Cytogenetic studies: important but ignored tests for evaluation of diminished ovarian reserve patients

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Context: Pathologic diminished ovarian reserve (DOR) from cytogenetic viewpoint.
Objective: To evaluate possible chromosomal aberration that may lead to DOR, as well, establish the cutoff rates in females with low-level X chromosome mosaicism to help prevent detection escaping.
Methods: Retrospective observational study which was conducted on clinical features, biochemical parameters and cytogenetic findings followed by additional tests as complimentary (FISH on granulosa cells).
Setting: Medical genetics clinic in a research institute for reproductive biomedicine.
Patient(s): 284 individuals including 174 cases with DOR, and also 110 women were looking for elective sex selection for family balancing as control group.
Intervention(s): Conventional cytogenetic method and fluorescence in situ hybridization (FISH) on granulosa cells as second tissue; given its ovarian origin.
Main Outcome Measure(s): Karyotype, and FISH.
Results: The overall prevalence of chromosomal abnormalities was 10.34% which 2.3% of it belongs to normal variable chromosome features which was significantly higher than that of in control group (p=0.027). Regarding females with low-level X chromosome mosaicism, those who have 4% of frequency were evaluated in-depth using FISH on their granulosa cells, which surprisingly showed 13% of X chromosome mosaicism.
Conclusions: Our results emphasis on the critical role of chromosome abnormalities as one of the possible etiologies for pathologic DOR and an important diagnostic consideration for ovarian follicular attrition. Moreover, chromosomal aneuploidy cutoff rates should decreased, as 4%, to help prevent detection escaping in DORs.