INTRODUCTION

Polycystic Ovarian Syndrome (PCOS) is referred to some times as Sclerocystic Ovarian Disease, Stein-Leventhal Syndrome and Polycystic Ovarian Disease (PCOD). PCOS is a complex, heterogeneous, polygenic endocrine disorder in women of reproductive age and is considered as a multifactorial reproductive, cosmetic and metabolic problem. The etiology of PCOS is not well understood and its pathophysiological and molecular basis is still a puzzle. PCOS is likely to be the result of a number of both genetic and environmental factors. Some of the contributing factors to PCOS also include a low level of chronic inflammation in the body and fetal exposure to male hormones. However, androgen excess and insulin resistance leading to hyperinsulinemia are considered to be the basic defects in PCOS that was described way back in 1921 by Archard & Theirs as "diabetes of bearded women".

The world wide prevalence of PCOS syndrome is 6-10% and in its "classical" form may affect 5 - 7% of women. PCOS is quite common in Asian population. A high prevalence of up to 35% is reported for the Indian women and the incidence and prevalence of PCOS in overweight and obese women is greater than 20%. Women with PCOS are at a higher risk for a number of illnesses, including high blood pressure, diabetes, heart disease and other cardiovascular problems and cancer of the uterus, ovary and breast.

PCOS also presents with a variety of biochemical abnormalities. The most consistent abnormality is hypersecretion of androgens. Because of the high degree of
heterogeneity of PCOS, it is suggested best to consider PCOS as increased androgens clinically (acne, excessive hair on face, abdomen, or thinning of scalp hair) or in the blood (total or free testosterone, DHEAS), with oligo-ovulation (cycles greater than every 35 days, low mid-luteal progesterone, monophasic basal body temperature.

PCOS has yielded some positive results but the controversy on the mode of inheritance (eg. autosomal dominance, modified autosomal dominance, X-linked, multifactorial) still persists. Thus, there is a great need to identify the potential candidate genes that may have a modest effect individually and in groups in PCOS. Three general genetic models have been proposed namely,

- **Single gene Mendelian model** which predicts that there is single gene defect inherited in a recessive or dominant pattern and that woman who inherit this defect develop clinically evident PCOS.

- **Multifactorial model** where PCOS is considered as a multifactorial genetic disorder and women carrying this defect through inheritance or environmental factors will have increased risk of clinical PCOS.

- **Variable expression single gene model** where there is a combination of the above two models. It follows that a single gene defect is present but its expression is modified by environmental factors. Therefore, a lady who is genetically predisposed but not exposed to environmental factors may develop only subclinical forms of PCOS and not the full disorder. This theory explains the heterogeneous nature of the disorder.

Therefore, it can be said that what started initially as a gynaecological curiosity, over the years has become a subject of multisystem endocrinopathy known
as PCOS. Considering certain lacunae in the area of genetics of PCOS as pointed out earlier, we propose to study the family history of PCOS subjects, not only for PCOS but also for its related conditions. This would help us to determine the pattern of inheritance of PCOS and related clinical presentations.

**Rationale of the study:**

PCOS is a very common endocrine problem, However its pathophysiology, clinical features, multiple endocrine problems and associated metabolic conditions are poorly understood. There are very few studies on genetic polymorphismsmsims affecting steroid synthesis, Many of them lack clinical correlation hence this study was chosen to connect genetic polymorphisms in cortisol and androgen synthesis as the culprit for the syndrome with clinical correlation.