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Introduction

The Brittle Bone Society

Founded in 1968, the Brittle Bone Society is the sole charity for people with Osteogenesis Imperfecta (OI) in the UK and Republic of Ireland, offering support for them and their families. We work closely with NHS Healthcare Professionals in this specialist area. In 2012 we formed our Medical Advisory Board. In 2016 we were accepted into the Association of Medical Research Charities (AMRC). We launched our first research grants programme for OI in 2017 managed by our Scientific Committee and also held our first International Scientific Symposium.

The Brittle Bone Society regularly host consultation events and workshops to gather feedback to help formulate our operational programmes and activity, to gather opinions of various topical issues and influence strategic planning in general. This also helps the Charity to demonstrate outcomes to supporters and funders.

The objective of the Research Priorities Questionnaire was to formally identify key areas of interest within the members and supporters of the Brittle Bone Society. This will then help to inform the decisions made by our Scientific Committee in their selection of projects for the Brittle Bone Society Research Grant Programme.

- Patricia Osborne
  Chief Executive of the Brittle Bone Society
Background

Osteogenesis Imperfecta

Osteogenesis Imperfecta (OI) is a genetic condition present from birth. Its most striking feature is that fractures may occur easily. Therefore it is frequently called “brittle bone disease”.

OI is a disorder of collagen, the protein which forms the framework for the bone structure. In OI the collagen may be of poor quality, or there may just not be enough to support the mineral structure of the bones. This makes the bones weak and fragile and results in the bones being liable to fracture at any time even without trauma.

As the composition of collagen in the bone is not correct, even when there are no fractures there will be other problems connected to the condition: such as the ligaments stretch more easily, allowing dislocation of joints. The joint hypermobility can significantly affect the quality of life as it results in fatigue of many muscle groups.

As a result the mobility and performance of ordinary day tasks of everyday living are impaired. The majority of individuals with OI suffer from bone and muscle pain.

We held a consultation workshop during our Annual Conference in August 2018. The workshop was facilitated by Professor Nick Bishop, a professor of metabolic bone disease and Trustee of the Brittle Bone Society. We then followed this up with a questionnaire to further examine our audience’s attitudes to Research into OI.

The primary aim of this exercise was to assess the feasibility of eliciting thoughts and opinions with regards to research topics in OI. Secondary aims were to: gain insight into the vocabulary used by this group when discussing research; uncover some of the main research priorities; and to highlight some common themes which may be useful to discuss further within a focus group setting.
Workshop Activity Outline

Preliminary Exercise into determining research priorities in Osteogenesis Imperfecta

This document details the preliminary exercise to gain insight into the research priorities of young people and adults with OI, alongside their families and carers.

The Brittle Bone Society provides Research Grants to clinicians and researchers working in the field of Osteogenesis Imperfecta. To ensure the charity are awarding grants to relevant projects we consulted with members during 2018 to find out what people living with OI felt was priority to be researched.

We held a consultation workshop during our Annual Conference in August 2018. Discussions were carried out in small groups of 8 to 10 people. In total we had 149 participants from around 19 different countries. Following on from the feedback ingathered during this event we sent out a questionnaire to all UK and Irish members and received 79 responses.

Workshop on Research into OI

This workshop activity included young people and adults with varying types of OI, alongside their family and carers. The activities involved a short presentation, with some discussion, about current research in OI, the burden this research can place on patient and families, how the results are fed back, and what involvement in research patients and families would be comfortable with.

Discussion & Debate

This workshop session was facilitated by Professor Nick Bishop, a professor of metabolic bone disease and Trustee of the Brittle Bone Society. Dr Claire Hill, Advanced Physiotherapist from Sheffield Childrens Hospital observed the session, and Penny Clapcott a patient representative who serves on the Charity’s Scientific committee as a layperson provided assistance throughout the workshop.

Discussions were carried out in small group sessions of between 8 to 10 individuals. There were 149 participants in total. Groups were asked to document their thoughts surrounding research priorities in OI, highlighting the themes that they felt should be at the forefront of OI research.
**Workshop Activity Findings**

**Analysis of Answers & Themes**

Participants were given 10-15 minutes to discuss and document their thoughts and opinions on post-it notes and place them on A3 poster boards. These thoughts and opinions were then fed back to the whole group by Professor Bishop, and participants were given the option to discuss their themes with the group, and clarification was gained for any ambiguous themes to enhance understanding. The A3 posters, with post-its attached, were analysed by Claire Hill using thematic analysis; themes and subthemes were extracted.

**Activity Findings**

149 thoughts and opinions were documented, and then categorised into 14 main themes. The main themes which arose from the discussion are detailed to the right, with the numbers indicating the number of individual responses. Some of the vocabulary used in the discussion warrants further investigation and discussion, such as:

- What do individuals mean when using the term fatigue?
- Do people understand the difference between treatment and management?
- What do individuals mean when reporting sleeping problems? (Falling asleep? Waking in the night?)

In the diagram to the right, each circle includes a theme and a number. The number indicates the amount of participants who saw the theme as a research priority. These thematic results therefore indicate areas of interest to the adult OI population; further scoping may be needed to identify the themes for children.
The purpose of this questionnaire was to formalise the comments made during the workshop activity, and gain further information regarding participation preferences of those willing to get involved in research projects. 79 responses were received in total.

The questionnaire included questions to:
1. Obtain demographic information
2. Formally rank research priorities.
3. Gauge perceptions on collaborative research with conditions similar to OI
4. Identify attitudes towards different types of research participation from both adults with OI and parents of children with OI.

By including questions which set out the preferences for participation, an understanding of what projects might be more successful in recruitment within the OI community can be made quantifiable. No qualitative questions were included in the questionnaire, and no personal information was obtained, ensuring anonymity in the responses.

**Demographics**

**Gender**

- 82% Female
- 18% Male

**Age**

- Under 18: 8%
- 18-25: 8%
- 26-30: 13%
- 31-40: 18%
- 41-50: 18%
- 51-65: 33%
- 66+: 2%
Questionnaire participants were asked “What areas did they want the Brittle Bone Society research to focus on?” Unlike the workshop activity, Pain Management ranked highest, with 49% of respondents listing it as priority. Fractures ranked second in the questionnaire, despite not surfacing as a key theme in the workshop activity, but fatigue, quality of life, and soft tissue are a shared priority between the two.

- 79 responses were received, with respondents having on average 6 priority research areas.
- 87% would be willing to participate in a research project, and on average would consent to 6.7 different types of participatory methods.
- 27 participants were parents or guardians of a child with OI, and 75% would allow their child to participate in research activities. On average, they would consent to 6.5 different types of participatory methods.
Participants were asked “What would they be willing to do?”, and if they had a child dependent, they were also asked “What would they consent to for their child?”.

The findings for participation preferences in research projects demonstrate the willingness of Brittle Bone Society members and supporters to get involved in research. Over half of the respondents would consent to eight of the methods of participation for themselves.

As methods of participation become more invasive, or have a higher risk factor, fewer respondents say they would be willing to participate. Parents or guardians of child dependents were far less likely to consent to CT or MRI scans (between 12 and 20% less likely). The majority of other methods of participation remained at the same levels as personal consent.
Conclusion

Current Opportunities for the OI Community

Over the previous few years the Brittle Bone Society have increased their participation and co-operation where possible in studies and trials around OI. They are now routinely in touch with professional bodies such as The James Lind Alliance and have promoted numerous surveys through contacts via ERN-BOND and others. They are now recognised by the Society for Endocrinology as a patient support group in this area and are also reaching out to grow awareness amongst healthcare professionals networks.

There is a section on the Charity website listing current clinical trials (such as the Asteroid and TOPaZ trials) and research projects like the RUDY study, that may be of note and interest to people with OI, as well as projects funded by the Brittle Bone Society. You can find this by visiting: http://brittlebone.org/research-studies/get-involved-in-research

Summary of Findings

This paper provides an overview of what people living with Osteogenesis Imperfecta feel is priority to be researched. It should be noted that there was no differentiation made between the types of OI, nor the degree of severity.

There are certain themes which may need more understanding and clarification, for example what do individuals mean when they report sleeping problems (is this getting to sleep or waking up during the night?) Further consultation on specific areas may be required.

Although results from the Workshop and the Questionnaire differ slightly, it is clear that pain management, treatments and quality of life list high on the list of priorities with fracture management being of secondary importance.
Trustees of the Brittle Bone Society would like to acknowledge and thank all the funders who helped facilitate the Charity’s 50th Anniversary Family Conference Event with particular thanks to funds given and support from Alexion Pharmaceutical, Kyowa Kirin, and Mereo BioPharma.

The Brittle Bone Society would also like to thank the facilitators at the Workshop Activity during our 2018 Annual Family Conference, culminating in the research findings presented in this paper: Professor Nick Bishop, a professor of metabolic bone disease and Trustee of the Brittle Bone Society, Dr Claire Hill, Advanced Physiotherapist from Sheffield Childrens Hospital, and Penny Clapcott a patient representative who serves on the Charity’s Scientific committee as a layperson.