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Abstracts of Research Papers

1

Genetics of male infertility: Y chromosome microdeletion test plays important role

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Introduction: The worldwide burden of infertility is around 8–12% of all couples. The male factor is responsible for around 50% of infertile couple. Intracytoplasmic sperm injection (ICSI) has generated a hope for infertile man to have his own child. On one hand it has set a ray of hope to these infertile men while on the other hand doctor has the liability to properly guide these patients.

Material and method: On going through literature in Google Scholar, Pubmed and Cochrane library I found many publications on Y chromosome microdeletions in male infertility. Out of which I have selected recent ten years' papers for review.

Results: There are several tests available to diagnose male infertility. Cytogenetic analysis and Yq microdeletion analysis are mandatory tests. Azoospermia factor (AZF) region in Y chromosome is important gene responsible for infertility. Three non-overlapping regions responsible for spermatogenesis are present in AZF region on long arm of Y chromosome which are azoospermia factors (AZFa, AZFb, AZFc). AZFa is expressed in spermatogonial stem cells involved in early division of spermatogenesis. AZFb is expressed in primary spermatocytes i.e. premiotic germ cells. AZFc has variable heterogeneous phenotype. Y chromosome microdeletions test has an important diagnostic and prognostic value.

Conclusion: Before going for ICSI, the couples must be thoroughly investigated otherwise inadvertently spouse may transmit infertility to offspring.

Conflicts of interest

The author has none to declare.

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2

Karyotype findings in bilateral cryptorchidism and non palpable gonads with/without hypospadius: A preliminary study

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Background: Cryptorchidism (or undescended testis) is a condition seen in newborns where one or both of the male testes have failed to descend down into the scrotal sac. About 3-5% of males are born with undescended testis of which 10% are bilateral. About one-third of premature males are born with undescended testis which generally descends into scrotum by 6months of age, while rest may subsequently require hormone therapy and surgical interventions. All cases of bilateral cryptorchidism with hypospadius or non palpable gonads or both must be evaluated for intersex conditions. We studied the incidence of chromosomal anomalies in patients of bilateral cryptorchidism with hypospadius or non palpable gonads or both to determine the value of routine karyotyping in this population. In older children testicular ascent probably represents ectopic testis and does not require a chromosomal analysis. Peripheral blood samples from 30 cases of bilateral undescended testis with hypospadius or non palpable gonads or both were studied for chromosomal analysis by traditional karyotype at 450-550 band resolution.

Methods: 5 ml of venous blood was cultured for leucocytes and subsequently karyotyped using standard protocol of Trypsin Giemsa banding.

Results: Chromosomal anomalies were detected in 04 cases with bilateral cryptorchidism and isolated hypospadius and in 02 case of bilateral cryptorchidism associated with hypospadius and non palpable gonads.

Conclusion: All cases of bilateral cryptorchidism with hypospadius or non palpable gonads or both must be evaluated for intersex conditions. The aetiology of intersex conditions is variable. A chromosomal analysis helps to establish the genetic sex and provides surgeon with vital information required before proceeding on Hypospadius repair surgeries and orchidopexy.

Keywords: Cryptorchidism; Hypospadius; Gonads; Karyotype