



## PUBLIC HEALTH GENOMICS ; THE FUTURE AND ROLE OF AYURVEDA

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**ABSTRACT** Genomics is the branch of molecular biology concerned with the structure, function, evolution, and mapping of genomes. The national effort to use genomic knowledge to save lives is gaining public importance. The increasing demand of genomics in public health is reflected in the inclusion of genomics in key public health initiatives. Public health genomics has progressed to responsibly integrate advancements in genomics into the fields of personalized medicine and public health. Public health has an important and decisive leadership role in addressing the promise and hazards of human genomics for population health. Such efforts are needed to implement what is known in genomics to improve health, to reduce potential harm and create the infrastructure needed to derive health benefits in the future. There are many controversies about the role of genomics in public health practice and debates has been ongoing for quite some time. Ayurveda uses an entirely different holistic system, which includes terms such as dosha and Prakriti. The concept of genomics is already merged in Ayurveda under the umbrella of Prakriti. In spite of accelerating human genome discoveries in a wide variety of diseases of public health significance, the promise of personalized health care and disease prevention based on genomics has lagged behind. So appropriate, effective and sustainable integration of genomics into healthcare requires an organized approach. This paper outlines how genomics can be incorporated into public health practice, its challenges and ethical issues along with the role of prakriti and genomics into the new era of precision public health.

**KEYWORDS :** Public Health, Genomics, Prakriti, Public Health Genomics, Ayurveda

### INTRODUCTION

The questions are as old as humanity. Why do children resemble their parents? What is responsible for a person's grey eye, curly hairs? Why do certain diseases including psychological diseases run in families? Before the advent of molecular biology scientists approached such questions largely through the study of whole organisms. Then emerged the new genetic techniques which allow researchers to read an organism's genome and breakthrough in the field of genetics took the front page of various frontline and science and technology magazines.

Genomic science is developing rapidly and engagement of public health professionals will be necessary to appraise new technologies and use them effectively. Public health genomics has evolved to responsibly integrate advancements in genomics into the fields of personalized medicine and public health. Appropriate, effective and sustainable integration of genomics into healthcare requires an organized approach. Ayurveda uses an entirely different holistic system, which includes terms such as dosha and Prakriti. The concept of genomics is already merged in Ayurveda under the umbrella of Prakriti. Studies are needed in this field to establish the relationship between Prakriti and genotype. This paper outlines how genomics can be incorporated into public health practice and also the role of prakriti and genomics into the new era of precision public health.

### MATERIALS AND METHODS

MEDLINE (www.pubmed.com) or the PubMed database was screened using keywords like *Public Health, Genomics, Prakriti, Public Health Genomics, Ayurveda, Ayurgenomics* with their corresponding mesh terms in combination like OR, AND. The inclusion of articles in this review was done by the following process and eligibility criteria: The search was limited to only English literature. Articles published from 1999 to 2021 were included. This search resulted in a total of 80 articles from the PubMed source and 10 articles from other sources such as Google Scholar, MEDSCAPE, and Science Direct. After removing all the duplicates 30 articles were selected. From these 30 articles 14 articles were included as the other 16 articles were unrelated to the topic concerned. This search was undertaken in September 2020.

### Public health genomics

An emerging field that assesses the impact of genes and their interaction with behavior, diet and the environment on the population's health and use this information to develop strategies to prevent disease.<sup>1</sup>

The human genomic research has the potential to achieve a number of public health goals, such as to reduce global health inequalities by providing developing countries with efficient, cost-effective means of preventing, diagnosing & treating diseases. This global inequalities can be reduced by equitable economic investment, clinical research, use of genomic services and technologies globally.

### Core functions of public health<sup>2</sup>

The Institute of Medicine (IOM; now known as the Health and Medicine Division) of the United States of America's National Academies of Sciences, Engineering and Medicine in 1988 identified and defined the three core functions to be provided by all public health agencies.

- **Assessment:** to assess and monitor the health of communities and populations at risk, and to identify health problems and priorities. This requires the regular and systematic collection, analysis, and dissemination of information on the health of populations
- **Policy development:** to formulate public policies, plans, standards, guidelines, and resources in collaboration and partnership with stakeholders, and to solve identified local and national health problems and priorities.
- **Assurance:** to assure that all populations have access to appropriate and cost effective care (including health promotion and disease prevention services), and to evaluate the effectiveness of healthcare and public health interventions

### The 10 essential public health services<sup>3</sup>

1. Monitor and evaluate health status to identify community health problems.
2. Diagnose and investigate health problems and health hazards in the community.
3. Inform, educate, and empower people about health issues.
4. Mobilize community partnerships to identify and solve health problems.
5. Develop policies and plans that support individual and community health efforts.
6. Enforce laws and regulations that protect health and ensure safety.
7. Link people to needed personal health services and assure the provision of health care when otherwise unavailable.
8. Assure a competent public and personal health care workforce.
9. Evaluate effectiveness, accessibility, and quality of personal and population-based health services.

10. Research for new insights and innovative solutions to health problems.

**Table 1 | Public Health Genomics Activities In Relation To The 10 Essential Public Health Services.<sup>2</sup>**

Essential public health services	Public health genomics activities
1. Monitor health status to identify and solve community problems	<b>Assess the distribution and impact of modifiable and genetic risk factors to determine their contribution to health status and the burden of disease.</b> A better understanding of these risk factors could enable more precise decision-making about resource allocation and the prioritization and targeting of public health programs, and lead to new approaches to disease prevention and treatment
	<b>Promote the development of resources that enable monitoring of the genomic-related health status of populations. Key activities could include:</b> <ul style="list-style-type: none"> <li>assessing the inclusion of genomics information in the collection, management, and analysis of routine data</li> <li>working with national surveys and large epidemiology groups to maximize potential from databases</li> <li>exploring the potential for disease-specific, and population-based, registries to be used to conduct disease surveillance.</li> </ul>
2. Diagnose and investigate health problems and hazards in the community	<b>Identify and track infectious disease outbreaks using genomic technology</b> This involves utilizing genomic technology to improve the speed and efficiency of infectious disease surveillance and response
	<b>Assist with the redesign of diagnostic and laboratory services to incorporate new genome-based technologies.</b> Examples of these technologies include massively parallel sequencing such as whole exome and whole genome sequencing (39). There is potential for the incorporation of these technologies into diagnostic and laboratory services that can improve the diagnostic yield from genetic testing.
3. Inform, educate, and empower people about health issues	<b>Improve the genomic literacy of the public.</b> This involves providing education materials to communities that teaches them about genetics and genomics in understandable language
	<b>Empower all stakeholders, including health professionals and the public, to make informed decisions about the uses of genetic information with realistic expectations about the risks and benefits.</b> This includes the provision of relevant information on the uses of genomic information in disease prevention (22, 31), as well as on the associated ethical, legal and social issues.
	<b>Facilitate the integration of genomics into health promotion and disease prevention programs.</b> This will contribute to informing and educating people about genomics knowledge and technologies, as well as its limitations.

4. Mobilize community partnerships to identify and solve health problems	Foster collaborations between stakeholders. This encompasses capacity building, and developing networks and partnerships between diverse stakeholders including public policy makers, patients, the general public, academia, clinicians, researchers, and industry
5. Develop policies and plans that support individual and community health efforts	Policies and plans that could be developed include those relating to: <ul style="list-style-type: none"> <li>the appropriate use of genomic applications, through standards and guidelines that recognize the complexity of genomics and define when and how genome-based information and technologies should be used to promote health and prevent disease, including in the clinical setting</li> <li>equity and accessibility, to assure genomics knowledge and technologies are accessible across all segments of the population</li> <li>the use of family health history information to inform people of the role of inheritance in the development of disease and identify people at risk of disease</li> <li>reproductive decision-making, including prenatal screening, population-based carrier screening and pre-implantation genetic diagnosis</li> </ul>
6. Enforce laws and regulations that protect health and ensure safety	Contribute to: Laws and regulations for genomic applications. This could apply to genetic tests, including direct-to-consumer tests and related issues such as funding, data protection, insurance coverage for high-risk individuals and the prevention of genetic discrimination. Regulations for laboratories using genome-based technologies. An example of these technologies is massively parallel sequencing.
7. Link people to needed health services and assure provision	Support the appropriate integration of genomic knowledge and technologies into all aspects of healthcare and public health. <ul style="list-style-type: none"> <li>This may be operationalized in a number of ways, such as:</li> <li>Supporting the implementation of evidence-based genomic applications and discouraging the use of unvalidated applications, to prevent the premature use, misuse and overuse of genomic applications.</li> <li>Providing expert advice on the commissioning of services that use genome-based knowledge and technologies. This may relate to issues such as the appropriateness of the technologies for use; and the impact on, or requirements for, supporting functions such as counseling, education, and service coordination.</li> <li>Supporting the incorporation of genomic applications into existing</li> </ul>
	public health practice, such as: using pathogen and human genomic technologies to control and manage communicable diseases; expanding population-based screening programs to include the use of genetic information; and targeting interventions for preventing diseases in population groups based on genetic information. <ul style="list-style-type: none"> <li>Promoting the use of family health history to identify individuals at risk of disease. Family history is the most</li> </ul>

	<p>consistent risk factor for all diseases and reflects the complex interactions between genes, behaviors, cultures and environments that family members share. It can be used to identify families at high risk for disease and could be incorporated into tailored chronic disease prevention and health promotion messages.</p> <ul style="list-style-type: none"> <li>Ensuring equity and accessibility to genomic applications and services . This is especially important for population groups that traditionally face barriers to accessing health services, such as Indigenous and low socio-economic groups.</li> </ul>
<p>8. Assure a competent public and personal healthcare workforce</p>	<p>Contribute to training and education in, and development of, genomic knowledge, skills and capacity for health professionals. This is so that: genomics is appropriately integrated into their work; they can effectively communicate genetic information; and they can support informed decision-making by patients .</p> <p><b>Support the development of workforce capacity in genomics-related fields.</b> These fields include bioinformatics, genetic epidemiology, law and ethics, and health economics as applied to genetics and genomics</p>
<p>9. Evaluate the effectiveness, accessibility and quality of health services</p>	<p><b>Evaluate new genome-based knowledge and technologies to determine their evidence base, quality, appropriateness and readiness for implementation in healthcare and public health practice.</b></p> <p>The need for evaluation is based on concerns that the availability of genome-based tools and technologies, such as genetic tests, diagnostic equipment and therapies, are being driven more by technical feasibility and commercial potential than by evidence-based implementation. Such evaluations ensure that the benefits of genomic discoveries are realized efficiently, effectively and equitably, and are only implemented when it is in the public's best interest.</p> <p><b>Evaluate the use of genome-based knowledge and technologies in healthcare and public health practice.</b> Examples of evaluations include: the current use of genetic tests and services; the factors that influence utilization; cost-effectiveness; and the impact on service, intervention and patient outcomes</p>
<p>10. Research for new insights and innovative solutions to health problems</p>	<p><b>Monitor the results of human genome epidemiology studies.</b> This provides a population perspective on gene-disease associations, estimating the contribution of gene variants to the occurrence of disease in groups and the population overall. Monitoring these studies can help identify gaps in knowledge at the population level and could lead to changes in public health prevention interventions and disease management.</p> <p><b>Support the development of infrastructure for conducting genomic-related population research.</b></p> <p>Patient registries, population data sets and linked biobanks are key resources enabling the conduct of large population studies to assess gene-environment interactions. However, steps must be taken to ensure that databases reflect genomic reference ranges for the whole population, inclusive of minority groups, to avoid inequity of the applications of genomic technology and knowledge.</p> <p><b>Conduct and monitor translation research.</b></p>

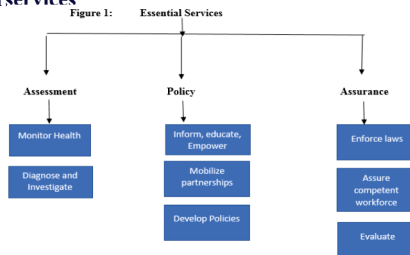
The aim of translation research is to move appropriate genomic technologies from the discovery phase to application in healthcare and public health practice, and to evaluate its use in practice for improving health outcomes

The integration of genomics into public health research, policy and practice The main objective was to examine the opportunities, responsibilities of the public health community in bridging the gap between gene discovery and application of genetic information to improve health and prevent disease.

The integration of genomics with the 3 core functions of public health are:4

- Assessment: The regular systematic collection, assembly, analysis, and dissemination of information, including genetic epidemiologic information, on the health of the community.
- Policy Development: The formulation of policies, in collaboration with stakeholders, that promote the effectiveness, accessibility, and quality of genetic tests and services.
- Assurance: The assurance to constituents that genetic tests and services meet goals for effectiveness, accessibility, and quality.

**Essential services**



**Essential services: assessment**

- Monitor Health: Monitor health status to identify health problems, including those that have a known genetic component, within the community.
- Diagnose & Investigate: Investigate the distribution of genetic and environmental risk factors within the community to determine their contribution to identified health problems and to improve health outcomes.

**Essential services: policy**

Inform, Educate, Empower: Facilitate communication and education about the integration of genetics into health promotion and disease prevention programs.

Mobilize Partnerships: Foster collaboration among public and private agencies and constituent groups to promote effective and efficient policy making.

Develop Policies: Establish policies and guidelines for when and how genetic tests should be applied to promote health and prevent disease.

**Essential Services: Assurance**

Enforce Laws: Promote the enforcement of policies and standards enacted to ensure the effectiveness, accessibility, and quality of genetic tests and services.

Assure Competent Workforce: Ensure that present & future health professionals have appropriate training and skills in the use of genetic tests and services to promote health and prevent disease.

Evaluate: Evaluate the effectiveness, accessibility, and quality of genetic tests and services.

**How Genomics Improved The Understanding Of Diseases.<sup>3</sup>**

“Genomic knowledge” refers to the information that is obtained from studying the complete genetic makeup of a cell or organism. In recent years, scientific research in this area has contributed significantly to our knowledge about the human genome, improving our ability to understand disease etiology, risk, prevention, diagnosis, and treatment. The ways in which these areas can be enhanced by genomic knowledge are outlined below. Based on these improved understandings, genomic tools, and technologies are being developed to enable better health not

just for the individual, but for populations as well.

### 1) Etiology:

Increased genomic knowledge about a disease can provide insights into how the disease may develop. This can occur through a better understanding of the function of genes that make up the genome, how different genetic variants contribute to the phenotype of diseases, the role of gene expression, and the role of the interaction between genes.

### 2) Risk:

Genomic knowledge is expected to improve our understanding of why some individuals remain healthy while others are more susceptible to disease. For example, information on the genetic variants associated with an increased risk of common diseases, such as cardiovascular disease and diabetes, might at some point be used to make predictions about the likelihood a person will get these diseases. This knowledge could then be applied to develop new tools for risk prediction or predictive testing in relation to the onset or recurrence of disease.

### 3) Prevention :

Understanding how the genome influences the etiology and risk of diseases may lead to improved understanding of how diseases, or the symptoms of disease, can be prevented. Genomic tools and technologies can also identify infectious diseases with greater speed and precision to enable rapid responses to disease outbreaks and more efficient surveillance.

### 4) Diagnosis:

Historically, clinicians generally used a set of observable or measurable characteristics as the basis for diagnosing disease. Genomic knowledge takes this one step further, by enabling clinicians to look at a person's genes to provide a molecular diagnosis. In line with this, diagnostic technologies have been developed that include a plethora of clinical genetic tests.

### 5) Treatment:

To date, genomic knowledge has mostly been used to inform disease treatments. Pharmacogenetics and pharmacogenomics are two fields where new and improved therapies and treatments have been developed, including hundreds of new drugs which are advancing disease management. The expectation is that genomic knowledge will further improve the ability to assess treatment responses, such as how different people metabolize drugs and which people are more likely to experience adverse drug reactions. Based on genetic profiles, tailored therapies may be developed for an individual and across individuals within specific patient populations to deliver the right drug in the right dose at the right time.

### Role Of Ayurveda

Ayurveda has a unique way of classifying humans, which is used in the clinical management of health and disease. Humans are classified into three fundamental types of constitution or prakriti, called vata, pitta, and kapha based on their anatomical, physiological, and psychological characteristics. According to Ayurveda, prakriti of a person is determined at the time of conception and does not change until death. Recommendations on diets, lifestyles, and drugs vary depending on the prakriti of the individual.<sup>5</sup> Since it gets determined at the time of conception, in the past decade, the hypothesis that prakriti has a genetic basis was tested by different groups of Indian scientists.

A correlation between specific prakriti and HLA-DRB1 polymorphism was demonstrated by Bhushan et al.<sup>6</sup> Prasher et al.<sup>7</sup> and Mukherjee and Prasher<sup>8</sup> have used prakriti-based classification and have demonstrated the genomic and biochemical correlates with specific prakriti types. They have termed this approach of classification of humans as Ayurgenomics and propose its potential use in personalized and preventive medicine. Frequency of association of CYP2C19 genotype was demonstrated to vary depending on the prakriti.<sup>8</sup> Differential expression of a high-altitude adaptation gene, EGLN1 as a response to hypoxia, was correlated to specific prakriti type.<sup>9</sup> Rotti et al.<sup>10</sup> found a significant correlation between dominant prakriti to place of birth and body mass index (BMI).

### Prakriti And Genome

The identification of genomic link to the theory of Prakriti led to a search for a possible classification of people on their Prakriti based on their genetic makeup. A significant correlation between various alleles of HLA genotype and Prakriti provides preliminary experimental support for the concept of association between HLA polymorphism and individual Prakriti types.<sup>11</sup> Association between Prakriti and gene-

related to drug metabolism such as CYP2C19 have been drowned out, where the genotype related to extensive metabolizer genotype was associated with pitta Prakriti, while the poor metabolizer genotype was highest in Kapha prakriti.<sup>12</sup> Assessment of Prakriti through genotype mapping is a method of objective assessment of Prakriti. On searching for links of Prakriti and genotypes is the need of the hour to bridge the gap between Ayurveda and current sciences with the development of common vocabulary

### Challenges<sup>13</sup>

The challenge to public health genomics is to overcome inequitable allocation of benefits, the tragedy that would befall us if we made the promise of genetics only for those who could afford it and not for all society. Understanding the need of the patient is the key for determining the right genetic test. The biggest hurdles faced by clinicians are genetic data interpretation, finding genetic links for complex conditions

### Ethical Issues In Genomic Research

It is the duty of the researcher to disclose the results of genomic research to the individual participating in the research. This could be a cause of anxiety or depression to the individual and his/her family.<sup>14</sup> The individual and the family could be subjected to unnecessary psychosocial harm. Issue of social stigma and discrimination—When not revealed. When the genomic data are publicly accessible, there is a risk of discrimination during enrollment for a job or obtaining health insurance<sup>15</sup>

### CONCLUSION

The emergence of “public health genomics” is a multidisciplinary field for the 21<sup>st</sup> Century. Public health genomics has been successfully integrated into existing paradigms for the provision of traditional public health services.<sup>16</sup> The continued alignment of genomics with public health promises to deliver more precise, personalized health care to benefit the population. Ayurveda classifies the whole human population in three major constitutions as Vata, Pitta, Kapha and their possible combinations. Their homologous relation to human genetic structure needs to be studied for validation. Advanced researches for links of Prakriti and genotypes is the need of the hour to bridge the gap between Ayurveda and current sciences and use this knowledge for the public health. Governments and policy makers in this arena have a unique role to play that ensures effective implementation of genomic knowledge and technologies into health systems.

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