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Human Genetic Response to Environmental Change: Biological Adaptation to Global Climate Change

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Abstract The study comprehensively explored the genetic response of humans to global climate change, particularly the genetic adaptation strategies revealed through genome-wide association studies (GWAS). The study conducted an in-depth analysis of the current situation of global climate change and its wide-ranging impacts on human society and health, emphasizing the importance of understanding human genetic response mechanisms. The biological basis of genetic adaptability was introduced, including the role of genetic variation and natural selection, as well as the impact of environmental stress. On this basis, the principles and methods of GWAS technology were further elaborated, as well as its role and significance in revealing human genetic adaptation to environmental factors such as temperature changes and increased ultraviolet radiation. The challenges faced in using GWAS to study human adaptability were discussed, and technological advancements, especially high-throughput sequencing technology and the application of artificial intelligence in data analysis, were explored to help overcome these challenges. This study aims to provide a new perspective for understanding human genetic adaptability and scientific basis and strategic recommendations for addressing the challenges of climate change in the future.

Keywords Global climate change; Environmental change; Genetic adaptability; Genome-wide association studies (GWAS); Biological adaptation

Global climate change is one of the most severe environmental issues facing the world today. The direct consequences include rising temperatures, an increase in extreme weather events, rising sea levels, and changes in ecosystems (Lamaoui et al., 2018), which have profound impacts on all living organisms on Earth, including human society and health. For humans, climate change not only threatens the security of food and water resources, but may also exacerbate the spread of infectious diseases, trigger health and socio-economic issues, and threaten the living environment and quality of life for humans. Therefore, understanding how humans respond to environmental changes through genetic adaptation is not only a scientific issue, but also an urgent issue that needs to be addressed at the social, health, and even policy levels.

Genome-wide association studies (GWAS), as a powerful genetic research tool, reveal the genetic basis of many human diseases and physiological traits by analyzing the association between genetic variation and specific traits (Uffelmann et al., 2021). In recent years, with the development of sequencing technology and advances in bioinformatics, GWAS technology has been widely applied to study human genetic responses to environmental changes, especially in the context of global climate change. By analyzing genomic data from different geographical and environmental backgrounds, researchers have begun to understand how humans adapt to specific environments through genetic variation, including high-altitude, low oxygen environments, extreme temperature conditions, and so on. These studies not only enhance our understanding of human evolution, but also provide scientific basis for predicting future population health trends, and even provide reference for formulating public health strategies to adapt to climate change.

The aim of this study is to integrate and analyze the results of GWAS research, identify key genetic markers that respond to climate change, explore the genetic response mechanisms of humans to environmental change, especially global climate change, and understand how these genetic variations help humans adapt to the constantly

changing environment. This study will provide a detailed introduction to GWAS technology and its application in genetic research, and then analyze how humans respond to environmental changes through genetic adaptation through specific research cases. We hope that this study can provide a new perspective for understanding human genetic adaptability, and provide scientific basis and strategic recommendations for addressing the challenges of climate change in the future.

1 The Current Situation and Impact of Global Climate Change

1.1 Current status and future trends of global climate change

In the past few decades, global climate change has become an irreversible global issue affecting the Earth's ecosystem and human society. Scientific evidence shows that global surface temperatures have risen by approximately 0.6 °C above the average of the previous century, consistent with model predictions of rising concentrations of carbon dioxide (CO₂) and other greenhouse gases. Extreme temperature events have become more frequent, causing increasing damage to ecosystems, agriculture, and human health. At the same time, the hydrological cycle will strengthen due to increased surface and ocean evaporation rates, leading to increased rainfall in tropical and high latitude regions, while large areas within the continent will become drier and warmer. More frequent climate extreme events will increase the frequency and intensity of drought and flood events worldwide. In addition, rising sea levels will put millions of people at risk and pose significant challenges to many low-lying areas (Rossati, 2016).

According to a report by the Intergovernmental Panel on Climate Change (IPCC), the global average temperature has risen by approximately 1 °C since the late 19th century. Although this number may sound small, it has led to significant climate change, including an increase in the frequency and intensity of heat waves, accelerated glacier melting, and an increase in extreme rainfall events. In the future, if greenhouse gas emissions continue to increase, it is expected that the global average temperature will rise by 2 °C to 5 °C compared to pre industrial levels by the end of this century. This temperature rise will further exacerbate the impact of climate change and have more serious consequences for both natural and human systems.

1.2 The widespread impact of global climate change

The impact of global climate change is multifaceted, including changes in natural ecosystems and various aspects of human society and economy. The high temperatures and extreme weather events caused by global climate change have had a wide-ranging impact on ecosystems. The habitats of organisms are undergoing changes, and some species are at risk of extinction, posing a serious threat to biodiversity. For example, coral reefs experience bleaching due to rising seawater temperature, losing their vitality (Yamano et al, 2011); The melting of the Arctic ice sheet poses a threat to the living environment of species such as polar bears (Stirling and Derocher, 2012) (Figure 1).

Climate change has also had a direct impact on agricultural production, leading to increasingly serious food security issues. Heat waves and droughts reduced crop yields, while floods and rainstorm damaged farmland and affected the food supply chain (Lamaoui et al., 2018). In addition, climate change has intensified the frequency and intensity of natural disasters, such as hurricanes, typhoons, and floods, which not only cause huge economic losses but also seriously threaten human life safety. At the same time, climate change also poses a significant threat to public health. Sargent et al. (2022) predicted that under climate change scenarios, the emergence of certain new suitable areas will put populations who were not previously exposed to such diseases at risk, while the suitability for dengue fever and Zika virus transmission is expected to increase in duration and geographical scope. In addition, heatwaves will increase the risk of heatstroke and cardiovascular disease; The deterioration of air quality will exacerbate respiratory diseases; Extreme weather events will also exacerbate mental health issues, including anxiety and depression.

Faced with the severe challenge of global climate change, the global community is striving to find strategies to adapt to and mitigate climate change. This includes reducing greenhouse gas emissions, protecting and restoring ecosystems, enhancing the resilience of socio-economic systems, and developing agricultural technologies that

adapt to climate change. At the same time, understanding the genetic response mechanisms of humans to climate change can provide us with deeper adaptation strategies, thus better preparing us to cope with the challenges brought by future climate change.

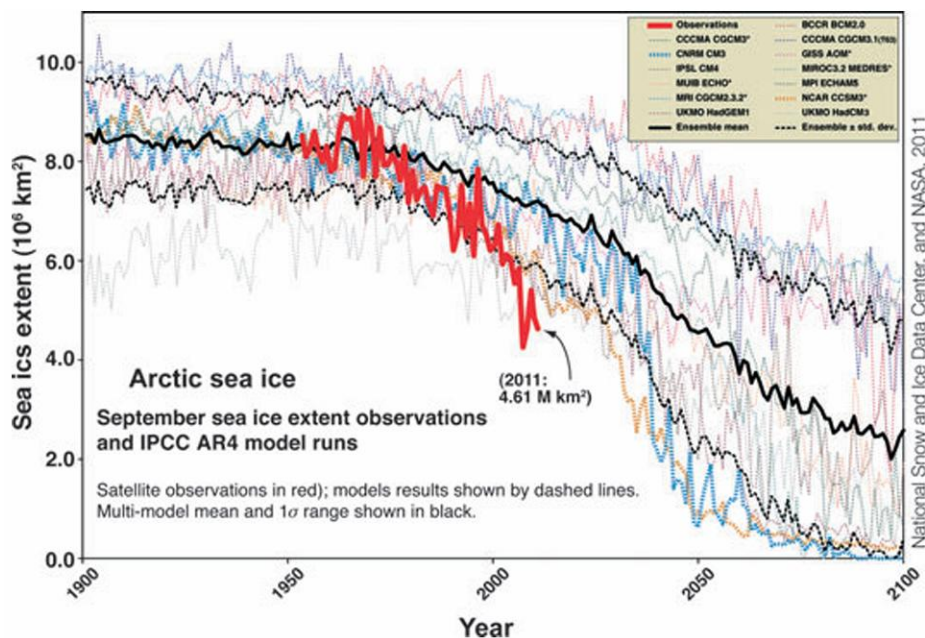


Figure 1 Projections of the minimum amount of sea ice remaining in September in the Arctic Ocean from 1900 to 2100 (Stirling and Derocher, 2012)

Note: The black line is the average of the models and the red line, showing direct observations made from satellite images, shows sea ice is actually being lost more rapidly than currently modeled

2 The Biological Basis of Genetic Adaptability

2.1 Concept and mechanism of genetic adaptability

Genetic adaptability refers to the ability of organisms to survive and reproduce in specific environments through genetic variation (Jerison et al., 2017). This concept is the core of Darwin's theory of evolution, emphasizing the interaction between organisms and the environment. The formation of genetic adaptability depends on genetic variation and recombination, which provide raw materials for natural selection. In the process of natural selection, genetic variations that can improve an individual's survival and reproductive success rate in a specific environment will gradually accumulate, while those that are not conducive to survival will be eliminated. In addition, gene flow (exchange of genes between different populations) and genetic drift (changes in gene frequency due to random events in small populations) can also affect the formation of genetic adaptability.

Genetic adaptability is the accumulation of genetic variations that occur during the long-term evolution of organisms in order to better adapt to environmental changes. This process involves complex biological mechanisms, including the generation of genetic variations, natural selection, and changes in gene frequency. The researchers fitted some models to estimate the narrow sense heritability of this adaptability (Jerison et al., 2017) (Figure 2).

2.2 The role of genetic variation and natural selection in adaptive evolution

Genetic variation is the cornerstone of adaptive evolution. Whether through mechanisms such as point mutations, gene recombination, or gene replication, genetic variation provides diversity for populations, enabling organisms to adapt to environmental changes. Natural selection is the process of determining whether these variations can be preserved. Under natural selection, beneficial mutations increase an individual's adaptability, gradually increasing their frequency within the population, while harmful mutations gradually disappear due to reduced survival and reproductive abilities.

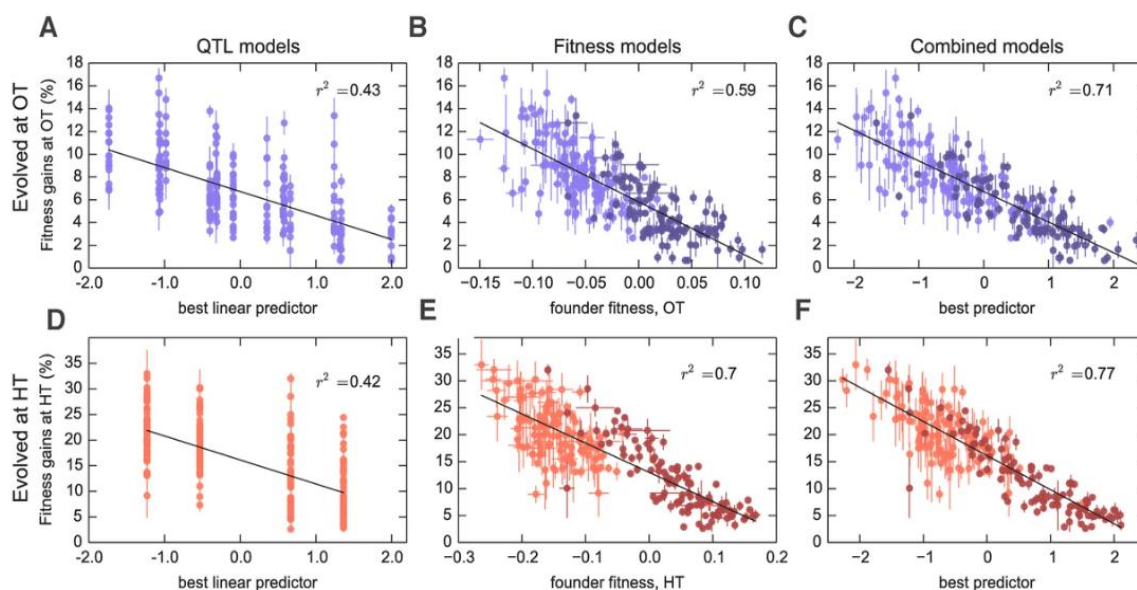


Figure 2 Models to predict adaptability (Jerison et al., 2017)

Note: Each point represents the average fitness change of four populations descended from the same founder after 500 generations of evolution in the (A–C) OT environment or (D–F) HT environment (Lines show the predictions of different models)

For example, there is a hemoglobin variant (sickle cell variant) that is resistant to malaria in populations located in Africa. In areas where malaria is prevalent, individuals carrying this variant have higher survival and reproduction rates due to their stronger resistance to malaria, resulting in a higher frequency of this variant in the population (Olukorede et al., 2022). This is an example of how natural selection can increase population adaptability to certain genetic variations in specific environments.

2.3 The impact of environmental stress on genetic adaptability

Environmental pressure is the main driving force behind genetic adaptive evolution. When environmental conditions change, the original biological adaptability may no longer adapt to the new environment. At this time, genetic variations that can increase biological adaptation to the new environment will be favored by natural selection (Guzella et al., 2017). Environmental pressure can refer to changes in the ecological environment, such as climate change and changes in food sources; It can also be changes in biological factors, such as the spread of diseases, the emergence of predators or competitors. These environmental pressures prompt organisms to continuously adapt, thereby driving the evolution of genetic adaptability.

Climate change is a typical example of environmental stress. As global temperatures rise, many species have to adapt to warmer climate conditions. Roth et al. (2014) found that due to climate warming, some species in low altitude areas have shifted towards adapting to warm conditions and have correspondingly moved up to a certain altitude. However, as altitude increases, the rate of change in butterfly communities slows down, while bird communities transition towards species that adapt to warmth at all altitudes. This indicates that there are differences in the response of different species to climate change.

Chen et al. (2011) demonstrated through meta-analysis that species are changing their distribution at an increasingly rapid rate in response to climate change. The migration rates of these species towards higher altitudes and latitudes are 11 meters and 16900 meters per decade, respectively, which are approximately two to three times higher than previously reported rates. The distance of species migration is related to the level of temperature change, indicating that the range of species depends on multiple internal and external factors.

Genetic adaptability is one of the core mechanisms of biological evolution, which enables organisms to survive in constantly changing environments. Genetic variation provides the raw materials for adaptive evolution, while natural selection determines which variations can be preserved, and environmental pressure is the main driving

force behind this evolutionary process. Understanding this process is not only crucial for revealing the formation and maintenance mechanisms of biodiversity, but also provides a theoretical basis for predicting and responding to adaptive changes in organisms under global climate change.

3 GWAS Technology Principles and Functions

Genome-wide association studies (GWAS) are an important method in modern genetic research, which reveals the genetic basis of many diseases and physiological traits by analyzing the association between genetic variations on the genome and specific traits. The application of GWAS has greatly promoted our understanding of genetic etiology, which is of great significance for analyzing the role of human genetic variation in adapting to environmental changes.

3.1 Principles and methods of GWAS

The core principle of GWAS is to search for associations between genotype (individual genetic information) and phenotype (observed traits or disease status) through statistical analysis (Uffelmann et al., 2021). This method typically involves two steps: first, collecting genomic and phenotypic data from a large number of individuals; Secondly, statistical methods are used to analyze these data to determine which genetic variations (usually single nucleotide polymorphisms, SNPs) are associated with specific phenotypes.

During the data collection phase, researchers typically select two groups of individuals: one group is individuals with a specific phenotype (such as a certain disease), and the other group is a control group (individuals without that phenotype). Through high-throughput sequencing technology, researchers can identify millions of SNPs in the genomes of these individuals. During the data analysis phase, researchers used statistical software to compare the frequency differences of SNPs between two groups, in order to identify SNPs significantly associated with specific phenotypes.

3.2 The role and significance of GWAS in revealing human genetic variations

The application of GWAS technology has greatly promoted the development of human genetics. Through GWAS, researchers have successfully identified thousands of genetic loci associated with human diseases and physiological traits (Visscher et al., 2017). These findings not only enrich our understanding of genetic diversity, but also provide new ideas for disease prevention, diagnosis, and treatment.

GWAS has made significant achievements in the field of disease genetics, especially in the analysis of genetic causes of complex diseases. For example, in the study of cardiovascular disease (Liu et al., 2014), type 2 diabetes (Hao et al., 2015), Alzheimer's disease (Ertekin Taner, 2010) and other diseases, GWAS revealed a large number of previously unknown risk genetic loci. These findings not only contribute to understanding the genetic mechanisms of diseases, but also provide possibilities for early prediction and personalized treatment of diseases.

GWAS has also played an important role in studying human genetic adaptability to environmental changes. By analyzing genetic variations associated with specific environmental factors, researchers can explore how humans adapt to different environmental pressures during long-term evolution. For example, GWAS research has found a specific genetic variation in Tibetan populations located in high-altitude areas (Han and Li, 2023), which helps them adapt to low oxygen environments. Similar studies also include exploring resistance to malaria and adaptability to changes in dietary habits.

At the same time, GWAS also provides a new perspective for understanding human genetic diversity and history. By comparing the genetic variation patterns of different populations, researchers can trace the pathways of human migration, explore genetic exchange between populations, and analyze the historical evolution of human populations. I believe that with the advancement of technology and the accumulation of big data, GWAS will play a more important role in future human genetic research.

4 Genetic Response of Humans to Environmental Changes

4.1 GWAS revealing human genetic adaptation strategies to global climate change

The environmental pressures brought about by global climate change, such as temperature changes and increased ultraviolet radiation, require humans to have corresponding genetic adaptability to ensure survival and reproduction. GWAS research helps us understand how humans adapt to these changes by identifying genetic variations associated with these environmental factors.

Williams et al. (2016) found that there is a genetic variation related to energy metabolism in populations living in high latitude regions, which helps improve the body's ability to generate heat in cold environments. Through GWAS analysis, researchers found that there are abnormally high frequencies of certain SNPs in populations in these regions, which are located on genes related to fat metabolism and energy production, indicating that these genetic variations may be the genetic basis for human adaptation to cold environments.

Similarly, GWAS research has also discovered the genetic basis for human skin color diversity in adaptation to ultraviolet radiation. Crawford et al. (2017) conducted a GWAS study on different races in Africa and found significant genetic variations associated with skin color variations, located in four genomic regions, explaining almost 30% of phenotypic variations. The most significant association was found near the SLC24A5 gene, which has previously been associated with skin color in European populations. The study also found that variations near MFSD12, DDB1, TMEM138, OCA2, and HERC2 were significantly associated with skin color. These genes are involved in various functions such as UV response, DNA damage repair, and melanocyte biology.

Yang et al. (2022) found that Tibetans in the Qinghai Tibet Plateau have a deeper baseline skin color and better tanning ability compared to Han Chinese in the lowlands (Figure 3). This study found that the adaptive variation of GNPAT may enhance the tanning ability of Tibetans through the interaction of genes such as CAT and ACAA1 related to melanin synthesis, in order to better protect the skin from UV damage.

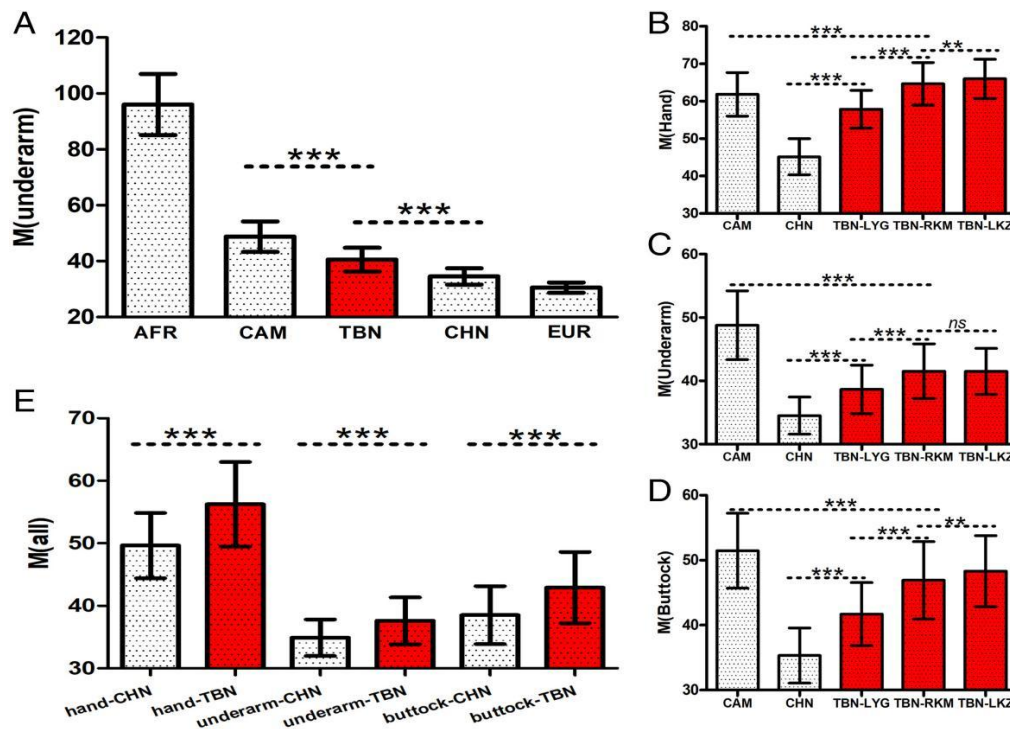


Figure 3 Skin pigmentation comparison among different populations (Yang et al., 2022)

Note: A: Comparison of skin darkness among world populations. The y axis indicates the M value of underarm and the error bars indicate SD; B–D: Comparison of skin darkness (hand, underarm, and buttock) among Tibetans from different altitudes; E: Comparison of the constitutive and facultative skin color of TBN and CHN at similar altitudes (three different locations in Qinghai Province of China, 3,712 ± 64 m). ns, not significant, $P > 0.05$; * $P < 0.05$, ** $P < 0.01$, *** $P < 0.001$

These studies confirm that the variation in human skin color is not only a genetic adaptation to ultraviolet radiation, but also reflects the richness of human genetic diversity. Through GWAS analysis, scientists have identified multiple SNPs related to skin color, deepening our understanding of the complex genetic mechanisms behind human adaptation to different environmental lighting conditions.

4.2 The significance of genetic adaptation strategies for human future survival and health

The genetic adaptability of humans to environmental changes is not only the result of evolutionary processes, but also of great significance for our future survival and health. Understanding human genetic adaptation strategies can help us predict potential health challenges in the context of global climate change. For example, as global temperatures rise, some diseases that originally only appeared in tropical regions may spread to temperate regions. Understanding human genetic adaptability to these new environmental pressures will provide guidance for public health strategies.

Moreover, research on genetic adaptability can help us design more effective medical interventions. By understanding the mechanisms of human genetic variation under specific environmental pressures, we can develop targeted drugs and treatment methods to address health issues caused by environmental changes. It is worth mentioning that research on human genetic adaptability to environmental changes emphasizes the importance of protecting genetic diversity. Genetic diversity is a key resource for humans to adapt to environmental changes. Only by maintaining a high level of genetic diversity can humans effectively cope with future environmental challenges.

5 Challenges and Opportunities

GWAS has become an important means of analyzing human genetic adaptability, but in practical applications, researchers still face many challenges, including the complexity of data and the difficulty of statistical analysis. GWAS involves a vast amount of data, with each participant's genome potentially containing millions of single nucleotide polymorphisms (SNPs), making the processing and analysis of this data extremely complex. Moreover, research on human genetic adaptability also requires consideration of the interaction between environmental and genetic factors, which increases the difficulty of data analysis (Welter et al., 2013). Meanwhile, GWAS requires the use of complex statistical methods to identify genetic variations associated with specific traits. Due to the large number of hypotheses tested, false positive results are prone to occur. Therefore, how to design a reasonable statistical model and how to correct the impact of multiple comparisons have become challenges in research.

At present, the development of high-throughput sequencing technology has greatly improved the efficiency and accuracy of GWAS, making it possible to sequence more samples in a shorter time and at a lower cost (Schmidt and Hildebrandt, 2017). This not only speeds up the discovery of genetic variations, but also makes diversity research, including smaller populations, feasible. In addition, the application of artificial intelligence and machine learning technology, especially in data processing and statistical analysis, provides powerful tools for processing complex data in GWAS. These technologies can help researchers identify meaningful patterns and associations from large datasets while reducing the occurrence of false positive results.

With the intensification of global climate change, understanding how humans can adapt to environmental changes through genetic adaptation has become particularly important. Genetic adaptability research can not only help us predict the potential impact of future climate change on humans, but also provide scientific basis for formulating long-term strategies to adapt to climate change. Although GWAS research on human adaptability faces many challenges, technological progress is continuously driving the development of this field, providing us with new opportunities to deepen our understanding of human genetic adaptability. Applying these research findings to public health and strategies to address climate change is expected to make significant contributions to protecting human health and promoting sustainable social development.

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