The RUDY Study: a novel approach to patient driven research in rare musculoskeletal diseases

Zhang L[1], Watts L[1], Turner A[1], Teare H[2], Gray D[1], Gray N[1], Popert R[1], Hogg J[1], Barrett [1]J, R Luqmani[1], Wordsworth P[1], Kaye J[2], Javaid MK[1]

[1]NDORMS, University of Oxford, UK; [2] HeLEX, University of Oxford, UK

Introduction

Rare disease research is more challenging that studying more common diseases as patients are often geographically dispersed. This means that they may not be able to travel long distances for traditional research studies. Recruitment via tertiary centres alone would result in a sample biased towards those with more severe disease. We describe the initial results of the RUDY (rare and undiagnosed diseases) study, a prospective cohort study of rare musculoskeletal diseases. RUDY uses a web based platform and patient involvement to maximise engagement and satisfaction.

Methods

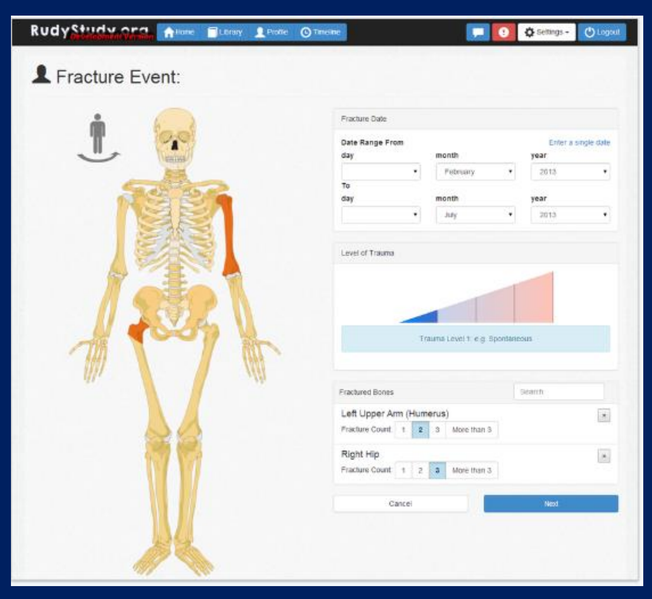
Participants are eligible if they are aged 0-100 and have a rare disease (defined as a prevalence of less than 1 in 2000) or are related to somebody with a rare disease.

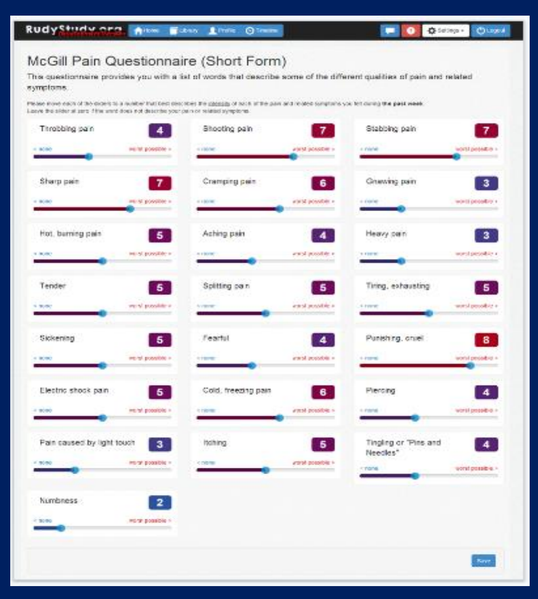
Participants can be referred to the website by their clinical team, patient group, web-search, Facebook or Twitter.

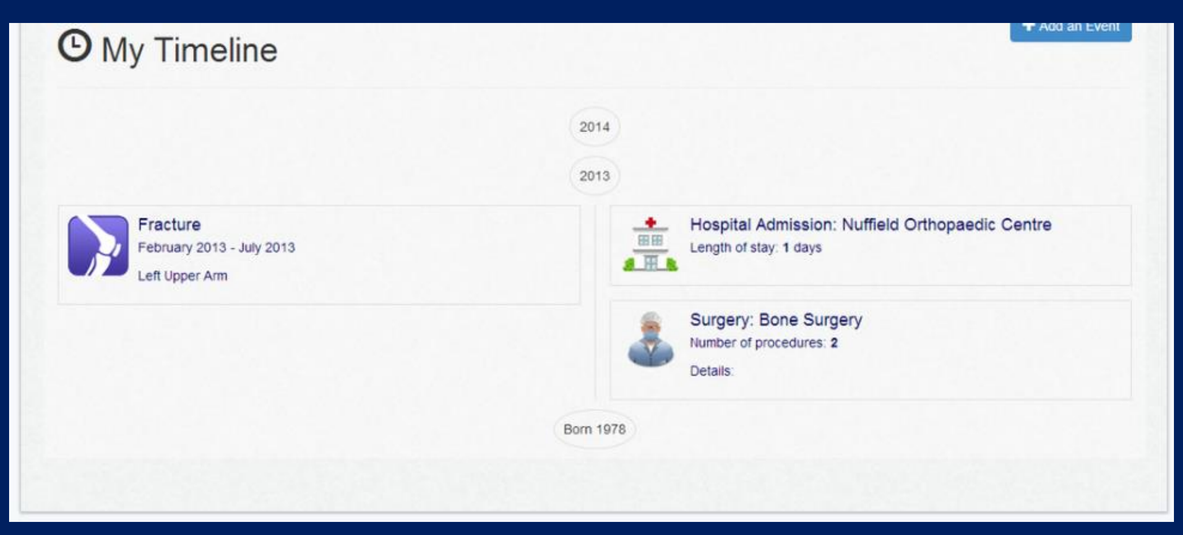
Recruitment occurs via online registration on a secure website followed by a telephone call from the research team and then sign electronic consent form.

Consent is dynamic, with participants able to alter their consent options online at any time.

Data is collected via the secure website, with a timeline of significant clinical events and six monthly validated questionnaires addressing quality of life, pain, sleep, function, fatigue and mood.

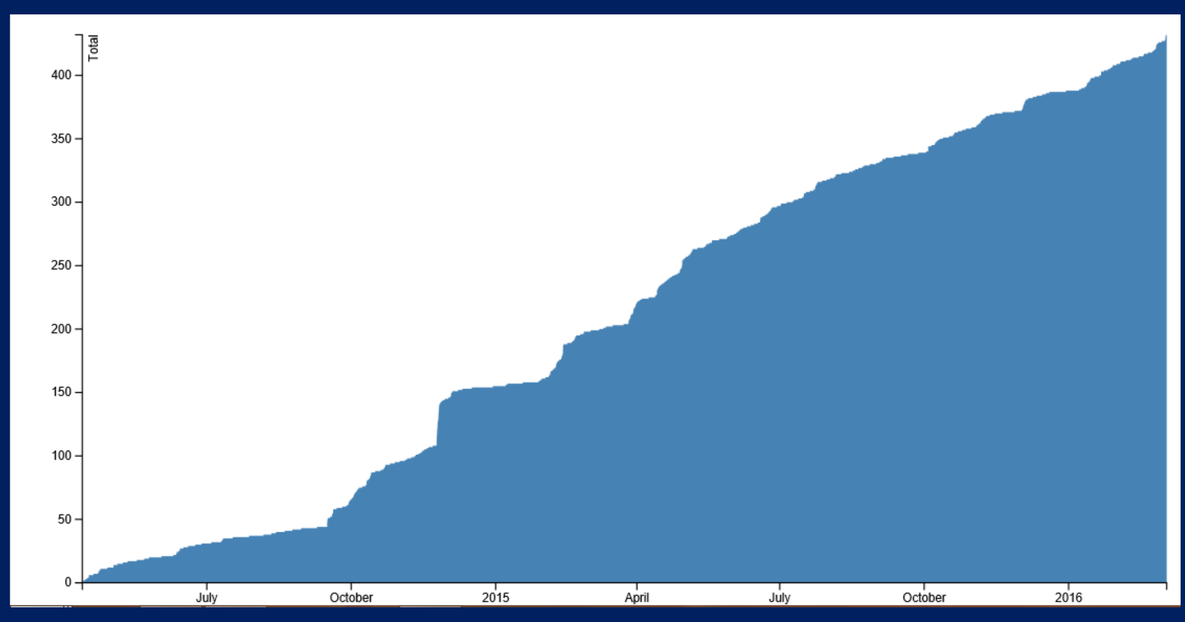


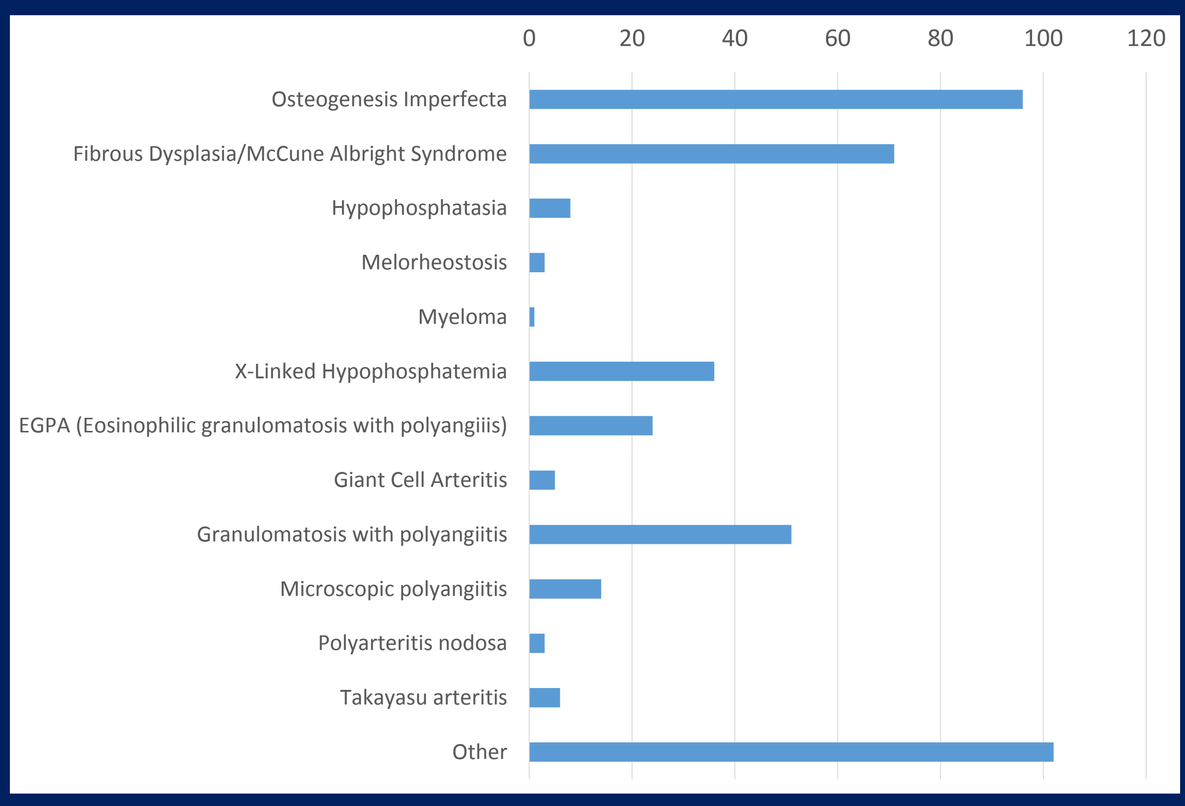




Results

To date, 420 patients with rare conditions have been recruited. This includes 96 with Osteogenesis Imperfecta, 71 with Fibrous Dysplasia/McCune-Albright Syndrome, 51 with granulomatosis with polyangiitis and 36 with X-Linked Hypophosphatemia. Baseline questionnaire completion is in excess of 50% for all conditions. One individual has withdrawn and two have amended their consent options. Participant involvement has been essential, with substantial changes to study design resulting from participant suggestions via the RUDY patient forum.





Conclusions

A web-based platform to recruit, consent and assessment patient reported outcomes has been successfully implemented in the UK using dynamic consent.

A high level of patient participation involvement in study design has facilitated recruitment of a prospective cohort of patients with rare musculoskeletal diseases.

The successful implementation of a language pack opens opportunities to implement this platform internationally.

In association with:

for more information please contact kassim.javaid@ndorms.ox.ac.uk