MITOCHONDRIAL DNA MUTATIONS AND ND1 GENE COPY NUMBER IN PATIENTS WITH POLYCYSTIC OVARY SYNDROME (PCOS)

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Present study analyze the possible mutations in mitochondrial encoded genes TRNL1 and ND1, the association of mtDNA copy number in PCOS women with and without diabetic mothers was evaluated. Blood samples were collected from all the individual participants of study and detailed anthropometric and demographic variables were collected. Sequence of each amplified product of RNA gene TRNL1 was analyzed in comparison with the updated Cambridge Sequence. The PCR amplified products of ND1 were digested using Hae III and TaqI and were run in electrophoresis in 4 % agarose gel electrophoresis. Relative quantification of mtDNA was compared with unique nuclear encoded gene based on the ratio of mtDNA to unique nuclear encoded gene using Real Time PCR. The group was effectively and closely similar for age and BMI. Aged varied between 24–25 years and 50–51 years for daughters and mother respectively. Results identified a five different type of mutation in study sample, among which only one was seen in non PCOS group, rest other four were found in both daughters and mother of PCOS group. All these nucleotide point mutation were verified by sequence analysis of both strands and appeared to be heteroplasmy Of the 36 PCOS samples analyzed one sample showed the G3316A mutation in the homoplasmy state. Nonsynonymous mutation leads to amino acid change from Alanine to Threonine. Analysis of the copy number in mothers revealed that mothers of PCOS women too had less copy number when compared to the mothers of Non-PCOS. Mtcopy number was significantly reduced in PCOS women with diabetic mother when compared to the PCOS women with non-diabetic mother and also the Mtcopy number was less in PCOS women of non-diabetic mother with diabetic family history. Though evidence is not confirmatory to say that PCOS is a phenotypic feature of MIDs. This study on mtDNA variants reported with small number of sample in association with PCOS by a single centre can be justified with more number of samples in multicentre studies. To explore more of hereditary PCOS, whole exome (genome) sequencing is suggested.

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