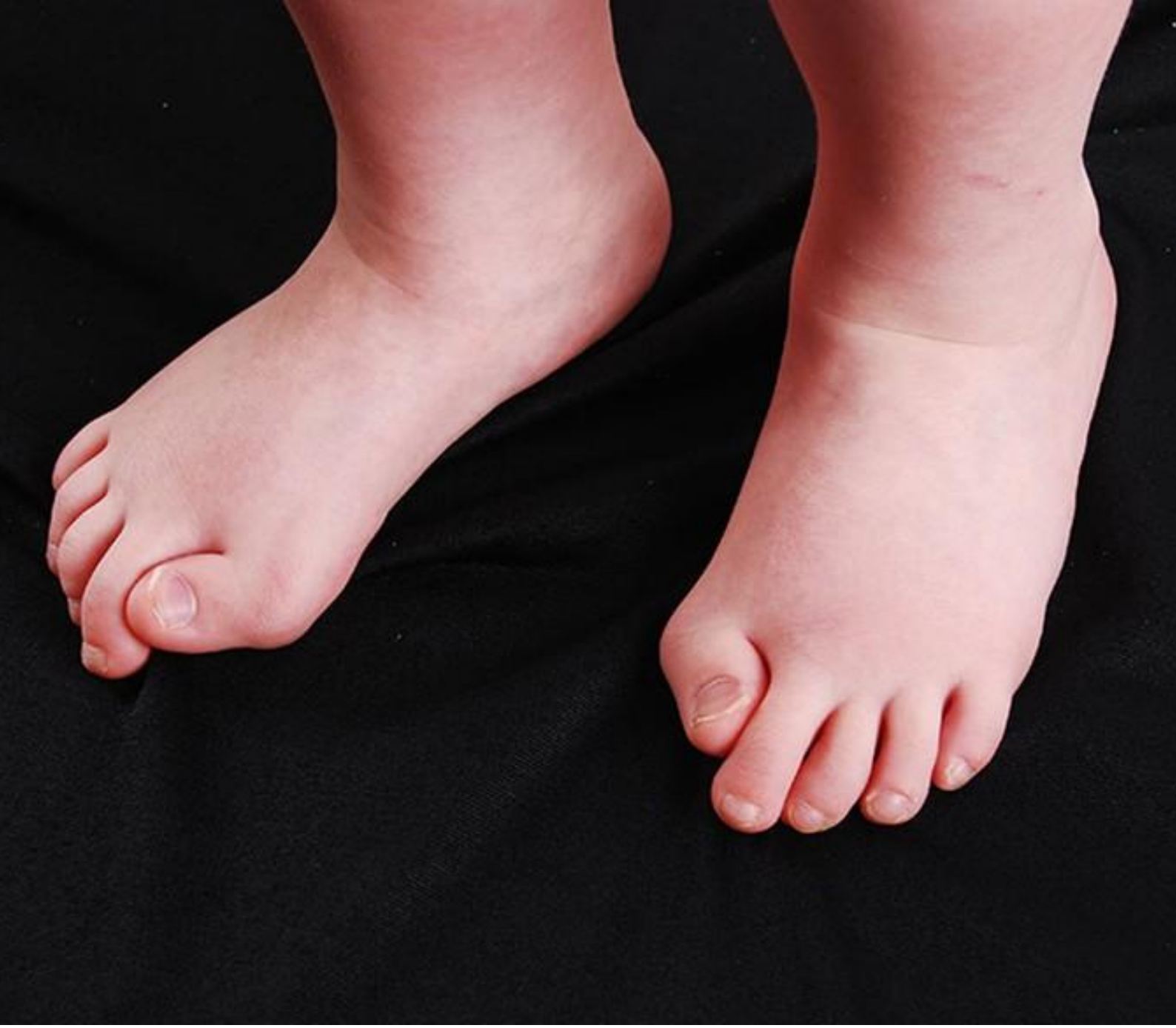

Community Report

Understanding the Needs of People Living with FOP in Australia and New Zealand

Prepared for FOP Australia
December 2025

ACKNOWLEDGEMENTS

Community and Patient Preference Research would like to acknowledge the valuable assistance provided by FOP Australia in raising awareness of, and providing funding for this study.

Importantly, the research team would like to thank the individuals who generously gave their time and shared their experiences of living with Fibrodysplasia Ossificans Progressiva, for the purposes of this project. The study would not have been possible without their support and participation.

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ABBREVIATIONS

Term	Definition
ANZ	Australia and New Zealand
USA	United States of America
BWS	Best Worst Scaling
CaPPRe	Community and Patient Preference Research
FDA	Food and Drug Administration
FOP	Fibrodysplasia Ossificans Progressiva
HCP	Healthcare Professional
HREC	Human Research Ethics Committee
IFOPA	International Fibrodysplasia Ossificans Progressiva Association
IV	Intravenous
NDIS	National Disability Insurance Scheme
PEI	Patient Experience Index
PBS	Pharmaceutical Benefits Scheme
PICF	Participant Information and Consent Form

EXECUTIVE SUMMARY

Background and Key Research Objectives: Fibrodysplasia Ossificans Progressiva (FOP) is an extremely rare and disabling genetic condition causing bone to form in muscles and other soft tissue, leading to ongoing and permanent restriction of movement. This study investigated lived experiences (including physical, emotional, social and financial impact, as well as impact on work and education) among people diagnosed with FOP, and carers of people diagnosed with FOP. Participant experiences with the healthcare system were also explored, to identify unmet needs.

Sample and Methods: This report includes data from a total of 12 participants (adults diagnosed with FOP, and parents of children diagnosed with FOP). To answer the research objectives, one-on-one in-depth exploratory interviews lasting 60 minutes were conducted. The interviews included a quantitative component via sharing of an online link to a survey that examined experiences with the healthcare system (i.e., Patient Experience Index; PEI), using Best Worst Scaling (BWS) methodology.

Analysis: Qualitative interviews were thematically analysed through NVivo software. Data from the patient experience exercise were computed as BWS scores for analysis. Relative scores were converted into absolute measures to form the Patient Experience Index (PEI).

Key Findings: Participants described significant physical and functional impacts from FOP, including frequent flare-ups and progressive bone growth, sometimes sudden and severe, leading many to rely on wheelchairs and specialised equipment, and to engage in extensive home modifications. Symptoms caused major restrictions in life, with participants largely being dependent on carers for basic tasks. Despite participants demonstrating strong resilience, the emotional burden was high, with reports of anxiety, frustration, grief, and social isolation. FOP had also negatively affected financial stability, due to impact on work prospects and out-of-pocket expenses for equipment and home modifications, when not covered by the National Disability Insurance Scheme (NDIS). Carers faced substantial physical demands, emotional strain, and disruptions to their own work and family life.

The PEI exercise revealed a mean score of 55.6, with gap analysis highlighting key unmet needs in the areas of access to effective treatments, time to diagnosis, treatment side effects, and access to other treatment and care. Key frustrations lay with misdiagnosis of FOP, lack of Healthcare Professional (HCP) awareness and allied health support, and navigating the NDIS for funding support. Participants emphasised the importance of ongoing education, holistic care (especially in the adult setting), and further investment and government approval of new therapies for FOP, that are safe and effective.

Conclusions: This research provides important supplementary evidence to inform treatment and funding decisions for current and future FOP therapies. While clinical trials provide valuable data on efficacy and safety endpoints, these often overlook other aspects of care that matter to patients and carers. Adopting a patient-centric approach that considers the broader humanistic burden of FOP on patients, carers, and society is critical for evaluating new therapies, and identifying key areas for improvement within the healthcare system.

BACKGROUND

DISEASE AREA

Fibrodysplasia Ossificans Progressiva (FOP) is an extremely rare and disabling genetic condition causing bone to form in muscles and other soft tissue, leading to ongoing and permanent restriction of movement. It is a progressive disease, and there is currently no cure with very limited treatment available. Most people with FOP experience severe physical disability and have a limited life expectancy. It is estimated that 1 in 1 million people have been diagnosed with FOP, with researchers being aware of approximately 800 cases worldwide.[1] At the time of this research, it is believed there are twenty-one diagnosed cases in Australia and three in New Zealand.

Established in 2015, FOP Australia is a volunteer-run charity that links people with FOP and their families and supporters, within Australia and New Zealand (ANZ). In 2025, FOP Australia received an independent research grant from Ipsen Biopharmaceuticals to engage in research that provides insight into FOP lived experiences. FOP Australia also wished to obtain specific insight into experiences of the healthcare system, to understanding how best to service the FOP community, including facilitating access to therapeutics through advocacy, and with regulatory bodies.

Community and Patient Preference Research Pty Ltd (CaPPRe) has conducted research with people diagnosed with FOP and carers of people diagnosed with FOP in ANZ, on behalf of FOP Australia. The study aimed to characterise, from the patient and carer perspective, the broader effects of FOP by identifying the physical, emotional, social, work/education, financial, and family impacts of the condition. The results of this research will be used to contextualise experiences within clinical care pathways and support structures, which will help in understanding the needs and priorities of people living with this condition, and their carers. FOP Australia also intends to publish results in a peer reviewed journal.

RESEARCH OBJECTIVES

This study aimed to understand experiences of people diagnosed with FOP, and parents of children diagnosed with FOP in ANZ. The key research objectives set out to determine:

- Lived experience and impact on quality of life with a focus on:
    - Physical burden
    - Emotional burden
    - Social burden
    - Impact on work/education/finances
    - Impact on carer
- Treatment received
- Awareness and perceptions of new treatment
- Experiences and perceptions of the healthcare journey
    - Identify key areas of unmet need

OUR APPROACH

METHODOLOGICAL APPROACH

CaPPRe used a mixed methods research approach which incorporated a qualitative and quantitative component. The research approach is outlined below:

1. Team Meeting

CaPPRe facilitated a team meeting with FOP Australia to agree on and finalise the research materials, including the qualitative interview guide and patient experience choice-based experiment prior to submission to a Human Research Ethics Committee.

2. In-depth Interviews Incorporating a Quantitative Patient Experience Exercise

CaPPRe conducted one-on-one in-depth interviews lasting 60 minutes with 12 FOP participants. These interviews were structured to include:

- An initial exploratory component:
    - This section holistically explored lived experiences of FOP including overall burden of disease, management and treatment, and awareness and importance of access to new FOP products.
- A Patient Experience Index (PEI) component:
    - In this component, the researcher shared an online link to a choice based instrument (scripted by CaPPRe in Forsta HX platform), whereby participants completed an exercise using Best Worst Scaling (BWS) methodology, to quantify satisfaction and importance with various aspects of the healthcare journey. This yielded an overall PEI score and an automatically generated individualised report which was discussed in the next section of the qualitative discussion.
- Discussion of PEI results:
    - A detailed discussion based on gaps identified in the PEI report, with a focus on understanding the areas considered highly important, yet for which there was low satisfaction. Reasoning for this dissatisfaction was explored, along with suggested improvements. These results will help provide insights into areas where resources should be prioritised within patient support.

RESEARCH DESIGN

QUALITATIVE DISCUSSION GUIDE

The qualitative discussion guide was designed by CaPPRe in collaboration with FOP Australia and was used to steer and encourage a smooth flow of conversation during the interviews. The discussion guide was structured so that spontaneous feedback on key topics was obtained, to allow for the evaluation of authentic and impartial opinions. Prompted questions were also asked to ensure all research objectives were addressed in detail.

PATIENT EXPERIENCE EXERCISE USING BWS METHODOLOGY

BWS is a survey technique that takes advantage of an individual's ability to reliably identify extremes ('best' and 'worst') in a set of three or more items, with respect to a continuum such as satisfaction and importance. This exercise elicits a discriminating ranking of items free of scale bias. Additionally it is simple and intuitive for participants to complete.

In this research, the BWS task was used to measure the satisfaction and importance of different aspects of the healthcare pathway, which were defined by a master list of 11 domains (i.e., domains were developed in previous CaPPRe research through expert review, qualitative research, pilot testing and statistical analysis to form a validated 'Patient Experience Index'). The domains were systematically divided into 11 sets of 6 according to an experimental design, resulting in a BWS exercise containing 11 scenarios. For each scenario, participants were asked to consider the subset of 6 displayed domains and select the best and worst domains across two dimensions, namely satisfaction and importance. Data collected detailed how important each domain was to an individual, as well as how satisfied they were with each domain.

Table 1 lists the 11 domains and the descriptions that were presented to participants. Figure 1 provides an annotated example of a BWS choice task.

Table 1: BWS Healthcare System Domains

	DOMAIN	Description (Hover-over text)
1	Time to diagnosis	The length of time from developing symptoms through to being diagnosed – whatever this looked like for you.
2	The quality of information available about your condition and care	Having clear, concise, relevant information in a format that works for you (e.g., provided to you by your healthcare team/online/Apps/podcasts).
3	Your involvement in decision making	How involved you are in decisions about your treatment and care, e.g., when selecting specific medication and/or when developing a treatment plan
4	The quality of your healthcare team – access to your key healthcare professional/s, consistency of care, and their communication with you and between each other	Suitable access to your key healthcare professional (e.g., haematologist), at regular intervals that you feel are most beneficial to you or in acute situations where urgent access is required. Being able to see the same trusted healthcare professional/s on-going for your treatment and care. How well your needs are met in any interactions with your healthcare team (including doctors, nurses, care coordinators). The extent to which different members of your healthcare team (e.g., haematologist/GP/clinical nurse specialist) communicate with each other about your condition and care. They may be healthcare professionals within the same service or in different services.
5	Treatment logistics	The broad impact that following a treatment and care plan has on you., i.e., day-to-day difficulties of arranging and attending treatment sessions.
6	Access to, and effectiveness of, medication	Your access to medication for your condition. How effective the medication prescribed by your healthcare professional/s is in treating your condition.

7	Side effects of medication	Side-effects you may experience from medication prescribed by your healthcare professional/s.
8	Monitor & identify progress/deterioration	The ability to monitor day-to-day and long-term changes in your physical and overall wellbeing, for yourself, and by your healthcare professionals (e.g., joint stiffness) and adjustments to treatment and care based on this.
9	Access to other treatments/services (including a care coordinator), to support physical health, mental health, overall wellbeing (holistic approach)	Other services could include seeing a psychologist or exercise physiologist. Complementary treatments could include acupuncture, massage, mind-body techniques, the ability to monitor day-to-day and long-term changes in your physical and overall wellbeing, for yourself, and by your healthcare professionals (e.g., joint stiffness) and adjustments to treatment and care based on this, and management strategies for increased wellness (e.g., access to dietitians/physiotherapists/occupational therapists/psychologists). Having someone who is assigned to you (e.g., social worker or peer support worker) to help you navigate the healthcare system and offer emotional support and guidance.
10	Support for your 'support person'	Information/websites specifically for significant others (e.g., spouse, partner, friend etc) and support groups where family members/friends can talk with others in similar situations.
11	FOP-related costs	The overall impact that having FOP has on your financial wellbeing, e.g., how much you are out-of-pocket, and the impact of loss of income.

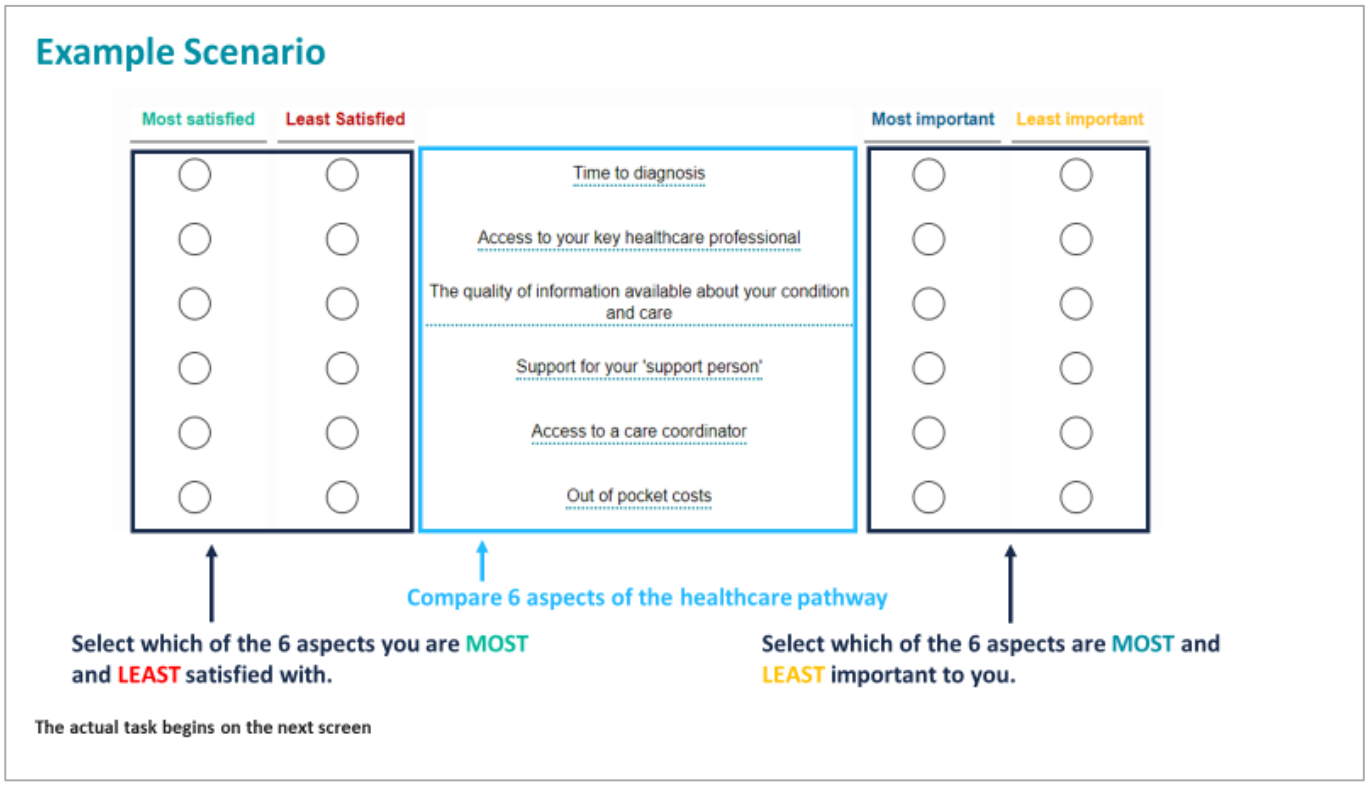

Figure 1: Annotated Example Scenario of a BWS Choice Task

RESEARCH SAMPLE

A total of 12 participants diagnosed with FOP, and parents of children diagnosed with FOP, participated in this research. Inclusion and exclusion criteria are outlined in Table 2.

Table 2: Participant Inclusion/Exclusion Criteria

Inclusion criteria	Exclusion criteria
• 18 years and over • Australian or New Zealand citizen/permanent resident • Diagnosed or parent of a child diagnosed with FOP • Provision of consent within the Participant Information and Consent Form (PICF)	• Participants working for a pharmaceutical, medical device or vaccine company

RECRUITMENT

All participants were recruited via FOP Australia, who were instrumental in raising awareness for and distributing information about the research. Email addresses of interested participants were forwarded to CaPPRe who then contacted these people with a registration link. Potential participants could click on the link anonymously to find out more information about the research. If they wished to participate, they were asked to complete the online PICF. Participants had the option of printing the consent screen (or printing/saving to PDF) if they wished to retain a copy for their records.

Upon providing consent, participants completed an online screening survey to check for eligibility. This screening survey identified FOP diagnosis, after which participants were invited to take part in the qualitative interview.

DATA COLLECTION

Interviews were conducted between 22nd August and 5th November 2025, with four interviews conducted face-to-face in Melbourne at the 2025 FOP Australia Conference (held on 22nd-24th August), and the remainder via video conferencing. The decision was made to close fieldwork with a sample of 12 participants reflecting both the rich information acquired from these interviews and the volunteers who were available during the fieldwork period. FOP Australia is mindful that some community members may not have been aware of, or had the opportunity to participate in this research and wishes to affirm that every experience within the community, whether captured in this research or not, is equally important and valued.

All interviews were digitally audio recorded and transcribed with identifiable information anonymised. Participants also completed the Quality of Life Questionnaire, EQ-5D-5L (a standardised instrument for describing and valuing health) in the online screening survey, prior to scheduling the exploratory interview.

In appreciation of their time and contribution to the research, participants received a payment to the value of AUD 100 in the form of an electronic gift card.

DATA ANALYSIS

Qualitative Analysis

Qualitative interviews were analysed in a rigorous and methodical manner by an experienced qualitative researcher using the methodology of thematic analysis. Thematic analysis places emphasis on the identification, organisation, and interpretation of themes (i.e., patterns of meaning) and is therefore an appropriate approach when determining participant views, knowledge, experiences, or values.[2]

NVivo software was used to assist in the sorting and organising of the themes identified in the interviews. NVivo software enables the research team to engage deeply with the data and develop a comprehensive coding framework when examining the large amounts of text generated in qualitative interview transcripts. This framework ensures meticulous and systematic analysis and allows for review by multiple researchers for quality assurance and sharing of information. Furthermore, researchers are able to record memos within the software throughout the analysis process to capture interesting aspects and emerging themes across the interviews.

BWS Analysis

Data from the BWS exercise were computed into BWS scores for analysis. The BWS exercise yielded scores reflecting the relative hierarchy of each domain versus another. The BWS scores were calculated for each domain by subtracting the number of times it was chosen as worst (least satisfied/important) from the number of times it was chosen as best (most satisfied/important), divided by the number of times it was shown throughout the task.

$$BWS\ score = \frac{most\ -least}{times\ appeared}$$

BWS scores were then mapped onto a scale ranging from 0 ("Not satisfied at all"/"Not important at all") to 10 ("Completely satisfied"/"Extremely important") describing the level of satisfaction and importance. These rescaled scores allow direct inference of how satisfied/important each individual healthcare domain was, rather than just their relative ranking.

Best-Worst Scaling (BWS) scores range from -1 to 1 and represent the relative ranking (ordering) of the domain

- A negative score indicates the domain was chosen as worst more often than best
- A positive score indicates the domain was chosen as best more often than worst
- A zero score indicates the domain was chosen as best and worst an equal number of times OR was never chosen

Rescaled scores range from 0 to 10 and represent the individual level of satisfaction and importance experienced. The scale was labelled at each extreme as follows:

- 0 = "Not satisfied at all"/"Not important at all"
- 10 = "Completely satisfied"/"Extremely important"

Patient Experience Index

Standard BWS scores cannot be used to build an index that is comparable between groups of participants because the scores represent a relative ranking. CaPPRe researchers have developed a new method to convert these scores from relative to absolute measures which can be combined to form a Patient Experience Index (PEI). An index was built to measure the overall satisfaction of the healthcare system among people with experiences of living with FOP in ANZ. The PEI is a combined score of the 11 BWS domains, accounting for both satisfaction and importance, and ranges from 0 to 100.

STUDY CONDUCT

While FOP Australia received a research grant from Ipsen Biopharmaceuticals to engage in this study, Ipsen Biopharmaceuticals had no role in the conception, design, data collection, analysis or reporting of this research. All research activities were planned and conducted independently by FOP Australia.

This study was conducted in compliance with the research protocol, the International Society for Pharmacoepidemiology (ISPE) Guidelines for Good Pharmacoepidemiology Practices (GPP), and the laws and regulations of Australia, such as the Medicines Australia code of conduct. In addition, the research was conducted according to the Research Society's Code of Professional Behaviour. All ethical considerations as outlined in the National Statement on Ethical Conduct in Human Research were considered and applied to the conduct of this research.

All written material used in the qualitative and quantitative component of the research received ethics approval by the HREC, Bellberry Limited (approval letter dated: 20th August 2025; Application number: 2025-08-1245). The review and approval by the Ethics Committee ensures the study was conducted according to the non-interventional design and protected the rights of participants (including data protection and participant privacy).

KEY FINDINGS

This report outlines the key research findings, drawing upon both the qualitative component of lived experiences, and quantitative outputs from the PEI online survey.

Ensuring credibility of these qualitative results has been prioritised through the accurate representation of first-hand experiences of carers and people diagnosed with FOP. This research has been conducted in a small sample of participants - it is therefore important to consider that the generalisability to a broader population arises from a conceptual transferability of the themes explored, rather than statistical representativeness of the sample.

Furthermore, the majority of quotations illustrating participants' lived experiences have not been included in this report to minimise risk of identification. The small number of quotations that have been retained, are based on researcher and FOP Australia's judgement that they will not identify the individual. Patient quotes are presented in *blue* and carer quotes in *purple*.

BACKGROUND ON PARTICIPANTS

Due to the small number of people affected by this ultra-rare condition in ANZ, it was decided to only include a brief summary of participant demographics in this report, to enhance participant anonymity and confidentiality.

Adults diagnosed with FOP and parents of children diagnosed with FOP took part in this research. In several cases, parents or carers of adult participants also joined and contributed to adult interviews. There was a wide variation in age amongst participants, and a broad geographic representation across Australian states. Most participants resided in a metro area.

Adult participants diagnosed with FOP were generally not working (with some exception) as a result of restrictions resulting from their condition. Children, and a few younger adults, were generally engaged in studies or employment.

FEEDBACK ON LIVED EXPERIENCES

The following section provides insight into the impact of living with FOP on quality of life. In particular, burden of disease was explored in terms of the physical, emotional and social impact, as well as on work/education and finances. The effect of FOP on family - in particular, key carers - was also explored.

PHYSICAL IMPACT

While there were variations in severity, nearly all participants had experienced a profound negative physical impact due to FOP. Participants had encountered vast restrictions due to symptoms including flare-ups and/or bone growth, which had led to loss of movement, especially when bone growth had occurred over joints in the body, such as the elbows or knees. Participants reporting a more minor impact, nonetheless had experienced negative physical effects, just to a lesser degree than described by others. Participants had usually experienced worsening of disease over time. However, several noted that disease progression was unpredictable and could be very sudden, with new bone formation causing severe stiffness or permanent immobilisation in that area of the body. This has enormous implications for people's mobility, ultimately

leading to an increased - or for some, complete - dependence on carers. The main symptoms experienced by people living with FOP and outcomes of these are outlined below:

Symptoms

Flare-ups

All participants had experienced flare-ups/flares (terminology was used interchangeably) which was a key physical burden with FOP. Flare-ups commonly marked the onset of further bone formation, resulting in additional restriction of movement and a permanent reduction in function. Flare-ups commonly occurred following minor injury resulting from a fall (particularly when young) or knock, or from surgical intervention (i.e., removal of lumps). However, flare-ups could also occur spontaneously, with no apparent trigger.

Participants defined flare-ups as swelling, lumps and nodes that occurred on various parts of the body including the back, neck, shoulders, head, elbows, ankles and legs. Flare-ups caused pain, sensitivity and intense fatigue, and were described by some as a 'burning sensation' (i.e., red, inflamed and hot to the touch).

> *"I had to go to hospital in an ambulance because the pain was so bad it wouldn't stop. It was in both my ankles [which] had swollen up like a golf ball, so they were massive."*
>
> *"Basically, my body will just shut down, and I go to sleep, then wake up and then the bones do [their] thing and fuse."*

The frequency and duration of flare-ups varied considerably among participants. Data from the quantitative research revealed that over the preceding three years, the average number of flare-ups experienced across the sample was 114 [please interpret this figure with caution due to the average (i.e., mean) being sensitive to a small sample and individual variation within the sample (i.e., range was 1-1000 flare-ups for the three year period). The greatest annual frequency of flare-ups experienced across the sample, averaged at 34.3 flare-ups, most commonly occurring during the teenage years (mean: 16 years, median: 15 years; again, please keep in mind the small sample size in interpretations).

Bone growth

All participants had experienced bone growth, where bone tissue had formed in soft tissues such as the muscles and ligaments. The extent of bone growth (i.e., heterotopic ossification) had been confirmed with a CT scan in 75% of participants. Bone growth commonly occurred at the onset of flare-ups, however could also develop independently. How and where bone growth occurred led to stiffness or immobilisation of that part of the body, and was particularly problematic when it occurred in joints involved in movement. Most participants reported significant loss of mobility, with a limited number being permanently fixed in certain positions (e.g., seated position with bent legs). There were variations for people regarding the areas of the body that had been most affected by FOP. Several had been impacted in the legs which had led to requiring a wheelchair, while others had experienced more mobility issues in the arms leading to logistical difficulties in completing tasks (e.g., getting dressed independently).

Malformed Toes

Nearly all participants mentioned having malformed toes, often present at birth, where toes were curved inwards, missing, or appeared short. This malformation had commonly assisted in diagnosing FOP as this represents a near-universal sign of the condition.

Locked Jaw

Several participants reported having had experienced progression in the jaw area. For some this had been gradual, yet had led to a severely reduced capacity to move or open their jaw. Some of these individuals mentioned that a locked jaw had led to swallowing and eating difficulties necessitating a transition to eat softer foods, or nutritional support via nasogastric feeding (i.e., gastrostomy tube). There was also limited reference to greater difficulties in speaking (i.e., being able to form certain words), due to difficulties in opening the mouth.

Difficulties Maintaining Weight

Many participants spoke about difficulties in maintaining weight, with some having experienced weight loss, especially those with a fused jaw, or following specific setbacks (e.g., undergoing surgery).

Dietary measures were commonly adopted to minimise weight loss. For example, participants spoke about eating a lot of food, or food that could be consumed easily (e.g., smoothies, thin food such as pringles) and using supplements (e.g., PediaSure shake) to increase their intake and nutrition. Some carers balanced oral hygiene with 'favourite foods' (e.g., pancakes with butter, maple syrup, ice cream) to encourage food intake and avoid weight loss.

Conversely, a smaller number of participants mentioned some weight gain resulting from medication, particularly corticosteroids (e.g., Prednisone).

Outcomes of FOP Symptoms/Progression

The negative impact on quality of life resulting from FOP symptoms and disease progression had been profound for many participants. Disease burden was high due to experiencing:

Loss of mobility

Approximately half of our FOP participants had lost most mobility, the remainder still had some ability to walk. However, a few mentioned only being able to walk short distances (e.g., mentions of 10 meters) or for a short period of time (10-15 minutes). Walking often caused fatigue and for some, pain and risk of falls. Most of the participants who were still walking mentioned there had been a change in their gait resulting from FOP, including dragging of the foot, limping, slanting while walking, or walking on their toes.

People who had experienced bone growth in the neck, chest, back and arms also had impaired movement, with some no longer being able to turn to the side or move their head up and down, leading to some visual impairment (i.e., lack of peripheral vision or ability to look down). There were mentions of limitations in shoulder movement due to arms being fused against the body, or bone formation preventing forward movement of the arms. Further fusing had occurred for people in the knees and hips, also restricting mobility. These physical impacts had led to broad functional consequences on daily life, with a small number of participants, referencing the inability to drive.

Reduced/loss of independence

Bone growth was a key burden with FOP, causing loss of independence and subsequent deterioration in quality of life. All participants in our research required assistance from carers in some form, to manage challenges with FOP. For some, this meant complete dependence on others, as these participants could not use the toilet, wash themselves (e.g., shampoo or brush their hair), dress themselves (i.e., put on clothes, socks or shoes), get out of bed or feed themselves. These duties fell to parents or support workers (acquired through the NDIS).

Other participants had been able to maintain a certain level of independence (e.g., walking, attending studies, shopping, work), however required assistance with some tasks that had become difficult due to restricted movement. Strategies for maintaining independence were implemented where possible, such as carers placing items (e.g., drinks) in the fridge where they could be reached, or wearing slip on shoes which did not require lacing.

> *"We just adapt, just adapt. There is nothing off the shelf really…, it's whatever we come up with."*

Restrictions in activities/hobbies

Loss of mobility had prevented capacity to engage in activities and hobbies previously enjoyed, such as playing sport (e.g., cricket) or walking.

> *"It's really hard, I can't do the things that I used to enjoy, that I did like before I got into a wheelchair and all that. So, I used to walk for like hours and miles, and used to be independent, and now the condition just made me like worse and I can't do all of that."*

Logistical Burden of FOP

The need for equipment and home modifications

Due to the physical restrictions caused by FOP, most participants mentioned requiring some form of equipment to assist with daily life. This included wheelchairs which were mostly electric and customised (e.g., custom seating, footrests and tilting/inclining positions, custom positions for the wrists, hand controls). Other equipment mentioned included hospital beds (with a pressure relief mattress), shower chairs, tables, grab sticks for picking things up off the floor, walkers used for greater stability during transfers, use of a ventilator (at nighttime), and eye gazing equipment.

Participants also spoke of necessary modifications made to the home, including removal of all steps, upgrading bathrooms (mentions of installing a full wet floor to be able to roll in and out of the bathroom using a shower chair), installing ramps and bars/rails, and accommodating for wheelchair access (e.g., installing hardwood floors, widening areas of the house including bedrooms, front doors and hallways).

Travel

Due to limited mobility, travelling to and from medical appointments often posed logistical challenges for participants. This was especially pronounced for those who relied on a wheelchair, as they frequently needed to organise specialised transport such as hiring a taxi with wheelchair access. Going on holidays also required much planning, time and coordination.

Time in hospital

Most participants reported they had not spent extensive periods in hospital as a direct consequence of FOP. Visits to the Emergency occurred intermittently, often for medication and monitoring, following accidents such as breaking bones due to falls, or when experiencing severe flare-ups, which had caused substantial weight loss or impacted on breathing. Some participants mentioned some longer stretches in hospital (i.e., three months, three weeks, regular stints lasting up to a week at a time). There was reference to experiencing adverse events while on a drug trial (including hair loss, weight loss and illnesses) which had led to extended time in hospital.

EMOTIONAL IMPACT

Participants in this research demonstrated resilience and acceptance of their or their child's condition, showing admirable attitudes in striving to maintain a sense of normalcy, and to maximise their current circumstances. There was clear evidence of a strong mental approach and mindset towards the condition, with a view that things could be worse.

> *"Most of the time I sort of deal with it… like there is always someone worse off in the world than you. Like I try and see that from their perspective."*
>
> *"You've just got to keep going, My attitude makes the biggest difference really."*

While people mentioned having 'good and bad days' and certain periods of the FOP journey being worse than others, the emotional impact of living with FOP was clear, with participants conveying sadness about their situation. For people living with FOP and carers, anxiety was evident, particularly when thinking about the uncertainty of the future, due to the high likelihood of disease progression. Clear disease progression had been evident and concerns were raised regarding what further progression will mean. Several participants were using anxiety and/or anti-depression medication to help manage the emotional impact of living with FOP.

> *"There are ups and downs… It's okay like most of the time, but yeah, sometimes thinking about too much, I do feel a little bit of like anxiety and depression."*

Frustration was evident, regarding the physical restrictions in life which hindered functioning. There were some mentions that the 'teenage years' were particularly frustrating as physical limitations hampered participation in 'normal teenage activities' such as going out with friends or playing sport, causing people to feel that they were missing out on life. There was also some evidence of being victimised due to the challenges caused by FOP, another example of the emotional burden associated with this condition.

"Just like reflecting back on the past, the things I used to be able to do and the things that I can't do now…very frustrating and annoying. Yeah, I used to think, you know, why us? Out of everyone."

Some carers acknowledged the challenge in really knowing the psychological impact of FOP on their child. They reported that outwardly their children were tolerant, claiming to be 'fine,' however parents suspected that the emotional burden of FOP may be greater than that communicated by their children, especially in terms of what the future will hold.

Others mentioned feeling frustrated with the lack of community understanding regarding FOP. For example, in a social context, people can be quick to judge a person with a disability (e.g., being in a wheelchair) and avoid engagement. There were mentions that people in the community sometimes assume that the physical restrictions visible with FOP also translate to intellectual disability.

SOCIAL IMPACT

Overall, participants had been heavily impacted socially due to living with FOP. Several participants spoke of a supportive environment, peer acceptance with some close friendships formed during the school years. However, severe physical restrictions had contributed to friendships breaking down and being lost, in part due to logistical difficulties in attending social outings (e.g., going out to a restaurant), with participants commonly being placed in the 'too hard basket.'

Living with numerous flare-ups had commonly led to participants feeling too unwell to attend social occasions. Some had deliberately chosen to isolate themselves, especially from new people, due to finding it tiring to constantly explain their condition, or feeling frustrated by evident assumptions that a physical disability also equates to an intellectual disability.

"I'm fairly isolated myself, I don't have many friends. But that's more of a choice than anything. Just having to try and explain everything to everyone you know every time, it gets a bit exhausting. So, yeah, socially I'm not much of a social butterfly."

There was also mention of COVID 19 having led to social isolation due to the need to minimise risk of illness, which had caused long term impact on friendships.

IMPACT ON WORK AND EDUCATION

Adult participants had generally not been able to work due to physical restrictions caused by FOP. Only a small number were working full time (mainly from home) or on a casual basis. There was mention of previous employment by a limited number of participants, however due to experiencing disease progression, this had stopped capacity to work altogether.

In thinking back to the school years (for adults) or discussing current school experiences (carers of children), participants spoke about not being able to engage in many school activities due to the need to avoid injury and resulting flare-ups. Most had not been allowed to participate in sport. School attendance had been

impacted for many where time off school had been required for hospital visits (e.g., for flare-ups), HCP appointments, and immense restrictions causes by the disease (i.e., losing the ability to walk).

However, participants were generally appreciative of the support provided in the school environment which had included accommodations; including teacher's aide, accompanying registered nurse to ensure all needs were met (e.g., administration of medication), additional breaks while completing school exams, not having to carry heavy textbooks around and installation of equipment (e.g., speaker system in the classroom for hearing the teacher). As mentioned, several participants reported positive experience during school years, having been accepted by peers and forming close friendships.

"The school was so accepting…to join and participate…which was fantastic."

FINANCIAL IMPACT

Nearly all adults in this research had been unable to work, highlighting the rarity of stable employment among individuals with this condition. There was also acknowledgement that financial opportunities had been significantly constrained by the physical limitations associated with FOP for those who could engage in work. Overall, people living with FOP appeared to be highly reliant on financial support from family members, as well as from disability funding and services (i.e., Centrelink and NDIS).

IMPACT ON CARERS

The significant functional limitations associated with FOP meant that participants were often highly dependent on others for support. Carers showed considerable resilience, and worked to maintain as much normalcy as possible for their child or partner, providing physical and emotional care which stemmed from their deep attachment and commitment. However, this ongoing support came with substantial personal sacrifice.

"[Mother] is helping with basically daily things and does the cooking, the laundry. As a family of a person who grew up with FOP it was obviously not easy, especially for the parent."

"There's the commitment and the sacrifice socially as well."

The physical burden for carers was high, particularly for those caring for a person with advanced progression. Physical assistance included toileting, showering, dressing, feeding, lifting in and out of bed and/or wheelchair, co-ordinating/taking to and from/attending doctor appointments, and helping with medications. Carers also described a heavy emotional toll, often marked by grief at diagnosis, throughout disease progression and when contemplating an uncertain future.

"It was quite heartbreaking to find out…and just to think what sort of life [child with FOP was] going to have."

There were mentions of relationship strain, with some marriage breakdowns due to challenges of raising a child with FOP. Positively, divorced parents had learned to co-parent well. Feelings of isolation had occurred due to loss of friendships, and a lack of knowledge among HCPs.

> *"It's been really, really hard, really stressful. We haven't had any support. The specialists have been useless, they don't know what is wrong or how to deal [with a] a flare-up. Yeah, it's just been a nightmare…I haven't seen my friends for years."*

Financial strain was also evident for carers, including loss of earnings, time off work, or deciding to retire early due to the high dependency of their child. Carers spoke of high financial outlay for home modifications, required equipment, purchase of suitable cars, additional expenses when going on a holiday, which added to financial burden.

> *"I've had to have so much time off work. Work's been sort of harassing me for taking so much time off."*

Carers also spoke of challenges in balancing the needs of their child with FOP, with those of other family members. A few parents had faced judgement by others (e.g., HCP fearing for the safety of the child brought into hospital with swelling to the side of the head). Feelings of guilt were also raised by a small number of carers, as they had the belief they had not done 'enough' for their child with FOP.

> *"I feel guilty about working and then feel guilty about not being at home to help [child with FOP]. I feel like I've let [child with FOP] down. I feel like I should have done more, if I had had more information I might have been able to do something more, got onto some different medicines sooner."*

MANAGEMENT OF FOP

KEY HEALTHCARE PROFESSIONALS

There were numerous mentions that it had been difficult for participants to come under the care of HCPs who had expertise in FOP, especially in the early stages of the condition. Some had reached out to HCPs based in the United States of America (USA) who were perceived to have more experience in managing FOP than their Australian counterparts.

> *"Finding the right people within the system, [that] seems to be the obstacle."*

However, most participants had eventually come under the care of a specialist who they highly regarded and praised for their experience and knowledge in this therapy area. Key specialist types included rheumatologists, general physicians, and endocrinologists. People who were regional/rural based, mentioned further difficulties in accessing specialists in these areas.

Patients were seeing their key HCP every 3 to 6 months for general check-ups and monitoring (e.g., physical examination including assessing movement, blood tests, blood pressure, weight, lung capacity testing, monitoring of medications such as liver functioning), or more frequently when required (e.g., when experiencing issues with flare-ups, locked jaw).

Allied HCPs were also seen for FOP, including psychologists, physiotherapists, neurologist, geneticists, registered nurses, dieticians for nasogastric nutrition support, occupational therapists, speech pathologist, and specialist dentists (due to the care required with fillings or teeth extractions to prevent flare-ups in the jaw area). GPs were also seen, primarily for the management of prescriptions and mental health.

DIAGNOSIS OF FOP

There were variations in age of diagnosis for participants in this research. Nearly all had been born with early signs and symptoms (i.e., malformed/improperly formed (i.e., stubs) toes, nodes on the head, stiffness in the neck), however due to the little known about FOP at the time, had not received the correct, official FOP diagnosis for several years, which had caused substantial frustration for many.

> *"Leading up to diagnosis was a very, very frustrating time… the podiatry people, 'No, we don't need to do anything about [the] feet yet.' So, they didn't really want to know anything about it."*

Some participants spoke of symptoms developing during the toddler years with intense lumps and swelling occurring following falls. Others had developed symptoms in infants or primary school or early teens, including flare-ups following immunisations (described as calcifications at the injection site), fatigue, lumps, pain (e.g., in hips), stiffness (e.g., in neck). A small number had been diagnosed at a later age (later teens/early adult).

Upon seeking medical advice, HCPs commonly performed biopsies on lumps. Some participants mentioned undergoing X-rays, ultrasounds, CT scans and MRIs, with most eventually receiving genetic testing (i.e., largely confirming a classic mutation). A small number of participants mentioned that at the time, blood tests were sent to the USA for diagnosis, with a 2 year turnaround time for results.

Several participants mentioned initially having been misdiagnosed, and others spoke of experiencing a long wait time before obtaining an official diagnosis, as specialists could not confirm the underlying cause.

> *"Like lots of people…gave me so many new names, so many things, and all of them were just interested in like seeing what's wrong with me, no one like cared…they were just like very amazed to see this new disease. Like I was an experiment mostly for them."*

> *"[Child with FOP] had been having like what would now consider classic FOP signs, but nobody sort of connected the dots and knew what they were."*

Several participants spoke about having undergone initial surgery to remove lumps, however, this had triggered bone growth, scar tissue formation, recurrence of lumps, and further disease progression.

> *"They took the lump out…then a few months later they sort of went, 'Oh oops no, we shouldn't have operated'….so they shouldn't have operated because that will just, like make it get worse."*

TREATMENT RECEIVED

During the FOP journey, participants had generally not accessed treatment that was specifically indicated for FOP, due to no treatment being available until very recently. Instead, regular use of pain relief (e.g., analgesic such as paracetamol and anti-inflammatories such as ibuprofen or piroxicam) and corticosteroids (to treat flare-ups; e.g., Prednisolone) were commonly taken.

> *"I haven't really been on any medication or anything like, not for FOP, other than if I start having a flare-up…I will like go on steroids straightaway."*

In September 2025, the first ever treatment for FOP (palovarotene) which slows disease progression was made available in Australia via the Pharmaceutical Benefits Scheme (PBS). Several participants in this research had experience with palovarotene (through clinical trial access). Other participants had experience with new potential therapies either through a clinical trial or off-label use (i.e., tofacitinib: Janus kinase (JAK) inhibitor indicated for rheumatic conditions; imatinib: tyrosine kinase inhibitor (TKI) used off-label for control of inflammatory and hypoxic flare-ups).

Other supportive medication mentioned by participants included:

- Pamidronate (bisphosphonate) infusion: for symptomatic relief for flare-ups
- Celebrex: for pain and inflammation
- Augmentin: antibiotics
- Morphine: for pain
- CBD oil/medicinal marijuana

Key Benefits with Treatment

Participants mentioned experiencing benefits with supportive medication taken for FOP in terms of providing pain relief and assistance in managing flare-ups.

Medications developed specifically, or used off-label for FOP were showing promising efficacy for some participants. Specifically, there was feedback that palovarotene reduced flare-ups, and that flare-ups noticeably increased when treatment was stopped by the Food and Drug Administration (FDA) who issued a partial clinical hold for patients under 14 years, due to a potential side effect of premature physeal closure (PPC)[3].

> *"I believe that [child with FOP] got this massive, new bone growth down [the] neck when…taken off it, when it was paused for kids that age. It wasn't until… 14, that we could*

get…straight on, but that bone had already formed. I wouldn't even want to think of what it would have been like if [not] on palovarotene."

Encouragingly, there was anecdotal evidence of reduction in flare-ups and disease progression with tofacitinib.

"[Tofacitinib] That seems to be working... [Regarding flare-ups] It not only stopped it, it kind of retreated a little bit…[there hasn't been] any significant bone growth or any new bone in probably six to eight months."

Key Challenges with Treatment

A lack of efficacy in controlling flare-ups and bone growth was a key challenge mentioned for palovarotene. Side effects were also experienced, including weight loss, stunted growth (for a child diagnosed with FOP), hair loss, dry and itchy skin, burns (at high chronic doses), lethargy, and reduced immunity, leading to illnesses. There was also mention of some initial difficulties in establishing the optimal dose for chronic use.

"[Child with FOP] had side effects from it…but we've been able to get…to a sweet spot with the medication [at] a chronic dose without it having side effects."

For imatinib, there was also mention of lack of efficacy, impact on the immune system, and difficulties swallowing this oral medication. This had led to discontinuation of the product.

Weight gain, difficulties sleeping, and impact on mood, including irritability and mood swings were side effects mentioned with corticosteroids. High cost was mentioned with supplementary treatment, CBD oil.

"[Corticosteroids] They make you hungry which makes you put on weight. You can't sleep, normally the first day it's okay, second day you basically just feel wired, like it's not a great feeling."

"I tried…a little bit of CBD oil years back. I'd like to give that another go but it's just very expensive."

AWARENESS AND PERCEPTION OF NEW FOP TREATMENT

Most participants were aware of a new treatment for FOP that has received approval in Australia (i.e., Therapeutic Goods Association approval of palovarotene, with recent PBS listing) or products undergoing clinical trials which have showed promising results overseas (e.g., mention of garetosmab).

[Garetosmab] "The results from this drug has just been amazing and that was the one that I always wanted."

People expressed gratitude for growing interest in this rare condition, displayed by greater awareness among HCPs compared to in the past, and greater investment in treatment by pharmaceutical companies. Participants expressed hope that there would be several FOP treatment options available in the future. This would mean that combination therapy could target specific components of the disease (e.g., specifically treating flare-ups or bone growth) for better outcomes.

> *"It wouldn't be one initial drug on its own. That could be a cocktail of drugs…one stopping flare-ups, one slowing bone growth, all these different things working together. That would be amazing."*

The Importance of Accessing New FOP Treatment in ANZ

All participants were of the opinion that the Australian and New Zealand governments' approval and funding of new FOP treatment is of extreme importance. Some frustration was expressed regarding longer wait times in ANZ for products to become accessible compared to other countries, such as the USA.

Participants also felt that access to treatment developed specifically for FOP will lead to improvements in quality of life, with the hope of slowing disease progression and subsequently the loss of mobility and independence. Early access, ideally upon diagnosis, was considered optimal to minimise progression from onset and thereby improve disease prognosis. The teenage years were also noted to be particularly high risk for disease progression, due to developmental/hormonal changes.

> *"[People with FOP] might not have a lot of hope and they'll be very restricted at a very early age and what hope do they have if there is nothing out there that can make a difference for them?…I think it's very important [for] these submissions to get support from government, to help people, because it will make a big difference to people's lives."*

> *"And then about a year ago we started tofacitinib which was proved…that it was reducing flare-ups. I was very keen to get [child with FOP] on it during this sort of puberty time, because a lot of evidence has shown that the hormones…can start setting off another flare cycle or it can make FOP again quite active."*

Participants voiced the importance of having a variety of treatments approved for FOP to offer choice. It was acknowledged that due to the heterogenous factors involved in managing and treating FOP (e.g., status of progression/bone growth, age, and risk profiles), there was likely to be substantial variation in terms of expected eligibility and efficacy for individuals.

> *"I think we need more options on the market…without a doubt. Because I don't think it's a one size fits all."*

> *"Even with something that we think may not be as effective as what we would like, I still think that having it available for people to use if that's what they choose, is ultimately the most important thing, because I think people should be able to have choices."*

Government funding of FOP treatment was considered essential. Participants voiced that new products are likely to be prohibitively expensive, with most people unable to pay for treatment themselves. As mentioned, most adult individuals living with FOP had low financial prospects. Carers had also been financially affected by missed work opportunities or expenses involved with ongoing management of FOP.

> *"I don't have to tell you that, some of these things can be exorbitantly expensive to the point where it's not within reach. Even people with…relatively decent incomes, it requires government support."*
>
> *"For funding, it's very important for the government to actually fund most of it."*

It was mentioned that government funding of new products will likely lead to cost savings for governments, as reduced disease progression will translate to less burden placed on the healthcare system, including accessing hospital and services such as the NDIS for ongoing support.

> *"I don't want little kids, to be told 'Hey this is going to stop progressing for you guys,' and that it's going to be $100,000 a year or something…It would save the government though. Like the amount it costs the government on NDIS for FOP patients, it will be so much cheaper."*

Ultimately, it was hoped that new treatment will provide more stability in disease progression. It was envisaged that new treatment will most likely be of benefit to those who have experienced less severe disease progression.

> *"There are a lot of people that are completely locked, that taking a certain drug probably has very little impact on them because they are not going to regain much movement. Yeah, so that's challenging."*

Participants with more severe progression [i.e., those who had experienced bone fusion in multiple areas of the body such as back, neck, shoulders, elbows, jaw, wrists, knees, ankles; those that had experienced greater loss of mobility (e.g., required permanent or frequent use of a wheelchair, could only walk short distances), those that experienced more frequent flare-ups; and those who had greater loss of overall function (inability to engage in activities of daily living, such as getting dressed, showering and thereby being reliant on others)] believed that they were unlikely to benefit as much, as it was unlikely new treatment could reverse current impairment. However, they still very much welcomed new treatment to help preserve the mobility they had left and provide protection against further flare-ups and potential bone growth.

> *"If we could get a drug that would sort of like stop FOP in its tracks… it would ease my mind that say I wouldn't cease up… getting any worse than what it is now…It would mean the world to me."*

These participants also displayed strong support for the whole FOP community, and emphasised the importance of new treatment being available for others, particularly younger or newly diagnosed patients.

> *"Oh, just for the new little kids, they've got a lot more hope which is awesome I'm so happy for them."*
>
> *"If we could stop excess bone growth, from day dot, from the first start of diagnosis [would be] amazing."*

A small number of participants expressed hope that new treatment may prevent bone re-growth following surgical removal of bone, which could mean reversal of damage (e.g., release a locked jaw) and regaining functionality. This might potentially include replacing damaged joints with artificial, prosthetic joints, although it was acknowledged that damage in the muscles may present a barrier. Several participants voiced how much they would welcome the ability to return to previous hobbies and increased independence.

> *"Hopefully to get back some of my former capacities…doing all the things I enjoyed and going places by myself…be more independent."*

New products would lead to improved mental well-being for the whole FOP community, in terms of providing hope, particularly in being able to preserve mobility. Benefits of new treatment would increase confidence in maintaining current function, thereby relieving stress and concern associated with new flare-ups or bone growth.

> *"Obviously something that would slow down the progression would be a good thing."*

Willingness to Try New Treatment

The decision to try new treatment was largely based on trading off perceived benefits, potential side effects, and logistics involved with receiving treatment (e.g., travel and testing requirements, if part of a clinical trial).

Several felt the burden of living with FOP was enough to warrant trying new medication and believed that potential benefits (i.e., slowing disease progression) outweighed possible risks (i.e., side effects). They expressed enthusiasm in receiving new medication. This included going to great lengths to be involved in drug trials, as had previously been the case.

> *"Nothing to lose and everything to gain…I mean everything is going to have a degree of danger to it, but it's worth it, I reckon."*

However, some participants voiced hesitancy due to potential debilitating side effects with new treatment, which could further exacerbate the burden of living with FOP.

"We'd have our line in the sand when we think there is a point. I guess [when] the impacts…are far greater than the outcome…[such as being] away from school and…friends or socialising, or unwell, losing weight, skin deterioration, mood swings, all those kind of side effects."

Preference for Oral Versus Intravenous Infusions

There was a strong preference for oral medication versus an IV infusion, with nearly all participants preferring an oral administration. An oral tablet was considered safer, with caution expressed with IV due to the potential for triggering flare-up and/or new bone growth if the IV damages muscles or causes an infection.

"I don't mind the pills, they are fine by me…Going in through the bloodstream can be a bit difficult for FOP patients, just finding the veins…can be difficult for some."

There was spontaneous mention by several participants about the convenience of being able to take medication at home, as opposed to having to having a nurse come and administer or travel to hospital, which involved logistical challenges.

"Like needles, which you obviously can't administer yourself, so you've got to go somewhere to that, which is slightly inconvenient. Talking about needles, not a lot of people like them… So that could be one of the like factors that would stop people from wanting that type."

"Oral is probably going to always be the easiest option…That would be preference to travelling to hospital."

Upon probing, people conceded they would consider an IV if it was the most effective option. This would warrant the effort of travelling to hospital (i.e., at a probed frequency of once every 2 weeks).

"It depends on how effective it is…and if the results actually show that it…would help, then I would do that, I would go. But if…it doesn't make any dramatic or significant change, then it would be quite hard to go."

People were also more open to an IV administration if administered by a nurse in the home, however concerns regarding potential to trigger disease progression with IV remained.

PATIENT EXPERIENCES WITH THE HEALTHCARE SYSTEM

The findings discussed in this section are based on data generated from the online survey, and follow up discussions in the qualitative interview. Patient experiences were specifically examined in terms of satisfaction and importance of various aspects (i.e., domains) of the healthcare system.

PATIENT EXPERIENCE INDEX

The PEI for FOP, a measure of overall satisfaction that accounts for the relative importance of each aspect of the healthcare pathway, is displayed in Figure 2. The overall mean PEI score was 55.6 (median 58.1). This score should be viewed as a benchmark of current satisfaction with the healthcare system for people living with FOP, and their carers. Future research could use the PEI to assess shifts in satisfaction and importance ratings, especially following access to newly approved and funded treatments, or upon implementation of new programs to address current unmet needs.

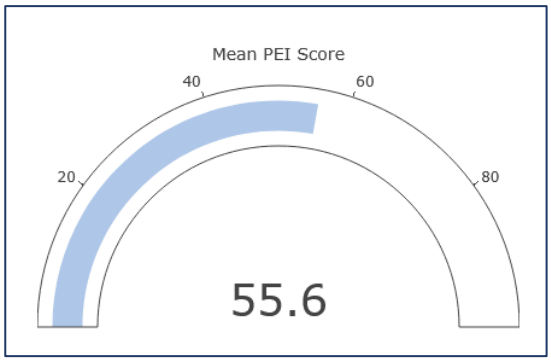

Figure 2: FOP Patient and Carer PEI

RESCALED IMPORTANCE AND SATISFACTION

Figure 3 displays the rescaled importance and satisfaction scores. The rescaled values directly correspond to the level of satisfaction and importance stated between "Not satisfied at all"/"Not important at all" (0) and "Completely satisfied"/"Extremely important" (10).

The magnitude of satisfaction and importance can be compared to identify differences between the level of satisfaction and importance of each aspect of the healthcare system (i.e., gap analysis). In other words, by identifying aspects of the healthcare system that were rated as highly important, but with low satisfaction, we can uncover key areas of unmet need and subsequently priority areas for improvement.

In Figure 3, the aspects of the healthcare system have been ordered from top to bottom by importance (i.e., the most important domain appears at the top of the figure). The orange arrows indicate where the biggest gaps between importance and satisfaction were found.

Figure 3: Rescaled Importance and Satisfaction

Participants found all the aspects of the healthcare system to be at least somewhat important with only 'FOP-related costs' scoring less than 5 out of 10. The areas of the healthcare system, *Access to, and effectiveness of medication and treatment* (8.01), *Access to other treatment and services* (7.84), *The quality of your healthcare team* (7.46) and *Your involvement in decision making* (7.12), appear to be especially important, on average. *FOP-related costs* (4.41) and *Support for your support person* (5.33) appeared to be the least important, on average.

Satisfaction levels were lower than importance levels for most aspects (8 out of 11). The areas of the healthcare systems for which there was highest satisfaction included *The quality of your healthcare team* (7.46), *Your involvement in decision making* (7.12) and *The quality of information available about your condition and care* (6.01). Areas for which there was the lowest satisfaction included *Time to diagnosis* (3.7), *Side effects of medication and treatment* (4.24) and *FOP-related costs* (4.41).

In determining the key areas of unmet need, it is important to assess the gaps between importance and satisfaction. For example, there was clear dissatisfaction among participants with FOP-related costs (4.48), however cost was of relatively lower importance compared to other domains (4.41). Gap analysis between importance and satisfaction scores illustrate the key areas of unmet need for people living with FOP (and carers). The top four areas are listed below (in order):

1. Access to, and Effectiveness of Medication/Treatment

PEI Scores: Importance: 8.01; Satisfaction: 5.10; Gap: 2.91

Having access to government approved and funded treatment is imperative to improve the quality of life for people living with FOP. It is hoped that new treatments will slow disease progression and reduce physical burden by preserving mobility. Emotional burden is also reduced as new treatment provides hope, reduces uncertainty, and relieves ongoing anxiety associated with FOP. Until recently, there had been no treatment indicated for FOP in Australia, with participants only having access to supportive therapy to reduce pain and

inflammation. Participants are urgently seeking medications specific to FOP, that can assist with symptoms, prevent or reduce flare-ups and bone growth, and minimise treatment side effects.

> *"Probably there is not enough medication there, like we should be doing more to get more out there, trying some more of it."*

There was also mention that HCPs must be diligent in conveying information about clinical drug trials to avoid missed opportunities in accessing treatment.

> *"We have basically been left to ourselves. We didn't know there were any trials going on, nobody has given us any information at all, it's been actually shocking. I feel guilty now because being on the trial, maybe [child with FOP] wouldn't have got so bad."*

Recommendations

Participants were grateful for the opportunity to be part of evidence-based research to ensure the 'patient voice' is included in government submissions for new treatment, and industry decisions to invest in research and develop new products (i.e., further clinical trials). Participants noted that ideally a variety of options are required to treat the heterogeneity of FOP. Some participants believed specialists were responsible for keeping up to date with new drug trials and inform patients and families of these, to ensure equity of access to latest treatments and advances in FOP.

2. Time to Diagnosis

PEI Scores: Importance: 6.30; Satisfaction: 3.70; Gap: 2.6

Generally, there was the perception that there had been a gradual increase in knowledge of FOP over time, however in recounting their journey, participants reported a harrowing path to diagnosis, stemming from a lack of knowledge about the condition among Australian HCPs.

Many had experienced symptoms from a young age (some from birth, others during toddler or teenage years) and several reported the progression experienced while waiting for a correct diagnosis. There was reference to being treated like 'guinea pigs' due to the rarity of the condition. This lack of knowledge about FOP among HCPs at the time, had led to several participants also being misdiagnosed and mistakes being made (e.g., unnecessary tests, surgery on broken bones triggering new bone formation). Some believed that an accurate and timely diagnosis would lead to better FOP outcomes through improved management of the disease (e.g., extra care during younger/toddler years to prevent falls, and associated flare-ups).

Recommendations

Participants stressed the importance of ensuring a timely diagnosis for new patients. This involved education of HCPs to recognise early signs and symptoms (e.g., malformed toes which are common at birth).

> *"Just that simple thing of looking at the toes to start with, we might have had a diagnosis at six months old instead of ten [years]."*

"Because it's such a rare disease, trying to get awareness out there is the biggest thing. Trying to get more doctors and nurses [aware] from…[the time of] being born."

Participants recommended that if in doubt, HCP should reach out to those with more experience and expertise in managing FOP (including Australian and overseas based HCPs).

"You can really do some damage and cause incurable flare-ups that would restrict and potentially lose movement…it's really heartbreaking seeing young kids that aren't even teenagers that have got restrictions in their movement…Some of that may have been because the medical knowledge wasn't enough, because the treating doctor made a call which they thought was the best, but had they made a phone call or Googled a website they may have been able to have a different result."

There was mention of the appeal to achieve more 'buy in' from HCPs to be part of the FOP community to further increase expertise in the area. This would improve the diagnostic pathway for new patients with a greater chance of accessing the correct healthcare team from onset.

"I think from an Australian point of view we need more doctors that can assist with patients and that's hard because it's so unique. You need to get them to buy into actually being part of the community… get more doctors interested in it. So, you can say to a new person, "The doctor in New South Wales is this," and give them the contact details. We don't have that."

There was also the perception that education on rare diseases should be provided during medical training to equip future doctors with greater knowledge of rare diseases, which would lead to greater proficiency in diagnosis and managing FOP.

"It should be part of their training I would say. Yeah, there is a lack there in the students. If I could get that awareness out there to these young and upcoming doctors, the better, because the more that people know…that then goes through for new babies, children don't go through what we went through to get the diagnosis, and you got early intervention of some sort."

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3. Side Effects of Medication and Treatment

PEI Scores: Importance: 6.38; Satisfaction: 4.24; Gap: 2.14

People living with FOP used supportive medications including corticosteroids and anti-inflammatories to manage symptoms. Several also had experience with specific FOP medication (e.g., palovarotene; imatinib). People spoke of experiencing various side effects with treatment, including; weight loss or weight gain, impact on mood and sleep, dry and itchy skin, lethargy, impact on the immune system that led to illness.

Side effects with treatment add an additional burden for people living with FOP, who must balance the efficacy of treatment with side effects. Participants spoke of the need to be able to access medication which have tolerable side effect profiles in order to maximise quality of life.

> *"I want the flares obviously to stop…and try to make like less side effects which include our skin, because FOP already has lots of skin conditions on its own, and I can't deal with another skin problem. I have a lot on [my] plate."*

Recommendations

Recommendations in relation to side effects with treatment rested on pharmaceutical company investment into research and development of new effective products with favourable side effect profiles.

> *"Minimise side effects of medications, provide best quality of life." Verbatim response from PEI survey*

4. Access to Other Treatment and Services

PEI Scores: Importance: 7.84; Satisfaction: 5.8; Gap: 2.04

Allied healthcare

FOP was described as a condition that impacts the whole body, and hence requires a multidisciplinary approach by incorporating several areas of allied health (e.g., occupational therapists, nurses, psychologists, physiotherapists, specialist dentists). People spoke of the need to be able to access a variety of HCPs for universal care, which had been challenging for some.

> *"I would say that adaption for home life, everyday life. So, people like occupational therapists could be quite valuable in this space to do home assessments a bit more."*

Furthermore, when accessing allied healthcare, participants believed there had been an absence of holistic care, where HCPs had focussed on treating various symptoms according to their standard approaches, rather than adapting to the unique needs of people living with FOP. Participants explain that disease status can change very rapidly (even mentions of overnight) due to new bone growth, with resulting adaptation sometimes required on a daily basis. Frustration was voiced, as HCPs are not always willing to listen to the ongoing and changing needs of FOP patients and carers, including insights into what is likely to work and what is not, in managing FOP. Patients and carers themselves are proficient in finding their own solutions to best meet these changing needs.

> *"So it was finding someone who could sort of understand [child with FOP is] not inside the box…is going to break the mould, everything you've used for that is going to change. So, you've got to come up with something extra."*

A number of participants mentioned that there were substantial differences in a multi-disciplinary approach between paediatric care and the adult healthcare system. The children's hospitals were referred to as a holistic care model with a multidisciplinary team collaborating within the one setting, including scheduling appointments and follow up care. Upon moving into the adult system, the continuity of care had reduced and the transition for some had been difficult (e.g., even deciding when to see HCPs and making appointments).

Recommendations

Greater education is required for allied HCPs who are involved in managing FOP, who should be encouraged to collaborate closely with patients and carers for insights into optimal management strategies. Participants would like to see a multi-disciplinary approach adopted, particularly in the adult healthcare system.

"I think it's important to have a team of people around you, but that is difficult to get."

"I feel like if there was that same level of care that they have with children was reflected in the adult system, that would absolutely make things easier and better in that world."

National Disability Insurance Scheme (NDIS)

Several participants had accessed the NDIS for provision of support workers, and funding for home modifications and equipment. Participants expressed appreciation for the support received.

"Ultimately having the support person and having the NDIS package to get the support people was like an absolute God send."

However, participants frequently referred to administration difficulties when seeking funding through NDIS in Australia or District Health Board (in New Zealand), such as for equipment (e.g., wheelchair), home modifications and/or access to holistic care (e.g., occupational therapist, physiotherapists). There was evident dissatisfaction among participants due to a lengthy and complicated application process, long wait times to receive funding, being allocated support people who were not a good fit, not having a holistic focus on the broader needs of living with, or caring for a person with FOP (e.g., family aspects) and being denied further funding (e.g., upon needing to update a new home to meet the needs of the patient).

"I think NDIS is an absolute struggle when it comes to…people with rare diseases or needs."

"It's very difficult to get NDIS access and even sometimes if you do, like the funding they give might not cover exactly what you need, and only…covers like certain type of…services."

Recommendations

There were mentions that having a support person who could help navigate the NDIS and healthcare system would be very helpful.

Support Organisations

As well as gaining support from family and close friends, most participants had accessed support though patient organisations including FOP Australia, and the international community, IFOPA, with some also connecting and sharing experiences via online platforms (e.g., with families overseas through Facebook).

> *"Now that we have FOP Australia, it's just brilliant and although it's a rare disease, we have a very supportive international community, and during that time where you wouldn't know if it was okay if [child with FOP] fell over like…you could quickly put it on Facebook and someone in the world would answer you back. So, they've been our support really."*

Participants praised FOP Australia and were appreciative of information on the website and connections formed with others.

> *"The beauty of some of the things that FOP Australia have done by bringing the community in together, because they are so rare, is that you can actually…work with each other and…know, people…that know about (FOP)."*

Furthermore, several participants were grateful for opportunities to attend FOP conferences (mentions of a Manchester and Swedish conference, and the recent conference organised by FOP Australia in Melbourne, Australia). Conferences allow people to learn more about the condition including management practices (i.e., tests and new treatment), as well as connect with others in similar situations.

Recommendations

Participants spoke of the need to continue with provision of support, particularly any new evolving information that could be helpful in managing FOP (e.g., an increased risk of progression during teenage years). This can help create awareness on how best to manage certain stages of development to minimise injuries and progression.

> *"I wasn't aware of the age group that I had to be extra careful between 10 to 15, so my condition got worse."*

Participants would also welcome greater knowledge about which Australian HCPs have expertise in FOP and suggested generating a list of Australian specialists that could be contacted for questions or management, particularly when newly diagnosed, to ensure patients can come under the care of HCPs who have experience with FOP from disease onset. It is also recommended that awareness be raised within the FOP community regarding the register of international HCPs available on the FOP Australia website, as this is an existing resource that would be helpful as additional support.

> *"It would actually be really nice to have some experts, kind of based in Australia as opposed to overseas."*

There was mention of the importance of ongoing support throughout the FOP journey.

> *"Like everyone was around you when they thought [child with FOP] might have had it…but then it kind of like dropped off and that's the bit where we're all dealing with it the most. Having to now live with that."*

QUALTIY OF LIFE MEASURE

Participants completed the EQ-5D-5L to measure overall health and quality of life. The EQ-5D-5L assesses five areas of health status across five levels of severity (for each dimension, participants chose one of five options ranging from 'no problem' to extreme problem' or 'unable to'), creating a health state profile which is represented by a number score.

The five quality of life dimensions measured in the EQ-5D-5L include:

1. Mobility: How easy or difficult it is to move around, like walking or climbing stairs.
2. Self-care: The ability to manage personal care, such as washing and dressing.
3. Usual Activities: How well someone can carry out daily tasks, including work, hobbies, or family responsibilities.
4. Pain/Discomfort: Whether someone experiences physical pain or discomfort and how severe it is.
5. Anxiety/Depression: How someone feels emotionally (i.e., feeling worried, sad or stressed)

The EQ-5D Index Score is a single number that summarises a person's overall health based on the answers on the above dimensions. The score ranges from 1 (perfect health) to 0 (state similar to being dead), and in some cases, it can be negative (a health state worse than being dead).

Based on 11 participants who completed the measure, the mean index score was 0.31 (median: 0.38). Scores for each dimension are included in Table 3.

Table 3: EQ-5D-5L Scores

EQ-5D-5L Domain Frequency Distribution	N=11	
Mobility	N	%
No problems walking about	1	8.3
Slight problems walking about	1	8.3
Moderate problems walking about	3	25.0
Severe problems walking about	1	8.3
Unable to walk around	5	41.7
Self-care		
No problems washing or dressing	1	8.3
Slight problems washing or dressing	1	8.3
Moderate problems washing or dressing	1	8.3
Severe problems washing or dressing	3	25.0
Unable to wash or dress	5	41.7
Usual activities		

No problems doing usual activities	1	8.3
Slight problems doing usual activities	2	16.7
Moderate problems doing usual activities	3	25.0
Severe problems doing usual activities	3	25.0
Unable to do usual activities	2	16.7
Pain/discomfort		
Slight pain/discomfort	1	8.3
Moderate pain/discomfort	6	50.0
Severe pain/discomfort	4	33.3
Anxiety/depression		
Not anxious/depressed	3	25.0
Slightly anxious/depressed	3	25.0
Moderately anxious/depressed	4	33.3
Severely anxious/depressed	1	8.3

The EQ-5D-5L provides further evidence of the severe burden of FOP on people's lives. Most participants reported moderate to severe impact on all quality of life dimensions, with several being highly dependent on support people or equipment in terms of their mobility, self-care and ability to engage in usual activities (i.e., 'moderate' or higher problems). Most participants reported moderate to severe pain and discomfort, with several being moderately or severely anxious or depressed.

CONCLUSION

The key objectives of this research were to develop a holistic understanding of lived experiences with FOP, and examine experiences with the healthcare system to identify unmet needs.

Findings showed that FOP significantly reduces quality of life for people diagnosed with this condition, and their carers. Participants described severe symptoms, especially flare-ups and progressive bone growth, that impose major physical limitations, restrict independence, and hinder participation in hobbies and daily activities. Many also reported negative emotional, social, and financial impacts. EQ-5D-5L scores supported participant feedback, with moderate to severe problems being reported for all five health dimensions (mean score: 0.31; median 0.38 on a scale where 1 = perfect health and 0 = equivalent to being dead).

In addition, participants reported a difficult healthcare journey due to the rarity of FOP, including delayed or incorrect diagnoses and, in several cases, unnecessary surgeries that worsened disease progression. Despite recent improvements in HCP awareness, lack of effective and accessible treatment remains a major challenge. Through government approval and funding of new treatment, there is real opportunity to make a positive impact on the quality of life for people living with FOP in ANZ. New treatments have the potential to stabilise the condition, lessen dependence, and improve emotional wellbeing. With the PBS approval of palovarotene in Australia and new clinical trials underway, participants expressed hope for options that slow progression with tolerable side effect profiles. Participants called for continued pharmaceutical investment and for government bodies to incorporate 'patient voice/lived-experience' research when assessing disease burden and funding decisions.

The key gaps in the healthcare system - areas where importance significantly exceeded satisfaction - include access to effective treatment, time to diagnosis, treatment side effects, and access to supportive services. Even modest improvements in these areas could enhance patient experiences. Participants emphasised the need for better HCP education in terms of increased awareness of early symptoms to ensure a timely diagnosis, proactive communication to FOP patients about clinical drug trials to ensure access opportunities, and consistent multidisciplinary care, similar to paediatric models. Given the variability of FOP, HCPs should also recognise the rapidly changing patient needs, where circumstances can change from one day to the next (e.g., further permanent loss of mobility upon new bone formation). Repeating the PEI following approval of more FOP medication and/or upon implementation of new programs/system changes, is recommended to assess progress in meeting patient needs across various aspects of the healthcare system.

Overall, this research provides important supplementary evidence to inform treatment and funding decisions for current and future treatment for FOP. While clinical trials provide valuable evidence of efficacy and safety endpoints, these often do not address other aspects of care that matter to patients and carers. Increasingly (and crucially) a patient-centric approach is being adopted, that considers humanistic factors such as burden of disease and impact on quality of life, both on the patient and society, when assessing current and new therapies.

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About CaPPRe

Filling an important need in the community and patient preference research industry, Dr Simon Fifer, Laurie Axford and Professor John Rose have built on 15 years of experience to launch Community and Patient Preference Research (CaPPRe).

CaPPRe is an independent group committed to high quality, meaningful research leading to better engagement and understanding of community and health consumer needs. We use choice-based research to transform our clients understanding of their business and their clients, by understanding why people make the choices they do and predict the choices they'll make in the future. This helps our clients make better business decisions and create effective policies based on what people value.

The CaPPRe partners unique approach enables them to use the latest academic advancements in qualitative research and choice-based methodologies to provide clients with practical, easy to understand and actionable results via insightful reports and advanced dashboards.

We conduct research across the following sectors:

- ✓ Infrastructure
- ✓ Technology
- ✓ Transport
- ✓ Environment
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- ✓ Health.

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