



The High Bone Mass Study
Musculoskeletal Research Unit
Learning & Research Building (Level 1)
Southmead Hospital
Bristol BS10 5NB
Tel 44 (0)117 4147861
Fax 44 (0)117 4147924

10th December 2015

The High Bone Mass Study Newsletter

Hello, and welcome to the 6th edition of the annual High Bone Mass study update letter, coming to you from the University of Bristol. It has been another busy year of bone activity! Our team has seen four scientific papers published this year based upon the data collected as part of the original study visits to which you contributed, and a further four conference presentations.

Our principal paper this year related to the first of our genetics results; after many years of painstaking genotyping (checking for changes in specific genes), we have found that just a handful (<2%) of people with High Bone Mass have changes in the established High Bone Mass genes (these genes are called *LRP5* and *SOST*) (if this applies to you then we will have already contacted you to explain this), which means the majority of people with High Bone Mass require further genetic study (in progress). We have also been able to make a small, but important contribution to a large international effort to better understand the genetics of bone. In this study, scientists were able to pool summary results from many studies around the world (in all totalling 53,236 people who had had a bone density scan), and in doing so the team discovered a gene (called *EN1*) in which some rare changes can significantly affect bone density. This work helps us better understand the biology of bone. If you would like to receive a copy of either of these papers, please do let us know.

This year we have learnt more regarding the relationship between High Bone Mass and osteoarthritis, from the work conducted by Dr Sarah Hardcastle during her, now successfully completed, PhD. Sarah compared Hip X-rays taken in people with High Bone Mass with those from people with normal bone density. She has shown that people with High Bone Mass may, in later life, develop bony spurs where tendons insert into bone (these are called enthesophytes). This pattern, together with the tendency for people with High Bone Mass to be at increased risk of developing osteoarthritis in both their hip and knee joints, suggests that people with High Bone Mass are more likely to have a generalised 'bone-forming' state (two papers have been published on this topic, if you would like to receive copies then please do let us know). These findings have made us interested to look at how joints change over time in people with High Bone Mass. For this reason, we will be writing to you all next year (2016), as we would like to send you all a questionnaire to see how your bones and joints have been since you last took part in this study. We are also currently applying to research funding charities to see if we can set up a new formal follow-up study to evaluate bones and joints amongst you all in the future!

Aaron Murphy, a 3rd year medical student, working on his Bachelor's degree in Health Sciences joined

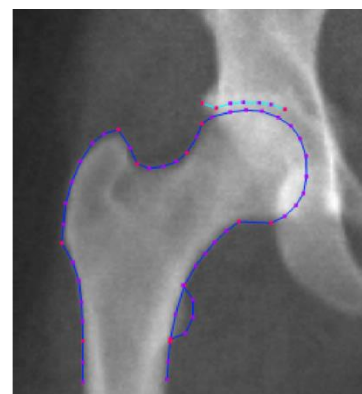


our team last year. He graded all the hand X-rays collected in the High Bone Mass study. He found people with High Bone Mass are more likely to have osteophytes (these are the bony lumps you see in finger joints when you get osteoarthritis). This suggests that the association we have reported to you before, about people with High Bone Mass being more likely to develop osteoarthritis later in life, reflects a generalized tendency, rather than only being seen in weight-bearing joints. Aaron was able to present his findings at the Bone Research Society conference in

Edinburgh in September, where he was awarded a prize for the best oral presentation, and Dr Celia Gregson was able to present this work at the American Society for Bone and Mineral Research in Seattle in October; we are currently finalising the full scientific paper.

We now have a new 3rd year medical student, Anjali Patel, who has just joined our team to work on her Bachelor's degree in Health Sciences. She is using some new computer software, developed by

our collaborators at the University of Aberdeen, to quantify hip shape based on the pelvis X-rays we collected as part of the original High Bone Mass study. We would like to know whether people with High Bone Mass have subtly different hip shapes, and whether this predisposes people to develop osteoarthritis in their hips. We will hopefully have some results this time next year!



We have also welcomed our new High Bone Mass Study administrator, April Hartley, to the team this year. April has been updating all your contact details and will be overseeing the postal questionnaires (mentioned above), which we plan to send out to you all next year. So, as long as you remain happy, you will be hearing from her in 2016.

Once again we would like to express our sincere thanks to you all for your time and interest – without our study participants, this exciting work would not be possible. We also continue to be grateful for the generous financial support we currently receive from Arthritis Research UK, as well as past funding from The Wellcome Trust and the National Institute for Health Research.

If your contact details change, then please do remember to let us know. From all of the team, Season's Greetings and all the best for the New Year.

With thanks and best wishes

Yours sincerely

Dr Celia L Gregson

On behalf of:

Chief investigator: Prof Jon H Tobias, email: jon.tobias@bristol.ac.uk

Study co-ordinator: Dr Celia L Gregson, email: celia.gregson@bristol.ac.uk

Clinical research fellow: Dr Sarah A Hardcastle, email: sarah.hardcastle@bristol.ac.uk

Study administrator: Miss April Hartley, email: april.hartley@bristol.ac.uk

University of Bristol, website: <http://www.bristol.ac.uk/clinicalsciencenorth/musculo/>

