Journal of the Anatomical Society of India 67S (2018) S26–S74

Contents lists available at ScienceDirect

Journal of the Anatomical Society of India

journal homepage: www.elsevier.com/locate/jasi

Abstracts of Research Papers

1

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

M.B. Sinha*

Department of Anatomy, All India Institute of Medical Sciences, Raipur, Chhattisgarh, India

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.

https://doi.org/10.1016/j.jasi.2018.06.002

2

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

Debasis Bandopadhyay*

Dept of Anatomy, Armed Forces Medical College, Pune 411040, India

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

Conflicts of interest

The author has none to declare.

https://doi.org/10.1016/j.jasi.2018.06.003

3

Mutational analysis of Tnnt 2 gene in dilated cardiomyopathy patients in north Indian population

Rubi Bhola*, Om Shankar, Rashmi Gupta, Preeti Kumari, Royana Singh

Department of Anatomy, Institute of Medical Sciences, Banaras Hindu University, Varanasi, India

Objective: This study was conducted to identify the possible genetic change in Dilated Cardiomyopathy in North Indian Population.

Material and Methods: Blood samples of dilated cardiomyopathy patients were collected from Cardiology OPD, Sir Sunderlal Hospital, Banaras Hindu University. DNA was isolated using salting out method. PCR was done to amplify exons 14 and 15 of TNNT2. The PCR product was sequenced to detect the mutational changes in Exon 14-15 of TNNT 2 gene.

Results: 39 intronic variations were reported. A frame shift mutation was found as insertion of A reported at 201330420_20133042 position in one patient. At 201330424_201330425 position insertion of C, at 201330417_20133041 position insertion of G reported, at 201331130_201331131 position insertion of T reported and at 201331093_201331093 position deletion of C reported in four subjects. in one patient original stop codon was reported at 201331044_201331044 position due to deletion of A. Several missense variant were also reported at 201330429T>C, 201330729G>A, 201330429T>C, 201331093C>G, 201331111T>G positions in more than 1 subjects. PolyPhen result in TNNT2 gene, there were, 4 were benign, 3 possibly damaging and 3 probably damaging.

Conclusion: Various synonymous and non synonymous variations had been reported. Several intronic variation, frameshift mutation and missense variation are reported, that suggest these variation may be responsible for pathogenesis of dilated cardiomyopathy in patients of North Indian population.

Conflicts of interest

The authors have none to declare.

https://doi.org/10.1016/j.jasi.2018.06.004

4

Genomic instability in workers occupationally exposed to cement dust

K.N. Krishna*, S. Ranjith, Ursula Sampson, P.T. Annamala, Kumudam M. Unni, Alex George

Department of Anatomy, Jubilee Mission Medical College and Research Institute, Thrissur, India

Introduction: Humans employed at high risk occupational conditions are recommended to undergo continuous bio-monitoring to suggest genetic risks related to genotoxicity. Cement is one of the most widely used construction materials and as a whole and its individual compounds are classified as chemical hazards. Chronic exposure to cement may result in genomic instability.

Purpose/Objective: To study the genomic instability in workers occupationally exposed to cement dust.

Materials and method: Thirty head load workers employed fulltime at cement godowns and thirty unexposed healthy individuals of same age and gender were considered as case and control groups respectively. Nuclear anomalies were assessed by CBMN Cyt assay in cultured lymphocytes. DNA damage was analyzed by COMET assay.

Result: A significant increase in the number of micronuclei is observed in exposed group (22.63 ± 7.45) compared to unexposed (2.96 ± 1.15) ($P < 0.0001$). Similarly an increase in Nuclear buds ($P < 0.0001$) and Nucleoplasmic bridges ($P < 0.0001$) were noticed. An increase in tail length were noticed in exposed (16.26 ± 7.79) compare to unexposed (7.40 ± 2.87) with a significant P value ($P < 0.0001$). Comet tail length showed a significant increase in the initial years of exposure whereas the number of micronuclei showed a steady increase with the years of exposure.

Conclusion: In present study we observed statistically significant increase in nuclear aberrations and an increased tail length of comets among workers occupationally exposed to cement dust which represents an increase in DNA damage. Hence the study indicates genomic instabilities among workers occupationally exposed to cement dust. Proper guidelines and safety measures have to be advised to avoid the ill effects of cement dust.

Conflicts of interest

The authors have none to declare.

https://doi.org/10.1016/j.jasi.2018.06.005

5

Chromosomal abnormalities in infertile men with azoospermia and oligospermia

A.Z. Drugkar*, S.D. Gangane, R.M. More, S.A. Drugkar

Department of Anatomy, C.C.M. Med. Col. Durg, India

The present study was carried out to find out frequency of chromosomal abnormalities in infertile males with azoospermia & oligospermia.50 males referred for complaints of infertility with azoospermia & oligospermia were included in the present study. The study was carried out in the following steps. 1) Selection of patients 2) Clinical examination of patients 3) Collection of blood and karyotyping 4) Photomicrography 5) Data tabulation and Analysis. Among the total 25 azoospermic males, 8 patients showed abnormal karyotype. Among these abnormal karyotypes, 3 patients showed 47XXY karyotype, 2 patients showed 46XX karyotype, 46XY(20%)/47XXY(80%) was found in 1 patient, 1 patient showed 47,X,i (Xq)Y & 1 patient showed a 45,XY,-22 t (14/22) karyotype. Seventeen patients had normal karyotype. Among the total 25 oligospermic male, 3 patients showed abnormal karyotype. Among these abnormal karyotype, 1 patient showed mosaic Klinefelter i.e. 46XY(20%)/47XXY(80%), 1 patient showed a karyotype of 46,XY, inv(9) and one patient showed 46,XY, large Y.

Keywords: Karyotype; Chromosome; Infertility; Azoospermia; Oligospermia