Rare Musculoskeletal Diseases in Adults: A Research Priority Setting Partnership with the James Lind Alliance

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For which topic were research priorities identified?
rare musculoskeletal diseases

In which location was the research priority setting conducted?
Europe - United Kingdom

Why was it conducted at all?
Osteogenesis imperfecta, fibrous dysplasia/McCune-Albright syndrome and X-linked hypophosphatemia are three rare musculoskeletal diseases characterised by bone deformities, frequent fractures and pain. Little high-quality research exists on appropriate treatment and long-term management of these conditions in adults. This is further worsened by limited research funding in rare diseases and a general mismatch between the existing research priorities and those of the patients.

What was the objective?
to identify the top 10 research priorities for rare musculoskeletal diseases in adults through joint patient, carer and healthcare professional collaboration

What was the outcome?
a ranking list of 10 research questions

How long did the research prioritization take?
December 2015 - November 2018

Which methods were used to identify research priorities?
JLA method

How were the priorities for research identified exactly?
Step 1: setting up PSP: defining scope. Step 2: collecting research questions: patients, their carers and healthcare professionals asked for any questions they had about one of the three rare musculoskeletal diseases (osteogenesis imperfecta, fibrous dysplasia/McCune-Albright syndrome and X-linked hypophosphatemia), 988 questions submitted. Step 3: data processing: cleaning of submissions, out-of-scope questions removed, grouping into overarching questions, remaining uncertainties checked against evidence, data cleaning resulted in 41 indicative questions, 2 questions identified as unknown knowns, 39 questions made it onto the longlist. Step 4: interim ranking: participants were asked to select and rank the ten most important questions from longlist, top 25 questions selected for workshop. Step 5: final prioritization: workshop: several small group discussions, consensus on final top 10 research priorities

Which stakeholders took part?
Patients, their carers and healthcare professionals. Survey: 198 participants: 77% patients with one of the three rare musculoskeletal disorders, 11% carers, relatives or friends, 11% health and social care professionals, and 1% representatives of organizations. Interim ranking: 220 participants: 85% patients, carers, relatives or friends, 14% healthcare professionals, 1% representatives of organizations. Workshop: 18 participants: steering group and others.

How were stakeholders recruited?
The steering group committed to publicising the PSP surveys and outcome through their connections to stakeholders and relevant communities.

Were stakeholders actively involved or did they just participate?
Stakeholders not only participated but were also actively involved in the research prioritization process: They were part of a steering group. The steering group consisted of 6 patient representatives and 5 healthcare professionals. The members promoted surveys, supervised survey development and data processing and participated in the workshop.