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Paget's disease genetic markers

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Description of Technology

This technology focuses on genetic markers linked to individual prediction of susceptibility and severity of Paget's disease of the bone.

The technology consists of single nucleotide polymorphisms (SNPs) associated with prediction of Paget's disease susceptibility and severity. Identifying individuals who carry the mutations will allow for close monitoring of disease development, and subsequent therapeutic intervention. Similarly, predicted extent of severity will help inform on the level of patient monitoring and potential prophylactic therapy administration.

Features/Benefits

- Biomarkers shown to predict susceptibility to Paget's disease
- Biomarkers shown to predict the likely severity of the disease
- Biomarkers identified from large scale clinical studies of 737 to 1940 patients

Potential Application

- Diagnostic for identifying individuals at risk of developing Paget's disease as well prediction of disease severity

Key Publications

- Visconti *et al.* (2010) Mutations of SQSTM1 are associated with severity and clinical outcome in paget disease of bone. J. Bone Miner Res.; 25(11):2368-73. http://dx.doi.org/10.1002/jbmr.132
- Albagha *et al.* (2013) Common susceptibility alleles and SQSTM1 mutations predict disease extent and severity in a multinational study of patients with Paget's disease. J. Bone Miner Res.; 28(11):2338-46. http://dx.doi.org/10.1002/jbmr.1975

Open Technology

Patent publication document (Reference WO2011128646A2), detailing invention, methods and results, will be provided following acceptance of the University's Open Technology standard terms and conditions.

University Services Available

Additional consultancy services relating to this technology are available on a commercial basis. If required, please contact:

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