Precision Healthcare in India: Integrating Omics Science with Digital Health Innovation

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Abstract:

India is undergoing a transformative shift toward precision healthcare, driven by the convergence of large-scale omics initiatives and digital health innovations. This article reviews three landmark programs, Genome India, Phenome India, and the Bharat Cancer Genome Atlas (BCGA), that are systematically mapping the nation's genomic, phenotypic, and cancer-specific diversity. Together, these projects aim to enhance disease prediction, early diagnosis, and personalized treatment for the Indian population by building comprehensive and publicly accessible data resources. Simultaneously, India's digital health ecosystem, including the Ayushman Bharat Digital Mission, eSanjeevani, CoWIN, and AI-driven surveillance platforms, is enabling broader access, longitudinal data integration, and precision public health interventions. This synergistic integration of omics science with digital platforms exemplifies a shift from reactive to proactive, personalized, and equitable healthcare. The article highlights how these national efforts collectively advance the principles of 4P medicine: Predictive, Preventive, Personalized, and Participatory, and set a precedent for precision health implementation in diverse, resource-limited settings globally.

Keywords: Precision medicine, Omics, Digital health, India, Public health, 4P medicine, Artificial intelligence, Health equity.

In line with its mission to identify, monitor and evaluate emerging scientific developments, the IFCC Emerging Technologies Division (ETD) has undertaken a focused analysis of recent advancements in omics sciences, institutional initiatives, and strategic collaboration opportunities. This effort is particularly timely as the global laboratory medicine community prepares for the upcoming IFCC WorldLab in New Delhi in 2026.

India is rapidly positioning itself at the forefront of precision healthcare, driven by a convergence of scientific ambition, pressing public health challenges, and the extraordinary genetic and cultural diversity of its population. Since the launch of the National Biotechnology Development Strategy (2015–2020) by the Department of Biotechnology (DBT), Government of India, the country has witnessed a strategic shift toward data-driven, personalized healthcare.

Flagship national programs such as Genome India (2020), the Bharat Cancer Genome Atlas (2020), and Phenome India (2023) reflect a coordinated, government-led effort to integrate genomics into public health, spearheaded by national research institutions and primarily funded through public grants. These initiatives are not isolated research ventures but are embedded within a broader national vision that connects innovation, equity, and digital health aligning with India's goal of self-reliance (Atmanirbhar Bharat). By mapping the country's rich genetic and phenotypic diversity, these programs aim to uncover how biological and environmental factors influence disease risk and progression across populations. The resulting insights are expected to accelerate the development of predictive, preventive, and personalized healthcare solutions with wide-reaching implications for public health, research, and policy.

This article explores these landmark initiatives in detail, examining their goals, progress, and the transformative potential they hold for precision medicine in India and beyond. Genome India, launched by the Department of Biotechnology, is a flagship consortium project involving 20 leading institutions, created to build a comprehensive map of Indian genomic diversity. Its primary objective is to create a catalogue of genetic variations that reflects the unique ancestral and ethnic diversity of India.

Key achievements and goals of Genome India include:

- Nationwide Genome Sequencing: Collected over 20,000 DNA samples from 83 diverse populations across India, ranging from metropolitan areas to remote indigenous groups. Whole-genome sequencing has been completed for 10,000 individuals, creating the first large-scale reference dataset of Indian genomes. This provides an unprecedented representation of India's genetic richness and population structure.
- Reference Genome and Biobank: The project established a robust biobank of Indian samples and is assembling a representative reference genome for the Indian population. All genome data (10,000 genomes) are archived at the Indian Biological Data Centre (IBDC) for open access by researchers, under national guidelines.
- Insights for Health and Diagnostics: By analyzing this trove of genomic data, scientists can identify genetic variants associated with disease susceptibility and drug response specific to Indians. Genome India's data is expected to facilitate affordable genetic tests and diagnostic tools tailored to Indian populations for example, developing India-specific SNP arrays for disease risk screening. Such a reference will improve understanding of how genetic differences contribute to disease in various ethnic groups, paving the way for precision medicine in India.
- Public Health Impact: The Genome India dataset serves as a critical national resource for public health research. It enables epidemiologists and policymakers to study genetic risk factors for common diseases (like diabetes, cardiovascular disease, cancers) in different regions and communities. Ultimately, this empowers data-driven policy decisions, ensuring that preventive health programs and therapies can be tailored to India's genetic diversity, rather than relying on one-size-fits-all approaches developed in Western populations.

Genome India marks a milestone in India's scientific landscape. It lays the foundation for genomics-based health interventions by cataloguing how one's genetic makeup may influence disease risk and treatment outcomes. By sequencing 10,000 genomes across India, this project has created the first holistic reference that will inform everything from new drug targets to personalized treatment guidelines for Indian patients.

While Genome India focuses on the genome, Phenome India (also known as the CSIR Phenome India-CSIR Health Cohort Knowledgebase, PI-CHeCK) targets the "phenome", the full set of observable characteristics and clinical parameters in individuals. Launched in late 2023 by the Council of Scientific & Industrial Research (CSIR), Phenome India is the first ever pan-India longitudinal health study aimed at understanding how lifestyle, environment, and biology together influence major diseases in Indians. Its emphasis is on cardio-metabolic diseases, conditions like diabetes, heart disease, and liver disorders that are rising in India.

Key aspects of Phenome India include:

- 10,000-Strong Health Cohort: In its first phase, the project enrolled nearly 10,000 adult participants across 17 states and 24 cities of India. Uniquely, this cohort includes people from diverse regions and backgrounds (initially CSIR employees, pensioners and spouses, as volunteers). Each participant contributes extensive health data, creating a rich longitudinal dataset for analysis.

- Comprehensive Health Data Collection: Phenome India collects a wide array of phenotypic data for each participant. This includes detailed clinical questionnaires, lifestyle and dietary profiles, physical measurements (anthropometry), imaging and scanning results, and a battery of biochemical and molecular tests on blood samples. By tracking individuals over time, the study captures how risk factors and health indicators change and potentially lead to disease onset.

- Aim: India-Specific Risk Prediction Models: A core objective is to develop better algorithms to predict and prevent non-communicable diseases (especially diabetes and cardiovascular illnesses) tailored to the Indian population. Currently, many health risk calculators and guidelines are based on Western data (primarily Caucasian populations). Indian populations have different genetic backgrounds, diets, and lifestyle patterns, meaning Western risk factors or thresholds may not apply directly. Phenome India addresses this gap by identifying India-specific risk factors and biomarkers. For example, what triggers diabetes in an Indian context might differ from the West – the study is poised to find such nuances, ensuring that "one-size-fits-all" models are replaced with more accurate, ethnicity-specific risk stratification.

- Precision Medicine and 4P Healthcare: Phenome India exemplifies the Predictive, Preventive, Personalized, and Participatory (4P) medicine model in action. By generating a comprehensive phenome database of Indian individuals, it allows researchers and clinicians to identify early warning signs (predictive), engage participants in their own health monitoring (participatory), devise targeted prevention strategies (preventive), and ultimately tailor interventions to individual risk profiles (personalized). The project's success in collecting nationwide data also encourages similar large-scale studies, setting the stage for a new era of precision public health in India.

Phenome India is creating an invaluable health dataset for India, linking lifestyle and clinical information with molecular data. Its longitudinal design (following participants over time) will shed light on how diseases develop and progress among Indians, and why certain populations are more vulnerable. Insights from this cohort will directly feed into public health planning; for instance, informing dietary guidelines, screening recommendations, or community interventions that are tailored to India's diverse population and its specific needs.

- The Bharat Cancer Genome Atlas (BCGA) is another landmark project, focused on mapping the genomic landscape of cancers in the Indian population. Initiated in 2020 and spearheaded by IIT Madras in collaboration with national research and clinical partners, BCGA was conceived to address a critical gap: India's cancer patients have been under-represented in global cancer genomics studies, meaning many genetic mutations common in Indian cancers are not recorded in international databases. BCGA aims to build a comprehensive cancer genome database for Indian patients, which will inform better diagnosis and treatment. Key highlights of BCGA include:

- **India's First Cancer Genome Atlas:** BCGA is the country's first comprehensive cancer genome mapping effort, analogous to the international Cancer Genome Atlas but focusing on Indian demographics. Its pilot focus has been on breast cancer, one of the most prevalent cancers in India. As of early 2025, the program has completed genomic sequencing of 480 breast tumor samples (performing 960 whole exome sequences, tumor and normal) collected from patients across India. This dataset represents the largest collection of Indian breast cancer genomes to date.

- **Publicly Accessible Database:** The anonymized genetic data from these tumors have been compiled into the Bharat Cancer Genome Atlas database, which was launched in February 2025 and is openly accessible to clinicians and researchers at bcga.iitm.ac.in. This atlas provides a compendium of genetic variants observed in Indian cancer patients, filling a crucial knowledge gap. It allows scientists to catalog which mutations are frequent in Indian breast cancers and how they might differ from Western patients. Ultimately, it helps classify genetic variants in terms of their relevance to early diagnostics, disease progression, and treatment outcomes in Indian contexts.

- **Insights for Early Detection and Treatment:** By analyzing the BCGA data, researchers have begun identifying cancer-specific biomarkers in Indians. These biomarkers (e.g., particular gene mutations or signatures) could enable earlier detection of cancers – for instance, blood tests or screenings that catch breast cancer at an earlier, more treatable stage, based on genetic markers prevalent in Indian women. Moreover, the data reveal potential novel drug targets, guiding the development of therapies that specifically target mutations common in Indian patient. This is especially important as some genetic drivers of cancer might differ in frequency from those seen in Western populations, necessitating different therapeutic strategies.

- **Toward Personalized Cancer Care:** BCGA is a steppingstone toward personalized medicine in oncology for India, Understanding the genetic basis of tumor development and drug response in Indian patients means oncologists can eventually tailor treatment plans to the genetic profile of each patient's cancer. For example, if a subset of Indian patients has a unique mutation that responds well to a specific targeted drug, this can

be incorporated into treatment guidelines. The BCGA initiative is also expanding to other cancer types, it invites researchers nationwide to contribute genomic data across various cancers. By federating data from multiple institutions, BCGA will grow into a pan-Indian cancer genomics resource to identify high-risk genetic profiles, monitor how cancers progress in different populations, and optimize treatment strategies for Indian patients.

- Through BCGA, India is addressing health disparities in oncology, ensuring that the genetic understanding of cancer is not biased toward Western countries alone. As one oncologist noted, genetic heterogeneity across ethnicities means "we can't absolutely rely on Western data" for treating Indian patients. BCGA's Indian-specific cancer genome data thus equips doctors with more relevant knowledge to improve cancer outcomes, from earlier diagnosis to choosing the most effective, personalized therapies.

- Together, Genome India, Phenome India, and BCGA represent a comprehensive approach to integrating genomic and phenotypic information for improving health outcomes. By mapping both the genetic blueprint and the observable health traits of Indians at scale, these initiatives are enabling a shift toward data-driven, predictive, and personalized healthcare. Below we discuss the broader implications and how these efforts support India's healthcare innovation and policy goals:

- **Enabling Precision Medicine:** The integrated genomic-phenomics approach directly fuels precision medicine in India – delivering the right intervention to the right patient at the right time. Genomic data from Genome India provides insights into genetic predispositions (e.g., variants that affect drug metabolism or disease risk), while Phenome India's longitudinal data add context about lifestyle and environmental exposures. The result is a more complete picture of individual risk profiles. For example, a person might carry a genetic variant that mildly raises diabetes risk but combined with certain dietary habits uncovered in the phenomics data, their risk could be substantially higher. By accounting for both, doctors can predict and prevent disease more effectively, advising personalized lifestyle changes or surveillance for those at high risk, long before illness develops. This embodies the preventive and predictive aspects of the 4P healthcare model.

- **Early Disease Detection and Public Health Planning:** These initiatives improve our ability to catch diseases early and guide public health strategies. The BCGA's identification of genetic biomarkers for cancer means screening programs can be more targeted, for instance, if certain mutations associated with early breast cancer are common in a region or community, screening tests in that area can incorporate those markers, leading to earlier detection and treatment. Similarly, phenotypic risk models from Phenome India can flag warning signs of cardio-metabolic disease in specific demographics (say, unanticipated risk in younger urban populations), prompting public health officials to implement targeted interventions or awareness campaigns. In essence, the data from these projects allow India's health planners to move from reactive care to proactive care, using risk maps and health forecasts to allocate resources and design programs tailored to the needs of each region and community. This is crucial for a country as diverse as India, where health disparities exist between different ethnic groups, rural and urban populations, and socioeconomic strata.

- **Tailored Treatment and Drug Development:** As India builds its own genomic databases, treatment guidelines can be customized for its population. For example, the Genome India data can inform pharmacogenomics, understanding how genetic differences affect drug response. If certain populations in India have variants that make standard doses of a drug less effective or more toxic, clinicians can adjust prescriptions accordingly, improving safety and efficacy of treatments. In cancer care, insights from BCGA mean oncologists can choose therapies that target mutations found in an Indian patient's tumor genome, rather than relying solely on protocols developed from Western patient data. Additionally, these initiatives spur local drug and diagnostic development: pharmaceutical researchers can use the identified genetic targets (from BCGA or Genome India) to develop new medications or repurpose existing ones for Indian-specific cancer subtypes or genetic disorders. Diagnostic companies can create affordable genetic tests (for example, a genotyping chip with variants common in India. to aid in routine clinical decision-making. Such innovations not only personalize treatment but also make healthcare more inclusive of India's genetic diversity.

- **Informing Healthcare Policy and Reducing Disparities:** The knowledge generated by these projects supports evidence-based policymaking. Health authorities can utilize the findings to update screening guidelines (e.g., start colon cancer screening at an earlier age if genomic data shows higher early-onset risk in Indians) or to introduce population-specific recommendations (such as different BMI cut-offs for obesity in South Asian populations, if phenomics data suggests different body composition). Importantly, by capturing data from under-represented and tribal communities (a priority stated by Genome India's future these initiatives ensure that health disparities can be identified and addressed. If certain genetic diseases or risk factors are found to be more prevalent in marginalized groups, resources can be directed to those communities (for example, targeted genetic screening or specialized clinics). Over time, this helps bridge the gap in health outcomes between different groups, aligning with India's policy goals of equitable healthcare access.

- **Research and Innovation Ecosystem:** These large-scale projects are creating a rich resource for the scientific community, sparking innovation in biomedical research. The open data sharing (Genome India's data via IBDC, and BCGA's public portal) means researchers anywhere can mine this information to uncover new disease associations or develop AI algorithms for risk prediction. Indian researchers and startups can build on this data to create home-grown healthcare solutions, supporting the country's vision of self-reliance (Atmanirbhar Bharat) in science and technology. Moreover, training the next generation of scientists and clinicians in genomics and data science is an explicit goal, Genome India, for instance, hopes to "inspire the next generation of genomic innovators" in India. This capacity building will ensure that India not only gathers data but also has the expertise to translate it into clinical practice and health policy.

Digital Health and ICT Innovations in India

In parallel with its omics initiatives, India is also pursuing a range of digital health and information communication technology (ICT) innovations to strengthen healthcare delivery. These programs provide a digital backbone that complements personalized and precision medicine by enhancing data availability, connectivity, and intelligent analysis in healthcare. Key government-led digital health initiatives include:

- **Ayushman Bharat Digital Mission (ABDM):** Launched in 2021, ABDM aims to create a unified national digital health infrastructure. It provides every citizen with a unique digital Health ID (now called ABHA – Ayushman Bharat Health Account) to link their electronic health records across providers. This interoperability enables longitudinal patient records and easier sharing of medical data, which is foundational for personalized care. For example, as of 2024 ABDM has generated over 670 million (67 crore) health IDs and linked more than 420 million health records, illustrating its massive scale. By making an individual's complete health history accessible to clinicians (with consent), ABDM supports more tailored treatment decisions and continuity of care across different facilities.

- **eSanjeevani Telemedicine Service:** India's national telemedicine platform eSanjeevani has dramatically expanded access to healthcare, especially for remote and underserved populations. Since its launch in 2019, eSanjeevani has facilitated over 340 million doctor-to-patient consultations as of early 2025, offering free online medical advice across the "length and breadth" of the country. The service operates in both hub-and-spoke clinic-to-clinic mode and a direct-to-public OPD mode, which proved crucial during the COVID-19 pandemic for continuity of care. By enabling patients to consult doctors via video from their homes, eSanjeevani reduces the need for travel and overcrowding at hospitals. This tele-healthcare model makes healthcare more accessible and patient-centric, allowing personalized medical consultations (including follow-ups and specialist advice) regardless of geographic location.

- **Aarogya Setu:** This mobile application, launched in April 2020 for COVID-19 contact tracing and health status monitoring, became one of the fastest-adopted digital health tools in the world. Aarogya Setu reached 100 million users within 41 days of its release, reflecting its rapid nationwide uptake. The app uses Bluetooth and GPS data to alert individuals if they may have been exposed to someone with COVID-19, effectively providing each user a personalized risk assessment. It also offers a self-assessment quiz and up-to-date health advisories, empowering citizens to take personalized precautions and seek testing or medical help early. Aarogya Setu demonstrated how smartphone technology can be leveraged for precision public health, delivering timely, individualized alerts and guidance to millions of people during a pandemic.

- **CoWIN Platform:** The COVID-19 Vaccine Intelligence Network (CoWIN) is India's digital platform for managing its unprecedented vaccination drive. CoWIN served as the digital backbone for administering COVID-19 vaccines to India's 1.3 billion people. Through a website and app, it enabled citizens to register, schedule appointments, receive reminders for second doses, and download digital vaccination certificates. CoWIN's real-time dashboards allowed for efficient logistics and minimized gaps in coverage. By March 2023, India had delivered over 2.2 billion vaccine doses via this platform, covering 95% of the eligible population with at least one dose. The individualized tracking ensured that each person got the right vaccine at the right time, exemplifying precision at population scale. CoWIN's success has shown how a robust ICT system can orchestrate a personalized healthcare intervention (vaccination with proper follow-ups) for hundreds of millions of people.

- **National AI Mission (#AIforAll):** Recognizing the transformative potential of artificial intelligence, the Government of India (through NITI Aayog) launched a National Strategy for AI in 2018, branding it "AI for All." Healthcare is a priority sector in this strategy, which envisions deploying AI to improve both access and quality of care. Under this mission, India is investing in AI-based healthcare solutions — for example, **AI-driven diagnostics, predictive analytics, and decision support tools** that can assist doctors. Pilot projects have applied AI for tasks like screening medical images (e.g., detecting diabetic retinopathy or tuberculosis from scans) and analyzing large health datasets to identify risk patterns. Such tools augment clinicians' capabilities, enabling more accurate and personalized treatment by rapidly processing patient-specific data (like scans, health records, or genomes). The National AI Mission thus supports precision medicine through technology, aiming to democratize advanced care (e.g., early detection of diseases and tailored therapies) across India's health system.

- **Integrated Health Information Platform (IHIP):** Launched nationwide in 2021 by the Ministry of Health's NCDC, IHIP is a next-generation digital disease surveillance system. It integrates data from hospitals, laboratories, and community health centers in near real-time under the Integrated Disease Surveillance Programme. By aggregating and analyzing this data (including trends on 30+ diseases) on a single platform, IHIP enables early detection of disease outbreaks and health trends across India. Health officials receive **timely, geo-tagged alerts** of anomalies, allowing swift, targeted public health responses. This facilitates **precision public health** – interventions (such as containment, vaccinations, or resource deployment) can be directed to specific locations or populations as soon as a threat is identified. As an example of the "One Health" approach, IHIP even plans to incorporate animal health and environmental data to predict zoonotic disease risks. Overall, the IHIP's data-driven surveillance enhances India's ability to monitor health threats and respond with pinpoint accuracy, protecting communities through informed decision-making.

India is advancing toward precision healthcare through a powerful integration of omics research and digital health technologies. Genome India, Phenome India, and the Bharat Cancer Genome Atlas (BCGA) are landmark initiatives capturing the country's genetic, phenotypic, and cancer-specific diversity. These efforts enable population-specific disease risk prediction, early diagnosis, and the development of personalized therapies tailored to India's unique genomic landscape.Simultaneously, digital platforms like the Ayushman Bharat Digital Mission, eSanjeevani telemedicine, Aarogya Setu, and CoWIN are revolutionizing healthcare access and delivery. With the support of AI-driven tools and real-time disease surveillance through platforms like IHIP, India is equipping its health system for data-driven, inclusive care.

Together, these initiatives represent a strategic shift toward predictive, preventive, and personalized medicine—ensuring healthcare is both evidence-based and equitable, and setting a strong foundation for India's long-term health innovation and resilience

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