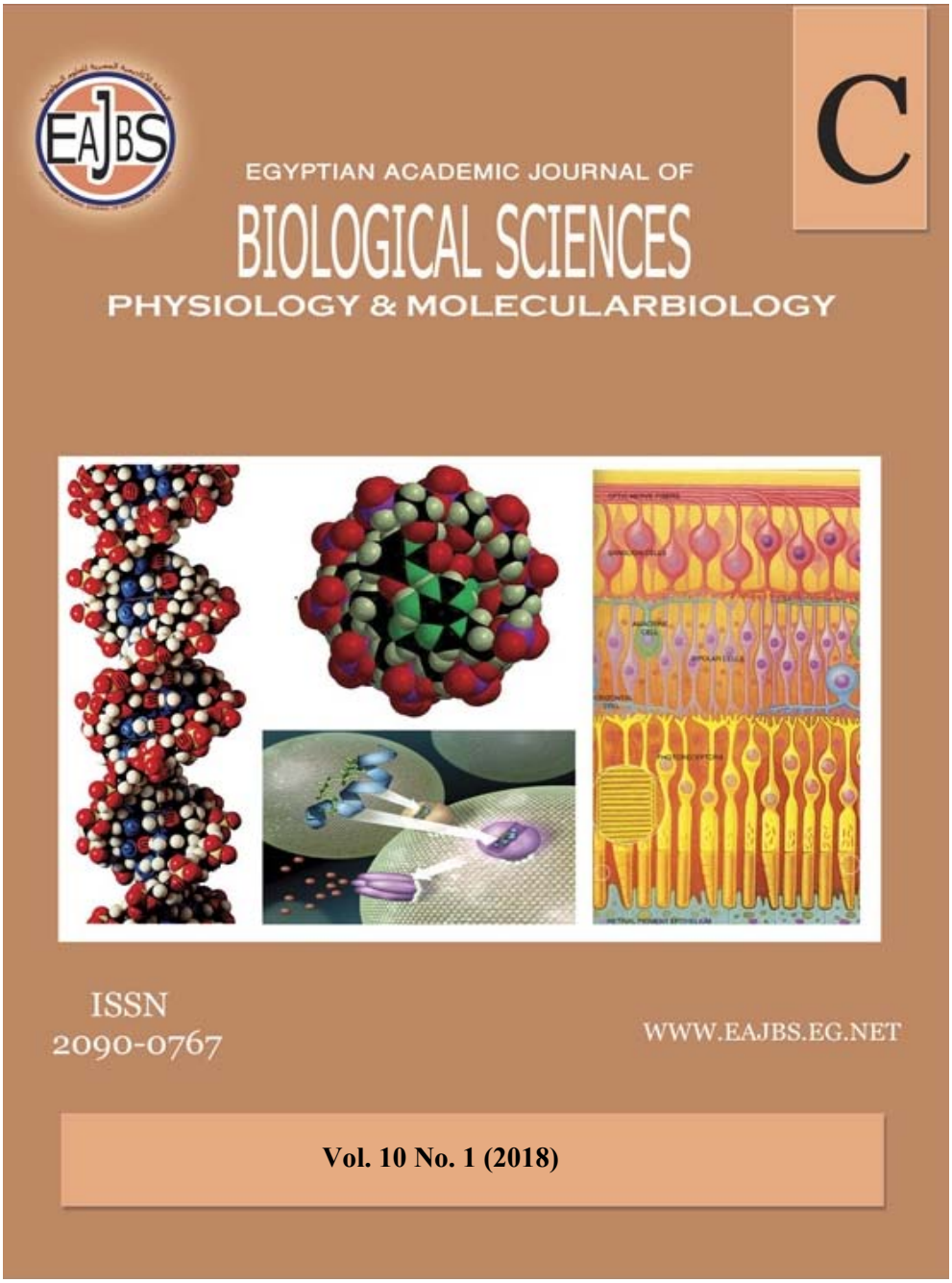

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Frequency of *XRCC1* exon 10 G>A gene polymorphism in the Saudi Arabian Population: Inter-individual study from different ethnic groups

Shafiul Haque
Research and Scientific Studies Unit, College of Nursing and Allied Health Sciences, Jazan University, Jazan-45142, Saudi Arabia
E-mail: shafiul.haque@hotmail.com

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ABSTRACT

Introduction: The failure of DNA repair genes is the main reason for cancer development in human beings. The normal X-ray repair cross-complementing Group1 (*XRCC1*) gene play a key role in base excision repair (BER) pathway. The occurrence of *XRCC1* exon 10 G>A polymorphism varies in different ethnic groups and the data on the allelic distribution of the same is lacking in Saudi Arabian population. The objective of the current study was to delve deep into the documented studies and analyze the frequency of genetic polymorphism *XRCC1* exon 10 G>A in Saudi Arabian population and further do the comparison with other ethnic groups of the world. **Methods:** PUBMED (Medline), web-databases was searched for the required epidemiological studies of different ethnic group. **Results:** The frequency of *XRCC1* exon 10 variant allele (A) was found to be 22.3%. Further this frequency was compared with various others ethnic groups and a significant difference was found for Thailand (p=0.001), Iran (p=0.025), Japan (p=<0.001), North India (p=<0.001), Poland (p=0.010), France (p=0.001), Norway (p=<0.001), USA (p=<0.001), Pakistan (p=<0.001), Spain (p=<0.001), Belgium (p=<0.018), Australia (p=<0.001), and Portuguese (p=<0.045) population. **Conclusions:** The overall results of this study suggest that frequency of this DNA repair genes demonstrates distinctive pattern in Saudi Arabia population, which might be possible because of ethnicity variation. This could assist in high-risk screening of humans exposed to environmental carcinogens and cancer predisposition in different ethnic groups.

INTRODUCTION

The area wise geographic variations in the incidences of cancer and its mortality indicates the role of genetic and environmental factors in the pathogenesis of cancer. The different environmental agents like ionizing radiations result in damage to DNA of the exposed cells. This causes the loss of integrity of the genetic information, which further leads to manifold increase in cancer risk.

The inability of the host system to repair such defective or destructive DNA may lead to carcinogenesis in the individuals. The person to person variations in their susceptibilities to cancer can be attributed to genetic alterations which affect DNA repair capabilities (de Jong *et al.* 2002)**.**

Single-nucleotide polymorphisms (SNPs) are the DNA base variants present in the human population. They are present in at least 1% population (Collins *et al.* 1997) and are usually less penetrant, but their study in cancer is very important (Perera *et al.* 2000). The inherent difference in the capacity of individuals to repair DNA lesion is induced by endogenous and exogenous carcinogens. These genetic polymorphisms and adverse genotypes have been suggested to reduce DNA repair capacity (DRC) and modify individual susceptibility for cancer than general population (Wu *et al.* 2004). Hence, the results obtained from the studies done on inter individual variations in different ethnic groups could possibly give further insight to discover candidate susceptibility alleles in the etiology of carcinogenesis.

The X-ray cross-complementing gene 1 (*XRCC1*) is mapped on chromosome 9q13.2. It consists of 17 exons and encodes a protein of 633 amino acids. *XRCC1* plays an important role in multi-step base excision repair (BER) pathway to remove the 'non-bulky' base adducts produced by methylation, oxidation, reduction or fragmentation of bases by ionizing radiation or oxidative damage (Yu *et al.* 1999). It is a scaffolding protein consisting of three DNA repair enzymes: DNA ligase III, DNA polymerase b and poly(adenosine diphosphate (ADP)-ribose) polymerase (PARP) involved in excision and recombinational repair pathways (Petermann *et al.* 2006). The significance of *XRCC1* came into light with null mutant mice experiment. The null mutant mice with error in *XRCC1* gene demonstrated embryonic development arrest (Tebbs *et al.* 1999). Different polymorphisms have been reported in *XRCC1* gene, but the extensively studied is at codon 399 in exon 10 (Arg to Gln). It is located in the BRCT-I interaction domain of *XRCC1* gene within a poly (ADP-ribose) polymerase binding region (Shen *et al.* 1998). In the year 1999, a team of scientists linked the above mentioned polymorphism to change in DNA repair capacity of the said gene (Lunn *et al.* 1999). These changes in the conserved sites of the protein were believed to amend base excision repair capacity. This negative amendment was found to downgrade the ability of human beings to fight adverse health conditions including cancer (Hung *et al.* 2005). The current study deals to investigate the frequency distribution of *XRCC1* exon 10 Arg399Gln, G23591A, rs25487 polymorphism in normal healthy individuals from Saudi Arabia, wherein different epidemiologic studies performed globally were considered for comparative analysis.

MATERIALS AND METHODS

Prevalence of gene variants:

PUBMED (Medline) web-database was searched for pertinent articles using ''*XRCC1*'' and ''polymorphism''. It was restricted to human subjects but covered all the languages. The genotype frequencies for the control population were considered for case-control studies. Those studies focusing only on allele frequencies and no genotype frequencies were excluded straightaway. In case of more than one hits for the same study population, the most recent publication was included. The number of publications identified and considered for the current study were 22 which were based on the prevalence of *XRCC1* exon 10 (G>A) polymorphism, and were later used for the comparison with Saudi Arabian population

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Statistical analysis:

Pearson's $\chi 2$ test was performed for the comparative analysis of the genotype and allelic frequencies of different populations by using SPSS (version 21) statistical software program. Court-Lab (web-based software program) was used to examine Hardy-Weinberg equilibrium. p-value 0.05 was maintained for statistically significant outcomes.

RESULTS

The distribution of *XRCC1* (exon 10, G>A) genotype frequencies in Saudi Arabian population has been given in Table 1.

Table 1. Observed and expected genotypic frequencies of *XRCC1* Exon 10 G>A polymorphism in the control group.

Gene	***Genotype***	***Observed (n) %***	***Expected (n) %***	***p-value (HWE)***
XRCC1 *Exon 10*	GG	142 (62)	139(60.4)	0.34
G23591A	GA	72 (31.4)	79(34.7)	
(rs25487)	AA	15 (6.6)	11(5)	

Note: HWE: Hardy-Weinberg equilibrium

The genotype distributions were in accordance with Hardy–Weinberg equilibrium (HWE). The frequency distribution of three genotypes and alleles of this polymorphism of different populations with reference to our population were compared using $\chi 2$ tests (Table 2).

Table 2. Genotype and allele frequency distribution of *XRCC1* Exon 10 G>A gene variant in various populations and p-values in comparison to Saudi Arabian population

Gene	Country/ ethnicity	(n)	Age (years), Mean age ± SD	Genotype			P	A	Reference
				GG	GA	AA			
XRCC1 Exon10	Saudi Arabia	229	NA	142 (62)	72 (31.4)	15 (6.6)	Ref	22.3	Siraj *et al.* 2008
	Hungary	102	56.7±8.9	53 (51.9)	41 (40.2)	08 (7.9)	0.444	27.9	Csejtei *et al.* 2009
	Korea	157	45-60	86 (54.8)	64 (40.8)	7 (4.5)	0.585	24.8	Tae *et al.* 2004
	Thailand	164	46-75	67 (40.9)	74 (45.1)	23 (14.0)	0.001	36.6	Kietthubthew *et al.* 2006
	Iran	190	NA	83 (43.7)	87 (45.8)	20 (10.5)	0.025	33.4	Fard-Esfahani *et al.* 2011
	China	249	48.9±4.1	145 (58)	75 (30)	29 (12)	0.060	26.7	Hu *et al.* 2011
	Japan	93	66.1 ± 13.7	51 (54.8)	21 (22.6)	21 (22.6)	<0.001	33.9	Chiyomaru *et al.* 2012
	Finland	313	55–63	154 (49)	130 (42)	29 (9)	0.088	28.9	Misra *et al.* 2003
	North India	200	59.1±10.4	83 (41.5)	83 (41.5)	34 (17)	<0.001	37.8	Mandal *et al.* 2010
	Germany	246	NA	113 (46)	110 (45)	23 (9)	0.065	31.7	Sanyal *et al.* 2004
	Poland	150	52-83	64 (43)	68 (45)	18 (12)	0.010	34.7	Romanowicz *et al.* 2011
	France	312	54.5±6.7	127 (40.7)	146 (46.8)	39 (12.5)	0.001	35.9	Moullan *et al.* 2003
	Norway	391	NA	151 (38.6)	186 (47.6)	54 (13.8)	<0.001	37.6	Zienolddiny *et al.* 2006
	Taiwan	282	NA	152 (54)	109 (38.7)	21 (7.5)	0.453	26.8	Cho *et al.* 2003
	USA	197	63.3±10.4	79 (40)	92 (47)	26 (13)	0.001	36.5	Stern *et al.* 2001
	Pakistan	74	NA	27 (36)	32 (43)	15 (20)	<0.001	41.9	Gulnaz *et al.* 2012
	Spain	474	45.99±17.26	196 (41.4)	212 (44.7)	66 (13.9)	<0.001	36.3	García *et al.* 2011
	Belgium	109	61 ± 12.3	46 (42.2)	50 (45.9)	13 (11.9)	0.018	34.9	De Ruyck *et al.* 2007
	Turkey	108	56.3±1.3	50 (46.3)	49 (45.4)	9 (8.3)	0.239	31	Engin *et al.* 2011
	Australia	130	69.07 ±7.99	37 (34.5)	60 (64.9)	33 (30.5)	<0.001	48.5	Dhillon *et al.* 2011
	Portuguese	178	43.02±8.73	80 (44.9)	80 (44.9)	18 (10.1)	0.045	32.6	Varzim *et al.* 2003
	North Africa	477	NA	279 (58.5)	163 (34.2)	35 (7.3)	0.597	24.4	Laantri *et al.* 2011

Note: NA: Not available

The minor variant allele frequency in the studied population was 22.3%. A significantly different pattern of genotypes and allele frequency was observed in *XRCC1* Exon 10 polymorphism in Thailand (p=0.001), Iran (p=0.025), Japan (p=<0.001), North India (p=<0.001), Poland (p=0.010), France (p=0.001), Norway (p=<0.001), USA (p=<0.001), Pakistan (p=<0.001), Spain (p=<0.001), Belgium (p=<0.018), Australia (p=<0.001) and Portuguese (p=<0.045) population.

DISCUSSION

It is well known these days on the basis of accumulating evidences that genetic variations influence the risk of environmental carcinogenesis and genetic susceptibility plays an important role in the development of human cancer. The new studies on genetic association of cancer risks have assisted in identifying the effects of SNPs as candidate genes. Among the cancer repair genes are most studied as they responsible for maintaining genome integrity. The analyzed studies suggest that inherited mutations in DNA repair genes exposes the individuals to exceptionally high risk for the development of cancer (Au *et al.* 2004). Due to the well-known differences in the distribution of DNA repair gene polymorphisms among several worldwide ethnic groups, the data from 'normal healthy' populations are of special interest. It can be used to establish the relevance, which further could be used for the evaluation of the investigated genetic markers in the susceptibility, manifestation, prognosis or treatment of deadly diseases. Many reports published on DNA repair gene polymorphisms have already proved their potential in disease related clinical roles in determining both inter-individual and inter-ethnic differences in carcinogenesis (Kittles *et al.* 2003). Saudi Arabian population is considered to be one of the most diverse population all over the world, and hence can be used to study the genetic variation vis a vis other populations. Therefore, there is a potential that it may have future propositions for the preventions as well as early intervention strategies. The current article compares the frequency distribution of *XRCC1* exon 10 G>A genetic variant in Saudi Arabian population from other populations of the world based on the published articles.

The repair of damaged DNA is foremost parameter to protect the cells against cancer (Friedberg 2003). The *XRCC1* gene studied in this paper plays an important role in repairing both single-strand break and the base excision (Cappelli *et al.* 1997). The cells lacking *XRCC1* gene are genetically unstable and sensitive to DNA damaging agents such as Ionizing radiation, alkylating agents, ultra-violet light and hydrogen peroxide (Kubota *et al.* 1996). It is also known that variants of *XRCC1* gene contributes to ionizing radiation susceptibility, for which the prolonged cell cycle G delay is considered as one of the indicators (Hu *et al.* 2001).

In the present study, the interesting finding is the significance of mutant allele (A). The frequency of this variant allele (A) of Exon 10 in Saudi Arabia population was found to be 22.3%. It was significantly different from many other countries of the world. It was found to be lower from Thailand, Iran, Japan, North India, Germany, Poland, France, Norway, USA, Pakistan etc., whereas it was similar to Korea, china and Taiwan, respectively. The differences in allelic frequency in these studies can be attributed to several reasons such as ethnic variation, heterogeneity of study populations and different sample sizes. Earlier reports have demonstrated the distinct patterns of DNA repair genes in comparison with different populations of other countries (Mandal *et al.* 2010; Areeshi *et al.* 2013). The differences in the prevalence of DNA repair polymorphisms across different

populations indicate that susceptibility factor in one population may not hold true for another. These types of studies may be helpful in forming the basis for future establishment of epidemiological and clinical databases. It should also be emphasized that for most instances, the allele and genotype frequencies presented here do not always consider the complete spectrum of variants at a locus.

The current study concludes that *XRCC1* exon10 allelic variant in Saudi Arabian population is significantly different from other populations. This information may be used as basis that can act as a contributing factor in the screening of cancer. The differences in the distribution of these DNA repair genes in healthy Saudi Arabian population and other ethnic groups can be used in making a profile that may help in assessing the disease predisposition and prevalence. The identification of the susceptibility factors associated with individual predisposition to cancer could possibly give further insight in to the etiology of carcinogenesis. Also, future large studies and biological characterization is warranted to use this polymorphism as a biomarker for the screening purpose for cancer.

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ARABIC SUMMARY

مدى تكرار النمط الجيني **XRCC1 exon 10 G>A** في سكان المملكة العربية السعودية وعمل الدراسة بين مجموعة فردية من مجموعات عرقية مختلفة

شفيع الحق
وحدة الأبحاث والدراسات العلمية ,كلية التمريض ,جامعة جازان , مدينة جازان – ٤٥١٤٢ ,المملكة العربية السعودية

يعتبر فشل جينات إصلاح الحمض النووي هو السبب الرئيسي لتطوير السرطان في البشر. ويلعب الجين العادي للإصلاح بالأشعة السينية المكمل لمجموعة ١ (XRCC١) دورا رئيسيا في مسار إصلاح الاستئصال القاعدي (BER). ويتنوع حدوث التحور الجيني (XRCC1 exon 10 G>A) بين مجموعات عرقية مختلفة، كما أن البيانات عن التوزيع الأليلي لذلك التحور غير متوفرة عن المجتمع السعودي. وكان الهدف من الدراسة الحالية هو الخوض بعمق في الدراسات الموثقة وتحليل مدى تكرار التنوع الجيني الناتج عن هذا التحور (XRCC1 exon 10 G>A gene) في السكان السعوديين، وبالاضافة الى ذلك، المقارنة مع المجموعات العرقية الأخرى في أنحاء العالم. تم البحث على موقع PUBMED عن الدراسات الوبائية المطلوبة والخاصة بمجموعات عرقية مختلفة. وقد وجد أن تكرار النوع أللیل(أ) XRCC1 exon 10 variant allele (A) هو ٢٢.٣٪. وقد تم مقارنة هذا التكرار مع مجموعات عرقية أخرى، ووجد فرق كبير في سكان تايلند (p = ٠.٠٠١)، وإيران (p = ٠.٠٢٥)، واليابان (p = <٠.٠٠١)، وشمال الهند (p = <٠.٠٠١) وبولندا (p = ٠.٠٠١)، وفرنسا (p = ٠.٠٠١)، والنرويج (p = <٠.٠٠١)، والولايات المتحدة الأمريكية (p = <٠.٠٠١)، وباكستان (p = <٠.٠٠)، اسبانيا(p=<٠.٠٠١) ، بلجيكا (p=<٠.٠١٨)، أستراليا (p = <٠.٠٠١) والبرتغال (p = <٠.٠٤٥). وتشير نتائج هذه الدراسة إلى أن تواتر جينات إصلاح الحمض النووي هذه تظهر نمطا مميزا في سكان المملكة العربية السعودية قد يكون ممكنا بسبب الاختلافات العرقية. وهذا يمكن أن يساعد في الكشف عن المخاطر العالية من البشر المعرضين للمواد المسرطنة البيئية في مختلف المجموعات العرقية.