Genetic Approach to Osteoporosis

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Osteoporosis is the most common metabolic bone disease around the world. The most important manifest is the fracture after a low-energy trauma. Osteoporosis is expected to arise in half of women and in one fourth of men over 60 in rest of their lives. It costs billions of dollars in countries like the USA or England because of the need for rehabilitation, surgical intervention and care after fracture. Like other prevalent diseases, osteoporosis is genetically found in the group of 'complex diseases'. In other words, a lot of genes with lesser effects can result in the phenotype of the disease. The most powerful findings showing the risk of fracture is age, having a fracture before and low bone mineral density (BMD). Osteoporosis is a familial disease. The children of people with osteoporotic fractures are more prone to have fracture and have lower BMD. The risk of having low BMD is six times higher than in the normal population in siblings of people who have low BMD independent from gender. Inheritance of BMD is 60% to 90%. Similar inheritance pattern is acceptable for fracture risk, bone turnover and bone architecture. BMD is also affected by environmental factors and age. Genome studies related to bone fractures have shown that all of the identified fracture predisposition loci are correlated with BMD. The studies intended to enlighten the genetic reasons behind the complex studies has identified that more than 60 loci have relation with BMD and close to 15 loci have relation with fracture risk in entire genome. Even though this data opened new scopes, there are still limited applications to prevent the disease even before it occurs. Knowledge of the loci creating tendency to osteoporosis does not provide any further foresight than evaluation of clinical risk factors and BMD measurements, yet this knowledge can find an area of application with personalized life style changes and with pharmacogenetic approaches in the near future when whole exon sequencing and even whole genome sequencing are expected to be available and accessible.

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