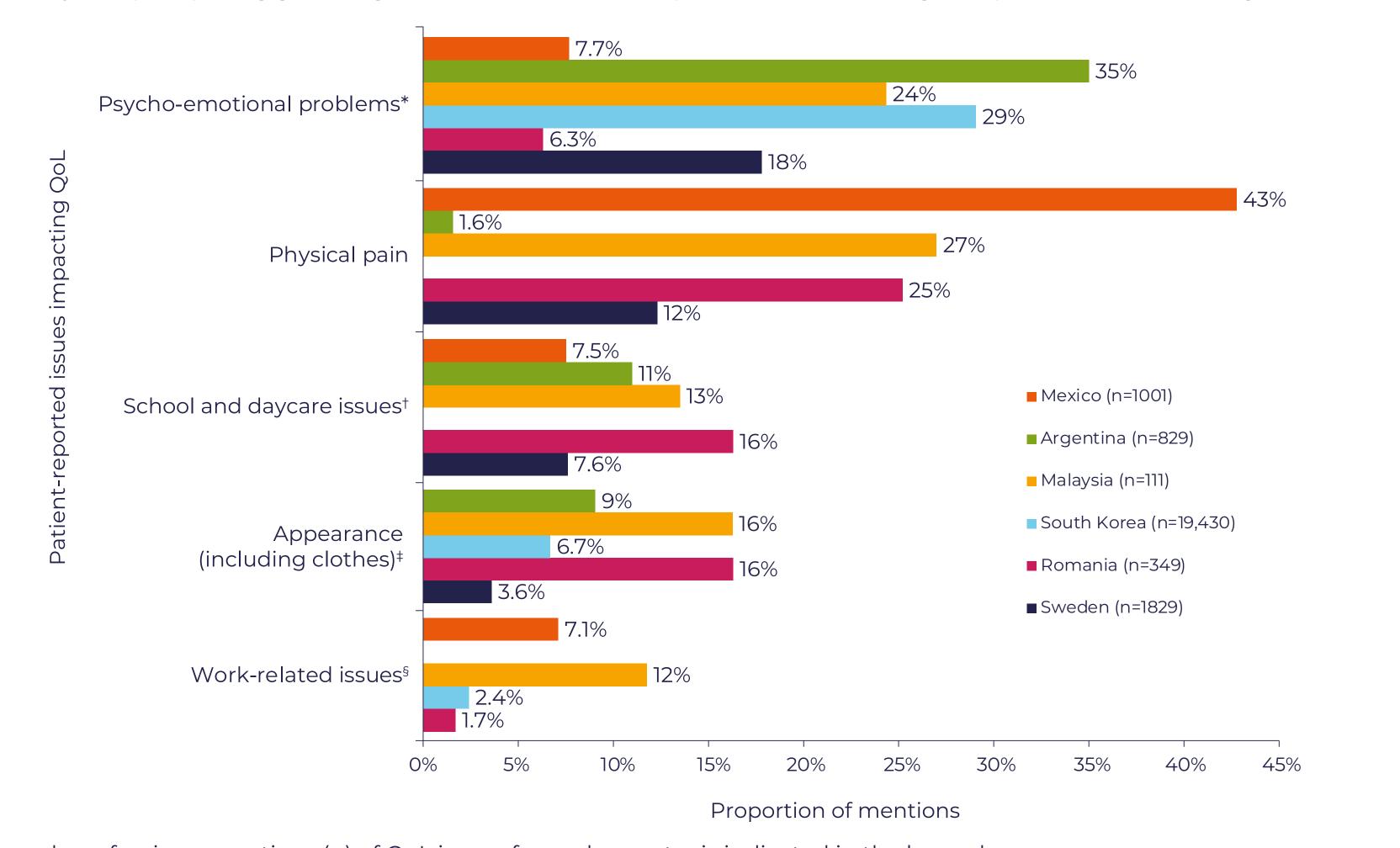NF1, neurofibromatosis type 1.

Figure 1. Quality of life

The analysis of topics related to QoL for patients with NF1 reveals significant regional differences:

- In Mexico, physical pain was the predominant concern, mentioned in 43% of cases, indicating a substantial impact on patients' daily lives
- In Argentina, psycho-emotional problems were more prevalent, with 35% of mentions, highlighting the mental health burden in this country
- In Europe (Sweden and Romania), concerns about school and daycare issues were significant in Romania (16%), reflecting the challenges faced by younger patients and their caregivers in educational settings
- In South Korea, there was a balanced distribution of concerns; however, physical pain was rarely mentioned. This may suggest better pain management or a lower incidence of pain-related issues
- Appearance-related concerns, including clothing, were more frequently discussed in Romania and Malaysia (16% each), indicating societal or cultural factors influencing these patients
- Work-related issues were less frequently mentioned overall but show the highest concern in Malaysia (12%), suggesting economic or occupational challenges specific to this region

The number of unique mentions (n) of QoL issues for each country is indicated in the legend.
*Work-related issues include inability to find a job or being forced to quit a job because of NF1; †Appearance (including clothes) includes public perception, difficulties selecting clothes (comfort vs the desire to hide visual manifestations of the disease); ‡School and daycare issues include cognitive issues in children, home schooling, nutrition, routine issues, problems with dressing and mobility; §Psycho-emotional problems include emotional burden, lack of support, depression and anxiety.
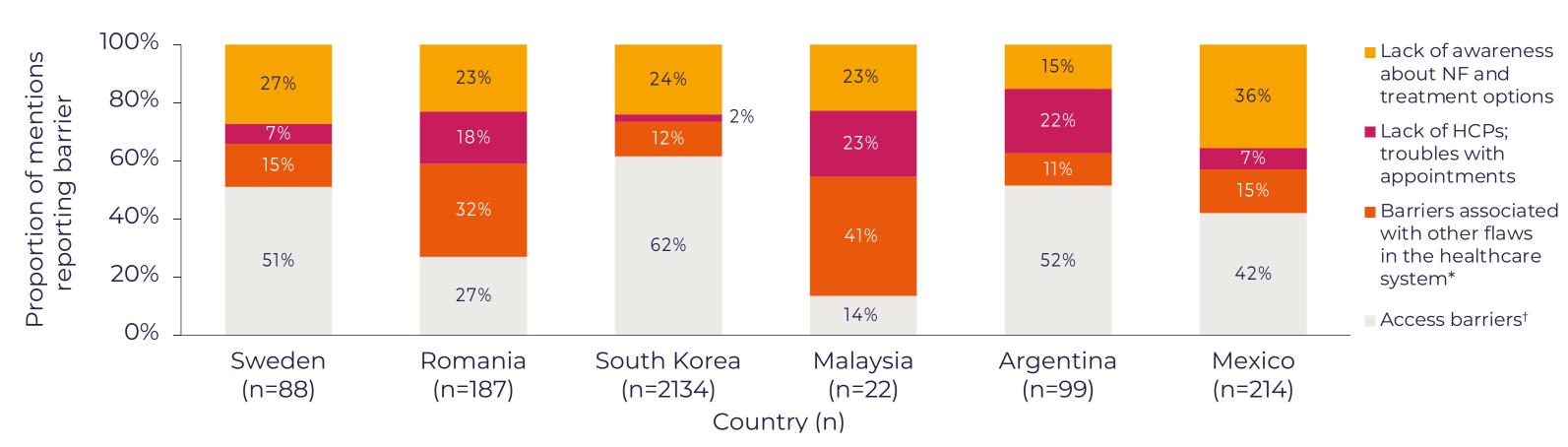QoL, quality of life.

Figure 2. Barriers for treatment and diagnostics

Barriers for patients with NF1 exhibit notable regional disparities:

- The majority of the countries faced considerable barriers due to the lack of information and awareness about NF1 (15–36% of cases), with the highest rate in Mexico, pointing to the necessity for improved patient and caregiver education
- Malaysia, Argentina, and Romania seemed to struggle with the lack of healthcare professionals (HCPs), clinics, travel issues to clinic centers and long wait for an appointment (23%, 22% and 18% of cases, respectively), indicating a need for enhanced medical education and training
- In all countries in this study, barriers associated with flaws in healthcare system were substantial (38–86%) with the highest rate observed in Malaysia followed by Romania (73% of cases)
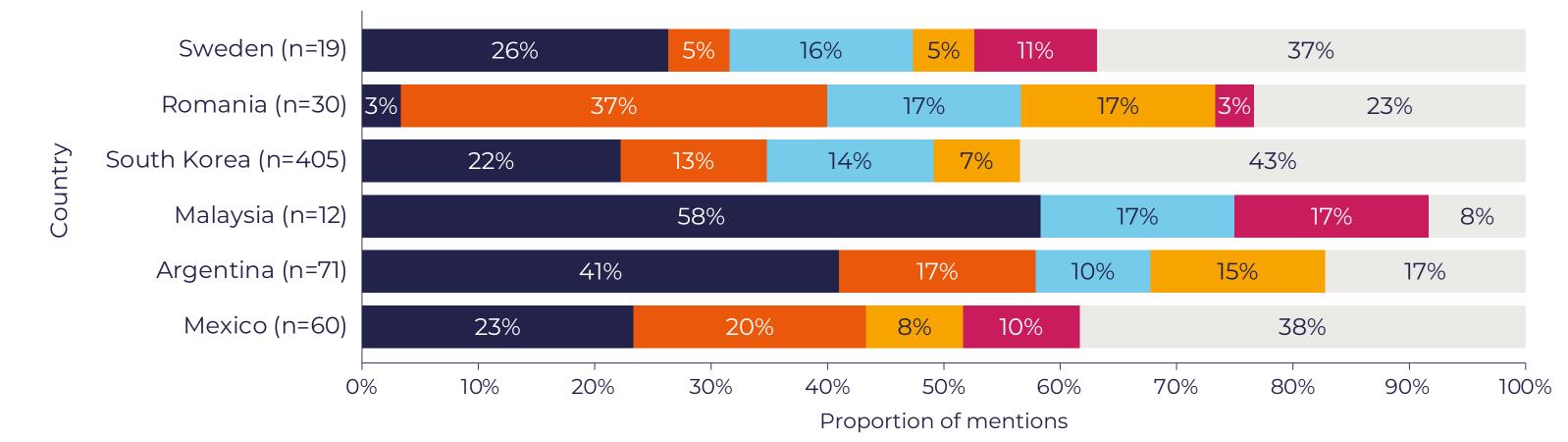

*Barriers associated with other flaws in the healthcare system include lack of high-quality help in diagnostics, clinics working with NF1, mistrust in HCPs, fears, etc.; †Access barriers include financial burden, such as high prices and insurance issues. HCPs, healthcare professionals; NF, neurofibromatosis; NF1, neurofibromatosis type 1.

Figure 3. Fears and concerns

Patient fears associated with NF1 show significant regional differences:

- Fear of disease progression was particularly acute in Malaysia, mentioned 58% of times, while in Romania this fear was among less frequently mentioned (3%) ones
- Caregivers in Romania exhibited heightened concerns for their children, including fears of bullying and poor academic performance (37%), as well as fear of inheritance (17%), indicating significant familial and societal pressures
- In Latin America, the fear of disease progression was pronounced in Argentina (41%) and Mexico (23%), highlighting the pervasive anxiety about the condition worsening
- Fear of treatment and procedures was a common concern across regions, with notable share of mentions in Romania (17%), Malaysia (17%), and Sweden (16%), suggesting apprehensions about medical interventions

These fears underscore the multifaceted psychological burden of NF1 on patients and caregivers, necessitating comprehensive support systems to address these concerns.
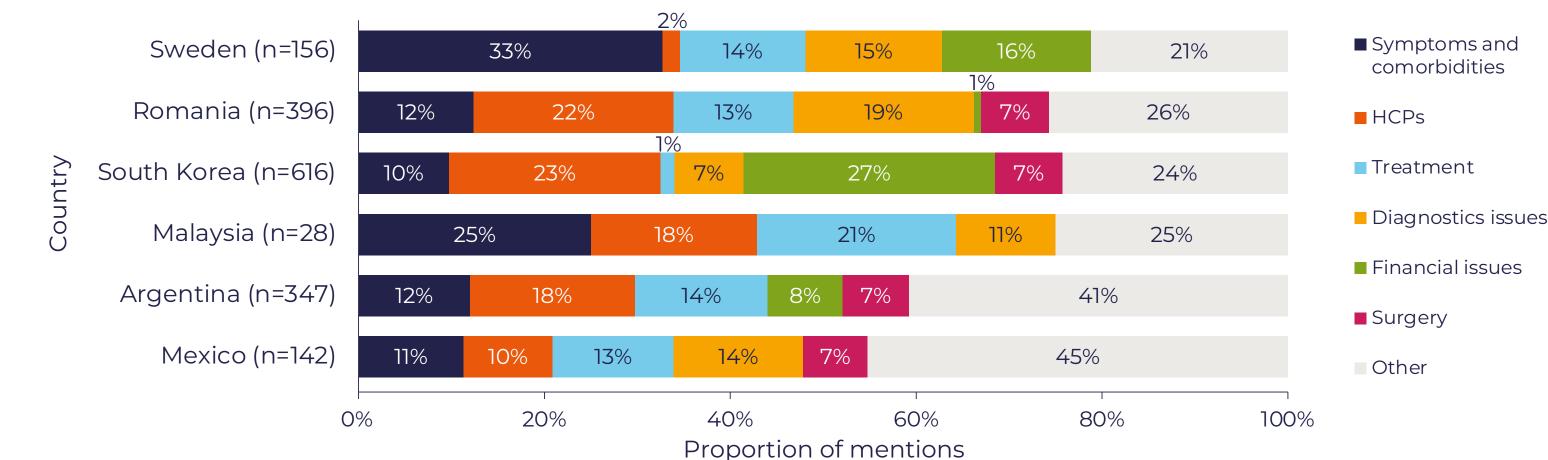

Figure 4. Unmet informational needs

The unmet informational needs of patients with NF1 and caregivers represent questions arising in patient discussions. The topics of questions vary in different countries:

- In Sweden and Malaysia, 33% and 25% of questions were about understanding symptoms and their progression, respectively, reflecting a demand for comprehensive disease education
- Patients and caregivers in Romania and South Korea frequently sought recommendations for expert HCPs (22% and 23%, respectively), underscoring the need for accessible specialized care
- Discussions in Romania often centered around diagnostic issues (19%), indicating difficulties in obtaining accurate diagnoses and timely interventions
- Patients in Malaysia and Mexico frequently inquired about treatment options (21% and 13%, respectively)
- Across all the countries, patients in South Korea were most troubled about financial issues (27%)

HCPs, healthcare professionals.

Using multilingual AI-driven techniques in understanding NF1 diagnostic journey globally

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María Carolina Erazo Muñoz[3], Marina Lutoshkina[3], Elizaveta Ugolkova[4]

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OBJECTIVES

The primary goal of this research was to uncover common patterns and identify the needs and challenges of patients with NF1 and their caregivers across various countries. Specifically, we aimed to:

- Comprehend the treatment and diagnostic journey of patients with NF1
- Identify the first points of contact for patients with NF1 within the healthcare system
- Examine barriers to treatment and diagnosis
- Utilize dSML to capture authentic patient voices and insights, offering a more accurate and comprehensive understanding of their experiences
- Perform a comparative cross-country analysis to investigate differences and similarities in NF1 treatment experiences worldwide

CONCLUSIONS

- The use of technology driven by artificial intelligence in healthcare has further demonstrated its potential
- This analysis provided valuable insights into the diagnostic journey of patients with NF1 from diverse perspectives and emphasized the unique characteristics of specific countries
  - Hyperpigmentation emerged as the prevailing initial symptom across all countries, often leading to the diagnosis of NF1
  - Patients with NF1 often first consulted pediatricians and neurologists. However, in some countries, like Malaysia and South Korea, dermatologists and surgeons were the initial point of contact
  - A consistent trend was observed across all countries, with most diagnoses occurring between 1 and 5 years of age
  - MRI played a significant role in diagnostics, with genetic testing being the second most frequent
- Such findings could aid in addressing knowledge gaps, enhancing communication between patients and specialists, and expediting NF1 diagnosis

PLAIN LANGUAGE SUMMARY

Why did we perform this research?
The aim of the study was to investigate the challenges encountered in various regions by patients with neurofibromatosis type 1 (NF1) throughout their diagnostic and treatment process; a specific focus was set on Sweden, Romania, South Korea, Malaysia, Argentina, and Mexico.

How did we perform this research?
We conducted the research by employing deep social media listening techniques to extract anonymized big data from various open social media platforms. The sources analyzed encompassed online groups on social media (such as Facebook, Twitter, Instagram, TikTok, Naver, etc.), open patient forums, and other sources of user-generated content spanning from 2010 to 2023.

What is deep social media listening?
Deep social media listening is collecting anonymized data from posts and comments in social networks in order to listen to patient voice. Its purpose is to assist various stakeholders in comprehending patients' needs, rather than solely relying on their own perceptions and knowledge.

What were the findings of this research and what are the implications?
Hyperpigmentation emerged as the prevailing initial symptom across all countries, often leading to the diagnosis of NF1. There was a consistent trend observed regarding the age of diagnosis overall. It is worth noting that pediatricians and neurologists were commonly the first healthcare professionals consulted by patients with NF1, serving as the entry point into the healthcare system. However, certain countries exhibited unique characteristics in this regard (e.g. dermatologists in Malaysia and surgeons in South Korea stand out). Among diagnostic procedures, MRI plays a significant role in all countries where the research was performed, while other methods (biopsy, genetic testing) may vary. A comparative analysis of the findings highlighted significant disparities in healthcare systems and NF1 awareness among patients across different countries.

BACKGROUND

- Neurofibromatosis type 1 (NF1) is a genetic condition that affects approximately 1 in 3000 people globally[1]
- Although NF1 is widespread, understanding of how multiple clinical manifestations affect pediatric patients with NF1 and their caregivers is still limited
- Conventional research methods frequently overlook the full scope of their experiences, challenges, and concerns, which are essential for enhancing care and support

METHODOLOGY

- This study employed deep social media listening (dSML) utilizing a proprietary tool that extracts anonymized big data from public social media platforms
- The sources analyzed included online groups, blogs, and forums on Facebook, Instagram, Twitter, Naver, and other platforms, covering the period from 2010 to 2023
- dSML involves gathering anonymized data from unstructured texts in social networks to capture the patient voice accurately, thereby providing stakeholders with a genuine understanding of patient needs beyond their preconceived notions and knowledge
- dSML technology is particularly effective for understanding the diagnostic journey, symptoms, and medical aspects of the community of patients with NF1
  - It employs advanced natural language processing (NLP) algorithms to analyze substantial volumes of real-time, authentic patient and caregiver discussions
- This allows the identification of emerging trends, prevalent concerns, and specific unmet needs that traditional research methods might overlook
  - Consequently, the gained insights are reflective of the actual experiences and challenges faced by patients with NF1 and their caregivers
- A comparative cross-country analysis was performed using data obtained through the same technology, ensuring consistent and reliable comparisons across different geographical regions (scan the QR code for detailed methodology in the **Supplementary Material**)

RESULTS AND INTERPRETATION

More than 90,000 messages were analyzed, and 8338 patients were identified in six countries (**Table 1**).

- South Korea was identified as the country with the largest percentage of digitally active patients
- In Malaysia, few patients were digitally active, but the number of posts per patient was high

The data illustrate four parts of a patient's diagnostic journey:

- Symptoms that prompted patients/caregivers to look for a medical diagnosis
- Entry point into the healthcare system – specialists who diagnosed NF1 or suspected the disease and referred the patient to a relevant specialist
- Age of NF1 initial disorder/suspicion
- Medical tests performed

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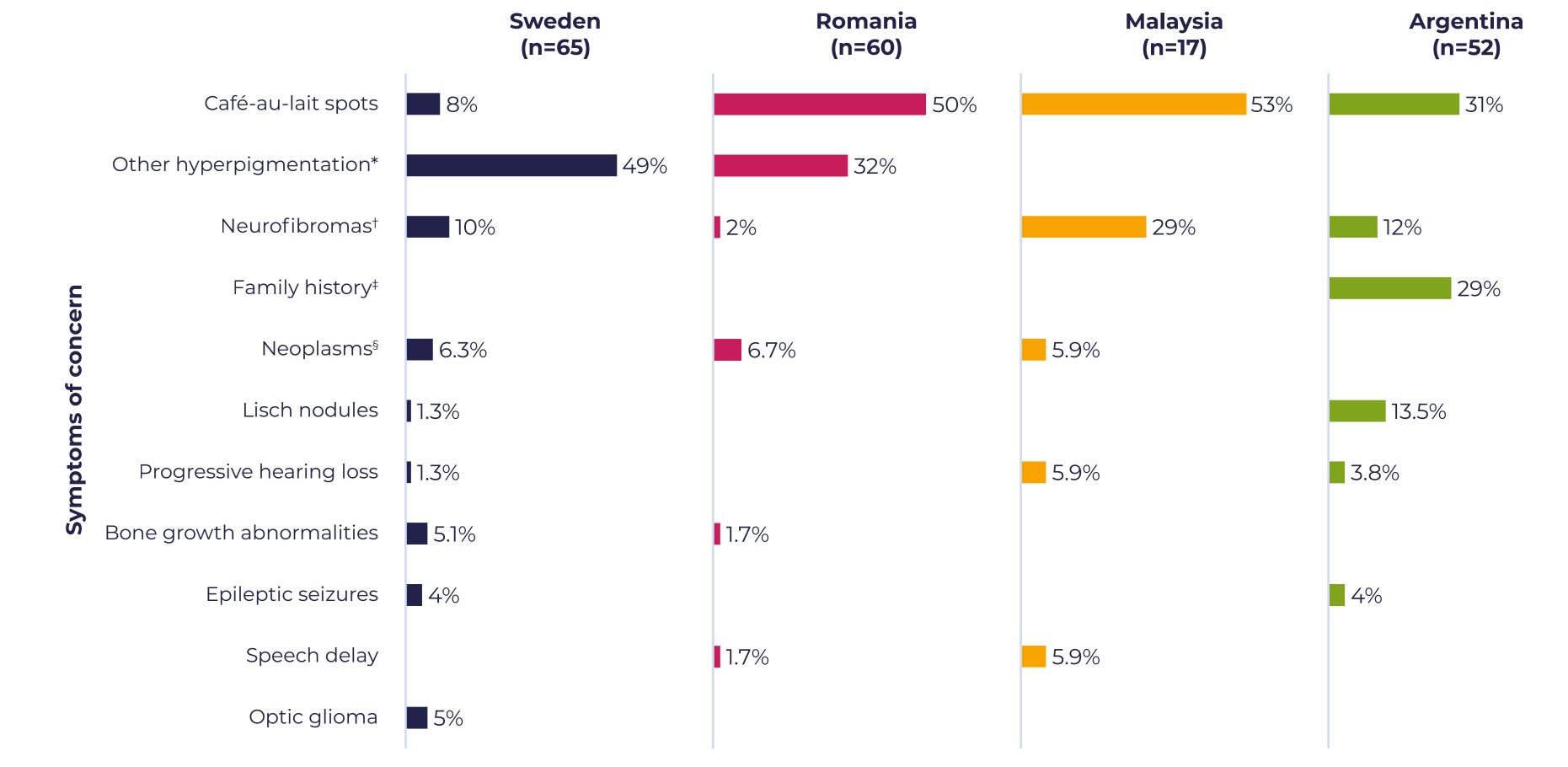Data collection occurred between 2017–2022 for Sweden, 2016–2022 for Romania and Argentina, 2016–2021 for South Korea and Mexico, and 2010–2023 for Malaysia.
NF1, neurofibromatosis type 1.

Figure 1. Symptoms of concern to patients

Top mentioned symptoms in children were hyperpigmentation (including café-au-lait-spots, spots on the skin, freckles, etc.), neurofibromas (including plexiform neurofibromas), neoplasms (benign tumors, brain tumors, and other malignant neoplasms), and in some cases, family history (including asymptomatic cases) (**Figure 1**).

Cross-country analysis, based on data obtained using the same approach, demonstrated that symptoms that cause major concerns vary slightly from country to country:

- Hyperpigmentation (general term) was one of the most frequently mentioned symptoms in all countries
- Café-au-lait spots most commonly triggered the diagnosis:
  - Mentioned in more than half of cases in Romania (50%) and Malaysia (53%)
- Neoplasms were one of the causes for diagnosis (5.9–6.7% of cases):
  - Whilst neurofibromas were frequently mentioned by patients in Malaysia (29%), fewer than 2% of patients mentioned this symptom in Romania
- In Argentina, almost 30% of cases were familial NF1 cases, not necessarily symptomatic

The number of unique mentions (n) on this topic for each country is indicated in each column.

*Hyperpigmentation includes spots on the skin, freckles, etc.; †Neurofibromas include all types along with plexiform neurofibromas; ‡Family history includes familial NF1 cases (including asymptomatic cases); §Neoplasms include benign tumors, brain tumors, and other malignant neoplasms.
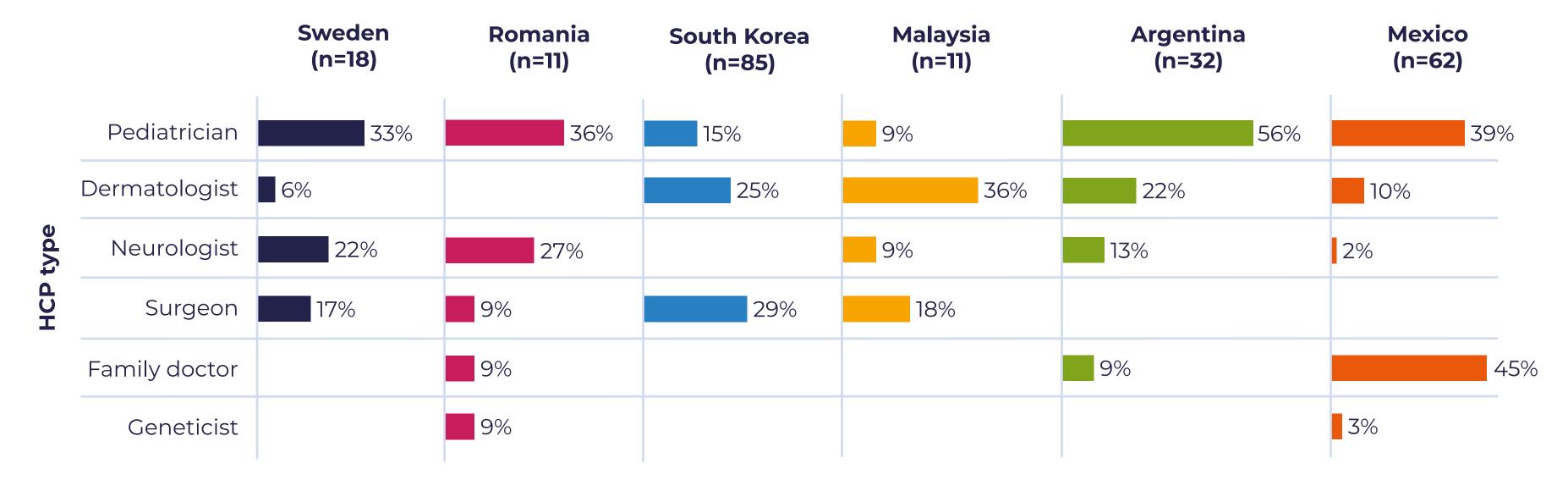NF1, neurofibromatosis type 1.

DIAGNOSTIC JOURNEY

Figure 2. Entry points/healthcare professionals

As entry points in the healthcare system (the specialists who were the first to whom patients and caregivers turned in search of a diagnosis), pediatricians are key (up to 56% of cases), as well as dermatologists (up to 36% of cases) and neurologists (up to 27% of cases) (**Figure 2**).
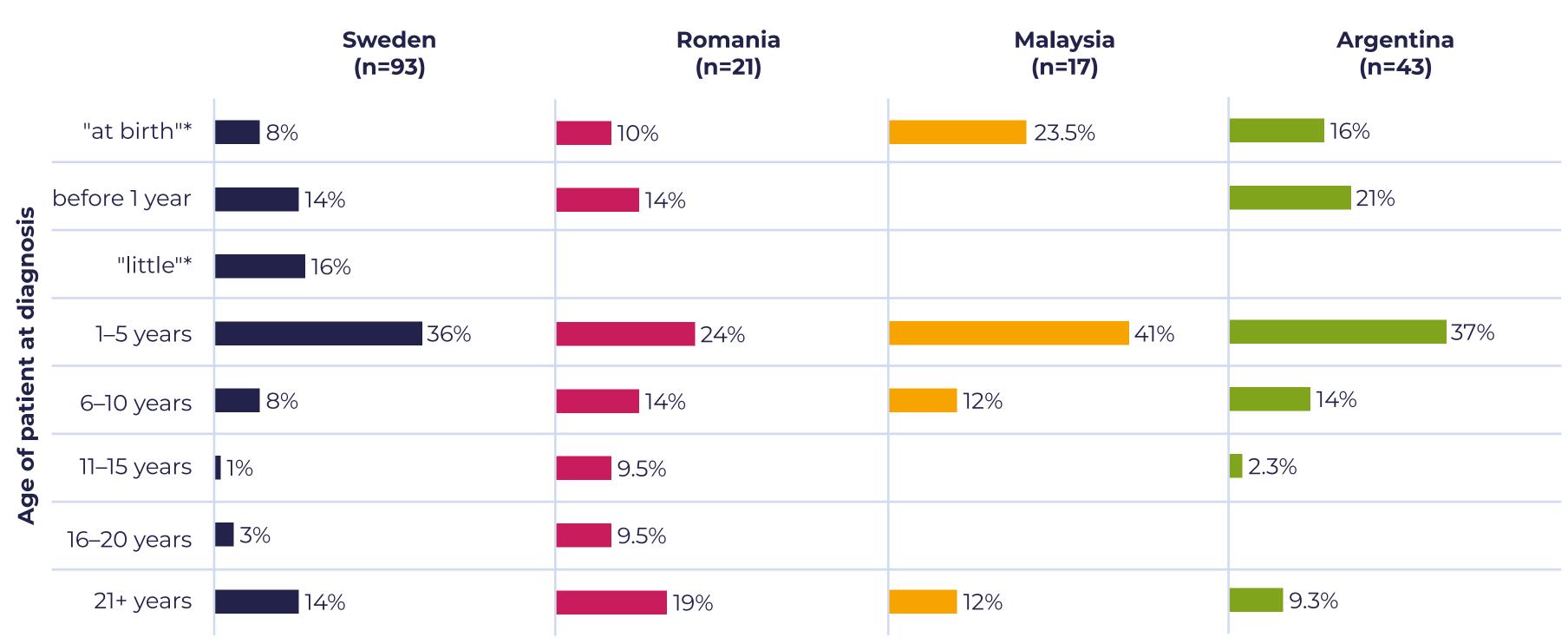
- In Sweden, Romania, Argentina, and Mexico, pediatricians play an important role
- In South Korea and Malaysia, surgeons and dermatologists are mostly mentioned, respectively
- In Mexico, family doctors were more often mentioned (45%) as the first healthcare professional consulted

Figure 3. Age of diagnosis

Despite significant differences between countries in the development of health systems and awareness of NF1, there are similar patterns in the age of diagnosis (**Figure 3**):

- In all countries, the peak of diagnosis occurred between 1 and 5 years of age
- In Malaysia, 23.5% of patients were diagnosed at birth
- In Romania, almost 20% of patients were older than 21 years by the time of diagnosis
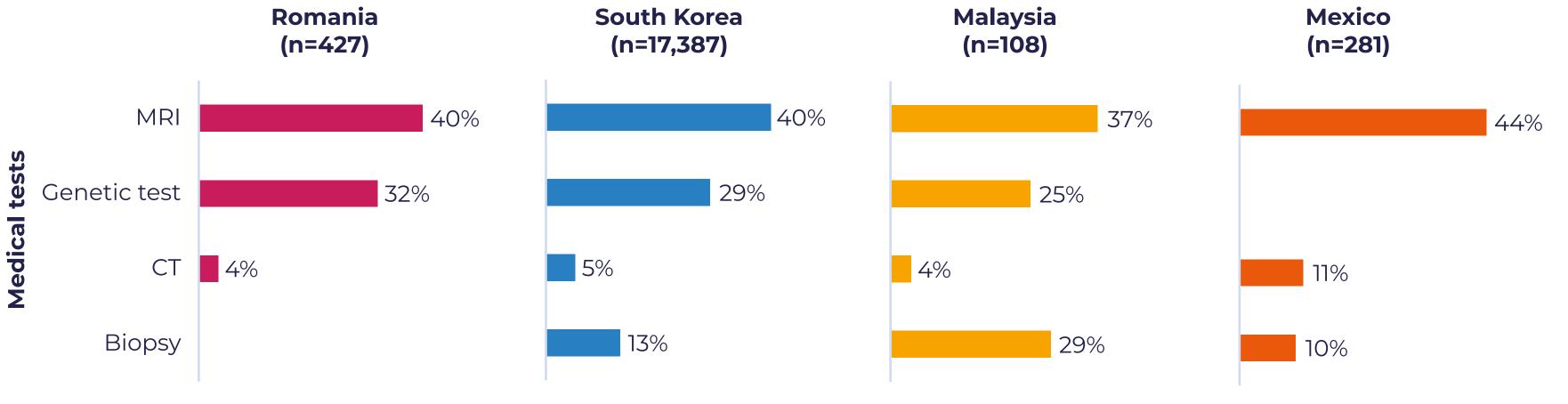
*In these cases, the patient or caregiver did not mention the specific age at which the diagnosis was made but referred to the time period with the words "at birth" or "(when the patient was) little."

Figure 4. Medical tests

A variety of different tests were mentioned (**Figure 4**):

- Magnetic resonance imaging (MRI) was the most frequently mentioned medical test (37–44% of mentions in all countries)
- Genetic testing was the second most frequent (up to 32% of mentions)
- Biopsy is not a widespread medical test for diagnosis in most countries
  - In Malaysia, almost 30% of patients mentioned they were diagnosed using biopsy

CT, computed tomography; MRI, magnetic resonance imaging.

Reference
1. Evans DG et al. *Am J Med Genet A* 2010;152A:327–332.
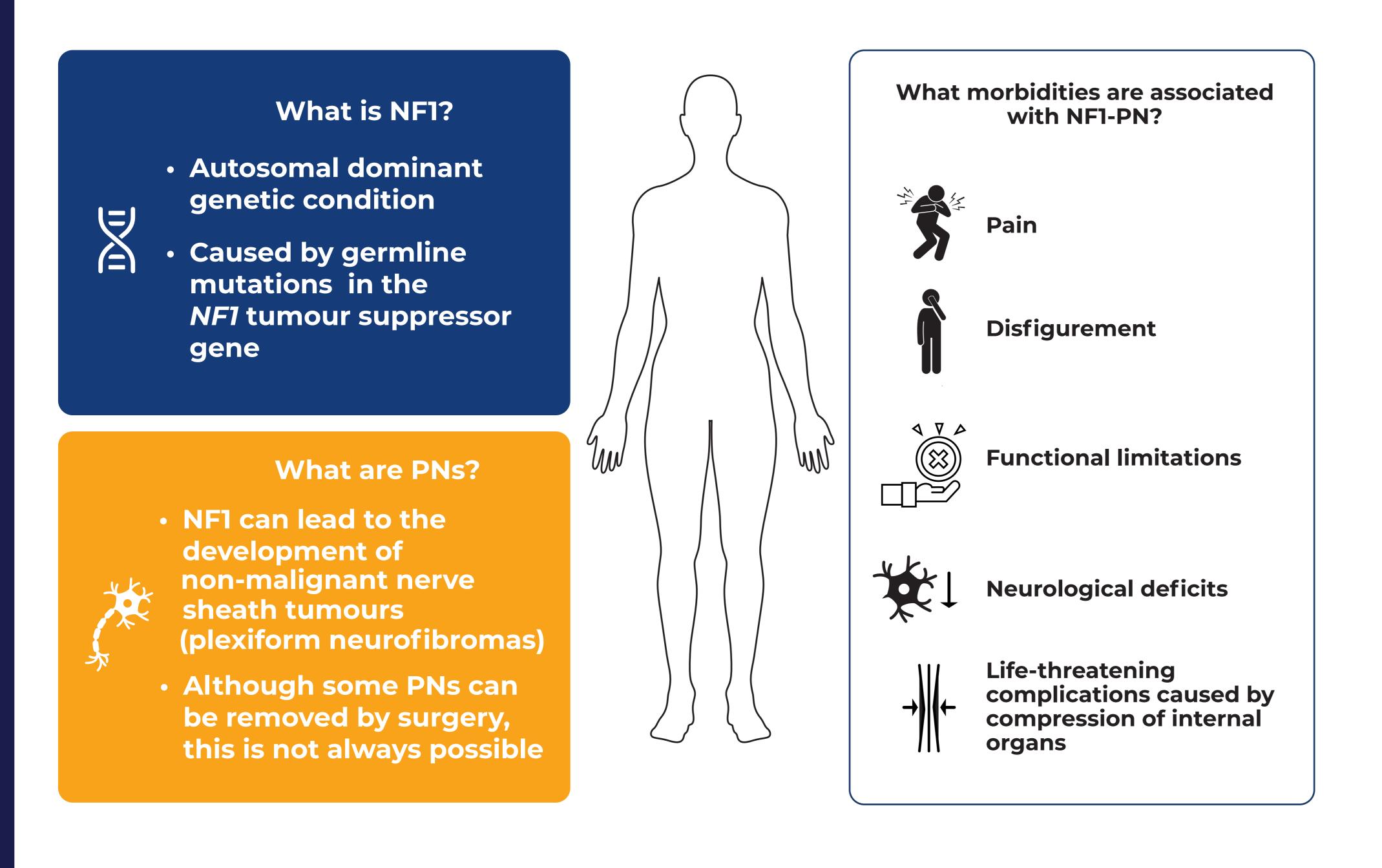
DIAGNOSTIC AND TREATMENT JOURNEY OF PATIENTS WITH NEUROFIBROMATOSIS TYPE 1 IN CANADA: AI BASED ANALYSIS

Susanna Burckhardt[1], Elizabeth Silayuv[1], Irina Efimenko[2], Anna Paleeva[2], Zlata Bezborodova[2], Vera Lipkovskaya[2]

[1]Alexion, AstraZeneca Rare Disease; [2]Semantic Hub

BACKGROUND AND OBJECTIVES

Knowledge of the impact of multiple clinical manifestations on NF1 paediatric patients and their caregivers remains limited. The aim of this research was to better understand the NF1 patient experience in Canada based on first-hand testimony extracted from "Big Data".

What is NF1?
- Autosomal dominant genetic condition
- Caused by germline mutations in the *NF1* tumour suppressor gene

What are PNs?
- NF1 can lead to the development of non-malignant nerve sheath tumours (plexiform neurofibromas)
- Although some PNs can be removed by surgery, this is not always possible

What morbidities are associated with NF1-PN?
- Pain
- Disfigurement
- Functional limitations
- Neurological deficits
- Life-threatening complications caused by compression of internal organs

CONCLUSIONS

These data highlight experiences and challenges faced by paediatric patients with NF1 and their caregivers in Canada; there appears to be a need to enhance diagnosis and, particularly, improve referral to specialists and Centres of Excellence. Besides the characteristics of the disease that are meaningful to HCPs and multidisciplinary teams to reach a diagnosis and manage NF1, this type of research allows an understanding of the perspective of the patients and the impact of the more common features and symptoms of NF1 on their QoL. These findings highlight the need for better communication with patients based on their actual needs (such as pain management) and may help shape awareness campaigns among patients, caregivers and HCPs, e.g. through the establishment of multidisciplinary teams.

PLAIN LANGUAGE SUMMARY

Why did we perform this research?
The goal of the research was to understand what kind of difficulties NF1 patients experience during their diagnostic and treatment journey particularly in Canada. To do so, we engaged an artificial intelligence tool that uses deep semantic analysis (drawing meaning from unstructured texts) to collect patients' and caregivers' honest and unbiased opinions on their patient journey and life with this condition.

How did we perform this research?
We performed the research using deep social media listening with a help of a proprietary tool extracting anonymised data from open social media. Sources analysed included online groups on Facebook, Twitter and Reddit for 2017-2022.

What is deep social media listening?
Deep social media listening (dSML) is collecting anonymised data from unstructured texts in social networks in order to listen to Patient Voice and to help various stakeholders understand what patients really need rather than rely on their own perceptions and knowledge. This goes in three main steps:

Step 1. Scanning open digital environment and collecting big (unstructured) data

Step 2. Processing data with the use of deep semantic analysis

Step 3. Building the knowledge base and generating results

What were the findings of this research and what are the implications?
Café-au-lait spots, the characteristic brown skin spots, most frequently led to NF1 diagnosis; notably, it was mentioned that referral to specialists or centres of excellence to confirm diagnosis or monitor disease progression was often difficult to achieve. Comparison with the outcome from other countries showed that the severity of various symptoms of NF1 may be perceived differently.

55th Annual Congress of the International Society of Paediatric Oncology, October 11-14, 2023, Ottawa, Canada

METHODOLOGY

This deep social listening research involved multilingual artificial intelligence-based analysis of Patient Voice in Canada obtained through anonymised stories from domestic and international social media. Comparative cross-country analysis was included.
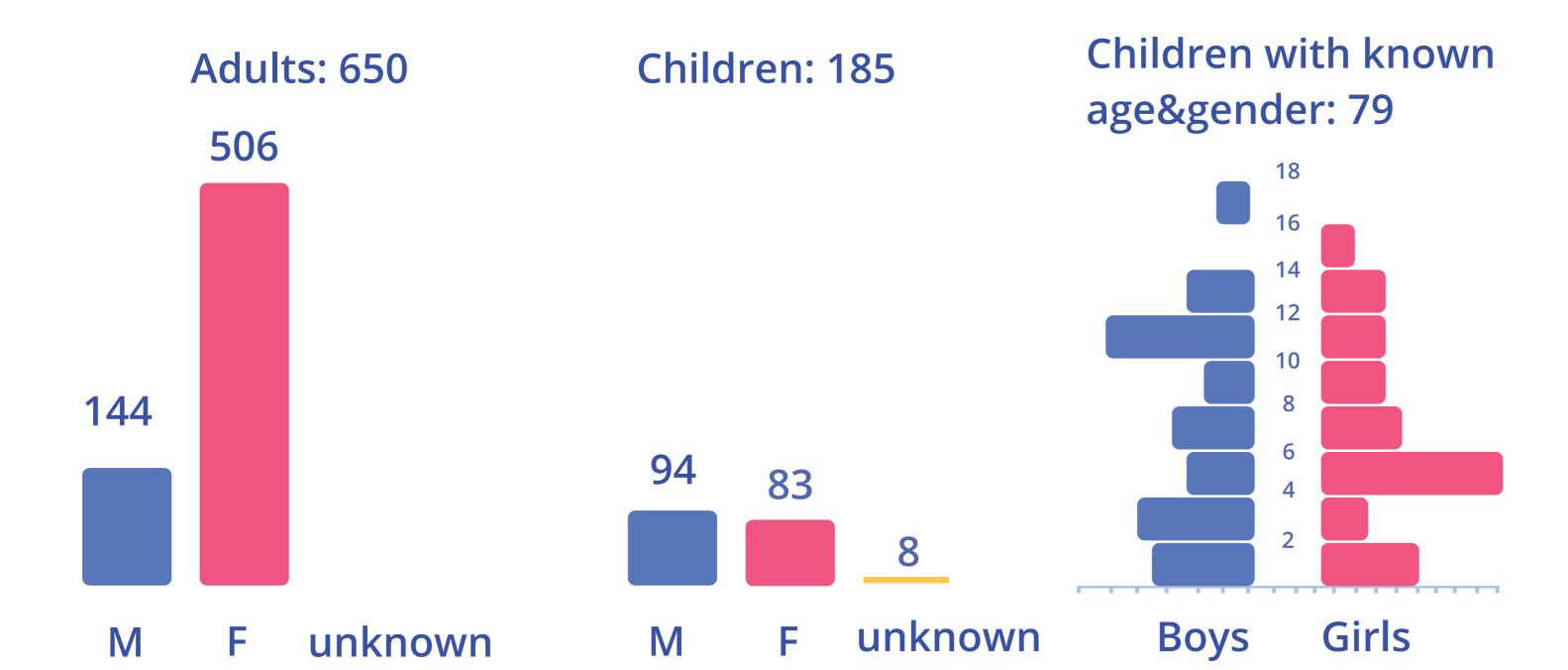
3,035 messages

835 patients

2017-2022 timeframe

RESULTS AND INTERPRETATION

The analysis allowed segmenting paediatric patients in the NF1 community present online: of the 835 patients identified, 185 were children (0 – 18 years old). Gender was indicated in 177 children (94 males, 83 females).

Patient characteristics

The analysis of collected data highlights multiple aspects related to the diagnostic and treatment journey of NF1 paediatric patients and their caregivers in Canada.

Over 85% of NF1 cases described online were diagnosed in childhood, mostly between 6 and 10 years. Most frequently mentioned first symptoms were café-au-lait spots (50%), followed by fibromas (12%). Patients and caregivers highlighted challenges in getting a definitive diagnosis as well as referral to specialists; paediatricians, neurologists and dermatologists were mentioned as most accessible among HCPs of all specialties (14 in total). Of all mentions (60), medical tests most often cited were MRI (48%) and genetic testing (18%).

Adults: 650
- M: 144
- F: 506

Children: 185
- M: 94
- F: 83
- unknown: 8

Children with known age&gender: 79
(Boys / Girls)

Treatment options
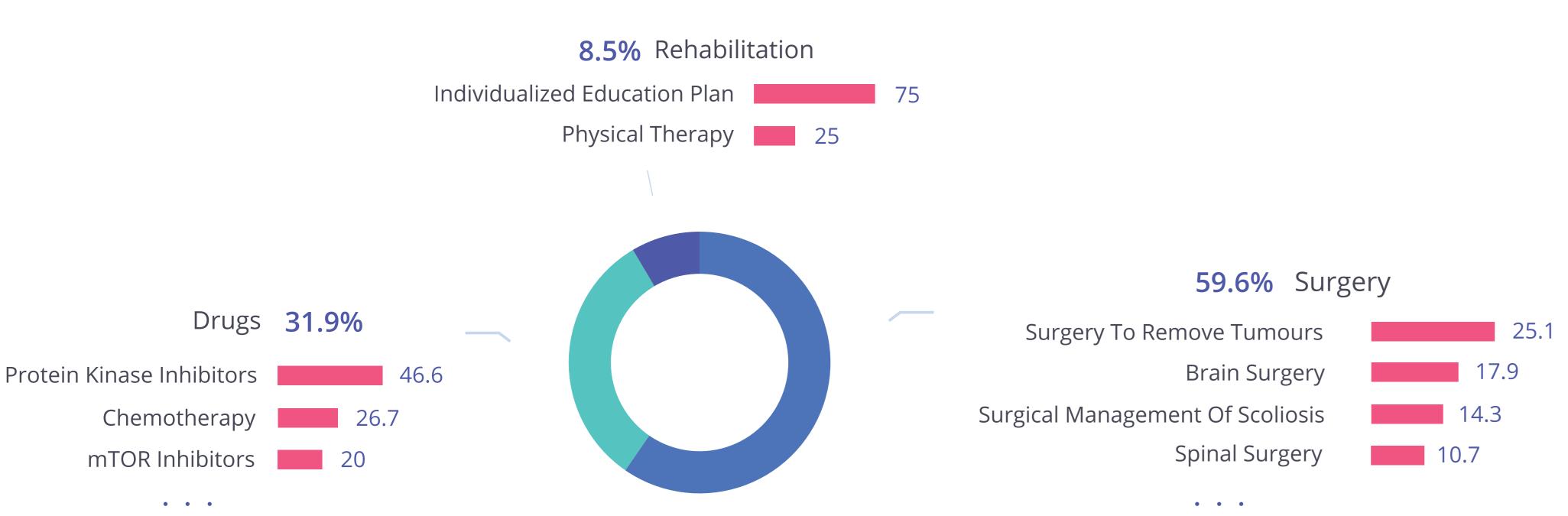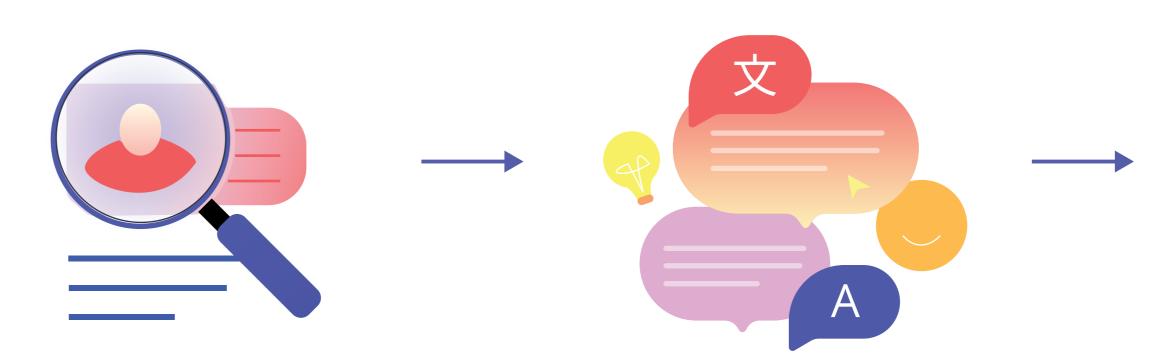
The treatment options most discussed for children were:
- Surgery (60%)
- Pharmacological therapy (32%, including protein kinase inhibitors, chemotherapy, mTOR inhibitors and anticonvulsants)
- Rehabilitation (8.5%)

Treatment-related messages highlighted a strong need for effective pain management.

8.5% Rehabilitation
- Individualized Education Plan: 75
- Physical Therapy: 25

Drugs 31.9%
- Protein Kinase Inhibitors: 46.6
- Chemotherapy: 26.7
- mTOR Inhibitors: 20

59.6% Surgery
- Surgery To Remove Tumours: 25.1
- Brain Surgery: 17.9
- Surgical Management of Scoliosis: 14.3
- Spinal Surgery: 10.7

Symptoms of concerns to patients
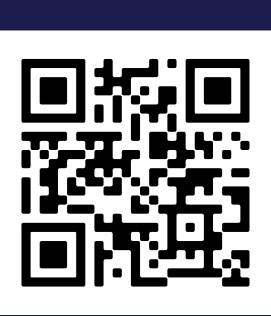
Top mentioned symptoms in children were hyperpigmentation, neurofibromas, neoplasm, neurodevelopmental abnormality, pain and brain neoplasm. Café-au-lait spots most commonly triggered the diagnosis but pain appeared to be the most burdensome.

Cross-country analysis, based on data obtained using the same approach, demonstrated that symptoms that cause major concerns vary from country to country. For example, in Canada neurofibromas were among most frequently mentioned symptoms, while in South Korea and Mexico hyperpigmentation and body pain were most relevant, respectively.

Canada
- Neurofibromas: 19.9%
- Neoplasm: 17.6%
- Physical pain: 13.7%
- Hyperpigmentation: 11.2%
- Brain neoplasm: 5.5%

Mexico
- Physical pain: 27.9%
- Neurofibromas: 16.4%
- Neoplasm: 13.9%
- Hyperpigmentation: 10.5%
- Café-au-lait spots: 6.5%

South Korea
- Hyperpigmentation: 45.9%
- Neoplasm: 29.5%
- Café-au-lait spots: 11.3%
- Physical pain: 2.7%
- Numerous pigmented freckles: 2.1%

Note: Categories of symptoms were classified based on patients' wordings.

References:
1. Bergqvist C et al. Orphanet J Rare Dis 2020;15:37.
2. Ferner RE et al. J Med Genet 2007;44:81–88.
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UNMET NEEDS OF FAMILIES WITH NEUROFIBROMATOSIS TYPE 1 IN CANADA: PATIENT VOICE BASED ANALYSIS

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BACKGROUND AND OBJECTIVES

Knowledge of the impact of the multiple clinical manifestations on NF1 paediatric patients remains limited. The aim of this research was to better understand issues that NF1 patients face when going through the diagnostic and treatment journey in Canada, using first-hand testimony extracted from "Big Data". The research scope included barriers to treatment, unmet needs of patients and caregivers, and quality of life (QoL) issues.

What is NF1?
- Autosomal dominant genetic condition
- Caused by germline mutations in the *NF1* tumour suppressor gene

What are PNs?
- NF1 can lead to the development of non-malignant nerve sheath tumours (plexiform neurofibromas)
- Although some PNs can be removed by surgery, this is not always possible

What morbidities are associated with NF1-PN?
- Pain
- Disfigurement
- Functional limitations
- Neurological deficits
- Life-threatening complications caused by compression of internal organs

CONCLUSIONS

These findings shed light on various aspects of life of NF1 patients and their families as well as their top unmet needs and concerns. Limited awareness of NF1, challenges to get referred to specialists as well as lack of educational resources were identified as key issues. These findings highlight the need for better communication, and they can help shape awareness campaigns for families and HCPs and develop educational materials that would address topics and concerns of NF1 patients and their families based on their actual needs.

PLAIN LANGUAGE SUMMARY

Why did we perform this research?
The goal of the research was to understand what kind of difficulties people affected by NF1 and their family members experience and what kind of unmet needs they have. To do so, we utilised an artificial intelligence tool that uses deep semantic analysis (drawing meaning from unstructured texts) to collect patients' and caregivers' honest and unbiased opinions on patient journey and life with this condition.
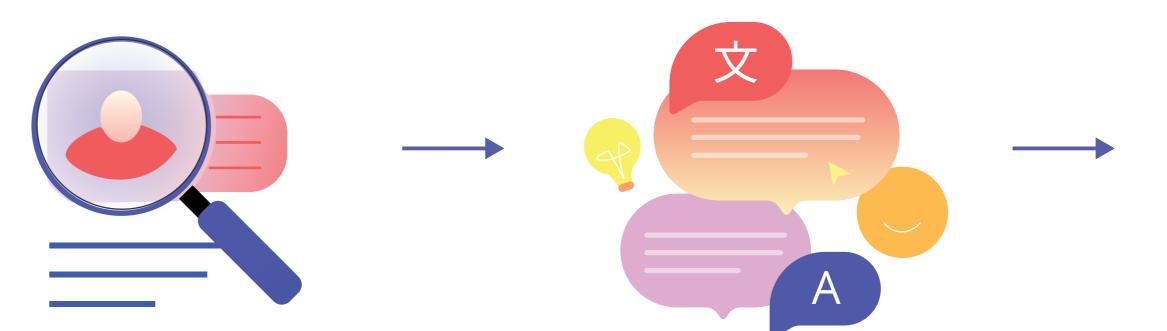
How did we perform this research?
We performed the research using deep social media listening with the help of a proprietary tool extracting anonymised big data from open social media. Sources analysed included online groups on Facebook, Twitter and Reddit for 2017-2022.

What is deep social media listening?
Deep social media listening (dSML) is collecting anonymised data from unstructured texts in social networks in order to listen to Patient Voice and to help various stakeholders understand what patients really need rather than rely on their own perceptions and knowledge. This goes in three main steps:

Step 1. Scanning open digital environment and collecting big (unstructured) data

Step 2. Processing data with the use of deep semantic analysis

Step 3. Building the knowledge base and generating results

What were the findings of this research and what are the implications?
Patients with NF1 and/or their caregivers communicate with each other in different groups on social media, reflecting the need to exchange experience, seek for information and mutual support. In this regard, patient support groups play an important role. Lack of information and awareness around NF1, even among doctors, was particularly prominent for French-speaking regions of Canada. Highest concerns of patients/caregivers were fear of existing symptoms worsening and new ones appearing.

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METHODOLOGY

This deep social listening research involved multilingual artificial intelligence-based analysis of Patient Voice in Canada obtained through anonymised stories from domestic and international social media. Comparative cross-country analysis, using data obtained with the same technology, was included.

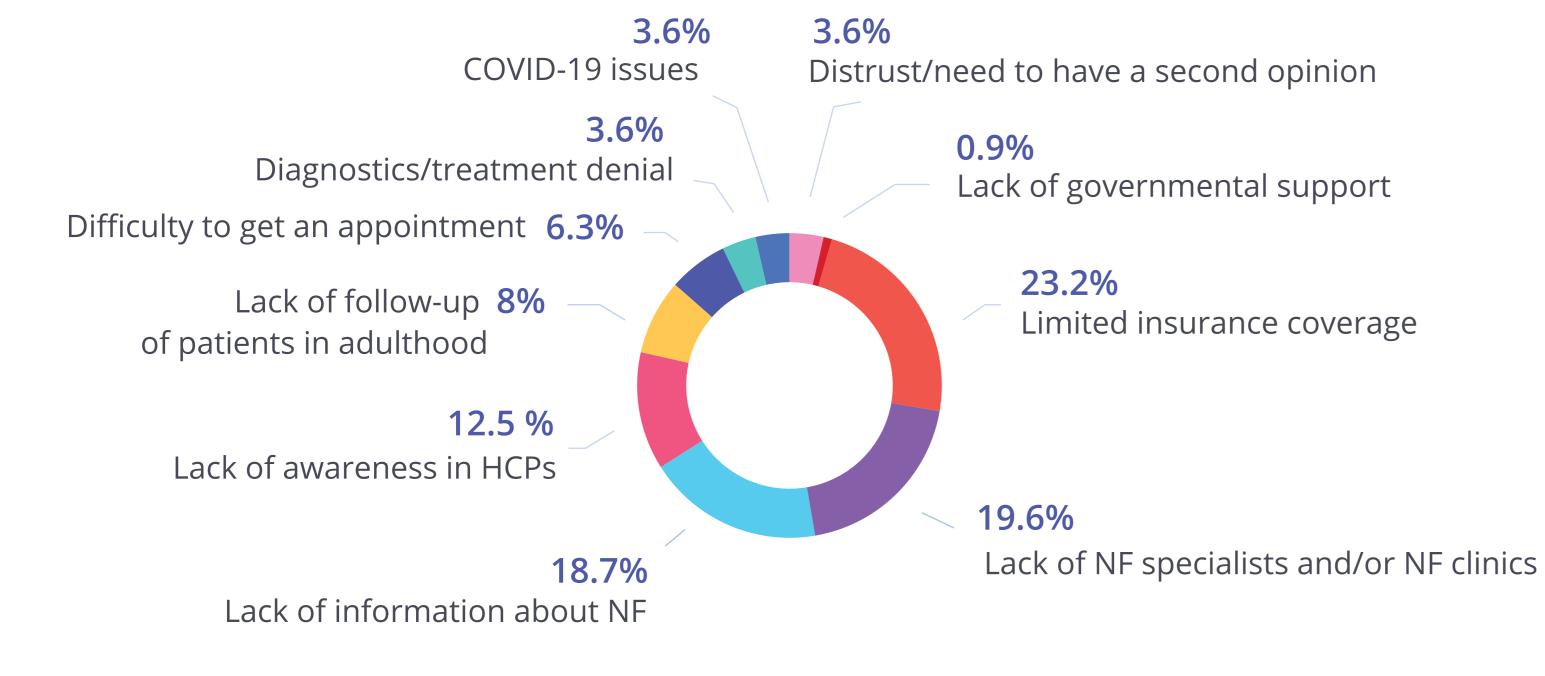**3,035 messages**

835 patients

2017-2022 timeframe

RESULTS AND INTERPRETATION

This analysis demonstrates a wide range of unmet needs and burden experienced by patients with NF1 and their caregivers in Canada.
We identified the main barriers to diagnosis and treatment as seen by patients and caregivers, their top concerns about the disease, and the outcomes with the biggest impact on their QoL.
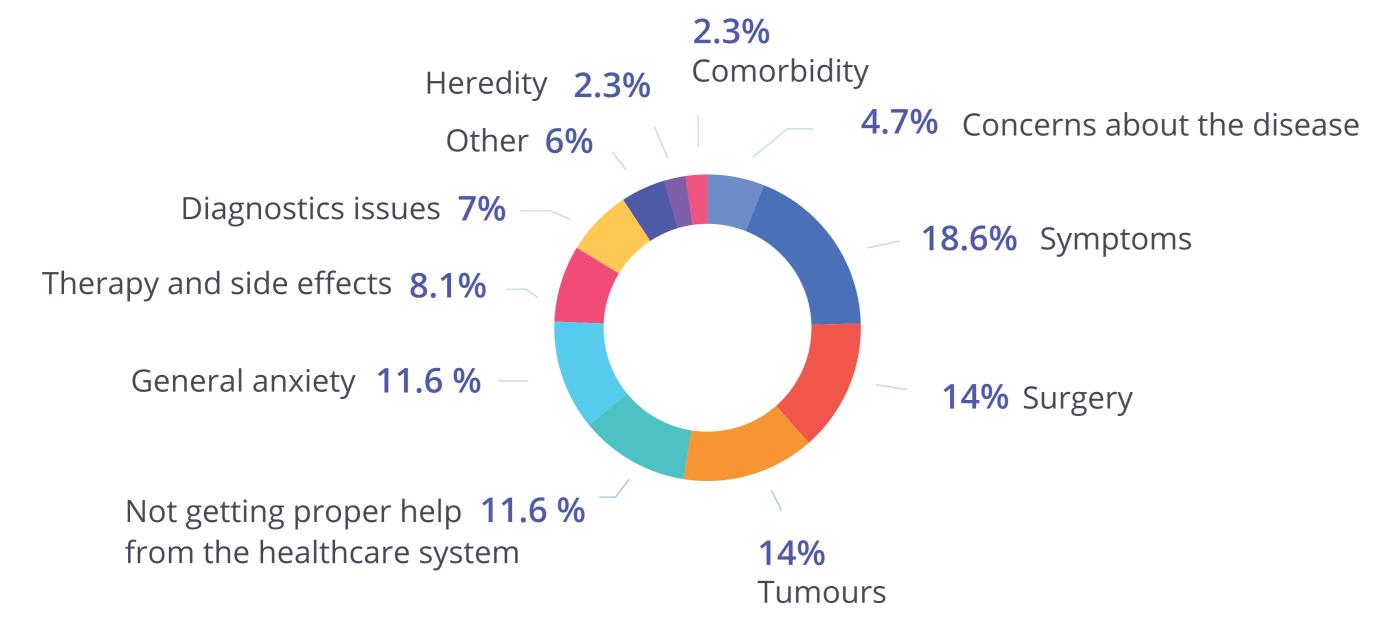
Barriers to diagnosis and treatment

Top identified barriers to diagnosis and treatment included insurance policy issues (e.g. difficulties of getting fibromas removal surgeries covered), lack of NF1 specialists, lack of information, especially for French-speaking Canadians; poor awareness among physicians, and issues with transition of care into adulthood. A general lack of awareness of treatment options was also mentioned.

- 3.6% COVID-19 issues
- 3.6% Distrust/need to have a second opinion
- 3.6% Diagnostics/treatment denial
- 0.9% Lack of governmental support
- 6.3% Difficulty to get an appointment
- 8% Lack of follow-up of patients in adulthood
- 23.2% Limited insurance coverage
- 12.5 % Lack of awareness in HCPs
- 19.6% Lack of NF specialists and/or NF clinics
- 18.7% Lack of information about NF

Top concerns of patients and caregivers

Top concerns of patients/caregivers were fear of existing symptoms worsening and new ones appearing, tumour growth, fear of surgery, and inadequate healthcare support.

- 2.3% Heredity
- 2.3% Comorbidity
- 4.7% Concerns about the disease
- 6% Other
- 7% Diagnostics issues
- 18.6% Symptoms
- 8.1% Therapy and side effects
- 14% Surgery
- 11.6 % General anxiety
- 11.6 % Not getting proper help from the healthcare system
- 14% Tumours

Top QoL topics discussed by patients and caregivers

Disease outcomes with the biggest impact on patients' QoL: disease-related fears, emotional / psychological burden, physical pain, lack of support, appearance and skin issues. Many QoL issues were similar in children and adults (e.g. lack of support), whereas some were related to children specifically (e.g. school issues).

Upon comparing the results on QoL topics most discussed by Canadian patients and caregivers with those obtained from NF1 families from other regions, we identified that QoL aspects vary greatly country-wise and they are dependent on multiple local factors.

Canada
- 15.2% Disease-related fears
- 13.1% Emotional/ psychological burden
- 12.3% Physical pain
- 9.8% Lack of support
- 8.4% Appearance and skin issues

Mexico
- 25.8% Physical pain
- 15.7% Comfort in faith
- 11.7% Family issues
- 9.8% Fear of progression
- 4.6% Emotional support

South Korea
- 53.8% Family issues
- 29% Emotional/ psychological burden
- 5.7% Trying to stay positive
- 4.4% Clothes
- 2.4% Work-related issues