

ASSESSMENT OF THE EFFECTS OF RADIATION ON THE STABILITY OF THE GENOME OF THE POPULATION FROM THE TERRITORIES ADJACENT TO THE SOURCE OF POLLUTION

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ABSTRACT

Relevance: The increasing introduction of new mutagenic factors into the human environment increases the frequency of hereditary human diseases associated with exposure to environmental mutagens. In the case of radiation exposure, the problem arises of the need to identify risk groups of people with hypersensitivity since the effect of radiation on the body, in addition to direct effects on its functional subsystems, induces or activates protective systems (repair, adaptation). If the DNA repair system is damaged, the risk of induction of mutation frequency increases. The above highlights the relevance of the research topic and the results obtained.

The study aimed to assess the impact of radiation pollution on the stability of the genome and human health, considering the long-term genetic consequences.

Materials and methods: Field and laboratory methods were used, such as creating a system of sites for sampling environmental objects, human peripheral blood samples, methods for measuring the radiation activity of objects, and cytogenetic and molecular genetic methods.

Results: Gamma activity measurements showed that the radiation level in the surveyed territory of the landfill and adjacent settlements was 0.6 to 0.14 mSv/h. Of particular importance in this regard is the study of the mechanisms of individual sensitivity to radiation and the role of the DNA repair system. Molecular genetic studies of the DNA of blood cells and cytogenetic analyses of people living in the zone of influence of the radioactive waste landfill revealed the spread of several mutant genotypes, which indicates the likelihood of an increased risk of environmental diseases in persons with pronounced genome instability.

Conclusion: The radiation level on the territory of the landfill and adjacent settlements was 0.6 to 0.14 mSv/h. The analysis of the distribution of people by genotypes in the examined groups showed an increase in the frequency of heterozygous alleles of the XRCC1 repair gene to 35% compared with the control group (10%), and the frequency of homozygous alleles for the Trp/Trp allele does not exceed the control level (3%). In turn, for the XRCC3 gene, there is a slight excess in the frequency of heterozygotes, and the homozygous Thr/Met allele remains at the control level – 21% compared with the control of 2%.

Keywords: mutagens, radiation, environment, genes, hereditary diseases, genome.

Introduction: Remote genetic consequences of radiation factors pose a real danger to biota humans and are important for environmental protection and human health. Knowledge of the mechanisms of radiation exposure is associated with individual radiosensitivity of organisms and the activity of the DNA damage reparation system (repair). In this regard, the increased frequency of human diseases associated with environmental mutagens determines the need to identify risk groups of people with increased sensitivity to radiation exposure. In addition to direct exposure, radiation also has an indirect effect on the body, particularly through protective systems, which in most cases leads to disruptions in the structure and function of the DNA molecule [1]. If the reparation systems do not work properly, the risk of mutation increases sharply [2]. The above emphasizes the relevance of the research topic and the results obtained.

The study aimed to assess the impact of radiation pollution on the stability of the genome and human health, considering the long-term genetic consequences.

Materials and methods: The types of radiation sources are given in Tables 1-4: these are settlements adjacent to the radioactive waste disposal site and located, taking into account the wind rose, on the leeward side, mainly in the Munaylinsky district and partially in the Tupkaragansky district, from whose territories soil, plant and water samples were taken (Figure 1).

The names of the surveyed settlements are also provided in Tables 1-4. The locations with elevated radiation levels are indicated in the description of the results of radioecological studies.

The studies of radioactivity of soil and vegetation samples were carried out by the gamma spectrometric method using the Multirad-gamma device (TD Avtomatika, Smolensk, Russian Federation) MKS-OTA No. 1935 (VA.17-

04-46889 dated September 15, 2023) and laboratory studies - molecular genetic analysis of DNA by the RAPD and ISSR methods on human peripheral blood samples [3]. The selected soil and plant samples in rural areas of the Munayli district of the Mangistau region were examined by the gamma spectrometric method in the radiological laboratory of the branch of the RSE on the Right of Economic Management "National Center of Expertise" of the State Public Enterprise of the Ministry of Health of the Republic of Kazakhstan for the Mangistau region following the Order of the Minister of Health of the Republic of Kazakhstan dated August 2, 2022 No. KR DSM-71 "On approval of the research procedure." Water samples were analyzed using the radiometric method on the UMF-2000 radiometer No. 1169, verification certificate No. A.17-04-46969 dated September 11, 2023, and also in the radiological laboratory of the branch of the RSE on the Right of Economic Management "National Center for Expertise" of the Ministry of Health of the Re-

public of Kazakhstan in the Mangistau region. Blood samples were taken from 60 residents of settlements adjacent to the landfill (38 women and 22 men). The control group consisted of 55 Almaty region residents with no contact with radiation. Genomic DNA was isolated from peripheral blood samples using a genomic DNA purification kit (GeneJET, Thermo Fisher Scientific, USA). Quantitative and qualitative assessment of the isolated DNA was performed using spectrophotometry (NanoDrop One, Thermo Scientific, USA) and agarose gel electrophoresis. Then, specific primers for the genes XRCC1 Arg194Trp, XRCC1 Arg399Gln, and XRCC3 Thr241Met were synthesized. The primers were synthesized on an automatic synthesizer ASM-800 (RF). The synthesized primers were tested in test reactions of PCR genotyping. The finished lyophilized primers were stored in freezers (-20°C) for PCR analysis. The polymerase chain reaction methods included restriction fragment length polymorphism (PCR-RFLP) and gel electrophoresis.



Figure 1 – Overview map of the current state of the tailings storage facility

Results: Below are the results of radiological studies of the total radioactivity of soil, plant, and water samples (tables 1-4).

Tables 1-4 indicate the sampling points of soil, water, and plants and the results of the determination of the level of radiation activity.

Table 1 – Specific effective (total) activity of soil from the solid waste landfill

No. of samples	Sample description	Sampling point	Cs-137	Ra-226	Th-232	K-40	Sr-90
11	Soil	Solid waste landfill	1.2±1.1	23.4±3.9	10.5±2.5	382±74	-*

Note: * – the activity was below the device sensitivity

Table 2 – Specific effective (total) activity of soil with plants from the flour terminal area

No. of samples	Sample description	Sampling point	Cs-137	Ra-226	Th-232	K-40	Sr-90
10	Soil with plants	From the flour terminal area	<5.9	<8.8	<20	820±230	.*

Note: * – the activity was below the device sensitivity

Table 3 – Total effective activity of drinking water

No. of samples	Sample description	Sampling point	Activity indicators, Bq/L	Detected value	Permissible value
4	Decentralized drinking water	Mangistau region, rural area – 1	Total alpha activity	0.044±0.012	0.2
			Total beta activity	0.062±0.014	1.0

Table 3 – Total effective activity of sea water

No. of samples	Sample description	Sampling point	Activity indicators, Bq/L	Detected value	Permissible value
7	Sea water from the channel, entrance to MAEK, point 1	Mangistau region, coastal sea zones	Total alpha activity	0.15±0.034	not rated
			Total beta activity	0.09±0.018	not rated

Site No. 1: Akshukyr Village. Coordinates: N 43° 0' 46.089", E 51° 0' 5.311". The elevation of the region is 7 meters. Ground radiation level: 65 nanosieverts per hour.

Site No. 2: Baskudyk Village, Biotope 1. The elevation of the region is 15 meters. Coordinates: N 43° 0' 41.960", E 51° 0' 12.232". Ground radiation level: 67 nanosieverts per hour. The radiation level at the sampling site is 103 nanosieverts per hour.

Site No. 3: Mangystau-1 Village. Site No. 4. Coordinates: N 40° 0' 42.529", E 51° 17' 707". The elevation above sea level is 10 meters.

Site No. 4: Mangystau-5 settlement, coordinates: N 43° 0' 41.649", E 51° 0' 17.797". The elevation of the region is 14 meters. Radiation level: 68 nanosieverts per hour.

Site No. 5: Mangistau Nuclear Power Complex – Chemical and Hydrometallurgical Plant (CHMP) located in the southern region of Aktau. The radiation level in the water samples from the discharge channel of the Mangystau Nuclear Power Engineering Plant is 0.08-0.09 mSv/h. Unusually high levels of gamma radiation were recorded near CHMP and the Aktau Foundry Limited Liability Partnership (LLP). The absolute maximum recorded is 1.98 µSv/h near CHMP [4]. Numerous investigations have demonstrated that the levels of radionuclides in environmental components collected from identical locations in areas surrounding the contamination source (radioactive waste disposal site) conform to sanitary and hygienic norms. The data about the buildup of radionuclides in plant samples aligns with the documented total alpha and beta activity in the assessed villages around the dump. The study area exhibits a negligible background radiation level, with an average environmental radiation dosage of 0.12 µSv/h. The peak measurement of 1.98 µSv/h was documented at site No. 5.

According to the results from the radiological survey of the area, essential preparatory work was conducted for molecular genetic research to evaluate the effects of radiation exposure on public health [5].

Blood samples from 60 individuals residing near the test location were utilized to assess the condition

of the body's repair mechanisms in the Mangystau region occupants. The polymorphisms of the XRCC1 Arg194Trp (rs1799782), XRCC1 Arg399Gln (rs25487), and XRCC3 Trp241Met (rs861539) genes were examined. Table 5 presents the exact primers for the examined repair genes, including the sequences of the primers and the endonucleases utilized for analysis. A commercial GeneJET kit (ThermoScientific, USA) was employed for DNA isolation. A qualitative evaluation of the extracted DNA was conducted by agarose gel electrophoresis (Figure 2).

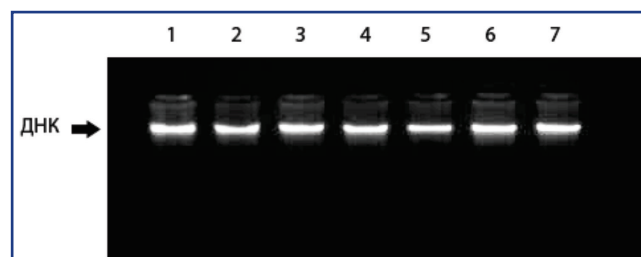


Figure 2 - Electrophoregram of DNA isolated from human peripheral blood: 1-7 - genomic DNA samples

Figure 2 illustrates the electrophoregrams of various DNA samples, showing that the electrophoretic analysis confirmed the isolated DNA's high quality and absence of degradation throughout the isolation procedure.

Table 5 presents the sequences of specific primers for the analyzed genes, the name of the restriction endonuclease used in restriction analysis, and the corresponding fragment sizes.

The diversity of the XRCC1 Arg194Trp, XRCC1 Arg399Gln, and XRCC3 Thr241Met repair genes was examined in individuals residing in places adjacent to radiation-contaminated sites to assess the condition of the body's repair mechanisms. The polymorphism induction of the XRCC1 Arg194Trp, XRCC1 Arg399Gln, and XRCC3 Thr241Met genes was assessed using the PCR-PDRF method. The DNA repair gene polymorphism frequently influences individual susceptibility to environmental stressors, such as radiation.

Table 5 - Primers used and conditions for amplification, restriction, and target products of XRCC1 and XRCC3 repair genes

Gene	Primers	PCR conditions	Restriction enzyme	Restriction products (polynucleotides)
XRCC1 Arg399Gln	(F) 5'-CAA GTA CAG CCA GGT CCT AG-3' (R) 5'-CCT TCC CTC ATC TGG AGT AC-3'	40 cycles: 94°C - 15 s 55°C - 30 s 72°C - 45 s	NciI	Arg/Arg: 89+59 Arg/Gln: 248+159+89 Gln/Gln: 248
XRCC1 Arg194Trp	(F) 5'-GCC CCG TCC CAG GTA-3' (R) 5'-AGC CCC AAG ACC CTT T-3'	40 cycles: 94°C - 15 s 57°C - 45 s 72°C - 45 s	PvuII	Arg/Arg: 490 Arg/Trp: 490+294+196 Trp/Trp: 294+196
XRCC3 Met241Thr	(F) 5'-GCC TGG TGG TGG TCA TCG ACT C-3' (R) 5'-ACA GGG GGG CTC CTC TGG AAG GCA CTG CTC AGC TCA CGC ACC-3'	40 cycles: 94°C - 15 s 60°C - 30 s 72°C - 45 s	NcoI	Thr/Thr: 136 Thr/Met: 136+97+39 Met/Met: 97+39

Note: M is a molecular DNA marker. Heterozygotes for XRCC3 241Thr/Met - 1-3, 5, 7-15; homozygotes for mutant allele XRCC3 241 Met/Met - 4

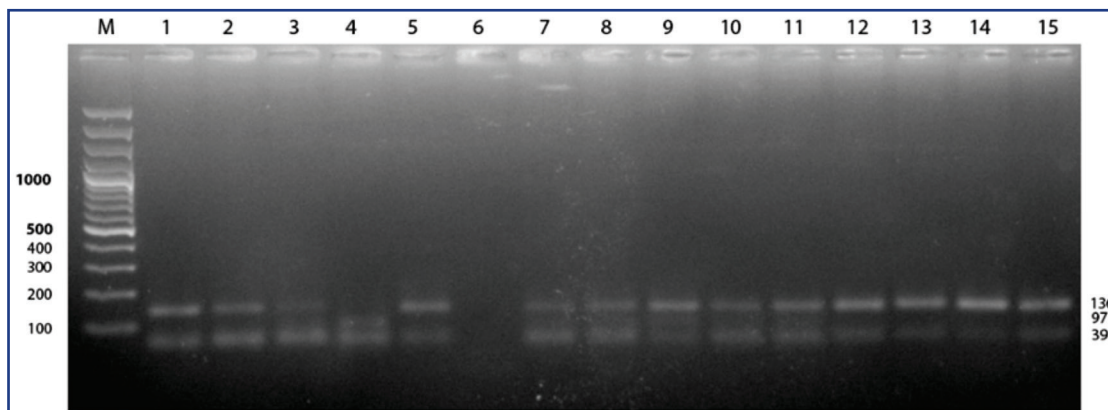
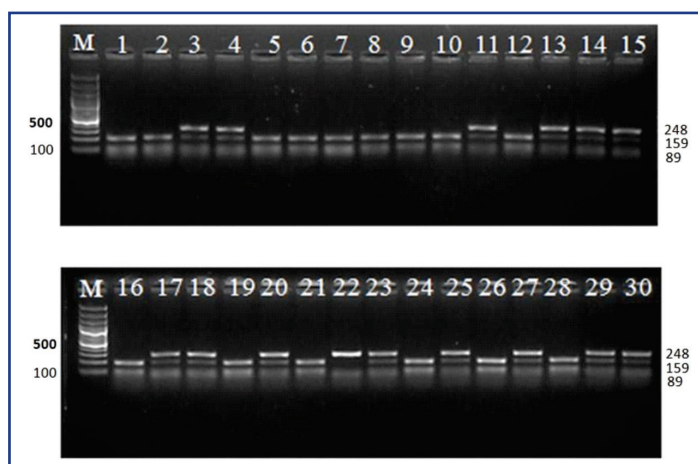


Figure 3 - Electropherogram of restriction products at the 241Thr/Met polymorphic site of the XRCC3 gene

Figure 3 illustrates the outcomes of the electrophoretic analysis. The genotypic distribution of DNA repair genes - XRCC1 (Arg194Trp) and XRCC3 (Thr241Met) was assessed. For the XRCC1 gene (Arg194Trp): homozygous genotype 194Arg/Arg - 44.8%, heterozygous genotype 194Arg/Trp - 48.3%, and homozygous mutant genotype 194Trp/Trp - 6.9%. The XRCC3 (Thr241Met) gene analy-

sis showed no presence of the homozygous genotype 241Thr/Thr, while the heterozygous genotype 241Thr/Met was observed at a frequency of 73.4%. The homozygous genotype for the 241 Thr/Thr mutant allele constitutes 26.6%.

Figure 4 displays the findings of electrophoretic analysis of restriction products following PCR-PDXRF analysis.



Note: M - molecular DNA marker; homozygous Arg/Arg (89+159) - 1, 2, 5-10, 12, 16, 19, 21, 24, 26 and 28; heterozygous Arg/Gln (248+159+89) - 3, 4, 11, 13-15, 17, 18, 20, 23, 25, 27, 29 and 30; homozygous Gln/Gln (248) - 22

Figure 4 – Electropherogram of restriction products at the polymorphism site of the XRCC1 gene - Arg399Gln

The allele frequencies and genotypes of the studied polymorphic markers in the human samples adhered to the Hardy-Weinberg equilibrium ($p > 0.05$). For the XRCC1 gene, the Arg194Trp variant in the control group exhibits

$\chi^2 = 0.243$, $p = 0.622$; in the “case” group, $\chi^2 = 0.398$, $p = 0.427$. For the XRCC3 gene, Trp241Met exhibits $\chi^2 = 3.491$ in the control group ($p = 0.062$) and $\chi^2 = 0.203$ in the case group ($p = 0.382$).

The subsequent analysis focused on the distribution of genotypes for the XRCC1 Arg194Trp and XRCC3 Trp241Met genes in the studied (Mangistau region) and control (Almaty region) groups. The distribution is represented as the percentage of individuals with a specific genotype within the group (Table 6).

The analysis of genotype distribution demonstrated that in the study group for the XRCC1 gene, the frequency of Arg/Arg homozygotes was 62%, Arg/Trp heterozygotes constituted 35%, and Trp/Trp minor allele homozygotes

accounted for 3%. In contrast, the control group exhibited frequencies of 90%, 10%, and 0%, respectively. The distribution of donors by genotypes for the XRCC3 gene was observed as follows: Thr/Thr – 77%, Thr/Met – 21%, Met/Met – 2% in the study group, and 67%, 33%, and 0%, respectively, in the control group. The data indicates that there are no people with homozygous genotypes for the minor alleles of both genes (XRCC1 Trp/Trp and XRCC3 Met/Met) among the examined population in the Almaty region. It may result from an inadequate sample size for the study.

Table 6 – Distribution of the XRCC1 and XRCC3 gene genotypes in the examined groups

Genes	Genotypes	“Case” group, people (%)	“Control” group, people (%)	χ ²	P
		N=60	N=55		
XRCC1 Arg194Trp	Arg/Arg	62%	90%	20.728	0.000
	Arg/Trp	35%	10%		
	Trp/Trp	3%	0		
XRCC3 Trp241Met	Thr/Thr	77%	67%	4.849	0.089
	Thr/Met	21%	33%		
	Met/Met	2%	0		

Discussion: A mutation in the genes of the DNA repair mechanism has significant implications for both the individual cell and the organism overall [5]. Microsatellite polymorphism in repair genes is correlated with increased radiosensitivity and some malignancies. Specifically, variation in this gene influences the risk of lung cancer [6]. The significant function of this gene is further underscored by the observation that homozygous mutations in XRCC1 when inactivated, result in embryonic lethality in mice. Polymorphism in this gene has been conclusively demonstrated to cause substantial deficiencies in DNA repair mechanisms, which markedly elevates the risk of early-onset carcinoma of the colon when combined with urban living. Mutations in this gene present a specific hazard to smokers and individuals with continuous radiation exposure [6]. The classification of surveyed groups into categories to assess the actual risk of radiation exposure to the population will be conducted via a questionnaire after the project, under the Work Schedule in 2025.

Moreover, single nucleotide polymorphisms (SNPs) in DNA repair genes might influence the efficacy of transcription and translation processes and predisposition to various diseases. Consequently, the analysis and identification of the distribution of DNA repair genes by genotypes is critically significant [7, 8].

Generally, the study indicates that radiation risk assessment relies on diversity in individual radiosensitivity [9, 10]. International scientists assert that the threshold dose for assessment of the acute effects of radiation exposure is 0.2 Gy. Consequently, when evaluating the findings regarding the potential effects of radiation on the human body in the study area, at lower doses, the sole radiation effects are stochastic (delayed) effects [11, 12]—oncological and hereditary diseases observed within the population of the study area. Nonetheless, disparities exist in evaluating outcomes attributable to the interaction of dosage functions

[13]. Numerous research studies have thoroughly examined hyperradiosensitivity to low radiation doses following in vitro irradiation of cells with charged particles and its correlation with adaptive response and induced radioresistance [14]. All knowledge regarding the long-term consequences of human exposure to low doses of radiation has been derived either by extrapolating experimental data from animals or direct radiation-epidemiological studies. The primary origin of the latter is acute, singular exposure to elevated doses resulting from nuclear catastrophes (Hiroshima and Nagasaki, Chornobyl, Fukushima, etc.) [15-17]. The quantitative parameter “probability of stochastic effects from low doses of radiation” is defined by multiple significant radiobiological factors; however, due to insufficient specific data, these effects have not been accurately established and continue to be contentious. The results can inform the implementation of strategies to enhance the region’s environmental quality and the population’s health.

The influence of detrimental environmental factors in ecologically unfavorable regions on the human body can be assessed through clinical examination, incorporating both quantitative and qualitative assessments of minor developmental anomalies, which also stem from alterations in the overall genetic equilibrium of the body. Consequently, evaluating the genetic impacts of external factors on human somatic cells may serve as a valuable adjunct to observing clinical outcomes.

Conclusion: A genomic study of persons residing in regions with significantly elevated gamma radiation levels (sites 1-5) demonstrated the development of mutations characterized by the amplification of DNA repair genes, resulting in gene polymorphism among those exposed to radiation.

The analysis of genotype distribution among the studied groups showed an increase in the frequency of hete-

rozygous alleles of the XRCC1 repair gene to 35%, in contrast to the control group's 10%, while the frequency of homozygous alleles for the Trp/Trp allele remained at or below the control level of 3%. For the XRCC3 gene, there is a marginal increase in the frequency of heterozygotes (21%) relative to the control, whereas the homozygous Thr/Met allele persists at the control level (2%).

The findings of this study align with existing literature regarding the genetic impacts of radiation exposure on the human genome in instances of nuclear power plant accidents and nuclear weapons testing. Upon completion of the design study, these results can be utilized to evaluate the actual risk for the specific population under investigation.

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АНДАТПА

ЛАСТАНУ КӨЗІНЕ ІРГЕЛЕС АУМАҚТАРДАН ПОПУЛЯЦИЯ ГЕНОМЫНЫҢ ТҰРАҚТЫЛЫҒЫНА РАДИАЦИЯНЫҢ ӘСЕР ЕТУ ЗАРДАБЫН БАҒАЛАУ

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Өзектілігі: адамның тіршілік ету ортасына жаңа мутагендік факторлардың көбеюі қоршаған орта мутагендерінің әсеріне байланысты адамның тұқым қуалайтын ауырлататын ауруларының жиілігінің артуына әкеледі. Радиациялық әсер ету жағдайында сезімталдығы жоғары адамдардың тәуекел топтарын анықтау қажеттілігі туындайды, өйткені радиацияның ағзаға әсері, оның функционалды ішкі жүйелеріне тікелей әсер етуден басқа, қорғаныс жүйелерін индукциялайды немесе белсендіреді (репарация, бейімделу). ДНҚ репарация жүйесі зақымдалған кезде мутация жиілігін индукциялау қаупі артады. Жоғарыда айтылғандар зерттеу тақырыбының және алынған нәтижелердің өзектілігін көрсетеді.

Зерттеудің мақсаты – созылмалы генетикалық әсерлерді ескере отырып, қоршаған ортаның радиациялық ластануының геномның тұрақтылығы мен адам денсаулығына әсерін бағалау.

Әдістері: далалық және зертханалық әдістерді қолданып: қоршаған орта объектілерінің сынамаларын алу үшін алаңдар жүйесін құру; адамның перифериялық қанының үлгілері, объектілердің радиациялық белсенділігін өлшеу әдістері, цитогенетикалық және молекулалық-генетикалық әдістер.

Нәтижелері: гамма-белсенділікті өлшеу полигонның зерттелген аумағының және оған іргелес елді мекендердің радиация деңгейі 0,6-0,14 мкЗв/сағ шегінде екенін көрсетті. Қан жасушаларының ДНҚ молекулалық-генетикалық зерттеулері және радиоактивті қалдықтар полигонының әсер ету аймағында тұратын адамдардың цитогенетикалық талдаулары бірнеше мутантты генотиптердің таралуын анықтады, бұл геномның айқын тұрақсыздығы бар адамдарда экологиялық аурулардың даму қаупінің жоғарылауы ықтималдығын көрсетеді.

Қорытынды: полигон аумағындағы және оған іргелес елді мекендердегі радиация деңгейі 0,6-0,14 мкЗв/сағ шегінде. Зерттелген топтардағы адамдардың генотиптік таралуын талдау XRCC1 репарация генінің гетерозиготалы аллельдерінің жиілігінің бақылау

тобымен (10%) салыстырғанда 35%-ға дейін жоғарылаганын көрсетті, ал Ttr/Ttr аллелі бойынша гомозиготалы аллельдердің жиілігі бақылау деңгейінен (3%) аспайды. Өз кезегінде, XRCC3 гені үшін гетерозиготалар жиілігінің шамалы асып кетуі байқалады, Thr/Met гомозиготалы аллель бақылау деңгейінде қалады – 21% бақылаумен салыстырғанда – 2%.

Түйінді сөздер: мутагендер, радиация, қоршаған орта, гендер, тұқым қуалайтын аурулар, геном.

АННОТАЦИЯ

ОЦЕНКА ПОСЛЕДСТВИЙ ВЛИЯНИЯ РАДИАЦИИ НА УСТОЙЧИВОСТЬ ГЕНОМА НАСЕЛЕНИЯ ТЕРРИТОРИЙ, ПРИЛЕГАЮЩИХ К ИСТОЧНИКУ ЗАГРЯЗНЕНИЯ

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Актуальность: Всё возрастающее введение в среду обитания человека новых мутагенных факторов приводит к увеличению частоты наследственно-отягощенных заболеваний человека, связанных с воздействием мутагенов окружающей среды. В случае радиационного воздействия встает проблема необходимости выявления групп риска людей с повышенной чувствительностью, так как влияние радиации на организм, помимо прямого воздействия на его функциональные подсистемы, индуцирует или активирует защитные системы (репарацию, адаптацию). При повреждении системы репарации ДНК возрастает риск индукции частоты мутации. Вышеизложенное подчеркивает актуальность темы исследования и полученных результатов.

Цель исследования – оценка воздействия радиационного загрязнения окружающей среды на стабильность генома и здоровье человека с учетом отдаленных генетических последствий.

Материалы и методы: Использовались полевые и лабораторные методы: создание системы площадок для взятия проб объектов окружающей среды, образцы периферической крови человека, методики измерения радиационной активности объектов, цитогенетические и молекулярно-генетические методы.

Результаты: Измерения гамма-активности показали, что уровень радиации обследованной территории полигона и прилегающих населенных пунктов находится в пределах 0,6-0,14 мкЗв/ч. Особое значение в этом отношении имеет изучение механизмов индивидуальной чувствительности к радиации и роли системы репарации ДНК. Молекулярно-генетические исследования ДНК клеток крови и цитогенетические анализы людей, проживающих в зоне влияния полигона радиоактивных отходов, выявили распространение нескольких мутантных генотипов, что свидетельствует о вероятности повышении риска экологических заболеваний у лиц с выраженной нестабильностью генома.

Заключение: Уровень радиации на территории полигона и прилегающих населенных пунктов находится в пределах 0,6-0,14 мкЗв/ч. Анализ распределения людей по генотипам в обследованных группах показал увеличение частоты гетерозиготных аллелей гена репарации XRCC1 до 35% по сравнению с контрольной группой (10%), а частота гомозиготных аллелей по аллелю Ttr/Ttr не превышает уровень контроля (3%). В свою очередь, для гена XRCC3 отмечается незначительное превышение частоты гетерозигот по сравнению с контролем – 21%, а гомозиготный аллель Thr/Met остается на уровне контроля – 2%.

Ключевые слова: мутагены, радиация, окружающая среда, гены, наследственные заболевания, геном.

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