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GENOTYPING IN CLINICAL PRACTICE: COMMON PURPOSES AND EMERGING APPLICATIONS

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Abstract: Genotyping has rapidly evolved from early PCR-based assays and Sanger sequencing to advanced next-generation sequencing (NGS) and AI-assisted variant interpretation, becoming an indispensable tool in modern clinical practice and biomedical research. Its applications extend across pharmacogenomics, carrier screening, disease risk prediction, oncology, infectious disease monitoring, forensic analysis, and population genetics. Clinical integration of genotyping has enabled personalized medicine, improving drug safety and efficacy, predicting disease susceptibility, and guiding targeted therapies. Globally, genotyping is well established, with widespread adoption of pharmacogenomic panels, expanded carrier screening, and tumor profiling. In India, however, clinical genomics remains limited by high costs, lack of awareness, infrastructural constraints, and insufficient clinician training. Ethical, legal, and privacy concerns further challenge large-scale implementation. Emerging innovations such as rapid point-of-care genotyping, AI-based variant interpretation, liquid biopsy technologies, and integration with electronic health records (EHRs) promise to overcome these barriers and expand accessibility. With decreasing costs, regulatory support, and national initiatives like GenomeIndia, genotyping is poised to play a transformative role in advancing precision medicine and public health in India and worldwide

Keywords: Genotyping, Pharmacogenomics, Oncology genomics,; Next-generation sequencing (NGS)

INTRODUCTION

The process of analyzing a person's DNA sequence to find genetic variants that could affect health and disease outcomes, such as single nucleotide polymorphisms (SNPs), insertions, deletions, and structural alterations, is known as genotyping [1]. In order to identify particular mutations in targeted genes, genotyping in medicine initially depended on low-throughput, comparatively easy methods such restriction fragment length polymorphism analysis and PCR (Polymerase Chain Reaction) [2].By allowing base-level analysis of DNA sequences, Sanger sequencing transformed the field in the 1970s and established itself as the gold standard for clinical diagnostics for many years [3]. Due to Sanger sequencing's limited scalability, high-throughput systems such DNA microarrays and, later, Next-Generation Sequencing (NGS) were developed [4]. Massively parallel sequencing of entire genomes or specific panels has been made possible by NGS, which has significantly reduced time and expense while opening up a wide range of therapeutic applications [5]. A larger transition from descriptive genetics to precision medicine, where genetic information is incorporated into therapeutic decision-making, is reflected in the historical development of genotyping [6]. This integration has enabled the identification of actionable therapeutic targets, the prediction of illness risk, and the customization of medicines to a person's genetic profile [7]. These days, genotyping is essential in many therapeutic settings, from infectious illnesses and carrier screening to pharmacogenomics and oncology [8]. Crucially, genotyping's use goes beyond straightforward clinical applications; it also forms the basis for extensive population research, ancestry analysis, and genome-wide association studies (GWAS) [9]. As the discipline develops, it is anticipated that the increasing use of liquid biopsy techniques, AI-based variant interpretation, and integration with electronic health records (EHRs) will increase the value of genotyping and make it a key component of the next era of precision healthcare [10].

PURPOSES OF GENOTYPING IN CLINICAL PRACTICE

2.1 Pharmacogenomics

To find genetic variations influencing drug metabolism, pharmacogenomic testing use next-generation sequencing panels, microarrays, or PCR-based techniques [11] .For instance, doctors might transfer poor metabolizers to ticagrelor or prasugrel by using CYP2C19 testing on blood or saliva samples to predict responsiveness to clopidogrel [12]. In order to prevent decreased efficacy or toxicity in psychiatric medications, CYP2D6 genotyping is utilized to advise codeine and tramadol dosage [13]. Furthermore, in order to prevent serious hypersensitivity responses, HLA-B1502 and HLA-B5701 tests—which are normally performed using PCR-based HLA typing—are now required before prescribing abacavir or carbamazepine [14].

2.2 Carrier Screening

Couples are increasingly being offered carrier screening panels, which are carried out by microarray analysis or sequencing, either before or during pregnancy [15].For instance, carriers of sickle cell disease or thalassemia can be identified using hemoglobin electrophoresis and HBB mutation testing, allowing for genetic counseling [16] .In many Western countries, carrier screening for cystic fibrosis routinely involves CFTR mutation panels, which include the prevalent ΔF508 deletion [17].Similarly, carriers of spinal muscular atrophy are identified by SMN1 copy number testing, which is frequently carried out by quantitative PCR or MLPA (Multiplex Ligation-dependent Probe Amplification) [18].

2.3 Disease Risk Prediction

Disease-predictive genetic testing is widely used for preventive care and involves targeted sequencing of known high-risk genes [19]. For example, *BRCA1/2* testing using NGS panels identifies women at risk of breast and ovarian cancer, allowing them to consider prophylactic surgery or enhanced surveillance [20]. *APC* gene sequencing is performed for suspected cases of familial adenomatous polyposis, often initiated after colonoscopy reveals multiple polyps [21]. For neurological risk, *APOE* genotyping is available as a blood test to assess susceptibility to late-onset Alzheimer's disease, although its clinical use is still debated [22]

2.4 Oncology Applications

Tumor genotyping in oncology is commonly accomplished on biopsy samples by FISH (fluorescence in situ hybridization), PCR, or targeted NGS panels [23]. For instance, EGFR inhibitor therapy is typically guided by EGFR mutation testing by PCR in non-small cell lung cancer [24]. To ascertain eligibility for cetuximab or panitumumab, KRAS mutation testing in colorectal cancer is frequently carried out using PCR or sequencing [25]. Similarly, to choose BRAF-targeted treatments, melanoma patients are examined for BRAF V600E mutations using PCR or immunohistochemistry [26].

2.5 Infectious Disease Genotyping

Genotyping techniques in infectious diseases use PCR-based resistance panels or sequencing to target pathogen DNA or RNA [27]. Whole-genome sequencing and line probe tests are used to quickly identify rifampicin and isoniazid resistance variants in tuberculosis [28]. Before beginning or altering antiretroviral treatment for HIV, drug resistance genotyping is frequently carried out, frequently with the use of reverse transcriptase and protease sequencing [29]. Whole-genome sequencing of SARS-CoV-2 variants proved essential for worldwide surveillance and directing vaccine development during the COVID-19 pandemic [30].

2.6 Forensic and Identity Testing

The main technique used in forensic DNA testing is STR profiling, which looks at highly polymorphic areas of the genome [31]. The majority of criminal labs around the world match biological samples to suspects using STR testing based on capillary electrophoresis [32]. Paternity and kinship testing frequently uses both STR and SNP-based assays, while criminal justice applications use commercial genotyping kits like CODIS (Combined DNA Index System) panels [33].

2.7 Population Genetics & Research Applications

Genome-wide association studies (GWAS) in research employ high-throughput genotyping techniques such SNP arrays and next-generation sequencing [34]. To find risk loci for complicated illnesses like diabetes and schizophrenia, GWAS examines thousands to millions of SNPs in sizable populations [35]. SNP genotyping panels are also used by direct-to-consumer ancestry services like 23andMe and AncestryDNA to offer data on migration history, ethnic background, and population relatedness [36].

COMMONLY USED GENOTYPING TESTS AND METHODS

In clinical practice, public health, and biological research, genotyping has become a crucial tool. There are numerous test panels and technologies available, each with a specific function, such as tracking infectious disease variations, identifying carriers of inherited illnesses, profiling malignancies, or directing medication therapy. One of the most used genotyping tools in clinical practice is pharmacogenomic panels. They identify variations in genes that affect how people react to drugs. CYP2C19 testing, for instance, aids in identifying patients who are unable to activate clopidogrel enough [37]. In Asian populations, HLA-B*1502 testing is essential to preventing Stevens-Johnson syndrome brought on by carbamazepine [38]. In patients receiving thiopurine treatment, TPMT/NUDT15 genotyping can prevent potentially fatal bone marrow suppression [39].International norms support these panels, which serve as the foundation for precision prescribing. Another crucial use is carrier screening, which looks for carriers of X-linked or autosomal recessive diseases. Genes linked to sickle cell disease and thalassemia, such as HBB [40], CFTR [cystic fibrosis], and SMN1 [spinal muscular atrophy,are frequently tested. In order to offer genetic counseling and reproductive alternatives, such screening is frequently advised during preconception or the early stages of pregnancy.

The purpose of oncology panels is to identify tumor mutations that can be treated. To direct targeted therapy, genes including EGFR, KRAS, BRCA1/2, and BRAF are frequently examined. Response to EGFR tyrosine kinase inhibitors is predicted by EGFR

mutations in non-small cell lung cancer [41]. The choice of targeted medications in development is influenced by KRAS mutations [42]. These panels are becoming more and more integrated into routine therapy and are essential to individualized oncology. Infectious illness genotyping facilitates outbreak surveillance and makes it possible to identify treatment resistance variants. BRCA1/2 mutations predict benefit from PARP inhibitors in breast and ovarian cancers [43]. Rapid molecular assays like Xpert MTB/RIF, which can simultaneously identify Mycobacterium tuberculosis and rifampicin resistance in a matter of hours, are recommended by the WHO for tuberculosis [44]. In order to monitor national resistance patterns and guide antiretroviral therapy, HIV medication resistance genotyping is recommended [45]. For identifying variants and tracking the efficiency of vaccines, SARS-CoV-2 sequencing has become essential[46].

One well-known use of genotyping outside of healthcare is forensic STR analysis. Criminal investigations, paternity testing, and individual identification all make use of short tandem repeat markers. A standardized panel of STRs is offered by the FBI-developed CODIS system, which is extensively used globally[47].High-density SNP microarrays or sequencing techniques are used by population genetics panels to investigate illness correlations and human diversity. Thousands of genetic loci have been connected to complex disorders by genome-wide association studies (GWAS) [48]. Additionally, commercial ancestry testing kits offer data on population migration trends and ethnic heritage. The technical basis for all of these applications is provided by generic genotyping platforms. In clinical and infectious disease testing, PCR and real-time PCR are frequently utilized for DNA amplification [49].Because of its excellent accuracy, Sanger sequencing is still the gold standard for confirmatory and small-scale sequencing. While next-generation sequencing (NGS) technologies have transformed the field by providing whole-exome and whole-genome sequencing, tumor profiling, and comprehensive pathogen genomics [50], microarrays provide high-throughput SNP genotyping throughout the genome [51].

GLOBAL SCENARIO

Globally, pharmacogenomic testing and next-generation sequencing (NGS) are now extensively incorporated into clinical practice for personalized medicine, rare illness diagnosis, and oncology [52]. In order to facilitate evidence-based prescribing in the fields of cardiology, psychiatry, and oncology, the U.S. FDA already lists more than 300 medications with pharmacogenomic biomarker information on their labels [53] .With businesses like 23andMe and AncestryDNA providing ancestry, health risk, and carrier status testing, the direct-to-consumer (DTC) genetic testing sector has grown dramatically in the US and Europe [54]. But issues with ethics, data privacy, and governmental supervision are still unresolved [55] .Large multi-gene panels are advised by guidelines to support reproductive decision-making, and expanded carrier screening has become the norm in many affluent nations [56]. Standard clinical guidelines in cancer now include genomic profiling for actionable mutations including BRCA1/2, EGFR, ALK, and KRAS. Additionally, top cancer centers throughout the world have created precision oncology and NGS-based tumor profiling programs [57].

INDIAN SCENARIO

High-prevalence illnesses are the main focus of genomic applications in India. The most prevalent carrier screenings are for thalassemia and spinal muscular atrophy (SMA), and Gujarat, Maharashtra, and West Bengal have state-level premarital and perinatal programs in place [58]. Pharmacogenomics is becoming more and more popular, particularly in cardiology, where CYP2C19 genotyping directs the use of clopidogrel medication in patients suffering from coronary artery disease [59] .Furthermore, among Indian patients administered carbamazepine, HLA-B15:02* testing is becoming more clinically significant in order to prevent serious drug responses such as toxic epidermal necrolysis and Stevens-Johnson syndrome [60]. A growing number of tertiary care cancer hospitals are implementing BRCA, EGFR, KRAS, and HER2 testing for precision oncology. Large private hospitals increasingly provide advanced technologies like liquid biopsy and NGS panels, although their availability is constrained by cost [61].In spite of these developments, India still faces a number of obstacles. Since genetic testing cost between ₹15,000 and ₹50,000 per panel, most people cannot afford them. There is currently a lack of knowledge regarding genetics in healthcare among both doctors and patients. Additionally, rural and semi-urban communities are underserved due to the concentration of infrastructure and expertise in metropolitan cities [62].

CHALLENGES AND LIMITATIONS

High cost of sophisticated genotyping: Certain genomic tests, such whole-genome sequencing (WGS), whole-exome sequencing (WES), and multi-gene NGS panels, might be unaffordable. Tests can cost anywhere from $1,000 to $5,000 in high-income nations, but in India, they usually cost between ₹15,000 and ₹50,000, with limited insurance coverage. This restricts application mostly to tertiary care hospital patients or urban privileged communities [63]. Limited access in low-resource and rural areas: In India, access to genetic testing varies. Most sophisticated diagnostic facilities are located in urban areas, but qualified staff, logistics, and cold-chain sample transportation are lacking in rural locations. In precision medicine, this leads to significant healthcare disparities [64]. Problems with interpretation (Variants of Uncertain Significance, or VUS) Variants of Uncertain Significance (VUS), which are challenging to categorize as benign or pathogenic, are commonly discovered by sequencing. Due to the absence of population-specific genetic datasets, this is particularly difficult in India, where many variations are still uncharacterized and clinically inactive [65].

Legal and ethical concerns: discrimination, consent, and privacy. There are serious ethical concerns with genetic testing. Data privacy violations, the absence of uniform frameworks for informed consent, and the possible abuse of genetic information in insurance and employment are among the issues. Unlike the U.S. (which has the GINA Act), India currently lacks specialized laws protecting citizens against genetic discrimination [66]. Lack of training and education for clinicians: Healthcare workers still have low levels of genomic literacy. Clinical integration is hampered by the fact that many doctors lack knowledge of drug-gene interaction guidelines, actionable variations, and pharmacogenomic test interpretation. This disparity highlights the pressing need for ongoing education initiatives and curriculum change in the medical field [67]. Infrastructure and regulatory gaps: India lacks a national regulatory framework and accreditation system to guarantee the consistency and quality of genetic testing. India's labs frequently use inconsistent quality assurance procedures, which leads in disparities in test results and erodes physician confidence, in contrast to the US (CLIA) or Europe (ISO standards) [68].

FUTURE PERSPECTIVES

Creation of fast genotyping point-of-care kits For bedside genotyping, CRISPR-based and lab-on-chip diagnostic technologies are being developed that can provide results in less than an hour. By increasing access to primary care and rural clinics, these devices have the potential to democratize pharmacogenomics, particularly in nations with limited resources like India where centralized lab infrastructure is hard to come by [69]. Variant interpretation powered by AI In order to prioritize drug-gene interactions, forecast the pathogenicity of uncommon variations, and harmonize multi-omics information, machine learning and deep neural networks are being utilized more and more. It is anticipated that these methods would shorten turnaround times and lessen the need for extensive expert panels, particularly in situations with a shortage of qualified genomicists [70]. Additionally, recent research emphasizes AI's contribution to the development of genomic databases tailored to India, which could increase the accuracy of interpretation for marginalized groups [71].

Electronic Health Record (EHR) Integration Safe prescribing increasingly depends on the incorporation of pharmacogenomic data into EHRs with real-time decision assistance. By integrating actionable drug-gene alerts into clinical processes, initiatives such as the U-PGx project in Europe and the IGNITE Network in the United States are setting the standard. With pilots under the National Digital Health Mission (NDHM) seeking to integrate genomic profiles in longitudinal patient data, India is just getting started [72].Liquid biopsy (circulating tumor DNA genotyping): Non-invasive monitoring of tumor mutations and treatment response is made possible by liquid biopsy methods that use circulating tumor DNA (ctDNA), exosomes, and cell-free RNA. In colon, breast, and lung cancers, where resistance mutations occur during therapy, they are very helpful. Although tertiary hospitals in India are still conducting trial programs, adoption is still restricted due to cost [73].

Greater use of prescribing that is guided by pharmacogenomics Genomic testing is anticipated to be incorporated into standard prescriptions in cardiology, psychiatry, and oncology as costs decline and regulatory pressure increases. More than 250 medications currently have pharmacogenomic information on their labels, according to the FDA, and India's ICMR clinical guidelines are probably going to follow suit soon [74]. expansion of national programs and public-private partnerships Large-scale population genomics databases are being constructed by nations such as the United States (All of Us Research Program) and the United Kingdom (100,000 Genomes Project). Similar initiatives have been started in India, such as GenomeIndia, which intends to gather genomic information from 10,000 people in order to enhance population-specific pharmacogenomic insights and aid in drug development [75].

CONCLUSION

Genotyping has become a cornerstone of precision medicine, enabling tailored therapies, risk prediction, and improved patient outcomes across diverse clinical domains such as pharmacogenomics, oncology, carrier screening, and infectious disease management. While high-income countries have already integrated next-generation sequencing and pharmacogenomic testing into routine healthcare, India’s adoption remains uneven due to high costs, infrastructural limitations, and lack of awareness among both clinicians and patients. Addressing these barriers requires investment in local genomic infrastructure, clinician education, regulatory frameworks, and equitable access strategies. Looking ahead, innovations such as rapid point-of-care genotyping, liquid biopsy technologies, AI-driven variant interpretation, and integration of genomic data with electronic health records will accelerate the mainstreaming of genomic medicine. With decreasing costs, public–private partnerships, and national initiatives like GenomeIndia, genotyping is poised to reshape healthcare delivery, bridging global disparities and ushering in a new era of accessible, equitable, and personalized medicine.

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