Karyotype Polymorphism and its significance in Idiopathic and in isolated male or female factor infertility

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There are certain influences, which have been documented to have an effect on the genetic conduct of a conception, be it natural or assisted. These are namely consanguinity, monogenic or single gene disorders; sex linked disorders, hormonal imbalance and environmental factors. The end result of any of these could be repeated miscarriages, phenotypic abnormalities, metabolic syndromes or carcinogenesis.

In the era of genetics and assisted reproduction it becomes imperative to assess couples chromosomally to eliminate any genetic cause in the pre-IVF workup such as to chromosomal variants or polymorphisms.

Karyotype polymorphisms are found in men and women, trying to conceive, with no other implicating factors. We performed a retrospective study on 4649 infertile patients having polymorphisms in the following groups Idiopathic, Bad obstetric history, male factor, Polycystic ovarian syndrome and endometriosis. The idiopathic group acted as control, to determine if the additional presence of a polymorphism added to fertility failure and if so which type was commonly found. Outcomes in all groups were determined.

Translocations in both or either partners have been known to contribute significantly especially in miscarriages. One case opted for PGS where none of the embryos were found fit for transfer.

The commonest findings especially in idiopathic group was interstitial deletions of long arm of chromosome Y(Yqh), translocations and increase in heterochromatin or satellite on either long or short arms of autosomes or Y chromosome.

Literature survey has shown that in patients who have fertilization failure termed idiopathic, the proportion of women with chromosomal abnormalities was higher than their male counterparts, about 6.4%.

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Stern et al noted the rate of balanced reciprocal and Robertsonian carriers were 3% in those who had failed at least ten embryo transfers.

Hence our study attempts to determine the significance of karyotype polymorphisms in above mentioned groups and their outcomes.