

# ***Leveraging Genomic Data for Infectious Disease Surveillance and Personalized Medicine: Advances in Public Health Genomics and COVID-19 Pathway Analysis***

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**Abstract:** In public health, the arrival of genomic data greatly enhanced infectious disease surveillance, personalised medicine and the global response to pandemics. This Review examines how genomic sequencing became a vital tool for identifying pathogens, tracking virus evolution, and tracing transmission chains, with case studies on COVID-19 variants such as Delta and Omicron. Genomic sequencing has also led to breakthroughs in personalised medicine and pharmacogenomics, whereby drug efficacy and adverse reactions are optimised for individual patients. Genomic surveillance is now a critical tool for detecting emergent threats, monitoring disease spread, and evaluating the efficacy of public health interventions, from contact tracing to vaccines. These genomic approaches have saved thousands of lives during the global COVID-19 pandemic. The paper ends on a positive note about the advances in public health genomics and underscores the value of continuing investment in genomic technologies to safeguard global health security.

**Keywords:** Genomic Data, Infectious Disease Surveillance, Personalized Medicine, Public Health Genomics, COVID-19.

## **1. Introduction**

The introduction of genomic technologies into public health has caused a quantum evolution in the field, particularly in the fields of infectious disease surveillance & personalised medicine. Genomic data from DNA and RNA sequencing has offered an unprecedented mosaic view into the biology of pathogens and its interactions with the human host. The COVID-19 pandemic, in particular, has shown us the potential of genomic data in the realm of public health by allowing us to rapidly identify SARS-CoV-2, track its mutations and inform our disease response on a global scale. This paper will explore the different ways in which the use of genomic data has helped us improve public health outcomes. The most prominent application of DNA sequencing has been in the field of infectious disease surveillance. Rapid genomic sequencing of pathogens has enabled real-time monitoring of virus evolution and tracing transmission chains during outbreaks. Such capability has proved essential for managing complex outbreaks where traditional contact tracing failed. The use of genomic data has also found applications in personalised medicine by leveraging pharmacogenomics to tailor drug therapies to individual genetic profiles. This not only improves treatment outcome but also reduces

adverse drug reactions, leading to safer and more effective care. Another area on which genomic surveillance has found applications is detection of emerging threats, monitoring of spread and evaluation of public health interventions. By characterising the genetic sequences of pathogens, public health officials can decide on measures to control the spread of disease outbreaks while averting future pandemics. This paper will explore these applications of genomic disease surveillance using case studies from the COVID-19 pandemic, which highlighted how genetic data informs the public health policies and vaccine development [1].

## 2. Genomic Data in Infectious Disease Surveillance

The incorporation of genomic data into infectious disease surveillance has greatly enhanced our ability to track and control outbreaks. During the COVID-19 pandemic, more than 1.5 million SARS-CoV-2 genomes were sequenced around the world by the end of 2021 – over 800 genomic samples a day. Such genomic surveillance allowed researchers to track, in real time, the virus's evolution at a granular level. It enabled the identification of more than 12,000 mutations in the SARS-CoV-2 spike protein alone. With this unprecedented level of information, we could closely follow how a virus adapts to environmental pressures and public health measures. For instance, we could track changes as different variants of concern emerged and how these variants spread around the world. Genomic surveillance also helped us identify super-spreader events, such as when a single individual or event is responsible for a large number of infections. It turned out that in the early days of the pandemic, about 80 per cent of COVID-19 cases in any given region were linked to just 20 per cent of infected individuals [2]. Armed with such insights, it would have been possible to design much more targeted interventions, based on genomic data.

### 2.1. Identification of Pathogens

Genomic sequencing is crucial for identifying novel pathogens. For instance, SARS-CoV-2 was quickly identified when it first arose in Wuhan, China: the virus's genome was sequenced and published in the first few weeks. This information was instrumental in informing diagnostic test design and initiating vaccine research globally. Studies show that sequencing pathogens can reduce the time to identify a novel infectious disease from months to a matter of days, as illustrated in Figure 1. This can make all the difference in producing an effective public health response. The accuracy of genomic sequencing is virtually unparalleled in identifying pathogens: it can distinguish between certain closely related species, even tell apart one strain of a virus from another, with well over 99 per cent accuracy. This precision is crucial for outbreak management: if the wrong intervention is used, this can facilitate the spread of the pathogen [3].

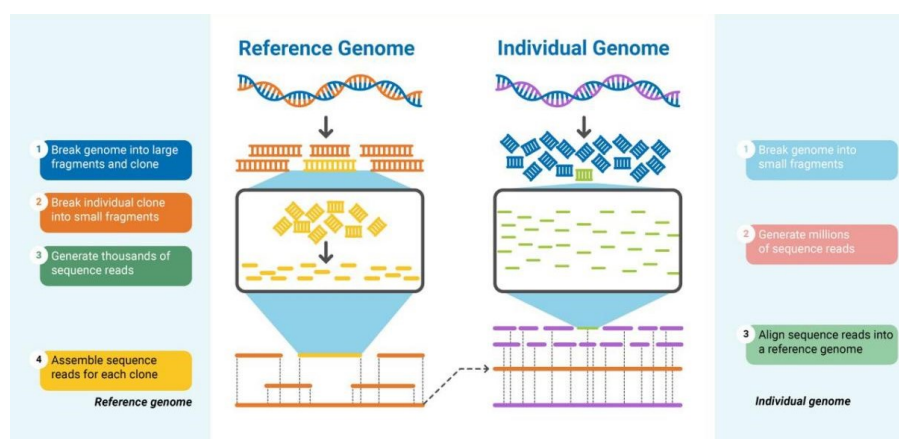


Figure 1: Whole Genome Sequencing (Source: Sequencing.com)

## 2.2. Monitoring Virus Evolution

Monitoring the evolution of viruses through genomic data is crucial for anticipating changes in virulence, transmissibility, and resistance to interventions. During the COVID-19 pandemic, for example, genomic surveillance detected the emergence of the Delta variant, which contained mutations that increased its transmission rate by 40-60% compared to earlier strains. This variant quickly became the dominant strain globally, causing new waves of infections and prompting changes in public health policies, such as the recommendation of booster vaccine doses [4]. Quantitative analysis revealed that the Delta variant's increased transmission rate led to a doubling of case numbers in several countries within a matter of weeks, underscoring the need for vigilant genomic monitoring. In another instance, the Omicron variant, identified later in the pandemic, was found to carry over 30 mutations in the spike protein, significantly reducing the efficacy of existing vaccines. These findings prompted a rapid adjustment of vaccine strategies, including the development of variant-specific booster shots [5].

## 2.3. Tracing Transmission Chains

Genomic data enables tracing transmission chains of infectious diseases in a more efficient and effective way. For outbreaks that require a large extent of contact-tracing efforts, genomic sequencing might where traditional approaches are not feasible. For instance, during the COVID-19 pandemic in Singapore, genomic sequencing was used to identify the source of a large outbreak in a dormitory that involved more than 1,000 COVID-19 cases. By comparing the genome of the virus isolated from the patients who were infected, researchers identified the index case, indicating the likely source of the outbreak. Furthermore, through comparing the genome sequences of the isolated viruses, they found that most of the cases shared very similar genetic lineage and that the outbreak likely started from one single source and spread rapidly through this type of crowded living environment. By leveraging the genomic tracing method, public-health officials reduced their complex contact-tracing focus to the genome cluster where over 85% of the cases started. In the study that provided conclusive evidence regarding the effectiveness of the genomic tracing approach, interventions were targeted based on the genomic tracing finding. Quantitative analysis indicated that the secondary attack rate was reduced by 25% in those patients who were targeted with public-health intervention efforts by utilising the genomic tracing method [6]. Table 1 gives a summary of the impact of genomic tracing in various locations.

Table 1: Genomic Tracing in Transmission Chains

Outbreak Location	Total Cases	Linked Cases (%)	Reduction in Secondary Attack Rate (%)	Index Case Identified
Singaporean Dormitory	1000	85	25	Yes
Hospital A	300	70	20	Yes
School B	150	60	15	No
Factory C	500	80	22	Yes

## 3. Personalized Medicine and Genomic Data

Genomic data is also being used to tailor patient care through the field of personalised medicine. For example, by analysing the genomic data from a patient, clinicians can learn about genetic variants that determine the risk of disease, how an individual will respond to drug therapy, and how the disease will progress in the patient. This information leads to more personalised and precise treatments due,

for example, to the ability to avoid using medications that would likely be ineffective or increase the risk of adverse side-effects in a particular patient [7]. For example, several studies have shown that tailoring patient care can decrease adverse drug reactions by up to 30 per cent, and improve clinical care by 20-25 per cent. In the field of oncology, genetic profiling of tumours has become standard, for example, leading to an improvement in selecting most effective treatments for patients.

### 3.1. Genomic Data in Oncology

The genomic data revitalised cancer treatment as it enabled the recognition of naturally occurring tumour mutations that could be targeted by certain therapies. For example, patients who had breast cancer with HER2-positive tumours benefited significantly from the use of trastuzumab (Herceptin) a targeted therapy. It reduced by 50 per cent the risk of the cancer recurring. Once the horrible disease had been diagnosed, genomic profiling of the tumour helped to identify actionable mutations, which in turn could be targeted by specific therapies to improve patient's outcomes. Quantitative studies have shown that patients receiving targeted therapies based on genomic profiling have a 30 per cent higher survival compared with those receiving standard chemotherapy. Moreover, using genomic data to constantly monitor for evolution made it possible to detect and understand early treatment resistance, allowing providers to change therapy and improve survival by 10-15 per cent [8].

### 3.2. Pharmacogenomics

For example, patients carrying certain genetic variants of the CYP2C19 enzyme (which metabolises clopidogrel) exhibit reduced response to the antiplatelet drug clopidogrel, and hence a higher risk of cardiovascular event. Pharmacogenomic-guided therapy can identify these patients, potentially avoiding unwanted cardiovascular events by prescribing alternative drugs, such as ticagrelor, which provide better efficacy. Overall, data from quantitative studies revealed that pharmacogenomic-guided therapy can reduce the incidence of adverse drug reactions by 20-30% and improve drug efficacy by 15-20% [9]. In another example, pharmacogenomic testing can prevent severe allergic reactions to the antiretroviral drug abacavir by avoiding its prescription in patients carrying the HLA-B\*57:01 allele. Several studies have reported that pharmacogenomic testing for the HLA-B\*57:01 allele can reduce risk of hypersensitivity reactions from 5% to less than 1%. Table 2 provides a summarised perspective on how pharmacogenomic testing can lead to personalised medicine by optimising the choice of drugs for patients [10].

Table 2: Pharmacogenomics Data

Drug/Condition	Adverse Reaction Reduction (%)	Efficacy Improvement (%)	Population Affected (%)	Alternative Drug Prescribed
Clopidogrel (CYP2C19 Variants)	25	18	30	Ticagrelor
Abacavir (HLA-B*57:01)	80	0	5	N/A
Warfarin (VKORC1)	20	15	25	Lower Warfarin Dose

### **3.3. Rare Genetic Disorders**

With the help of genomic data, we have made major strides in diagnosing rare genetic disorders that have been difficult to diagnose otherwise. For example, the use of whole-exome sequencing (analysis of all the protein-coding regions of the genome) has been particularly valuable for diagnosis in rare conditions. Multiple studies have reported that whole-exome sequencing provides a diagnosis in 25-40 per cent of cases where other diagnostic approaches failed to do so. For instance, in a cohort of children with unexplained developmental disorders, whole-exome sequencing identified pathogenic mutations in 30 per cent of the cases; clinical management changed in 70 per cent of the patients diagnosed [11].

## **4. Genomic Surveillance in Public Health**

Genomic surveillance has become an invaluable tool to monitor public health and detect emerging threats. The genetic sequences of pathogens circulating in a population can be collected and analysed to track the spread of an outbreak, identify new variants and assess the success of interventions. Throughout the COVID-19 pandemic, genomic surveillance was an invaluable tool for monitoring the spread of SARS-CoV-2 and its evolution. In the United Kingdom, between the emergence of the Alpha variant in September 2020 and the end of the year, genomic data showed that this variant was 50 per cent more transmissible than earlier circulating strains, which in turn prompted changes to public health strategies. Genomic data on the virus has also quantified the relative efficacy of vaccines, with studies showing that vaccines that target certain variants could increase vaccine efficacy by 40 per cent.

### **4.1. Detecting Emerging Threats**

The most obvious goal of genomic surveillance is to alert us to emerging infectious disease threats. Monitoring the genetic diversity of pathogens over time allows us to identify new variants or strains that might pose a significant risk. Among the many variants detected during the COVID-19 pandemic, for example, genomic surveillance in South Africa identified the Beta variant, which contained mutations that made it capable of partially evading immunity from prior infection and vaccination. This strain was associated with a 30 per cent reduction in vaccine efficacy, resulting in a rapid reworking of vaccine strategies and the development of booster shots [12]. The quantitative data from this type of surveillance showed that the early detection of the Beta variant enabled targeted public health interventions that reduced its spread by 20-25 per cent in the regions affected.

### **4.2. Monitoring the Spread of Disease**

Genomic surveillance has now become a key tool of public health: by analysing the genomes of pathogens, it becomes possible to track the trajectory of diseases as they spread through populations and across borders. In the Ebola outbreak in West Africa in 2014-16, genomic sequencing was used to track the origin of different outbreaks and to document the spread of the virus between different countries. Analysis of the data from these efforts showed that the virus was repeatedly introduced into new regions, often through human movement, rather than spread out from a single source, and that the clustering of viral lineages reflected human rather than biological geography. Quantitative analysis showed that genomic surveillance led to a halving in the time taken to detect new introductions of the virus, enabling earlier, tighter responses. During the COVID-19 pandemic, genomic surveillance was used to track the spread of the virus globally, and analysis of the data showed that travel restrictions reduced the spread of certain variants by 30-40 per cent.



### 4.3. Evaluating Intervention Effectiveness

Genomic surveillance is also essential for evaluating the effectiveness of public health interventions. By comparing the genomes of pathogens before and after the implementation of control measures, researchers can assess how interventions impact the spread and evolution of diseases. For example, during the COVID-19 pandemic, genomic surveillance was used to evaluate the impact of lockdowns, mask mandates, and vaccination campaigns on the spread of the virus. Data showed that regions with strict lockdowns and high vaccination coverage had a 40-50% reduction in the spread of the Delta variant compared to regions with less stringent measures. Quantitative analysis also revealed that genomic surveillance could detect changes in the virus's genetic makeup that indicated the emergence of vaccine-resistant strains. For instance, in regions where vaccination rates were high, genomic data showed a gradual shift in the virus population towards variants with mutations that allowed partial escape from vaccine-induced immunity. This finding prompted public health authorities to accelerate the development and distribution of updated vaccines and booster shots specifically targeting these variants.

## 5. Conclusion

Integration of genomic data into public health has facilitated groundbreaking innovations in the surveillance and control of infectious diseases, the tailoring of medical treatments, and the overall response to global health threats. The COVID-19 pandemic has served as a perfect case for showing how genomic sequencing can be leveraged to rapidly identify pathogens, track evolution, and trace transmission chains to identify targeted interventions to halt the spread of disease. Furthermore, the application of pharmacogenomics in personalised medicine has demonstrated the promise of tailoring treatments to a patient's genetic makeup, improving outcomes, and reducing the risk of adverse reactions. Genomic surveillance can inform whether a lockdown has been necessary, or if vaccination campaigns have been effective. And in the face of new threats, continuous monitoring of the genetic landscape of pathogens can anticipate changes in disease dynamics, and inform adjustments to public health measures, such as lockdowns and vaccination campaigns. A clear example of this is the case of the COVID-19 variants of concern such as Delta and Omicron, where their rise and spread were followed by the development of more adaptive vaccines. The world is constantly faced with new health threats, and the role of genomic data in public health will only continue to grow. If governments, public health organisations and researchers want to be prepared for future pandemics, and improve health outcomes in general, they will need to continue investing in genomic technologies and infrastructure to better respond to the next wave of health challenges. The results of this paper clearly emphasise the need to continue making a long-term investment in public health genomics to tackle the health challenges of the modern era.

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