The Association Between the rs2234693 and rs9340799 Estrogen Receptor Alpha Gene Polymorphisms and Risk Factors for Cardiovascular Disease: A Review


Although estrogen is primarily thought of as the hormone involved in female reproduction, it also plays a role in many additional physiological and pathological processes. Recent studies have demonstrated an association between estrogen and clustered risk factors for cardiovascular disease (CVD), such as lipid and glucose metabolism and obesity-related phenotypes, as well as the occurrence and severity of CVD.

Evidence suggesting a genetic basis for this link is accumulating. Several polymorphisms of the estrogen receptor alpha (ESR1) gene exist that may influence the impact of estrogen, leading to clinically relevant phenotypes. Based on the relationship ESR1 seems to exhibit with CVD risk factors, these polymorphisms may play a role in the mediation of vasoprotective effects, modulation of cardiovascular physiology, and development of risk factors for CVD. The two most frequently studied polymorphisms located in ESR1 are often identified by their restriction: endonucleases Pvull (rs2234693) and Xbal (rs9340799).

In this review, we have evaluated and summarized the results of studies involving rs2234693 and rs9340799 and clustered risk factors accompanying development of CVD. Despite inconsistent findings, these studies provide some support for a relationship between polymorphisms in ESR1 and risk factors for CVD. These summarized findings do not yet support inclusion of ESR1 genotypes in genetic testing algorithms for predisposition to CVD, but they do indicate that further investigation into the potential connection between ESR1 and risk factors for CVD is warranted.