Conflicts of interest

The authors have none to declare.

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93

Ovarian tissue cryopreservation as an option for fertility preservation in young unmarried girls suffering from cancer

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Introduction: Due to various social and economic reasons women in the reproductive age group delay their marriage and the age of bearing their first child. This leads to an important health problem, as women steadily lose their oocytes from birth to menopause, with an accelerated loss of oocyte quantity and quality from the age of 35 years onwards. The situation becomes grim when a unmarried girl is diagnosed with cancer. Fertility preservation presents a peculiar challenge in young unmarried girls suffering from malignancies. It is now well established that the chemotherapy and/or radiotherapy is gonadotoxic and hence the need for preservation of fertility. The multiple factors affecting the fertility potential include the drug or size/location of the radiation field, dose, method of administration, disease, age and sex of the patient, and combination chemotherapy and pretreatment fertility of the patient. The ovarian cortex contains primordial follicles which are undifferentiated and not active metabolically.

Materials and methods: The primordial follicles in the ovarian cortex can be cryopreserved by offering ovarian cortex freezing as a method of fertility preservation. The ovarian tissue is obtained by performing laparoscopy on the same day. The cryopreserved ovarian cortical tissue is intended to be thawed and implanted after completion of chemotherapy and/or radiotherapy.

Result and conclusion: This method of ovarian tissue cryopreservation can be offered as a method of fertility preservation to children who survive childhood malignancies. In young unmarried girls suffering from cancer where in vitro-fertilization-embryo transfer (IVF-ET) is contraindicated, ovarian tissue cryopreservation and transplantation could become the technique of choice in the future.

Conflicts of interest

The author has none to declare.

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94

Association of *pai-1* promoter sequence variations in idiopathic avascular necrosis of head of femur

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Background: Avascular necrosis of femur head (ANFH) is considered a multifactorial disorder mainly associated with intravascular thrombosis or occlusion of already meager femoral head blood supply. Pertinent to its etiology, lot of causative factors have been elucidated in literature but derangement in fibrinolytic mechanism have been focused with more concern. Though various genes and genetic factors play role in maintaining harmony in coagulation and fibrinolytic system, *PAI-1* gene plays a crucial role. Hence aim of our study was to find out any polymorphism in this gene in respect to AVN hip.

Methods: Two SNPs of the *PAI-1* gene (rs2227631, −844G/A; rs 1799889, −675 4G/5G) were genotyped in 12 patients diagnosed with idiopathic AVN of head of femur and 13 control subjects, using direct sequencing. Subsequently, association analysis was performed for the genotyped SNPs.

Results: In −844G/A genotype GG (normal) was found in 10/13 controls and 10/12 cases (AVN). Similarly, GA (polymorphism) was noticed in 3/13 controls and 2/12 cases (AVN). Also in −674 4G/5G genotype: 4G/4G (normal) was found in 3/13 controls, 4G/5G in 4/13 controls, and 4G/4G + A in 6/13 controls while 4G/4G in 4/12 cases, 4G/5G in 2/12 & 4G/4G + A in 6/12 cases of AVN. Hence, cases and controls had equal frequency of polymorphism association of *PAI-1* gene.

Conclusion: Equal frequency of genetic polymorphism of *PAI-1* gene in cases and controls, suggests that sequence variation in promoter region is not associated with AVN. However, larger study group is warranted to validate our findings.

Conflicts of interest

The authors have none to declare.

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95

Molecular diagnosis of sickle cell anaemia based on SNPs in β-globin gene

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Introduction: Sickle cell anaemia is an autosomal recessive disorder caused by a point mutation in the 6th codon of the β-globin gene on chromosome 11. The substitution of a single amino acid (glutamic acid → valine) decreases the solubility of the deoxyhaemoglobin molecule making the erythrocytes assume irregular shapes. The sickled erythrocytes become trapped in the microcirculation and cause damage to multiple organs.

Aims and objective: To standardize a DNA based diagnosis of sickle cell anaemia in adolescent tribal girls of selected regions of Odisha so as to create awareness among the tribal communities to avoid consanguineous marriages.

Materials and methods: 40 Blood samples were collected from microscopically diagnosed cases of sickle cell anaemia in adolescent tribal girls who were from western Odisha. The blood samples were collected in 0.1% EDTA treated vials and stored in fridge till DNA isolation. DNA isolation was done from 200 μl of blood from each sample using the conventional phenol–chloroform method. Primers were designed from human β-globin gene using Primer 3 software. Primers were then synthesized commercially. PCR amplification, Sanger sequencing and analysis was done using BIOEDIT sequence editor.

Results: A DNA based detection of sickle cell anaemia could be done, thus showing mutation after the 6th codon where the nucleotide 'A' has mutated to 'T', i.e., adenine is replaced by thymine.