Relationship between BMP-4 polymorphisms and hirsutism in polycystic ovary syndrome: a pilot study


Introduction: Polycystic ovary syndrome (PCOS) is an endocrine disorder characterized by hyperandrogenism and chronic anovulation. The hirsutism is the most common clinical manifestation of the hyperandrogenism. Growth, differentiation and regression of the hair follicle are modulated by a broad range of factors, but exact mechanisms that control it are not fully known. Recent works have pointed that Bone Morphogenetic Protein 4 (BMP4) has a crucial role in hair growth. Thus, we hypothesized that BMP4 polymorphisms might be related to hirsutism in PCOS. Objective: To assess two polymorphisms of BMP4 (rs4898820 and BMP4-538T/C) in PCOS and correlate genetic variations with presence of hirsutism. Methods: Sixty women were enrolled for this study and sorted in three groups: G1) PCOS and moderate to severe hirsutism (Ferriman and Gallwey score >= 15), G2) PCOS without hirsutism (Ferriman and Gallwey score <= 7), G3) controls without hirsutism. PCOS diagnosis was done according to Androgen Excess Society criteria. All participants had blood samples taken for genetic analysis. After blood DNA extraction, Restriction Fragment Length Polymorphisms was performed using enzymes Tas1 and Hph1 for analysis of rs4898820 and BMP4-538T/C polymorphisms, respectively. Results: In rs4898820 polymorphism, the prevalence of the wild GG genotype was similar in all groups, while the prevalence of heterozygous genotype GT and recessive TT varies among groups. The G1 had a frequency of 40% for recessive genotype, while the G2 and G3 showed only 16% and 19%, respectively (P<0.05). In the other polymorphism, the frequency of heterozygosis in G1 (23.53%) is increased in comparison to G2 (12.5%) and G3 (15.79%) (P<0.05). Conclusions: In both polymorphisms, G1 exhibited most differences compared to another groups, indicating that these polymorphisms might be related to BMP4 function in hirsutism.