

Mutations Analysis of the Growth Differentiation Factor 9 Gene in Syrian Women with Ovarian Failure

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ABSTRACT Premature ovarian failure (POF) is a primary ovarian defect characterized by absent menarche or premature depletion of ovarian follicles. Growth differentiation factor 9 (GDF9) plays an important role in normal growth, differentiation, and proliferation of granulosa cells surrounding the oocyte in the ovary. The present study was to verify the involvement of GDF9 variations in a POF woman in Syrian. POF cases (n= 80) consist of (primary amenorrhea PA n=55) and (secondary amenorrhea SA n=25) compared with 200 controls. All cases with POF had a normal karyotype analysis (46XX). Genetic analysis of the GDF9 gene were showed, four variants in 23 patients. Two of novel variants were observed in two patients, the first was [c.1231G<A] and the second novel variant was [c.531T<G]. The document variants [c.447C>T] was observed in 17 patients and 15 controls. But the variant [c.546G>A] was showed in one patient and in one control. The variant [c.447C>T] appeared associated with the variant [c.546G>A] in 3 patients [c.447C>T]+[c.546G>A] (compound heterozygous genotype). this compound variant didn't detect in the controls. The researchers' findings are consistent with the critical role played by GDF9 in human folliculogenesis. The presence of these variants might indicate a higher risk for POF.