

Estrogen receptor gene polymorphism

Estrogens act through binding to two different estrogen receptors (ERs), **Estrogen receptor alpha** (OMIM133430) and ER beta (OMIM601663), which are members of the nuclear receptor superfamily of ligand-activated transcription factors. Different **polymorphisms** have been described in both the ER alpha and ER beta genes. Although a large number of association studies have been performed, the individual contribution of these polymorphisms to the pathogenesis of osteoporosis remains to be universally confirmed. Moreover, an important aim in future work will be to define their functional molecular consequences and their interaction with the environment in the causation of the osteoporotic phenotype.

Park et al investigated the association between degenerative **lumbar scoliosis** (DLS) and **estrogen receptor alpha** (ER α) **gene polymorphisms** in 184 patients with a diagnosis of DLS, by determining the presences of the Pvu II and Xba I polymorphisms, measuring bone mineral densities at the lumbar spine (LSBMD) and femoral neck (FNBMD), and by investigating biochemical markers of bone turnover and comparing these results with those of 220 healthy normal controls.

Genotype frequencies in DLS patients and controls revealed a significant difference for the Pvu II polymorphism only ($p = 0.0287$). No significant difference was found between the DLS and control groups with respect to the Xba I polymorphism, bone mineral density (BMD), or biochemical markers. Furthermore, no significant association was observed between the Pvu II polymorphism and BMD, lumbar scoliosis, lateral listhesis, or biochemical markers in patients with DLS.

These results suggest that the ER α Pvu II polymorphism influences the prevalence of DLS ¹⁾.

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Park YS, Suh KT, Shin JK, Lee JS. Estrogen receptor gene polymorphism in patients with degenerative lumbar scoliosis. Br J Neurosurg. 2016 Jul 11:1-4. [Epub ahead of print] PubMed PMID: 27399961.

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