Management of osteogenesis imperfecta (OI): self-reported access challenges to consumables and services across the EU5 and Nordics

Taco van Welzenis[1], Lena Lande Wekre[2], Ingunn Westerheim[1], Cathleen Raggio[3], Michael B Bober[4], Oliver Semler[5], Tracy Hart[6], Ruby Dadzie[7], Maria Rapoport[7], Samantha Prince[7], and Frank Rauch[8]

[1] Osteogenesis Imperfecta Federation Europe, Heffen, Belgium; [2] Sunnaas Rehabilitation Hospital, Nesoddtangen, Norway; [3] Hospital for Special Surgery, New York, USA; [4] Alfred I. duPont Hospital, Wilmington, Delaware, USA; [5] University of Cologne, Germany; [6] Osteogenesis Imperfecta Foundation, Gaithersburg, Maryland, USA; [7] Wickenstones Ltd, Abingdon, Oxfordshire, UK; [8] McGill University, Montreal, Canada.

Background

- Osteogenesis imperfecta (OI) is a rare, heritable connective tissue disorder with multiple manifestations, and variable severity. Individuals with OI typically have low bone mass and skeletal fragility, and are susceptible to morphometric vertebral fractures and compression, bone deformities, scoliosis, and growth deficiency[1]
- The IMPACT Survey aimed to describe the clinical, humanistic and economic impact of OI on the OI community
- Here we present the data on access challenge relating to consumables and services of adults with OI (non-caregiver [CG]) across the UK, France, Italy, Spain, and Germany (EU5), and Nordic countries

Methods

The IMPACT Survey

- Developed by the Osteogenesis Imperfecta Federation Europe (OIFE), the Osteogenesis Imperfecta Foundation (OIF) and an international steering committee of OI clinical experts

Methods (continued)

- Aimed at adults with OI, CG (with or without OI) of children or adults with OI, adolescents with OI and relatives
- Included up to 102 questions on the clinical, economic, and humanistic impact of OI
- Was professionally translated from English into French, European Spanish, Latin American Spanish, Portuguese, Russian, German, Italian, and Dutch, and fielded online July–September 2021

Recruitment

- Advertised through emails, meetings and social media engagement by the OIFE, and OIF

Analysis

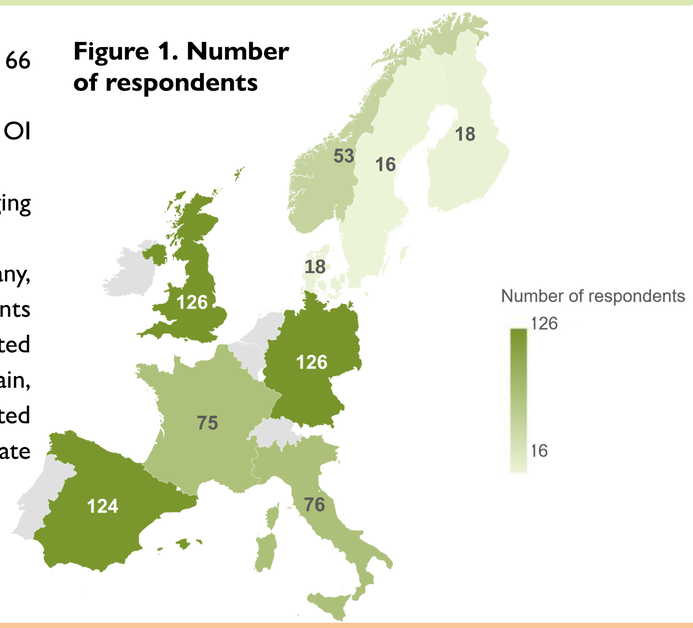
- Responses from all languages were professionally translated into English
- Microsoft Excel was used to clean, code, and analyse data
- Proportions are relative to the proportion of adults with OI reporting the need for a device or service

Results

Demographics and respondent characteristics

- Overall, 2,208 individuals responded across 66 countries, of whom 1,440 were adults with OI
- Across the EU5 and Nordics, 632 adults with OI (non-CG) participated (Figure 1)
- Women were more highly represented (ranging from 50%–73%)
- In six countries, (UK, France, Italy, Germany, Sweden, and Denmark) most respondents reported moderate OI (43—62%); fewer reported mild (17—39%) or severe OI (13—23%). In Spain, Norway, and Finland, most respondents reported mild OI (44—50%), fewer reported moderate (33—38%) or severe OI (15—17%)

Figure 1. Number of respondents



Quotes from respondents

"It took years to find an alternative to my NHS wheelchair that was no longer available. Standard wheelchairs are too cumbersome. I had to approach a company 25 miles away to make a wheelchair and maintain the wheelchair. Each repair is £85 plus parts. Wheelchair was £3,238." - ***UK***

"I need a pain specialist, but it's very hard to access. There is also a waiting list for occupational therapy." - ***Norway***

"Several documents to put together in order to get financing; time-consuming, intrusive and tiring procedures." - ***France***

"Being on income support I have to sleep on a sofa bed. When you have triple scoliosis and osteoarthritis, sleeping on a sofa bed worsens the pain. So, having good bedding would make life so much easier and greatly improve one's energy levels." - ***France***

"There are aids for which a private contribution is needed […]. To replace an aid, it often has to be destroyed, not just worn out. If a corset is made badly/mistakenly by the orthopaedic workshop, the Italian National Health System does not want to pay for a new one until 5 years have passed." - ***Italy***

"I have always received family help to buy a wheelchair and hearing aids, without that help I would not have been able to afford it." - ***Spain***

"In my experience, you can get any help you need. However, it isn't offered, rather, I have to find information about it myself, apply for it, and above all remain persistent and not take no for an answer. This is expensive and takes time and effort which not everybody has." - ***Germany***

"My type of OI doesn't fit in any of the "boxes" of health care provided by the National healthcare provision." - ***Denmark***

Access challenges

- Up to 56% of respondents who needed a queried consumable or service experienced access difficulties
- Difficulty in accessing manual wheelchairs, vehicle modifications, personal care/support assistance, and dental work were experienced in all countries (Figure 3 and 4)
- Prevalence of access difficulties to powered wheelchairs was highly variable between countries (0% [Sweden] to 64% [UK]), and was the most commonly reported challenge in UK, France and Germany
- Access to dental work was the most commonly reported challenge in Spain (46%) and Sweden (27%)

Figure 3. Proportion of respondents who experienced access challenges in the EU5 countries

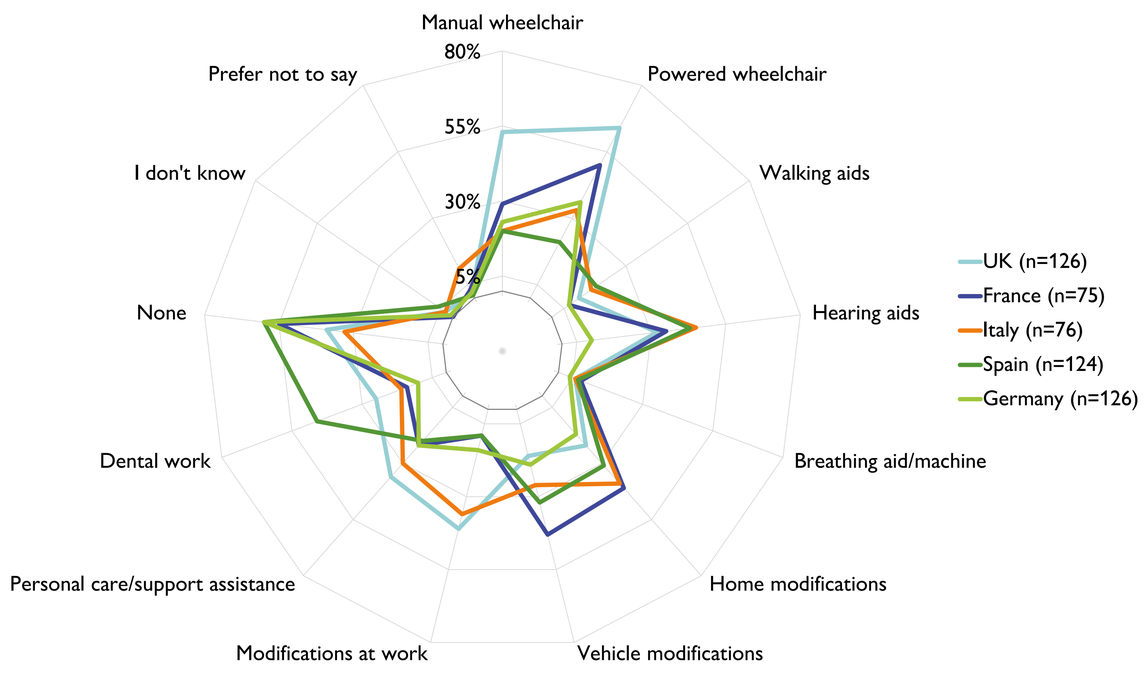
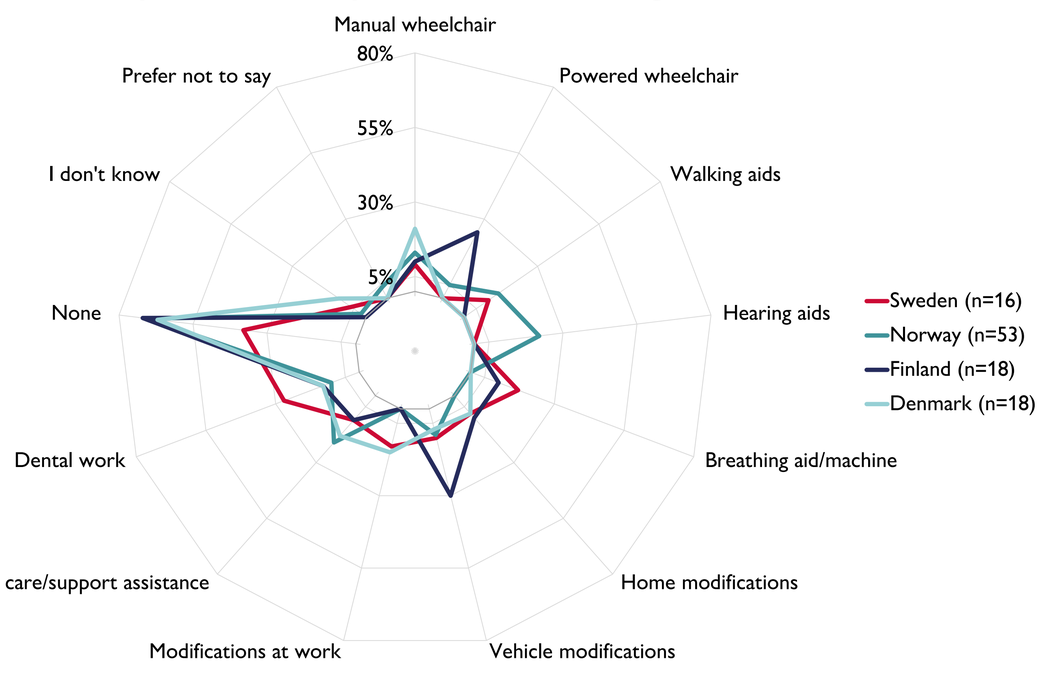
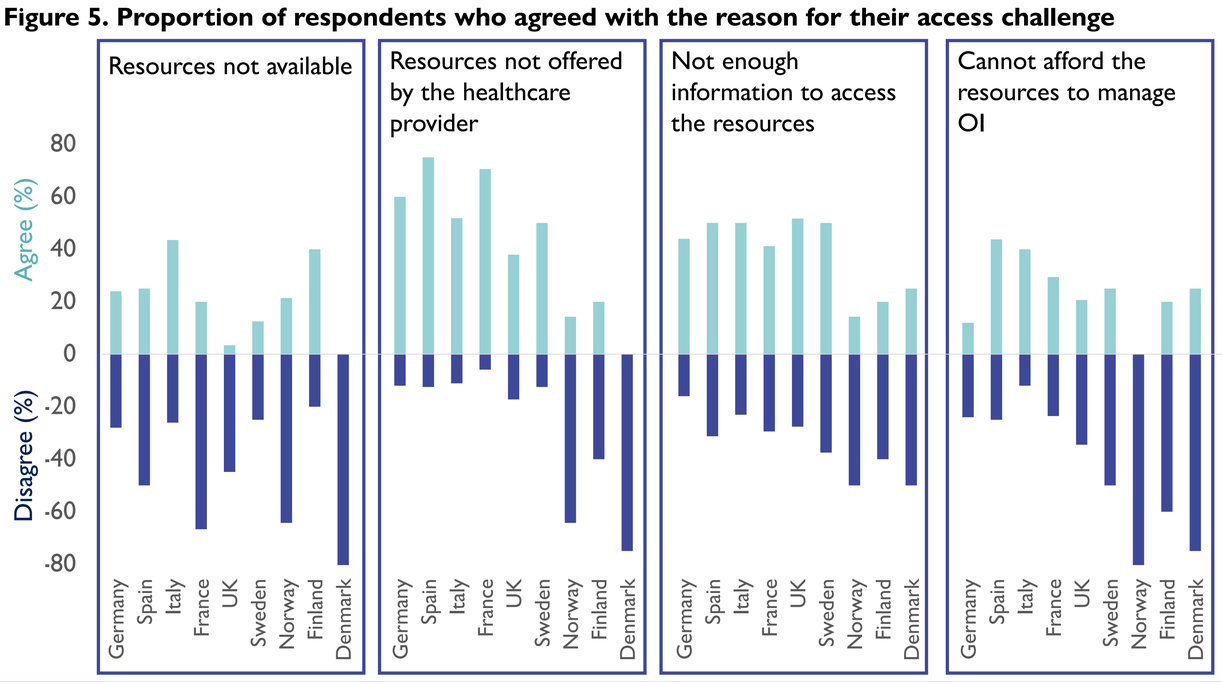


Figure 4. Proportion of respondents who experienced access challenges in the Nordic countries



Reasons for access challenges

Figure 5. Proportion of respondents who agreed with the reason for their access challenge



- Resources were generally seen as available by a high proportion of respondents (20–100% disagree with unavailability), particularly in Germany, Spain, France, UK, Sweden, Norway, and Denmark
- 'Resources not offered by healthcare providers' impacted accessibility for the highest proportion of respondents (20–75%) in six countries (Germany, Spain, Italy, France, Sweden, and Finland)
- Respondents from all countries (14–52%) reported that they lacked information to access resources (Figure 5)
- Affordability was considered less of a barrier in the Nordics (50–85% of respondents) than in the EU5 (12–34%)

Conclusions

- A high proportion of respondents in the EU5 and Nordics experience access challenges to obtain the consumables and services they need to manage their OI; wheelchairs (both manual and powered) were difficult to access across geographies
- While resources were mostly available and affordable to individuals, a lack of information, and lack of provision by healthcare providers hampered access
- The respondent's comments demonstrated that access challenges lead to substantial additional, personal expense, increased pain, and increased administrative burden
- Access difficulties are widespread and systemic indicating policy reform may be required
- Improved information and resource provision by healthcare providers may help individuals with OI to obtain required healthcare and support

References

1. HOYER-KUHN, H., REHBERG, M., NETZER, C., SCHOENAU, E. & SEMLER, O. 2019. Individualized treatment with denosumab in children with osteogenesis imperfecta - follow up of a trial cohort. *Orphanet journal of rare diseases, 14,* ***219-219***

Acknowledgments

Funding for the IMPACT Survey was provided by Mereo BioPharma Group, London, United Kingdom.
The authors would like to thank the OIFE, OIF and national member organisations for their support in the development of the survey and their sustained role in recruiting respondents.