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Abstract Title:- Genetics of Polycystic Ovary Syndrome (PCOS): our experience

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Aims:- PCOS is a common endocrinopathy among women of reproductive age, with a worldwide prevalence of 8 to 13%, depending on the criteria used for diagnosis. It is characterized by a constellation of features, including oligo/anovulation, clinical and/or biochemical hyperandrogenism, and polycystic ovarian morphology. PCOS is one of the common causes of female infertility. It is also associated with metabolic derangements, including obesity, insulin resistance, and compensatory hyperinsulinemia, which increase the likelihood of developing type 2 diabetes mellitus. Despite extensive research, the etiology of PCOS remains largely unknown. It seems likely that the hypothalamic-pituitary-ovarian axis dysfunction, partial folliculogenesis arrest, insulin resistance, and ovarian and adrenal androgen secretion may play a role in the pathogenesis of PCOS. Familial clustering of the cases of PCOS points to a genetic component linked with it.

Methods:- The initial genetic studies suggest an autosomal dominant pattern of inheritance of the disorder in some families; however, most studies support multifactorial origin. Since PCOS is a complex trait, the typical form of inheritance of PCOS follows a non-Mendelian pattern and involves complex genetic mechanisms. Studies involving linkage and association have suggested a connection between genetic variations and the risk of developing PCOS in certain families or populations. Through genome-wide association studies and next-generation sequencing techniques, several candidate genes have been identified that play a role in the etiopathogenesis of the disorder.

Results:- Pathogenic variants of various genes such as INSR, IRS1, GHRL, LDLR, MC4R, ADIPOQ, UCP1, UCP2, UCP3, FTO, PCSK9, FBN3, NEIL2, FDFT1, PCSK9, CYP11, CYP17, CYP21, HSD17, STAR, POR, AKR1C3, AMH, AMHR2, INHBA, AR, SHBG, LHR, FSHR, FSH β, SRD5A, GATA4, THADA, YAP1, ERBB2, DENND1A, FEM1B, FDFT1, NEIL2, TCF7L2, etc. in some PCOS cases are linked as underlying etiologic associations.

Conclusions:- This presentation aims to provide insight into the current genetic knowledge about PCOS, including ours. Discovering the genetic factors and pathways involved in the disorder will help us better comprehend the underlying mechanisms of the disorder.

Keywords: Polycystic Ovary Syndrome, Variants, Genetic Associations