

THE ROLE OF THE FRAGILE X SYNDROME IN THE DEVELOPMENT OF

PREMATURE OVARIAN FAILURE AND IVF INEFFICIENCY

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Key words: Fragile X syndrome, FXS, POF

Introduction: Fragile X syndrome (FXS) is a rare genetic disease associated with mental retardation and autism or autism spectrum disorder, females with premature ovarian failure (POF), and older males with progressive development of intention tremor and ataxia with or without dementia.

Methods: DNA was used by standard isolation of lymphocytes from peripheral blood of 30 women with reproductive disorders. Molecular genetic analysis was performed by PCR method with electrophoresis on the 310 Applied Biosystem by using the TP-PCR Primers test system (AbbottMolecular).

Results: The national composition is represented by Kazakhs - 17 (56,7%), Russians - 6 (2%) and other nationalities - 7 (23,3%). 11 women (36,7%) have somatic diseases such as chronic pyelonephritis, chronic bronchitis, etc. Analysis of pregnancy outcomes revealed severe reproductive disorders. The average number of pregnancies per 1 surveyed woman is only 0.7 ± 0.09 ; the average number of births is 0.34 ± 0.04 ; the average number of spontaneous early miscarriages is 0.36 ± 0.04 .

The average number of children was 0.33 ± 0.06 . 20(66,7%) of women have infertility (13 women have primary infertility and 7 have secondary). 25 programs of embryo transfer were carried out for the surveyed women. 20 programs (80%) of them were ineffective, pregnancy was confirmed only for 4 (20%) cases, which in 100% resulted in spontaneous early miscarriages. Studied women altogether have 10 children (30% of them with *undifferentiated* forms of *mental retardation*).

18 of the subjects ($60 \pm 5.6\%$) have a normal number of CGG-repeats; premutation in 7 cases ($23.3 \pm 4.4\%$); in the gray zone - 5 ($16.7 \pm 4.2\%$). Full mutation of the FMR1 gene was not detected.

Conclusion: The high frequency of premutation and the high risk of having children with mental retardation confirm the recommendations of ASOG that it is necessary to carry out molecular genetic test for the first ineffective attempt of IVF and for egg donors.