642: Molecular screening for FMR1 expanded allele in Indian females for fragile X carrier detection

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Objective
Premutation (PM) carriers are reported to be more frequent in population in comparison to FM (Full mutation) with a frequency as high as 1 in 113–259 women and are unstably transmitted to the offspring as FM, when passed onto offspring through female. In spite of a high frequency of PM carriers and its known risk of expanding to FM in subsequent generation, most PM carriers are unaware of their condition. Thus depicting the importance of tracing potent women PM carriers in order to decrease the disease load in the society. The main objective of the study is molecular screening of females of reproductive age for presence of PM expanded FMR1 (Fragile X mental Retardation 1) alleles.
Design
The study was conducted in 500 reproductive age females of Indian origin and extended family screening will also be offered to identified PM positive cases during the period from 2015 to 2019.
Materials and Methods
A previously validated laboratory-developed test using triplet-primed polymerase chain reaction (TP-PCR) was used to identify PM alleles in 500 Indian reproductive age females.
Results
We identified 2 (0.4%) asymptomatic PM and 1.6% (8/500) GZ (grey zone) carrier females in reproductive age cohort. Extended family screening was possible for 4 of 8 (60%) GZ and for both (100%) PM females.
Conclusions
In spite of clinical significance of conducting screening program for identification of the carrier status of reproductive age women, such objective had greatly suffered due to unavailability of accurate, cost effective and rapid molecular techniques. The indigenously developed TP-PCR in our study is cheaper and in comparison to available commercial kits and thus it has proved to be economically more feasible to be used in screening program.
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Disclosure
Authors have no conflict of Interest