

# PERSONALISED MEDICINE AS THE PROMISING IMPLICATION FOR HEALTHCARE

## Abstract

Recently the field of medicine and healthcare is experiencing a boom in a concept called personalized medicine. Personalized medicine involves customizing a treatment to fit the needs of an individual and disease patterns. It helps to gain a deeper understanding of how a person's unique genomic profile or proteomic profile makes them susceptible to specific diseases by identifying genetic, epigenomic, and clinical data. Personalized Medicine serves not only as potential treatment but also as a preventative measure. Our chapter explores the impetus for personalized medicine, its historical instances, developing technologies that enable it, and latest examples. Moreover, it discusses the roles of different omics technologies in emergence of personalized medicine; pharmacogenomics playing a key role by providing an explanation for the variability and uncertainty in drug responses.

**Keywords:** Medicine, Healthcare.

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## I. INTRODUCTION

Personalized medicine is a growing branch of medicine that implements genetic profiling to guide decisions regarding disease prevention, diagnosis, and treatment based on an individual's genetics. An individual's genetic profile can assist doctors in selecting the proper medication or therapy and administering it in the proper dosage and regimen. A large amount of data from the Human Genome Project is providing insights into personalized medicine. An individualized approach to diagnostics and drug therapy and prevention could explicitly replace a "one size fits all" approach.

We all are alike; however, we differ in terms of our genetic combination. Genomics is the key role player in the field of personalized medicine as it enables us to make specific estimates about disease risk that can assist someone in selecting the most appropriate anticipation plan for them based on their specific molecular differences [1]. It also permits the probability in some instances of selecting the precise drug at the accurate dose for the right- person as an alternative to the generalized approach to drug therapy.

Through personalized medicine, a medical treatment is tailored to each patient's individual characteristics. The hypothesis is perceived on the basis of how molecular and genetic profiles contribute to a person's susceptibility to certain diseases backed by several scientific studies [2]. As a result, we are gaining greater insight into which treatments will be safe and effective for each patient, as well as which ones will not. Personalized medicine can be considered a complement to conventional methods of disease diagnosis and treatment. With more precise tools, physicians can customize a treatment based on the molecular profile of a patient [3]. This approach not only helps in reducing detrimental side effects and ensuring a positive outcome, but also controls costs compared to a "trial-and-error" approach to disease management. Personalized medicine has the potential to change our perception, identify and manage health problems. Our understanding and technologies are improving, and this will have an even greater impact on clinical research and patient care.

Personalised medicine is based on the core research related to pharmacogenetics wherein it acts as a model of implementing individualized therapies for diverse patients or groups. Every individual differs based on their distinct heredity, health related behaviour and metabolism. Additionally, the elder population suffer from several illnesses. Until now, drugs are prescribed solely based on clinical overview of the disease irrespective of the individual make-up of distinct patients. Consequently, treatments with established efficacy frequently induce (adverse) side-effects or may be unable to induce any effect in a subgroup of patients. This approach not only have considerable impact on personal well-being, but also leads to economic costs which can be saved by certain targeted therapies. The advances of pharmacogenetic research since the late 1950s has provided deep insights into the future potentials of discretely designed drug therapies. The spectrum personalised medicine according to several researchers includes [4]:

1. Medicine, based on the utilization of distinctive therapeutic measures including the Regenerative Medicine - Tissue Engineering or Stem Cell Therapy
2. Pharmacogenetics
3. Other areas of research wherein biomarkers are used to improve prediction of disease and/or the course of disease.

Personalised medicine incorporates various strategies to tailoring healthcare. Till date various factors has been considered for diagnosis and treatment of various diseases such as age, sex and family history, co-morbidities and considering patient's psycho-social, lifestyle and perhaps family and economic conditions.

Over 60 years medical field has been struggling to become more 'scientific'. Medical practice has prominently focused on proof-based approach considering narrative accounts by patient, tactual exploration, demographic and societal aspects related to the patient, including others. The process operation has become more systematic and automated with respect to collection, processing and analysis of data which can be further easily aggregated and extracted [5].

The present concern in personalised medicine is related to customizing the science of medicine. As a result, it has the potential to go beyond producing evidence of the most effective interventions for the 'average' patient to produce scientific data and 'individual evidence' of the optimal treatment for the individual. Since everyone has distinct features of their disease, medicine is moving away from 'one size fits all' to 'personalised interventions'. It is already common practice to select antimicrobial drugs based on the sensitivity of the organism infecting the disease.

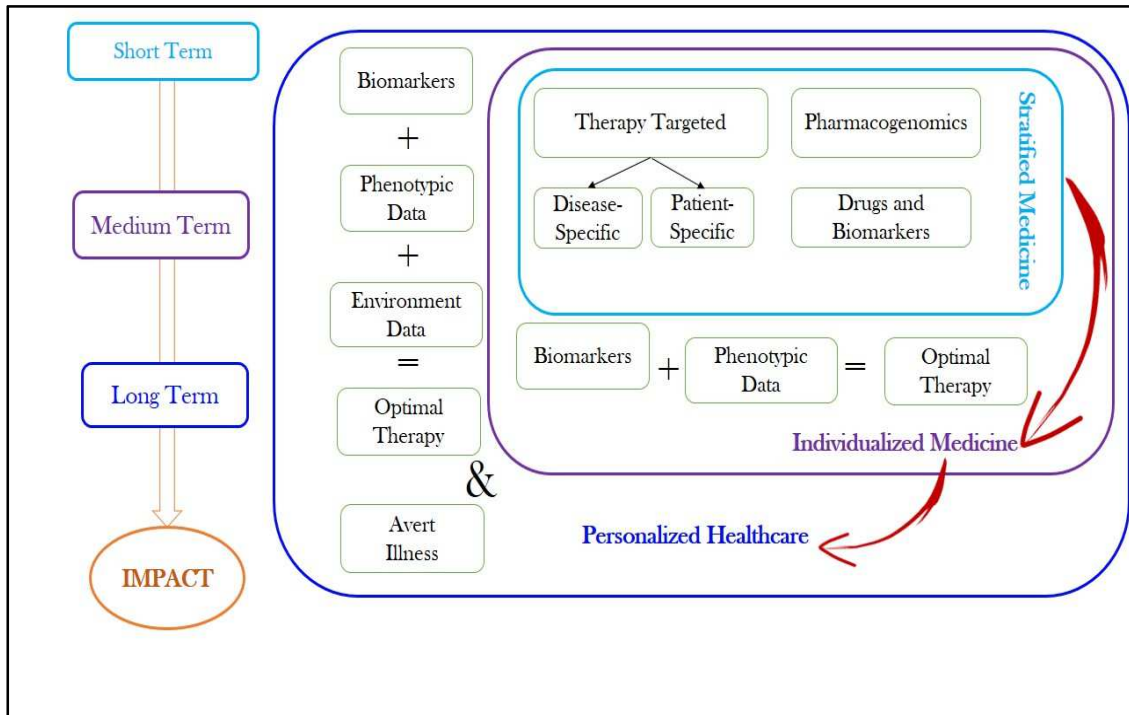
With the advancements in pharmacogenomics, the concept of personalised medicine is already becoming realistic for humans. For instance, research identifying genetic determinants of drug response is making it feasible to estimate which diverse therapies will benefit which individuals (Cetuximab and panitumumab fail to treat colorectal cancer patients with KRAS gene mutations), or who will respond adversely (hypersensitive reactions to the anti-AIDS drug Abacavir is linked to the existence of the HLA-B\*5701 allele) [6].

Pharmacogenomics customizes medication to the degree that it delineates patients having particular illness into subgroups. This definition brings about a fitting that is practically equivalent to having a decision of little, medium or enormous, as opposed to one-size-fits-all solution; it isn't the arrangement of a custom-tailored ensemble. Through extension of pharmacogenomics, and with research, by clarifying the sub-atomic premise of pathologies, reclassifying would be thought of by most to be a solitary sickness element into various infection subtypes (as has changed the therapy of leukaemias) or without a doubt various ailment, re-classifying normal complex illness into numerous intriguing infections [7].

Nevertheless, besides stratification, individualisation is also an important direction for tailoring advancement [8,9]. This means valuing the individual as an individual and as a totality. In silico representations of the virtually simulated patients can be built using omics-type analytic techniques, such as sequencing the entire genome and transcriptome of a cancer and comparing it to the patient's genome. This information can then be used to find a personalised treatment plan.

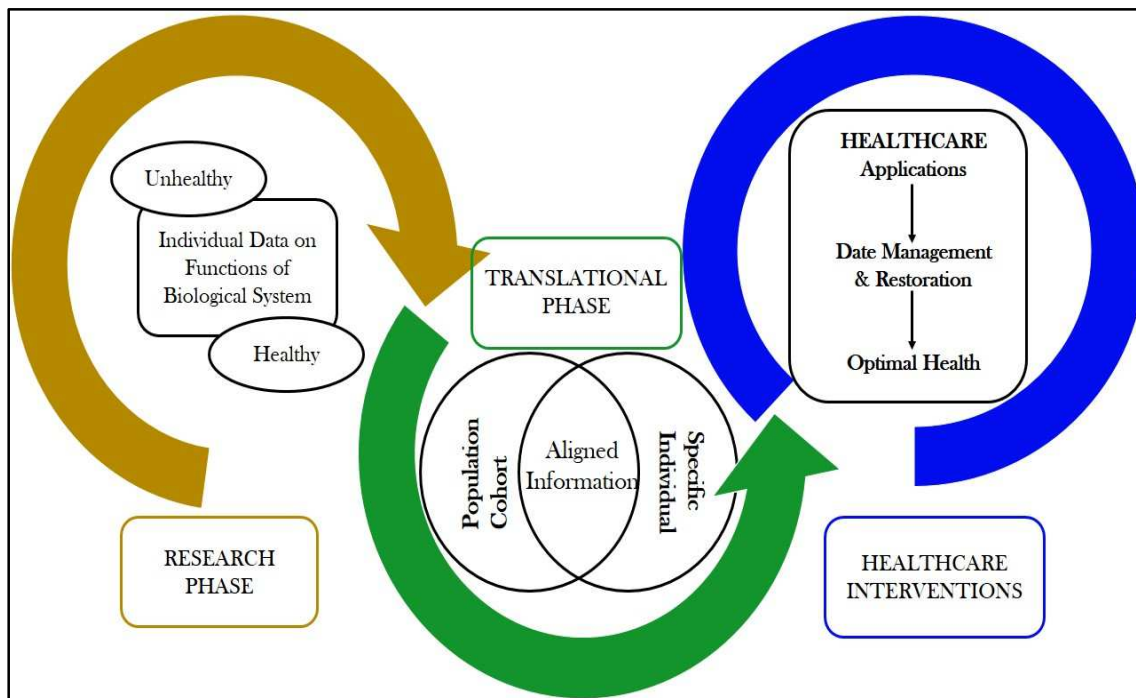
A comprehensive knowledge of the biological processes in the body as an intricate and dynamic entity is produced through systems biology methodologies. The associated healthcare practises would use a holistic perspective, evaluating how each person's unique biological make-up interacts with their surroundings and devising treatments to promote

wellbeing. Doctors could choose the best treatment for a patient by integrating data pertaining to that person's biological composition using techniques like gene pathway studies and in silico modelling. Additionally, it aims to refocus efforts towards preventative healthcare, allowing for personalised illness prevention strategy recommendations (Fig. 1).



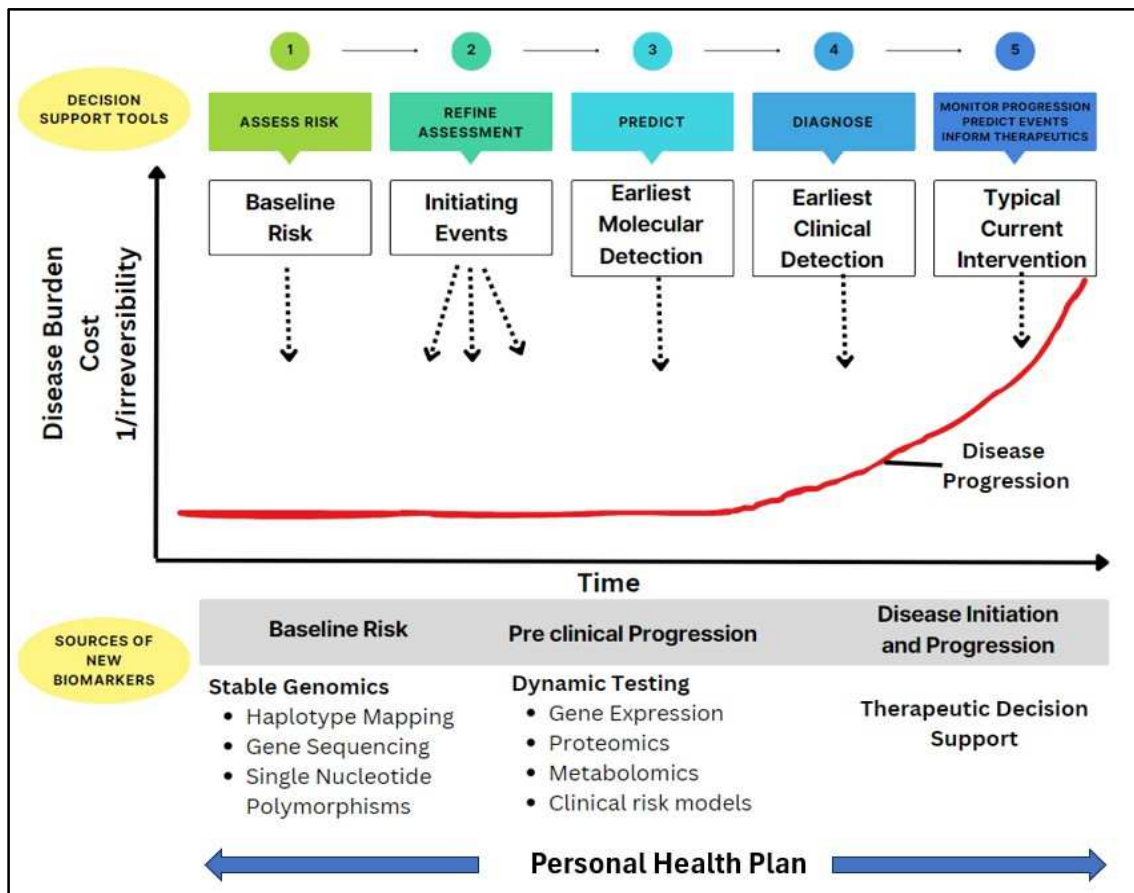
**Figure 1:** Impact of Personalized Healthcare-from Stratified to Individualized Health System

Personalised medicine, whether stratified or individualised, is predicated on the disposal of additional information about individuals. It promises to provide better, more precise therapy for individuals. There is a need for quantitative growth: additional in-depth information on human biology that is obtained from "-Omic" data including diverse fields of genomics, epigenomics, transcriptomics, proteomics, metabolomics, amongst others in amalgamation with other categories of data like biological imaging and physiological measurements. Likewise, there is a need for qualitative improvement; new data types, such as information about a person's surroundings that includes nutritional aspects, the microbiome, toxin exposure etc.; are now considered crucial for comprehending the physiological processes. Both research and healthcare practice require additional information, and there must be a process for evaluating how knowledge derived in one domain can be applied to the other (Fig. 2). In order to shift from stratified medicine to individualised medicine and individualised healthcare, various aspects of biological functions need to be studied and presented. (Fig. 3).



**Figure 2:** Translational Process Research to Applications

The precise and accurate quantification of data is another necessity of the scientific community so that inputs from different platforms can be merged effectively. It may be particularly challenging to analyse biological imaging data in this way. Analysing broader categories of data like lifestyle or environmental factors will likely pose further challenges. In an era of increasingly complex environments, economics, and family circumstances, it is extremely difficult to capture the clinical phenotype with all its complication [10]. By incorporating deep phenotyping, these 'phenomes' can interact with disease linked pathways predicted through '-Omics' technologies by using the thorough information about an individual's disease. There is a considerable informatics challenge here, not to mention the difficult task of capturing sufficient phenome detail. New techniques or technologies may be required to standardize and quantify linear data. In addition to the data generation on integrated biological systems, data collection on dynamic ensembles also needs to be considered; thus, new technologies will be required to achieve this. Processing large quantities of linear data will require good IT infrastructure, in terms of both storage and processing. Data processing will also be facilitated by new algorithms. Handling longitudinal information from highly dynamic ensembles over time and space will require new tools, as will the integration of multiple data types. To improve our knowledge of human functioning in health and disease, bioscience researchers must first gather big volumes of standardised data and convert into biological information. As a result of this improved understanding of biology, interventions can be optimized that facilitate health restoration and maintenance on an individual basis [10]. It is imperative that such interventions are proven before they can be used in healthcare.



**Figure 3:** The Role of Genome-based Information across Continuum of Health to Disease

Additionally, we must keep in mind that the concept of "personalised medicine" incorporates its use in various contexts, considering various areas of healthcare. A personalized approach can assist in selecting optimal treatments by predicting the outcome of interventions, as is already possible through pharmacogenetic testing [11]. Future applications might include predicting someone's risk of ill health and guiding their treatment selection.

It is particularly challenging to corroborate the efficacy for preventive action, as has even been acknowledged by those who claim their necessity. Moving personalised medicine from the lab into healthcare will require addressing important questions about suitable methodologies for assessing validity and utility. Although providing adequate answers to the questions necessary for introducing a novel tool into healthcare in principle, addressing the cost effectiveness question is essential in practice. Personalised medicine will be in a position to answer the questions with respect to cost effectiveness. As a matter of fact, one of the main benefits of personalised medicine is that it will decrease expenses by focusing on treatments for the individual, thereby avoiding spending money on interventions that may not be effective for the patient, or may lead to adverse effects that require additional therapeutic care.

The goal of personalised medicine is to determine the best therapy for an individual based on the information gathered from him or her. In its early stages, this will involve

transforming a research methodology into a device for investigating and/or evaluating a biomarker or set of biomarkers that can be routinely implemented for healthcare. Personalized medicine can be applied in a wide variety of contexts and by different users, thus the device may also take a different form depending on the task. As has so far been recognised, personalised medicine occurs in a clinical setting and entails assisting the clinician in making decisions regarding drug therapy and preventative measures (pharmacogenomics). The development of companion diagnostics, or biomarker assays that are created concurrently with therapeutic products and are used to assess the efficacy and/or safety of those drugs for a specific patient, is being sparked by advances in pharmacogenomics.

In addition to diagnostics firms collaborating with pharmaceutical firms to create companion diagnostics for their products (Qiagen + Pfizer), pharmaceutical firms themselves are establishing companion diagnostics departments (Roche). These companion diagnostics have so far involved obtaining a patient's sample and forwarding it to the lab for examination using specialised tools. Nevertheless, even in genetic testing, there is a tendency towards point-of-care testing or near-patient diagnostics, which brings the test to the patient.

As an illustration of this, pilot tests have shown that a hand-held gadget (the "SNIP doctor") for examining single nucleotide polymorphisms is effective. A key step for personalised medicine will be to develop diagnostic modalities that are handy for health professionals, such as kits, machines and portable imaging devices. Over the long run, personalised medicine may require citizens to become health literate and to use health monitoring and management tools in their daily lives [12]. The concept of self-monitoring is not new: chronic conditions such as diabetes and asthma are managed using self-monitoring. Additionally, high street pharmacies now sell blood pressure monitoring devices, which are becoming increasingly available for self-monitoring. The development of an iPhone app to track food intake via barcodes is an example of how advanced technologies are implemented to simplify regular health monitoring. Physiological variables are being tracked and communicated to a mobile smart phone or other computerised device through increasingly sophisticated medical devices. Proteus Biomedical, for instance, manufactures the 'raisin personal monitor' which is worn under the skin like a sticking plaster to monitor and analyse heart rate, physical activity, body position, and patient-logged events. The company is also developing 'chip-on-a-pill' technology: ingestible microsensors that can be implanted in tablets and pills along with this monitoring tools. These markers are currently used to support adherence to medication, but could also feedback data from diagnostics or other measuring devices. As a result of these technologies and gadgets, functional status can be continuously monitored in real time, allowing therapy to be fine-tuned to control chronic diseases or lifestyle changes to achieve health goals.

## II. PERSONALISED MEDICINE AND THE HUMAN GENOME

Personalised medicine is an emerging field which is based on an individual's genetic profile to take decisions in regards of prevention, diagnosis and treatment of disease. Understandings of a patient's genetic profile can aid doctors in selecting the appropriate medication or the therapy and its administration in particular controlled dose. It is being advanced through data from the Human Genome Project. According to Sir Bruce Keogh, Medical Director NHS England, "In 19<sup>th</sup> and 20<sup>th</sup> centuries, microscope and x-rays

revolutionised medicine so in 21<sup>st</sup> century, the knowledge of the human genome will dramatically change medicines”.

Studies on the genome sequences of microorganisms, plants and animals have revolutionized many fields of science which includes microbiology, virology, plant biology and infectious diseases. Similarly, the Human Genome Project has remodelled biology by decoding a reference human genome sequence along with the whole genome sequences of key model organisms [13]. Scientist, Renato Dulbecco first of all advocated the idea of the Human Genome Project publicly in an article published in 1984 in which the knowledge of the human genome sequence would help in better understanding of cancer [14]. Initially, the main aim of Human Genome Project was to determine a human genetic map and then a physical map of the human genome [15].

The completion of The Human Genome project in 2001 helped a lot in deeper understanding of medicines. The knowledge achieved through further research has changed the field of genomics and helping into clinical medicines [16]. The combination and analysis of information about our genome with other clinical and diagnostic findings will help in determining our individual risk of disease, earlier detection of illness and finding the most effective way to improve our health; either by using medicines or changes in lifestyles or even simple changes in diet. By understanding the role of DNA in our health will help in:

- The prediction of disease
- The precise diagnosis and prevention of disease
- Personalised treatment plans
- A more involvement of patients

**1. Decoding the Genome:** The human genome comprises of approx. 3 billion DNA base pairs (data from National Human Genome Research Institute, 2003) [17]. Previously, individual genes were sequenced exon by exon using conventional method of Sanger sequencing but now the advancements in the field of sequencing have enabled a genome to be sequenced within hours and cost effectively resulting in numerous applications in the field of diagnosis and research (DNA sequencing cost data from National Human Genome Research Institute, 2016) [18]. There are around 3-5 million genetic variants in every human genome when compared with the reference sequence. Hence it is very challenging in understanding variation within the human genome and to differentiate ‘normal sequences’ from disease causing variants. Therefore, it is very important to take expert interpretation and caution in analysing genomic results [19]. Accuracy is vital as inaccuracy in interpretation may not only cause harm to the particular individual but also to the family members as well as the future generations.

**2. Pharmacogenetics and Pharmacogenomics:** The basic difference between pharmacogenetics and pharmacogenomics is that the previous one examines the relationship between drug response and genetic variances but the later uses a genome wide approach to understand the complete range of genes associated with drug response [20]. Both pharmacogenetics and pharmacogenomics are basis for personalized medicines. They provide opportunities to revolutionize drug therapy by designing drugs according to individual genotypes [21].



The adverse reaction to drugs and their lack of effectiveness in several patients require new methods to improvise drug therapy that can be influenced by knowledge of specific genetic makeup of individuals [22]. The metabolism and fate of any drug inside any individual along with its therapeutic and toxicological effect depend on complex process which involves protein coded by different genes. These proteins influence the transport of drug inside body, its metabolism and mechanism of action [21]. At the time of evolution, majority of genes comprise certain discrepancies in their nucleotide sequence. If these variations occur in the coding region, it may result in substitution of an amino acid in specific position of polypeptide chain and which will in turn affect protein function. In other words, variations in nucleotide sequence influences transcription and translation processes [23]. The ultimate goal of personalized medicine is to exactly match each therapeutic intervention with the patient's molecular profile [24]. Hence, pharmacogenomics encourages the advancement of targeted therapies and was demonstrated by the use of drug ivacaftor for the treatment of subset of cystic fibrosis patients which was approved by the US Food and Drug Administration (FDA) and the European Medicines Agency [25].

### III. TECHNOLOGICAL REQUIREMENTS FOR THE DEVELOPMENT OF PERSONALIZED MEDICINES

Researchers and scientists have a great hope in personalized medicines as a form of healthcare that is being designed based on individual needs. The main aim is to shift 'one size fits all' concept to a more objective based prediction, prevention and therapy for underlying illness [26, 27]. It allows scientists to develop customizable therapeutic formulations based on an individual's genetic composition, medical history and physiology. Many novel technologies are required for the personalized medicine to be realised in practice.

**1. Genomics and Personalized Medicines:** Genomics plays an incipient role in clinical and public health research. Technology related to genomics have abundant applications in clinical medicine and have the capability to transform public health. The battle against the COVID-19 pandemic which has imposed the implementation of genomic technologies to find the origin, transmission and evolution of the SARS-CoV-2 virus worldwide and also to understand different host response against virus, severity of infection and outcomes [28]. Genetics and genome technologies allow us to develop a link between our genes and health.

Genomics studies provide precision both in terms of the infectious agent and the host population that is affected. One of the genomic technology, whole genome sequencing (WGS) is an important standard assay for characterizing infectious disease and provide good source of information to guide public health interventions [29]. DNA sequencing helps in the identification of pathogen species or variants which may be linked to variety of risks and hence bring about diverse responses in terms of clinical management of individual patients [28]. Different genetic variants produce conditions that require different treatments yet they share similar symptoms. So, without knowledge of exact genetic cause of symptom, it is difficult to decide most effective treatment. For example, there are many causes of lung cancer but those people who have an alteration in the gene EFGR only respond to treatment with tyrosine kinase inhibitors [30, 31]. The main aim of personalized medicine to predict most appropriate course of action. Genetic and clinical data can be combined to find best action plan for a patient.

**2. Proteomics and Personalized Medicines:** Proteomics, coined by Mark Williams in 1994, is the characteristic investigation of the complete protein complement at cellular, tissue or organism level including protein interactions, localizations and post translational modifications at a particular condition [32]. Proteins are the building blocks and are directly responsible for structure, function and regulation of body's tissues and organs. Proteomics provide direct biological insights into physiological patterns while genetics contribute for basis characteristics. Proteins are the products of gene expression and are direct bridge between diagnostics and therapeutics [33]. There are several factors which influence a person response to different disease and treatment. These factors can be epigenetic, transcriptional, proteomic, posttranslational modifications, metabolic and environmental factors. Though personalized medicine has the same roots as genomic medicine but it goes far beyond genetics considering the full complexity of cellular physiology [34]. There are different proteins which are expressed in an individual before infection, after infection or in an infected person [35, 36] (Herberts et al. 2003; Wahl et al. 2010). Therefore, it is important to integrate a cellular physiology, environmental factors and medical history of the individual for the development of personalized medicine and to generate a customized treatment strategy based on that individual's need. Techniques like RNA microarrays help in determining gene expression and the association of various proteins depicting different disease conditions or levels. The consequent progression of proteomics has enabled scientists to use microarrays for studying expression, kinome and interactome profiling [37].

The key technique presently used for proteomic study is the mass spectrometry. This technique depends on the accomplishment in three main aspects: sample pretreatment and examination and data analysis. The techniques used for sample preparation before mass spectrometric analysis are 2-dimensional gel electrophoresis and sodium dodecyl sulphate polyacrylamide gel electrophoresis (SDS-PAGE) [32]. Chromatographic methods are of importance for high level sample purification, recovery and automation before mass spectrometric analysis [38, 39]. Other than this protein microarrays are being used to study protein-macromolecule interaction analysis and identification of drug target in a speedy, reproducible and economical way [40]. Intensive studies in the field of tumour biology have led to exploration of prospective therapeutic targets resulting in emergence of new drugs. Translational studies facilitate novel and improvised clinical assays that shall help in development of new personalized medicines helping oncologists to determine right treatment for individual patients [41]. Proteomics is a powerful tool for the development of personalized medicine.

**3. Metabolomics and Personalized Medicines:** Metabolites provide straight forward knowledge on the biochemical activity in a cell during diseased condition. Measurement and assessment of these metabolites in huge scale categorised under the omics technology known as "metabolomics" [42]. It is an emerging 'omics' science combining characterization of metabolites and metabolism in biological system [43]. The formulation of personalized medicine requires the combination of all molecular variations that may differentiate individuals. Metabolomics in association with genomics can be applied to distinguish patients with different treatment response and hence helps in the progress of host targeted therapy in contrast to the pathogen targeted therapy as pathogens are susceptible to variations causing antimicrobial resistance. Therefore, metabolomics

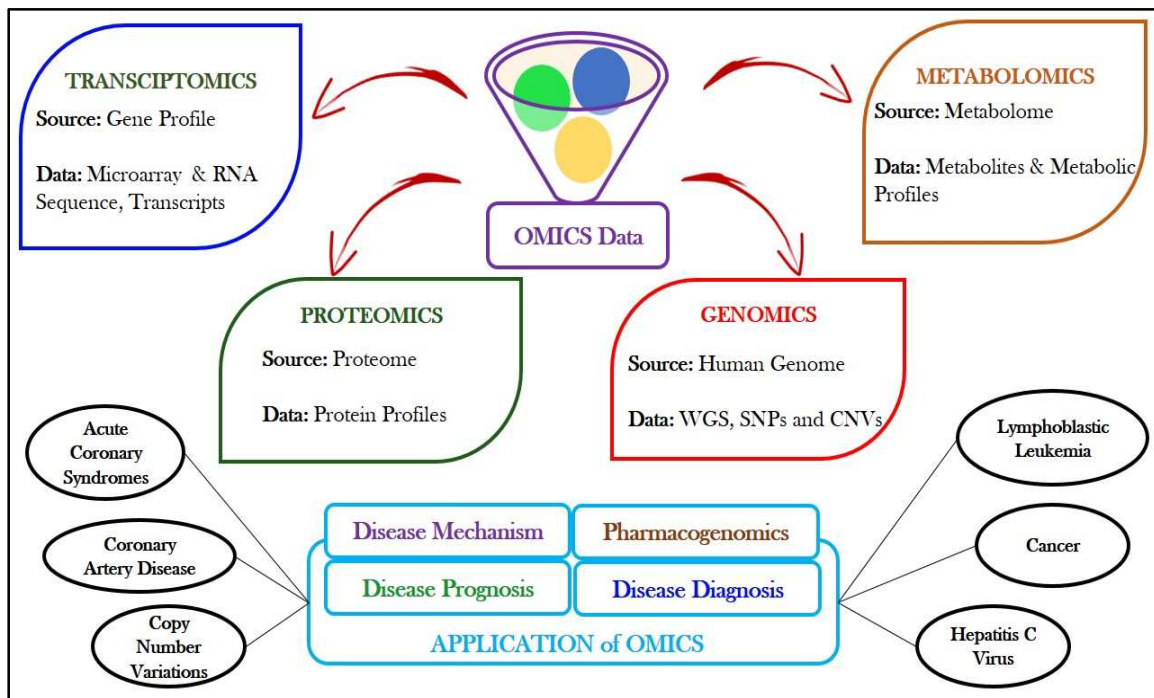
can be implemented for patient categorisation, personalized drug development and disease control and management [42].

The most commonly used analytical methods for metabolomics studies are the use of various chromatographic separation techniques, most commonly gas chromatography or liquid chromatography in association with various mass spectrometry detectors and Nuclear Magnetic Resonance. The application of metabolomics for the development of personalized medicine involves three basic steps [44]

- **Pre-Analytical Processes:** The important factors need to be taken into account at this step are type of sample containers for collection and storage of samples, sample handling and transport. It involves good experimental design for selection of sample, quenching solution and methods for metabolite extraction, separation and detection [45].
- **Detection of Analyte:** This involves biophysical methods that determines structure, characteristics and functions of biomolecules at atomic and molecular level. The commonly used techniques are X-ray crystallography, chromatography, spectroscopic techniques like nuclear magnetic resonance (NMR), mass spectrometry (MS) and surface plasmon resonance (SPR) [45, 46].
- **Validation of Result and Clinical Translation:** The type and concentration of metabolites varies depending on the type of sample taken. Hence, the selection of metabolite to be used for clinical inference or as biomarker require consent from regulatory bodies [47].

Metabolomic studies whose main aim is to identify biomarkers of disease requires complex technologies and well-designed roadmap for precise inference and representation of the patient related data. As evident, from the above information, ‘-Omics’ technologies project diverse applications in investigating the disease mechanism, disease prognosis as well as diagnosis along with pharmacogenomics studies, thus laying a strong foundation for ‘Personalized Medicine’ (Fig. 4).

4. **Epigenetics and Personalized Medicine:** ‘Epigenetics’, coined by Conrad Waddington in the 1940s is the study of reversible modifications to chromatin and their effects on gene regulation or the modifications in gene expression that are not due to changes in DNA sequences but due to changes in the internal and external environment [48]. The modification in gene expressions can be controlled by several mechanisms like DNA methylation, post translational modifications of histones, chromatin remodelling and microRNAs that act as regulatory molecules [49]. There are various methods to classify phenotypic or epigenetic modifications in biological system and patients with epigenetic alterations do not respond to conventional therapy. Epigenetic disease associations function as diagnostic biomarkers [50]. The two epigenetic tests that are currently available in US are ConfirmMDx for prostate cancer and AssureMDx for bladder cancer [51]. As epigenetic marks are reversible therefore drugs are developed with epigenetic mode of action. Hence, personalized medicines can be developed to efficiently deal these disorders on the basis of individual’s genetic profile.



**Figure 4:** Applications of OMICS

#### IV. PERSONALISED MEDICINE FOR CERTAIN DISEASES

1. **Personalised Medicine for Cancer:** Personalized Medicine has a promising impact in curing patients suffering from many diseases. One of the initial and prevalently used personalized medicine used for treating breast cancer is trastuzumab. Most of the patients with breast cancer (~30%) have a form that over-expression of a protein called HER2 [52], it is of a major concern due to its unresponsiveness to certain standardised therapies which otherwise play a major role in curing the disease. Hence, Trastuzumab was formulated and received approval in 1998 to treat the disease with HER2 overexpression. A research analysis conducted in 2005 had significant findings in reduction of breast cancer recurrence by 52% with personalized medicine along with chemotherapy [53].

In melanoma, BRAF is the human gene accountable for the production of a protein called B-Raf, which has a role in signal transduction related to direct cell growth and found to be mutated in cancers [54]. A breakthrough came up in 2011 to treat last stage melanoma using a drug called vemurafenib. It is a B-Raf protein inhibitor, hence, this drug and a treatment associated test known as BRAF V600E mutation test received approval for the treatment of late-stage melanoma. Vemurafenib worked well for treatment of cancer patients only with positive V600E BRAF mutation test [55].

2. **Personalised Medicine for Diabetes:** Personalized medicine for diabetes has gained significant importance as it is considered as most common form of disease now-a-days. It can be used to provide information about the genetic makeup of a patient with diabetes to formulate a strategy to prevent, detect, treat or monitor their diabetes [56]. Practicing personalized medicine for diabetes is a four-step process:

- The genes as well as biomarkers identification is crucial for diabetes and for obesity, as it is the prevalent risk factor for type 2 diabetes.
- After identification of these parameters, allocation of resources is essential to prevent or recognise the diabetes and/or obesity phenotype in high-risk people, whose risk depends upon their genotype.
- Selection of individualized therapies for affected persons is a third step. The drug will be selected based on which drug to prescribe, what dose of drug to use, and which diet to prescribe. Selection process also justifies on drugs having least side effects or toxicity.
- Evaluation of diabetes circulating biomarkers for monitoring the response for prevention or therapeutic use

## V. TECHNOLOGIES FOR PERSONALISED MEDICINE: FACTORS INFLUENCING THEIR DEVELOPMENT

- 1. Integration of Information:** Combining different sorts of information comes under integration which involves – biological, clinical, environmental, lifestyle – about the contributor in a research analysis or the patient in the clinic or the citizen determined to improve their health. This combining must transfer from the current linear, cumulative model to a truly integrative model for generation of system-level knowledge [57, 58]. Moreover, organisational integration is crucial. It is significant to have a framework for integrating science across countries, allowing scientists to collaborate on the same problems in different research sites.
- 2. Co-Ordination:** Generation of standardised data which speeds up research, there must be co-ordination in critical steps such as collection of data, verification for accuracy and storage of the data at suitable site. A practical measure for implementing this coordination involves imposing and monitoring quality assurance procedures for data assembly and data processing. Ideally, these processes should be implemented for all sorts of data (biological, clinical, environmental) as well as incorporating information about their origin [59, 60]. To integrate research output as suggested above, there needs to be coordination of research methods, in addition to data collection and analysis, so that information can be transformed into understanding of biological functioning and these insights can be converted into interventions to change that functioning in health-promoting ways. Communication tools that are more effective across different sciences, as well as between science and medicine, will be crucial to the coordination of efforts. In order for research to be targeted to priority areas and produce workable results, there needs to be better communication between bioscience and biomedicine about what health issues require attention and what remedies are implementable [59, 60].
- 3. Resources:** As anticipated, in order to develop technologies for personalised medicine, resources are an issue. Data processing and data storage needs access to computing resources. Bioscience funding needs to be re-distributed or re-prioritized by funding bodies like national governments in order to guarantee access to IT resources. The next-generation technologies have permitted cheap, rapid sequencing of DNA, similarly, it may also drive the advancements in new data processing tools [61]. Choosing the right investigator is crucial to the success of the investigation. However, lack of skilled and experienced people is a concern. It is essential that they be skilled and focused to succeed

in their jobs. Additionally, adequate grants must be available for translational and basic research. As the focus shifts from potent drugs to in vitro diagnostics, the pharmaceutical industry will have to develop innovative business models to implement new technology for personalized medicine and introduce it into healthcare practice. New partnerships between in vitro diagnostics companies and pharmaceutical companies are expected to emerge as a result. Translational research has been criticized for being less financially supported, whereas basic research is experiencing intense pressure [62]. The potentials of personalised medicine involve radical plus evolutionary progresses: as we move from linear to integrated and dynamic data, from explicating how biological components work to deciphering how biological systems function, from one-size-fits-all healthcare to one-tailored healthcare that is tailored to the individual, we must move from linear to integrated and dynamic data.

These transition press for a true originality which is accurately inventive, in compared to just novel provisions of what already exists. Such innovation necessitates a tremendous amount of effort along with the readiness to tolerate failure. Resource agencies need to be convinced of the value of this kind of "disruptive research."

## VI. CONCLUSION

Assessment of disease risk and prevention of the same is possible with the utilization of a promising approach of personalized medicine, which involve family history and genome information. Key clinical decisions can be easily taken with the help of data obtained from patients' genomic information which provides the foundation for genomic medicine. The incorporation of genomic research into the medical field needs to be optimized, standardized as well as reorganized. Personalized medicine is by now being practiced in the clinic, and the incorporation of genomic tools specifically in oncology and cardiology has improved patients' treatment. The additional integration of personalized medicine into the medical system needs seizing various barriers in learning, ease of access, regulation, and reimbursement.

## REFERENCES

- [1] Esplin, E. D., Oei, L., & Snyder, M. P. (2014). Personalized sequencing and the future of medicine: Discovery, diagnosis and defeat of disease. *Pharmacogenomics*, 15(14), 1771-1790. <https://doi.org/10.2217/pgs.14.117>
- [2] Goetz, L. H., & Schork, N. J. (2018). Personalized medicine: Motivation, challenges, and progress. *Fertility and Sterility*, 109(6), 952-963. <https://doi.org/10.1016/j.fertnstert.2018.05.006>.
- [3] Mathur, S., & Sutton, J. (2017). Personalized medicine could transform healthcare. *Biomedical Reports*, 7(1), 3-5. <https://doi.org/10.3892/br.2017.922>
- [4] Golubnitschaja, O., & Costigliola, V. (2010). Common origin but individual outcomes: Time for new guidelines in personalized healthcare. *Personalized Medicine*, 7(5), 561-568. <https://doi.org/10.2217/pme.10.42>
- [5] Bohr, A., & Memarzadeh, K. (2020). The rise of artificial intelligence in healthcare applications. *Artificial Intelligence in Healthcare*, 25-60. <https://doi.org/10.1016/b978-0-12-818438-7.00002-2>
- [6] Ferraldeschi, R., & Newman, W. G. (2011). Pharmacogenetics and pharmacogenomics: A clinical reality. *Annals of Clinical Biochemistry: International Journal of Laboratory Medicine*, 48(5), 410-417. <https://doi.org/10.1258/acb.2011.011084>
- [7] Brand, A. (2011). Public health genomics-- public health goes personalized? *The European Journal of Public Health*, 21(1), 2-3. <https://doi.org/10.1093/eurpub/ckq197>

- [8] Hood, L., Heath, J. R., Phelps, M. E., & Lin, B. (2004). Systems biology and new technologies enable predictive and preventative medicine. *Science*, 306(5696), 640-643. <https://doi.org/10.1126/science.1104635>
- [9] Galas, D. J., & Hood, L. (2009). Systems biology and emerging technologies will catalyze the transition from reactive medicine to predictive, personalized, preventive and participatory (P4) medicine. *Interdisciplinary Bio Central*, 1(2), 1-4. <https://doi.org/10.4051/ibc.2009.2.0006>
- [10] Oussous, A., Benjelloun, F., Ait Lahcen, A., & Belfkih, S. (2018). Big data technologies: A survey. *Journal of King Saud University - Computer and Information Sciences*, 30(4), 431-448. <https://doi.org/10.1016/j.jksuci.2017.06.001>
- [11] Stefanicka-Wojtas, D., & Kurpas, D. (2023). Personalised medicine—Implementation to the healthcare system in Europe (Focus group discussions). *Journal of Personalized Medicine*, 13(3), 380. <https://doi.org/10.3390/jpm13030380>
- [12] Brand, A., & Brand, H. (2011). Health literacy and public health genomics: Innovation management by citizens. *Public Health Genomics*, 14(4-5), 193-194. <https://doi.org/10.1159/000324237>
- [13] Hood, L., & Rowen, L. (2013). The Human Genome Project: Big science transforms biology and medicine. *Genome Medicine*, 5(9), 79. <https://doi.org/10.1186/gm483>
- [14] Dulbecco, R. (1986). undefined. *Science*, 231(4742), 1055-1056. <https://doi.org/10.1126/science.3945817>
- [15] Collins, F., & Galas, D. (1993). A new five-year plan for the U.S. Human Genome Project. *Science*, 262(5130), 43-46. <https://doi.org/10.1126/science.8211127>
- [16] Carrasco-Ramiro, F., Peiró-Pastor, R., & Aguado, B. (2017). Human genomics projects and precision medicine. *Gene Therapy*, 24(9), 551-561. <https://doi.org/10.1038/gt.2017.77>
- [17] Barbujani, G., & Colonna, V. (2010). Human genome diversity: Frequently asked questions. *Trends in Genetics*, 26(7), 285-295. <https://doi.org/10.1016/j.tig.2010.04.002>
- [18] The cost of sequencing a human genome. (2019, March 13). <https://www.genome.gov/about-genomics/fact-sheets/Sequencing-Human-Genome-cost>
- [19] Brittain, H. K., Scott, R., & Thomas, E. (2017). The rise of the genome and personalised medicine. *Clinical Medicine*, 17(6), 545-551. <https://doi.org/10.7861/clinmedicine.17-6-545>
- [20] Shastry, B. S. (2005). Pharmacogenetics and the concept of individualized medicine. *The Pharmacogenomics Journal*, 6(1), 16-21. <https://doi.org/10.1038/sj.tpj.6500338>
- [21] Nobili, S., & Mini, E. (2022). Special issue: “Gastrointestinal cancers and personalized medicine”. *Journal of Personalized Medicine*, 12(3), 338. <https://doi.org/10.3390/jpm12030338>
- [22] Wilke, R. A., Lin, D. W., Roden, D. M., Watkins, P. B., Flockhart, D., Zineh, I., Giacomini, K. M., & Krauss, R. M. (2007). Identifying genetic risk factors for serious adverse drug reactions: Current progress and challenges. *Nature Reviews Drug Discovery*, 6(11), 904-916. <https://doi.org/10.1038/nrd2423>
- [23] Cannon, J. G. (2006). Goodman and Gilman's the pharmacological basis of therapeutics. 11th edition edited by Laurence Brunton, John Lazo, and Keith Parker. McGraw hill, New York. 2005. xxiii + 2021 pp. 21 × 26 CM. ISBN 0-07-142280-3. \$149.95. *Journal of Medicinal Chemistry*, 49(3), 1222-1222. <https://doi.org/10.1021/jm058286b>
- [24] Cecchin, E., & Stocco, G. (2020). Pharmacogenomics and personalized medicine. *Genes*, 11(6), 679. <https://doi.org/10.3390/genes11060679>
- [25] Eckford, P. D., Li, C., Ramjeesingh, M., & Bear, C. E. (2012). Cystic fibrosis Transmembrane conductance regulator (CFTR) Potentiator VX-770 (Ivacaftor) opens the defective channel gate of mutant CFTR in a phosphorylation-dependent but ATP-independent Manner. *Journal of Biological Chemistry*, 287(44), 36639-36649. <https://doi.org/10.1074/jbc.m112.393637>
- [26] Ginsburg, G. S., & Willard, H. F. (2010). The foundations of Genomic and personalized medicine. *Essentials of Genomic and Personalized Medicine*, 1-10. <https://doi.org/10.1016/b978-0-12-374934-5.00001-5>
- [27] Wainwright, S. P., Williams, C., Michael, M., Farsides, B., & Cribb, A. (2006). From bench to bedside? Biomedical scientists' expectations of stem cell science as a future therapy for diabetes. *Social Science & Medicine*, 63(8), 2052-2064. <https://doi.org/10.1016/j.socscimed.2006.05.003>
- [28] Khoury, M. J., & Holt, K. E. (2021). The impact of genomics on precision public health: Beyond the pandemic. *Genome Medicine*, 13(1). <https://doi.org/10.1186/s13073-021-00886-y>
- [29] Gardy, J. L., & Loman, N. J. (2017). Towards a genomics-informed, real-time, global pathogen surveillance system. *Nature Reviews Genetics*, 19(1), 9-20. <https://doi.org/10.1038/nrg.2017.88>
- [30] Lynch, T. J., Bell, D. W., Sordella, R., Gurubhagavatula, S., Okimoto, R. A., Brannigan, B. W., Harris, P. L., Haserlat, S. M., Supko, J. G., Haluska, F. G., Louis, D. N., Christiani, D. C., Settleman, J., & Haber, D. A. (2004). Activating mutations in the epidermal growth factor receptor underlying responsiveness of non-

- small-cell lung cancer to gefitinib. *The New England journal of medicine*, 350(21), 2129–2139. <https://doi.org/10.1056/NEJMoa040938>
- [31] Paez, J. G., Jänne, P. A., Lee, J. C., Tracy, S., Greulich, H., Gabriel, S., Herman, P., Kaye, F. J., Lindeman, N., Boggon, T. J., Naoki, K., Sasaki, H., Fujii, Y., Eck, M. J., Sellers, W. R., Johnson, B. E., & Meyerson, M. (2004). EGFR Mutations in lung cancer: Correlation with clinical response to Gefitinib therapy. *Science*, 304(5676), 1497-1500. <https://doi.org/10.1126/science.1099314>
- [32] Nice, E. C. (2020). The separation sciences, the front end to proteomics: An historical perspective. *Biomedical Chromatography*, 35(1). <https://doi.org/10.1002/bmc.4995>
- [33] Ding, C., Qin, Z., Li, Y., Shi, W., Li, L., Zhan, D., & Yang, W. (2019). Proteomics and precision medicine. *Small Methods*, 3(7), 1900075. <https://doi.org/10.1002/smt.201900075>
- [34] Roberts, S., & Julius, M. (2016). Precision medicine. *Healthcare Management Forum*, 29(4), 158-161. <https://doi.org/10.1177/0840470416642773>
- [35] Herberts, C. A., Van Gaans-van den Brink, J., Van der Heeft, E., Van Wijk, M., Hoekman, J., Jaye, A., Poelen, M. C., Boog, C. J., Roholl, P. J., Whittle, H., De Jong, A. P., & Van Els, C. A. (2003). Autoreactivity against induced or upregulated abundant self-peptides in HLA-a\*0201 following measles virus infection. *Human Immunology*, 64(1), 44-55. [https://doi.org/10.1016/s0198-8859\(02\)00707-3](https://doi.org/10.1016/s0198-8859(02)00707-3)
- [36] Wahl, A., Schafer, F., Bardet, W., & Hildebrand, W. H. (2010). HLA class I molecules reflect an altered host proteome after influenza virus infection. *Human Immunology*, 71, 14–22. doi:10.1016/j.humimm.2009.08.012
- [37] Gupta, S., Manubhai, K. P., Kulkarni, V., & Srivastava, S. (2016). An overview of innovations and industrial solutions in protein microarray technology. *PROTEOMICS*, 16, 1297–1308. doi:10.1002/pmic.201500429
- [38] Nice, E. C., Rothacker, J., Weinstock, J., Lim, L., & Catimel, B. (2007). Use of multidimensional separation protocols for the purification of trace components in complex biological samples for proteomics analysis. *Journal of Chromatography A*, 1168, 190–210. doi:10.1016/j.chroma.2007.06.015
- [39] Duong, V.-A., Park, J.-M., & Lee, H. (2020). Review of three-dimensional liquid chromatography platforms for bottom-up proteomics. *International Journal of Molecular Sciences*, 21, 1524. doi:10.3390/ijms21041524
- [40] Ramiya Ramesh Babu, H. K., & Gheber, L. A. (2018). Fluorescence-based kinetic analysis of miniaturized protein microarrays. *Biosensors and Bioelectronics*, 122, 290–299. doi:10.1016/j.bios.2018.09.051
- [41] Li, A., & Bergan, R. C. (2020). Clinical trial design: Past, present, and future in the context of Big Data and Precision Medicine. *Cancer*, 126, 4838–4846. doi:10.1002/cncr.33205
- [42] Rahman, M., & Schellhorn, H. E. (2023). Metabolomics of Infectious Diseases in the era of personalized medicine. *Frontiers in Molecular Biosciences*, 10. doi:10.3389/fmolb.2023.1120376
- [43] Wishart, D. S. (2016). Emerging applications of metabolomics in drug discovery and Precision Medicine. *Nature Reviews Drug Discovery*, 15, 473–484. doi:10.1038/nrd.2016.32
- [44] Trivedi, D. K., & Goodacre, R. (2020). The role of metabolomics in personalized medicine. In M. A. van Aghoven & S. L. Bourke (Eds.), *Metabolomics for Biomedical Research* (pp. 227–244). Elsevier Inc. doi:10.1016/B978-0-12-812784-1.00011-6
- [45] Di Minno, A., Gelzo, M., Stornaiuolo, M., Ruoppolo, M., & Castaldo, G. (2021). The evolving landscape of untargeted metabolomics. *Nutrition, Metabolism, and Cardiovascular Diseases*, 31, 1645–1652. doi:10.1016/j.numecd.2021.01.008
- [46] Frédéric, M., Piroette, B., Fillet, M., & De Tullio, P. (2016). Metabolomics as a challenging approach for medicinal chemistry and personalized medicine. *Journal of Medicinal Chemistry*, 59(19), 8649-8666. <https://doi.org/10.1021/acs.jmedchem.5b01335>
- [47] Ramanathan, K., Antognini, D., Combes, A., Paden, M., Zakhary, B., Ogino, M., MacLaren, G., Brodie, D., & Shekar, K. (2020). Planning and provision of ECMO services for severe ARDS during the COVID-19 pandemic and other outbreaks of emerging infectious diseases. *The Lancet Respiratory Medicine*, 8(5), 518-526. [https://doi.org/10.1016/s2213-2600\(20\)30121-1](https://doi.org/10.1016/s2213-2600(20)30121-1)
- [48] Goldberg, A. D., Allis, C. D., & Bernstein, E. (2007). Epigenetics: A landscape takes shape. *Cell*, 128, 635–638. doi:10.1016/j.cell.2007.02.006
- [49] Rasool, M., Malik, A., Naseer, M. I., Manan, A., Ansari, S. A., Begum, I., Qazi, M. H., Pushparaj, P. N., Abuzenadah, A. M., Al-Qahtani, M. H., Kamal, M. A., & Gan, S. H. (2015). The role of epigenetics in personalized medicine: Challenges and opportunities. *BMC Medical Genomics*, 8. doi:10.1186/1755-8794-8-s1-s5
- [50] García-Giménez, J. L., Ushijima, T., & Tollefsbol, T. O. (2016). Epigenetic biomarkers. *Epigenetic Biomarkers and Diagnostics*, 1-18. <https://doi.org/10.1016/b978-0-12-801899-6.00001-2>



- [51] Partin, A. W., Van Neste, L., Klein, E. A., Marks, L. S., Gee, J. R., Troyer, D. A., Rieger-Christ, K., Jones, J. S., Magi-Galluzzi, C., Mangold, L. A., Trock, B. J., Lance, R. S., Bigley, J. W., Van Criekinge, W., & Epstein, J. I. (2014). Clinical validation of an epigenetic assay to predict negative histopathological results in repeat prostate biopsies. *Journal of Urology*, 192, 1081–1087. doi:10.1016/j.juro.2014.04.013
- [52] Iqbal, N., & Iqbal, N. (2014). Human Epidermal Growth Factor Receptor 2 (HER2) in Cancers: Overexpression and Therapeutic Implications. *Molecular Biology International*, 2014, 852748. doi:10.1155/2014/852748
- [53] Nahta, R. (2012). Molecular Mechanisms of Trastuzumab-Based Treatment in HER2-Overexpressing Breast Cancer. *ISRN Oncology*, 2012, 428062. doi:10.5402/2012/428062
- [54] Lin, K., Baritaki, S., Militello, L., Malaponte, G., Bevelacqua, Y., & Bonavida, B. (2010). The Role of B-RAF Mutations in Melanoma and the Induction of EMT via Dysregulation of the NF- $\kappa$ B/Snail/RKIP/PTEN Circuit. *Genes & Cancer*, 1(5), 409–420. doi:10.1177/1947601910373795
- [55] Shelledy, L., & Roman, D. (2015). Vemurafenib: First-in-Class BRAF-Mutated Inhibitor for the Treatment of Unresectable or Metastatic Melanoma. *Journal of Advanced Practitioner in Oncology*, 6(4), 361–365. doi:10.6004/jadpro.2015.6.4.6
- [56] McCarthy, M. I. (2017). Painting a new picture of personalised medicine for diabetes. *Diabetologia*, 60, 793–799. doi:10.1007/s00125-017-4210-x
- [57] Huang, S. (2000). The practical problems of post-genomic biology. *Nature Biotechnology*, 18(5), 471-472. <https://doi.org/10.1038/75235>
- [58] Kitano, H. (2002). Looking beyond the details: A rise in system-oriented approaches in genetics and molecular biology. *Current Genetics*, 41(1), 1-10. <https://doi.org/10.1007/s00294-002-0285-z>
- [59] Calvert, J. (2010). Systems biology, interdisciplinarity and disciplinary identity. In J. N. Parker, N. Vermeulen, & B. Penders (Eds.), *Collaboration in the New Life Sciences* (pp. 199–215). Ashgate.
- [60] Lewis, J. (2010). Matchmaking mechanisms: Collaborative arrangements in proteomics and bioinformatics. In J. N. Parker, N. Vermeulen, & B. Penders (Eds.), *Collaboration in the New Life Sciences* (pp. 219–233). Ashgate.
- [61] Partridge, W. M. (2003). Translational science: What is it and why is it so important? *Drug Discovery Today*, 8(18), 813-815. [https://doi.org/10.1016/s1359-6446\(03\)02823-x](https://doi.org/10.1016/s1359-6446(03)02823-x)
- [62] Osborne, T. (2003). Against ‘creativity’: A -philistine rant. *Economy and Society*, 32(4), 507-525. <https://doi.org/10.1080/0308514032000141684>