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Abstract Topic: - Epigenetics

Abstract Title: - Genome-wide DNA methylation analysis in blood identifies differentially methylated regions related to polycystic ovary syndrome- a pilot study

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Aims: - To investigate PCOS-associated differential DNA methylation regions

Methods: - A case-control study comprising eight women with PCOS diagnosed under the Rotterdam criterion and eight age and BMI-matched healthy controls were recruited. Subjects were classified into four groups based on BMI: Group 1 consists of controls with BMI <25, Group 2 consists of PCOS cases with BMI <25, Group 3 consists of controls with BMI ≥25, and Group 4 consists of PCOS cases with BMI ≥25. Group 1 and Group 2 were further considered as study comparison 2. Similarly, Group 3 and Group 4 were considered as study comparison 4. Genome-wide DNA methylation in whole blood had been evaluated using an Infinium Human Methylation 850k array

Results: - PCOS-associated differential methylation regions including sites, promoter region, and CpG sites considering ($p \leq 0.01$) significance level. Thirty-four hypo methylated and one hundred twenty-six hypermethylated sites encoded by genes were significantly associated with PCOS in comparison 2. In comparison 4, twenty-nine hypomethylated genes and fifty-nine hypermethylated genes were identified. Most of these genes are involved in estrogen biosynthesis, lipid biosynthetic process, and activation of matrix metalloproteinases (MMP). The genes which are common in both the comparison groups are mostly involved in the Inflammasome pathway.

Conclusions: - The study provides information about DMRs in the blood tissue which may be investigated further to understand underlying pathogenesis and as therapeutic targets in future.

Keywords: - PCOS-associated differential methylation regions including sites, promoter region, and CpG sites considering ($p \leq 0.01$) significance level. Thirty-four hypo methylated and one hundred twenty-six hypermethylated sites encoded by genes were significantly associated with PCOS in comparison 2. In comparison 4, twenty-nine hypomethylated genes and fifty-nine hypermethylated genes were identified. Most of these genes are involved in estrogen biosynthesis, lipid biosynthetic process, and activation of matrix metalloproteinases (MMP). The genes which are common in both the comparison groups are mostly involved in the Inflammasome pathway.