Final Report to Brittle Bone Society

18/NE/0230 - Assessing the Balance of Cost and Determining the Experiences of Families in Paediatric Osteogenesis Imperfecta Healthcare (ABCD) Study

PI: Dr Meena Balasubramanian, Consultant Clinical Geneticist, Highly Specialised Osteogenesis Imperfecta Service, Sheffield Children's NHS Foundation Trust, UK

Research Fellow: David Walker, Genetic Counsellor, Genetic Counsellor, Sheffield Clinical Genetics Service, Sheffield Children's NHS Foundation Trust, UK

Summary

Osteogenesis imperfecta is a highly variable, heritable connective tissue disorder, characterised mainly by a high susceptibility to skeletal damage due to bone fragility. Diagnosis is usually made on a clinical basis. Genetic testing may inform specific treatments or provide clarity for patients' relatives. Increased susceptibility to fracture may lead to safeguarding protocols being implemented for children affected with the condition. Treatment is not curative but can ameliorate symptoms. A combination of a diagnosis of a bone fragility condition and the potential involvement of social services is often highly distressing for patients and their families. In the UK, the Highly Specialised Severe, Complex & Atypical OI Service provides a multi-disciplinary approach to paediatric OI. Genetic testing is also provided as part of this service as one of the core interventions.

The aim of the study was to evaluate and develop a stronger understanding of the diagnostic experience in paediatric OI, and to contribute evidence in the consideration of local and national service development.

Semi-structured interviews were carried out with 8 participants, who were parents of children with OI, to explore their experience of obtaining a diagnosis. Thematic analysis was performed on the data to extract recurrent and shared experiences.

Three major themes were identified, each including several subthemes. These were: 1) time taken, challenges faced, and the emotional impact of obtaining a diagnosis was highly variable; 2) safeguarding concerns were largely unavoidable in the diagnostic experience; 3) the importance of specialist models of care, and the need for improved awareness and education, was highlighted.

Diagnostic experiences reflected the variability of the condition but demonstrated common themes. This study adds to the literature that earlier diagnosis and subsequent access to specialist care has a clear benefit for patients, their families, and their relationships with their doctors. The results of this study demonstrate a clear benefit to rapid diagnosis for the physical, emotional, and psychosocial wellbeing of families affected by OI.

Rationale for Research

Given the negative experiences associated with suspicions of non-accidental injury (NAI), and the significant implications of being affected by OI, it is reasonable to expect that the diagnostic pathway for OI should be quick and accessed early by patients and their families, in order to maximise the efficacy of treatment & management, inform family planning, and to reduce distress in families. A logical assumption would be that diagnosing the condition as early as possible (i.e. early in infancy/childhood) is also the most cost-effective method of managing these patients. However, no research group has yet evidenced that this is the case.

Although there are studies, which focus on an affected patient's quality of life in OI, there is a need to assess the impact of experiencing a diagnostic odyssey, especially in children, in terms of both to the families' emotional wellbeing, and the financial burden to health services.

One might expect that improving early diagnosis would positively impact the financial and emotional burdens, but there is no empirical evidence to show that this is the case. There are studies focused on health quality assessments but none have been identified on quantitative assessments in paediatric OI or how time taken to diagnosis affects families.

Aims and Objectives

The study aimed to evaluate the need for early diagnosis based on the experiences of patients and their families. This research will develop a stronger understanding of the experiences of the diagnostic journey, which was anticipated to suggest a desire, and need, for service users to receive a diagnosis as early as possible.

In collaboration with colleagues from the School of Health and Related Research at The University of Sheffield, these findings will then contribute to a larger pilot study, which intends to understand and compare the cost-effectiveness of early diagnosis in OI between current processes and newly-available rapid genetic testing for OI. This would theoretically have implications for future benchmarking, funding, and multi-disciplinary service development, both locally and nationally. This data will be obtained by interrogating the Highly Specialised, Severe, Complex, Atypical (HSSCA) OI Service national database to first obtain this data for the severe OI patients. Patients and families from this service will be recruited to complete the Child Health Utility-9D (CHU-9D) questionnaire (proxy or self-completion), which measures health-related quality of life for use in cost utility analyses. Together, this data should demonstrate burden of healthcare costs in paediatric OI and analyse cost-effectiveness of early genetic testing for diagnosis of OI.

The larger pilot study, entitled "*Assessing the Balance of Cost and Determining the Experiences of Families in Paediatric Osteogenesis Imperfecta Healthcare (ABCD Study)*", is currently ongoing. As such, this report covers qualitative findings from interviewing service users.

Funding and Ethics

A grant was generously awarded from the Brittle Bone Society. Ethical approval was granted by the North East Newcastle & North Tyneside 2 Research Ethics Committee.

Challenges Faced

The study had a number of limitations. By nature of being a qualitative study, the sample size was small, so it is reasonable to expect that not all views or experiences may be represented here. Data saturation was not met by the sample size. Purposeful sampling of families who had previously engaged in research was used in order to increase the likelihood of engagement, but this could have biased the sample. Further sampling bias could have been introduced by recruitment of participants from the Brittle Bone Society, who were self-selected, and may have had strong opinions or more extreme experiences compared to others.

All participants were married mothers of affected children. It is possible that the experiences of different family structures may vary from the sampled group. Additionally, only one parent was interviewed in each of the families, so it is possible that the individual experiences and opinions of their spouses could have been different. There could be an observable difference in male and female experiences, particularly with regards to raising safeguarding concerns, as there is evidence that male parents are more likely to be suspected of causing NAI than females given the same circumstances.

All of the participants were parents of affected children. This, of course, does not represent patients who receive a diagnosis in adulthood, adult patients who can recall their diagnostic journeys well, nor does it capture the viewpoints of patients in childhood. There is evidence in the literature that children tend to cope with and adapt to their diagnoses better than their

parents do, but there is little understanding of how children feel while trying to obtain a diagnosis for their problems, which was also outside of the scope of this study.

The sample group only included individuals known to the HSSCA, which, as the name suggests, cares for patients with the most severe, complex, or atypical cases of OI. It is therefore reasonable to hypothesise that the experiences of individuals who do not meet the criteria to be managed by this service may vary significantly.

On reflection and review of the transcripts, there was a tendency for participants to focus more on the process or procedural aspects of their diagnostic journeys, where the research also aimed to uncover more related to the individual emotional and psychosocial impacts related to trying to obtain a diagnosis. This may reflect a flaw in the methodology of the specific pre-planned questions. Interviews were designed, conducted, transcribed, coded, and analysed by the same single individual, which had the potential to introduce observer bias based on what the researcher assessed to be the most important or relevant to the research question.

As 7 of 8 interviews were conducted via telephone, physical cues could have been missed, so there may be additional psychosocial concerns which were not addressed in these interviews.

All participants were interviewed several years after they had gone through their diagnostic journey, so there is potential that recall or perceptions may have been altered due to hindsight or a prolonged period of reflection. As each of these participants had diagnoses several years ago, it is possible that more recent experiences are quite different, given the improved access to, and interpretation of, genetic testing in recent years.

Strengths of the Study

This study represents unique insights into the pathways and processes by which families were able to obtain a diagnosis of OI in their affected children. All participants described unique stories, but shared many inherent experiences throughout the process. Each experience was a sum total of both highly positive and negative experiences and, with a degree of retrospect in each of their journeys, some interviewees implied a sense of catharsis from being able to describe their experiences. Experiences appeared to vary depending on individual perceptions, core values, and the severity of the condition in their child. Three main themes were identified in the study, but it is clear that many of these themes have overlapping content with another.

The study findings are consistent with the literature with respect to the negative emotions associated with the diagnostic journey, including time points prior to diagnosis, at the point of diagnosis, and the period of adjustment following a diagnosis. There is literature to support the finding that parents of children affected with rare diseases experience relief in addition to these negative emotions as a result of concluding a prolonged diagnostic odyssey, which is reflected in participants' responses. Participants indicated that an earlier diagnosis would have been beneficial to their emotional wellbeing, which remains in line with previous research showing the impacts of timing and diagnosis.

This study also demonstrates the importance of the Metabolic Bone Disease Team (MBDT) as specialists in patient care but also highlights that there can be problems with initial access to that care, sometimes due to a lack of awareness or education of the disease. It is clear that diagnosis can be improved by inter-professional education and training – so there may be an additional role for the MBDT in forging or fostering improved links with the wider medical community, making their specialist knowledge more easily accessible.

Future Research

It would be useful to target patients at various "points" in the diagnostic journey in order to establish more contemporaneous views. Further and more thorough exploration of a larger sample of patients with milder forms of OI could also be carried out.

Further understanding of how socioeconomic status contributes to safeguarding concerns, and the impact this has on families, would be important to establish. Richer data could be obtained by interviewing families immediately following the conclusion of safeguarding protocols, although there is a concern that such interviews may add unnecessary burden to distressed families. Additionally, it would be interesting to study to what extent parents are suspected of NAI, and whether this has any relation to the severity of OI in their child.

Conclusions

OI is a highly heterogenic and phenotypically variable condition, ranging from very mild to lethal cases. The diagnostic experience for each patient and their family reflects this variability.

Regardless, this study suggests that common themes arising in each case include:

- the need for, and appreciation of, a timely diagnosis, with little delay after the onset of first symptoms;
- the burden that suspicions of NAI place on a family, which potentially delays diagnosis, and;
- the importance of access to specialist services for the diagnosis, management, and support of the condition.

This study adds to the literature that earlier diagnosis and subsequent access to specialist care has a clear benefit for patients, their families, and their relationships with their doctors. Future studies could focus on:

- larger samples, specifically including patients whose diagnosis took a prolonged time, with a deeper examination of demographics;
- comparisons experiences between severe and milder OI sub-types, and;
- evaluation of the effectiveness of attempts to improve education, awareness, and access to specialist OI services.