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

Conflicts of interest

The authors have none to declare.

https://doi.org/10.1016/j.jasi.2018.06.006

6

Lymphocytes DNA damage from panmasala, gutkha, kharrah, chewing tobacco users and chain smokers of central India, by using single-cell gel electrophoresis assay

D.D. Ksheersagar*, V.M. Paikrao

Department of Anatomy, NKP Salve Institute of Medical Sciences & Research Centre, Nagpur, India

Background: Oral submucous fibrosis (OSMF) is a chronic, complex potential potent pre-cancerous condition characterized by juxta-epithelial inflammatory reaction and progressive fibrosis of the lamina propria and deeper connective tissues. As the disease progresses, the jaws become rigid to the point that the sufferer is unable to open his mouth. These events are further influenced by exposures to carcinogenic agents including panmasala, gutkha, kharrah, tobacco consumption and smoking. Single-cell gel electrophoresis assay or comet assay is a sensitive and rapid method for DNA strand breaks; it further provides information on amount of damage among individual cells.

Aim and Objective: In this study, we are aimed to analyse the lymphocyte DNA damage from panmasala, gutkha, kharrah, chewing tobacco users and chain smokers of central India, by using Single-cell gel electrophoresis assay.

Materials & Methods: The peripheral blood samples from 60 addicted participants of age group 30–70 years were collected under sterile conditions in heparinised tubes used for Leukocytes culture and 30 healthy non-OSMF participants of same age group ware taken as control. The informed consent was obtained. The comet assay conducted using three well OxiSelect^{TM} Comet Assay Kit and stained with vista green dye, the slides were analysed by using Olympus$^{®}$ BX 51 fluorescence microscope. The results were statistically analysed.

Result: Mean age of participants were 45.31 ± 16.24 (SD). Obtained comets were analysed by the CometScore 1.5 Software. The Comet score analysis shows that the mean % TDNA (Tail DNA) of comet in Leukocytes of addicted participants is found to be 32.61 ± 18.19 (SD) than 6.09 ± 3.17 (SD) mean % TDNA of control participants.

Conclusion: It can be conclude that addiction to panmasala, gutkha, kharrah, chewing tobacco users and chain smokers can damage DNA of peripheral blood leukocytes.

Conflicts of interest

The authors have none to declare.

https://doi.org/10.1016/j.jasi.2018.06.007

7

Genetical basis of Down syndrome

G. Panneer Selvi*, K.R. Srinivasan, B. Senthil Kumar, V. Rajitha, G. Rekha, Deepti Shastri

Department of Anatomy, VMKV Medical College, Salem, Tamil Nadu, India

Objective: The study was designed to evaluate the karyotype pattern, maternal age, clinical features and other systemic anomalies of Down syndrome cases in Tamilnadu population.

Methods: Cytogenetic analysis was carried on 96 cases presented with clinical features of Down syndrome from various special schools of Tamilnadu and retrospective analysis was also done from their medical records.

Results: Out of 96 cases, 94 were true trisomy, one case had translocation and one case presented with 46, XY, der(21;21)(q10;q10) and mother of same case presented with 45, XX, rob(21;21)(q10;q10). In the present study, the mean maternal age was found to be 27.56 ± 5.35 years. In 73% of case mothers of Down syndrome the maternal age was $\leq$30 years; among them 41% were between the maternal ages ranging from 19 to 25 years and 32% ranging from 26 to 30 years. The maternal age $\geq$31 years were found in 27% cases. The most prominent clinical features observed in Down syndrome children greater than 60 percentages were: Epicanthic fold (97%), Mongoloid Slant (96%), High arched palate (89%), Flat Facial profile (83%), Small ears/Low set ears (75%), Short neck (69%), Furrowed tongue (61%), and Brachydactyly (67%) and Depressed Nasal Bridge (58%). Congenital heart disease was diagnosed in 36% among which 24% (VSD), 8% (ASD) and 4% (PFO) respectively. Gastrointestinal anomalies were noted in 3% and hypothyroidism in 14% of cases. Patchy Alopecia Areata was present in 2% cases.

Conclusion: A parental study is more important in the determination of the recurrence risk and to counsel them by providing available options.

Conflicts of interest

The authors have none to declare.

https://doi.org/10.1016/j.jasi.2018.06.008

8

Variations in the branching of cords of brachial plexus

Singh Rajani

AIIMS, Rishikesh, India

Brachial plexus is complex network of nerves, formed by joining and splitting of ventral rami of spinal nerves C5, C6, C7, C8 and T1 forming trunks, divisions and cords. The nerves emerging from trunks and cords innervate the upper limb and to some extent pectoral region. Scanty literature describes the variations in the formation of cords and nerves emanating from them. Moreover the variations of cords of brachial plexus and nerves emanating from them have iatrogenic implications in the upper limb and pectoral region. Hence, study has been carried out. Twenty-eight upper limbs and posterior triangles from fourteen cadavers fixed in formalin were dissected and rare and new variations of cords were observed. Most common variation consisted of formation of posterior cord by fusion of posterior division of upper and middle trunk and lower trunk continued as medical cord followed by originating of two pectoral nerves from anterior divisions of upper and mid-