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Application Number / Title
20217 - Pubertal timing variants across the trait distribution and musculoskeletal health parameters

Keywords
Puberty, menarche, musculoskeletal, osteoporosis

Application Lay Summary

1a: Alterations in puberty timing are associated with adverse adult health risks, including osteoporosis, a major health burden in the elderly. Pubertal timing is inversely related to bone mineral density, which tracks throughout life and predicts osteoporosis risk. While based in Finland, the applicant contributed to large-scale genetic discovery efforts revealing 130 genetic variants associated with puberty, using linear regression methods to model mean phenotypes like age at menarche. No studies have further tested these loci across percentile ranks. Thus, we aim to characterize puberty-associated loci across the trait distribution for pubertal timing and skeletal health measures and outcomes.

1b: The UK Biobank has a stated purpose to improve the prevention, diagnosis, and treatment of illness. Pubertal timing is highly associated with musculoskeletal health and disease, including osteoporosis, arthritis, and spondylitis. Despite strong evidence that puberty and adult health are interrelated, the underlying mechanisms remain to be elucidated; only by characterizing puberty loci in-depth will we gain a better understanding of how they impact later life health.

1c: Pubertal timing and skeletal outcomes such as bone mineral density and osteoporosis will be assessed by quantile regression for their association with pubertal timing variants.

1d: Full cohort of individuals with puberty, bone measures and outcomes, and genetic data.