Using the RUDY study platform to capture quality of life of adults with rare diseases of the bone

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Objectives: Developing novel therapies for rare diseases is an important research priority. Although current methods involve recruitment from hospital clinics, this may only include the most severely affected individuals and so overestimate the population level burden of rare diseases.
Further, any new therapy will require an economic evaluation before implementation in the UK. However, there is a paucity of data on health-related quality of life measures in patients with rare diseases. We therefore compared quality of life across three rare bone diseases in adult using the EQ5D-5L.

Method: Adults with osteogenesis imperfecta (OI), fibrous dysplasia (FD) and X-linked hypophosphataemia (XLH) were recruited via the Rare and Undiagnosed Diseases Study (RUDY), a web-based platform for patient recuritment and assessment of patient reported outcomes including the EQ5D. The EQ5D-5L utility scores of OI participants were used to generate a cost-utility simulation.

Results: 82 adults completed the EQ5D-5L questionnaire. Overall there was a wide distribution of quality of life with moderate/severe problems commonly reported in the pain and discomfort dimension (OI 60%, FD 56%, XLH 65%). A cost-utility simulation showed that a hypothetical intervention which increased the health utility of the lowest utility tertile of OI patients to the mean utility level of the overall group over 10 years and costing £79,000 would be found cost-effective for the English NHS based on a £30,000 per QALY threshold.

Conclusion: These findings confirm that RUDY is recruiting patients across a range of quality of life with pain and discomfort domains most commonly affected. This is the first study to estimate the cost required to improve quality of life for adults with OI who have the lowest quality of life. A greater understanding of health-related quality of life amongst this population could help guide novel therapy developments and resource allocation.