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Invited Plenary Abstract
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Genetics of osteoporosis – what are the questions? How to answer them?

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People have different bone mineral density (BMD) and different susceptibility to osteoporotic fractures (OF), in any given population or across populations. BMD has been used as a major measurable phenotype for defining osteoporosis and (OP) for genetics studies of OP.
A few questions central for genetics studies of OP are listed in the following, and a brief discussion or simplified answer is also given for each question listed. The presentation will discuss these questions in more detail.

Q1: How much of this variation is determined by intrinsic genetic (G) factors, extrinsic environmental (E) factors and the interplay (interactions) of G and E (GXE) factors?
Answer: more than 60% due to G, less than 40% due to E and an uncertain amount due to GXE.
Q2. How to quantify the contribution to BMD variation from G, E and GXE factors?
Answer: By using the information from related individuals, such as families, relative pairs and large pedigrees. Some simple analysis methods will be introduced.
Q3: How much variation in risk to OF is determined by G factors?
Answer: ~50%.
Q4: Are genes for BMD variation at different skeletal sites the same?
Answer: No, but some are shared.
Q5: Are genes for BMD variation and OF the same?
Answer: No, evidence shows that they are largely NOT shared.
Q6: How to identify individual specific genetic factors (genes) for osteoporosis?
Answer: Gene mapping, DNA microarray, and/or proteomics in humans as well as model organisms.
Q7: What are the samples of subjects necessary and how to design experiments for identifying susceptibility genes?
Answer: Families based (linkage, transmission disequilibrium test) or use unrelated subjects (association for candidate genes or whole genome).
Q8: What exclusion or inclusion criteria should be considered in recruiting subjects?
Answer: Some examples will be given for excluding subjects with known abnormal conditions for high or low BMD variation.
Q9: What phenotypes should be studied for genetics of osteoporosis?
Answer: risk phenotypes such as BMD AND OF.
Q10: Can we bypass studying osteoporotic fractures forever?
Answer: No.
Q11: What modern technologies are available to identify osteoporosis genes and their functions?
Answer: Those with whole genome approach such as genome wide linkage/association, DNA microarray and proteomics, plus those with candidate gene studies.