2019 Young Investigator Award Winner

643: Does the CGG repeat size and composition at FMR1 gene explain the unexplained recurrent spontaneous abortion?

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Objective
Recurrent spontaneous abortion is multifactorial disorder and till date various factors have been attributed in its pathogenesis. Still approximately 50% of RSA cases remain unexplained. Premutation (PM) expanded allele of fragile-X mental retardation 1 (FMR1) gene is known to contribute to ovarian dysfunction in 20% of the cases. Recently, link between expanded FMR1 allele and recurrent miscarriages have been reported.
Design
The present case-control study was conducted in women with RSA of Indian origin comparison to age matched healthy control women (N= 100 each) during the period from 2015 to 2019
Materials and Methods
We have investigated the status of CGG repeat size at 5'UTR of the FMR1 gene in all cases and control samples. The genomic DNA from these samples was subjected to molecular analysis for characterization of CGG repeat size and composition at FMR1 gene
Results
As compared to the control women, the RSA women cohort had a higher frequency of carriers with expanded alleles in grey zone (GZ) and PM range i.e. 2% (2/100) verses 5% (5/100) respectively. Also, the RSA cohort had a significantly higher number of normal alleles with ≥35 CGG repeats (24 out of 200 alleles) as compared to control group (8 out 200 alleles). The number of larger FMR1 alleles with pure CGG repeat tract was found to be significantly higher (P= 0.0058) in the RSA group (17 out of 200 alleles) as compared to that in control group (4 out of 200 alleles).
Conclusions
Henceforth, the CGG expanded uninterrupted FMR1 allele might be associated with recurrent abortions and may help to explain many of these unexplained cases.
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Disclosure
Authors have no conflict of Interest.