EURASIAN EXPERIMENT JOURNAL OF SCIENTIFIC AND APPLIED RESEARCH
(EEJSAR) ISSN: 2992-4146
©EEJSAR Publications Volume 7 Issue 2 2025

Engineering Genetic Testing: Improving Accessibility and Accuracy

Kabiga Chelule Kwemoi

Faculty of Engineering Kampala International University Uganda

ABSTRACT

Genetic testing has emerged as a critical tool in modern medicine, providing insights into inherited disorders, disease predispositions, and personalized treatment plans. Despite significant technological advances—such as next-generation sequencing and expanded variant analysis—barriers persist in terms of equitable access, interpretive accuracy, and regulatory consistency. This paper examines the historical development, current methodologies, and transformative technologies in genetic testing, emphasizing the dual imperatives of accessibility and accuracy. By evaluating ethical, infrastructural, and regulatory challenges alongside telehealth and data standardization opportunities, the paper outlines strategies for optimizing test reliability and reach. The integration of bioinformatics, global collaboration, and patient-centered frameworks holds promise for democratizing genetic testing and ensuring its safe, effective use in clinical and public health contexts.

Keywords: Genetic testing, next-generation sequencing, genomic medicine, accessibility, accuracy, telehealth, bioinformatics.

INTRODUCTION

Genetic testing, also known as genetic screening, examines genes or chromosomes for changes or mutations. Its primary aims are to identify inherited genetic disorders and assess the risk of having offspring with such disorders. Various forms of testing include carrier testing, prenatal diagnosis, newborn screening, presymptomatic testing, and susceptibility testing. Tests may analyze proteins, DNA, RNA, or chromosomes to detect mutations or gauge gene interactions with medications. Genetic tests can cover a wide range of conditions, from single-gene disorders like cystic fibrosis to chromosomal disorders such as Down syndrome and multifactorial disorders influenced by multiple genes and environmental factors, including late-onset conditions like breast cancer and type 2 diabetes. These tests provide crucial insights for diagnosing and managing congenital and acquired disorders. A spectrum of genetic tests exists, from simple diagnostic and carrier screenings to complex analyses for evaluating genetic risks, often limited to well-resourced laboratories due to high costs. Adequate laboratory infrastructure, including bioinformatics and skilled personnel, is critical for advancing genetic testing technologies. Furthermore, local resources are essential for international lab operations in genetic testing, often absent in developing contexts. Prior experience in DNA analysis is advantageous for utilizing next-generation sequencing effectively, highlighting the need for solid infrastructure and support systems [1, 2].

Overview of Genetic Testing

A genetic test is a medical test that examines for abnormalities in chromosomes, genes, or proteins. Genetic tests can identify changes in genes that may cause a genetic disorder, common disease, or even a predisposition to develop a condition later in life. Genetic tests vary in their complexity and can take the form of a simple blood or cheek swab (saliva) test to whole genome sequencing or array comparative genomic hybridization analysis. Genetic tests can provide information about a person's genes and chromosomes, which can lead to a diagnosis, inform prognosis (likely course of the disease), lead to prevention or management of disease, reproductive decisions, or health benefits, uncover some non-paternity situations, and pose a risk of significant distress. Clinical genetic testing aims to provide clinically relevant results to patients who have an established, suspected, or carrier access to relevant disease-causing variants. There are three starting points for a genetic test: an identified variant from a relative or a variant of uncertain significance (VUS), an established clinical condition with a known gene,

and a clinical suspicion based on family or medical history. Cancer is the most prevalent condition for which genetic testing is requested. Some tests are expected to return a positive result based on family history, while others may yield a VUS or negative results, which can lead to diagnostic odysseys. The genetic test results may be complicated by the problematic nature of the variant detected or the patient's clinical presentation, which may involve several or atypical phenotypes from what would be expected with a causative variant. This complexity makes it difficult to generate a comprehensive report using currently available software tools that outputs tangible information relevant to the patient's care. As the genetic testing landscape continues to evolve, including the surge of engineered gene editing and potentially curative genome-wide therapies, there is a need for ensuring fair access to and understanding of genetic tests, enabling their effective incorporation into care pathways, and preventing disease through genetic testing [3, 4].

Historical Context of Genetic Testing

Genetic testing has evolved significantly since the discovery of DNA, becoming widely utilized in clinics over the past two decades due to lower DNA sequencing costs, a result of advances in micro-scale devices and next-gen sequencing technology. This technology can sequence approximately 2 billion base pairs in hours at a cost significantly lower than traditional methods like PCR. Concurrently, advances in human genetics have revealed genetic causes for many diseases, prompting the development of biochemical technologies for genotyping patients. As next-gen sequencing became prevalent, the need arose to effectively communicate and interpret the vast amounts of data generated to facilitate informed medical diagnoses. Individuals with shared ancestry exhibit similar alleles, allowing for insights into susceptibility to tumors, rare diseases, and common ailments within populations. This data can inform predictions about phenotypes through machine learning models. Modern medicine increasingly incorporates genetic information to address pathophysiological functions and environmental factors affecting health. Genomic data is obtained via genotyping arrays, which detect numerous variants with low false positives, or through direct sequencing, which provides comprehensive genomic insights but may face availability challenges in diagnostics. Despite the growth and potential of genetic testing, significant discrepancies exist in its application regarding oversight, regulation, and accountability. Different statutes apply depending on the timing of tests marketed, with those before 2004 falling into a regulatory vacuum, complicating resolution without new legislation. These policy concerns differ in their regulatory approaches and usability, illustrating both strengths and weaknesses in the oversight landscape for genetic testing services in the United States as commercial offerings continue to expand [5, 6].

Technological Advances in Genetic Testing

Rapid advances in DNA sequencing technology and genome editing are revolutionizing genetic and genomic medicine broadly and creating opportunities and challenges for health care and society. New genetic tests and treatment modalities can now be implemented within the clinic. In addition to existing tests for pathogenic variants, a new generation of tests that assess variant effect on phenotype can now be offered. However, rapid changes present challenges in turn: new discoveries often outpace understanding and practice. These changes may be especially impactful for currently available and 'ready-for-use' tests and treatments. These trends are reviewed and their implications called to attention. Nucleotide sequence analysis is an expanding set of methods that offer information on gene structure and sequence. A variety of in-house and 'commercial' genetic tests are currently offered to identify base (point) substitutions. Those tests include codon-tuple sequencing tests to identify single variants. Testing modes range from large single tests for known mutations or large panels of genes down to individual tests for a specific mutation. Target capture acquisition methods are also available that allow screening candidates among panel genes and broader samples, ranging from targeted capture of groups of genes to exome to whole-genome capture 1. These testing technologies are readily implemented in medicine, but the interpretation and assessment of significance of layers of variants is still evolving. Interpretation is an evolving process that is most rapidly improving for single variants and often outpaces testing. Efforts to share a large portion of knowledge created are underway, with various databases. However, for many variants, data is sparse. The resulting uncertainty will take time to resolve, so that tests with uncertain significance must be offered until a growing consensus clarifies classification. This provides a major challenge for clinical geneticists interpreting tests and counseling families [7, 8].

Current Methods of Genetic Testing

Genetic testing is currently used for many applications, including single gene testing, carrier testing, congenital disorder testing, and hereditary cancer testing. Most of these tests are done based on a

previous discovery regarding a specific genetic disorder. Certain mutations can be looked for in specific genes based on personal family history or examination findings. Gene panels containing a set of specific genes related to hereditary cancers or congenital disorders are also widely available and vary in both cost and complexity. A variety of genetic tests have now been developed in clinical laboratories to identify a subset of genetic causes of neurodevelopmental problems in available patient populations. The vast majority of genetic tests in use today rely on next-generation sequencing technologies, comprising whole-exome or whole-genome sequencing, targeted-gene panels, copy number variations detection, and multiplex ligation-dependent probe amplification and array comparative genomic hybridization assays for microarray analysis for CNVs. Current genetic tests differ greatly in complexity, from one or two individual genes with preselected pathogenic mutation analyses to large gene panels containing a set of specific genes with varying permutations of patient capture technologies, bioinformatics, and laboratory validation. A great number of bioinformatic analyses have been developed and continue to be improved to both identify variants of novel candidate genes and classify channel variants in well-known genes. Current clinical procedures require confirmation of detected sequence variations by independent genomics methods in a clinical laboratory. Sanger sequencing or other lab-based methods with previously established follow-up tests can be utilized to confirm the vast majority of detected variants that can affect genome sequencing and this analysis is also necessary for quality control. Many gene mutations that were originally discovered in small family studies have been detected through later large-scale genome sequencing [9, 10].

Challenges In Genetic Testing

The rapid advancement of technology and scientific research has led to more genetic tests for consumers, but the US FDA and CMS struggle to keep pace. Genetic tests vary in analyses by laboratories, complicating quality assurance and patient safety. Other countries have enacted regulations, while the US sees a conflict between premarket review and self-regulation for laboratory-developed tests (LDTs), neither effectively safeguarding patients. The FDA has attempted to regulate LDTs but met resistance from labs, risking the release of tests with inadequate oversight, causing inaccurate results and harm. Key factors in testing performance include test selection based on analytes, laboratory quality, and costs. Genetic tests can differ in gene panels, with genes being added or removed over time. The type of genetic variant tested can vary, with some countries only testing for common mutations. Inclusion of introns might lead to false positives, and false rates are influenced by the testing technology and personnel expertise. The primary challenge for genetic testing companies is ensuring accurate detection of disease-related genetic variations; failure compromises reliability and liability. To enhance accuracy, companies may expand gene panels and some ethically send detailed reports to consumers. However, this doesn't always guarantee result accuracy. Variants of uncertain significance (VUSs) lack uniformity among companies; some include them in reports while others do not. To limit legal liability, companies often avoid false negatives in VUSs, although issues persist. Pathogenic variants pose a higher risk, prompting companies to be cautious to mitigate potential lawsuits [11, 12].

Ethical Considerations

The Technology, Evaluation, and Impact of Genetic Testing conference was hosted by the National Human Genome Research Institutes in 2018, and today's statistics on morbidity and mortality of tests speak to the need to ensure accurate genetic tests that bring appropriate benefits and do not harm individuals. In the 2018 Vision, Visionary Topic 3 emphasized the goal of "Ensuring that genetic tests are accurate and accessible." The subtopics were the "infrastructure for accurate genetic tests," "spanning from variant classification to patenting;" the "feedback loops for assessing how well tests perform," and "ensuring that tests are available to patients." They were promised that there would be a resulting white paper summarizing these discussions and issuing call for specific actions. Several features are specific to tests ordering and result interpretation, addressing concerns about test validity, safety, and efficacy, and ensuring patient and clinician-centric experience in the context of pre- and post-test counseling. Clinicians have a role in ordering and interpreting tests, and test developers and laboratories have a responsibility to ensure that tests are valid and provide clinically relevant information. Alternative healthcare models, populations, or societies may reasonably provide only limited test ordering, or interpretation more widely via certified laboratory personnel, internet tools or tele-health applications with some tests also being ordered without healthcare provider involvement. Considering some would prefer this, it would help drive the assessment of genetic tests forward and catch many tests that do not yet meet required standards for the above features. This requirement would result in a tipping point

where action needed to ensure accurate genetic tests shifts more toward the public sector in some situations defined above. It is understood that required standards would provide a hurdle for some expanding into the US market, requiring most genetic testing companies to unexpectedly and unpreparedly redesign their tests, testing choices, and platforms. Requiring verification of basic but broad criteria is proposed using a framework to assess tests' safety, efficacy, and claims, relying only upon good scientific evidence and the availability of information and results, acting in good faith and benefitting all but punitive [13, 14].

Regulatory Framework

The November meeting and scientific presentations of the INGENE ASIA group provided a productive forum for the participants to discuss ideas and findings in the area of genetic evaluation of dairy cattle. The wide diversity of environments and demographics of the countries represented in the group provided many perspectives on both issues to be overcome and strategies to succeed. While not all concerns during implementation of genetic evaluation systems were presented by participants from all countries, the scientific presentations addressed potential issues of importance to all groups and provided a starting point for future efforts. There is a belief within the industry in Asia that establishing a genetic evaluation system will provide immediate and long-term benefits. It is expected that further participation in the group will provide the tools and skills to evaluate the current genetic potential of dairy cattle in the various countries and to identify priority areas for further strategic steps toward improved dairy facilities. Collective work in such a forum will ensure that time and fiscal resources are shared to obtain a more effective outcome. In addition, the establishment of a technical alliance between countries in the Asian region is expected to be a further benefit from such activities. Despite the initially overwhelming task list, it is helpful that experiences and necessary tools are available from more developed countries. For various environmental, cultural, and socio-economic reasons, genetic evaluations and the development of dairy herdbook services differ sharply between countries participating in the Asian INGENE conference. ANC has been involved in the promotion of the genetic evaluation of dairy cattle through the establishment of an international reference database. A current focus is on the genetic evaluation of local breeds feasibility to assist and accelerate current genetic evaluations of local breeds. Maintaining the INGENE network of genetic evaluation representatives has been seen as a primary target. INCHEE was seen as a useful tool in expanding information exchange regarding pasture-based dairy [15, 16].

Improving Accessibility

Accessibility of genetic testing continues to be a critical challenge, with many eligible candidates faced with barriers to participation. Genetic testing accessibility can be broadly defined as the facilitation of systems, opportunities, or situations that result in genetic testing. In previous research, a conceptual framework for assessing the accessibility of health services was contextualized for genetic testing. This framework includes 8 components of accessibility: approachability, acceptability, availability, affordability, awareness, approvability, ability, and actionability. It is important to note that affordability, approvability, and actionability are defined separately from aspects of access that are focused specifically on patient insurance. Recent research on the potential applicability of the internet to enhance accessibility suggests that the internet may address many of the components of accessibility, including those primarily existing beyond patient insurance. An assessment of the usability of an internet-based health communication system was conducted. The purpose of this assessment was to evaluate accessibility of a multistep genetic testing communication system. In addition, several recommendations were utilized to reach an accessibility assessment more representative of the internet-wide genetic testing communication system. The recommendations generated through this assessment focused primarily on the organization and usability of the system. Further refinement is accordingly needed before use in the main study. The usability of the multistep communication system could effectively facilitate genetic testing implementation in the future, thereby increasing access to genetic testing, particularly among populations currently underserved. Current research highlights the potential for telehealth to increase access to genetic assessment, with recommendations to prioritize these options during the transition back to standard care. Applications of telehealth address critical barriers and improve accessibility. The potential of telehealth services, particularly in the context of genetic health, has gained renewed attention during the pandemic. Although telehealth has gained support from patients and became a standard of care among many health services during the pandemic, it remains uncertain whether telehealth will continue to be widely utilized once elective care resume [17, 18].

Enhancing Accuracy

Methods used to perform genetic testing can now accept unprecedented quantities of DNA sequence data in the form of multiple genetic variants or mutations. Most of the sequencing data analysis component can be somewhat automated, resulting in the potential for the same test to be processed on different facilities with different performance pressures and priorities. The output files vary in content, structure, and presentation, and multiple variations of these genetic variants can be given along with the diagnosis. Should this confound a test, the collection and reconciliation of available external clinical, pedigree and confirmatory test information should be considered as methods to anchor the test against its clinical purpose once again? The ability of the high-throughput testing technology to offer a similar test for application at the local level is daunting, and in the long run, a test will need external standards and reference samples made available in a quality-controlled, accessible, and indexed manner to assist in accurate interpretation, correct assay performance, and reproducibility. Presently, there is a fair amount of debate that public telecommunication is not ready; although this is an extreme view advocating local centralization of testing processes, it is hard to publicly reject the principle of testing distributed to where the analyst/effect person resides. It is also true that select molecular diagnoses are difficult to contest but relatively easy to either inaccurately configure or misinterpret qPCR type tests. It is possible that like clinical tests like antibody tests being performed at local facilities, diagnostic pathways will be designed in such a way that the greater test result impacts are anchored more into some reference facilities. The continual pressure to reduce the number of tests at the more theoretic breadth of the test while containing genetic uncertainties should be cognizant of the confounding lack of uniformity when designing or recommending comprehensive tests into new molecular areas of interest in specific conditions. A lesson learned is that reference test standards, at a level of knowledge both clinical and personal, should be built into the recommendations. Making the recommendations available to all interested parties early, prior to the creation of knowledge silos, should improve the chances of maintaining public trust in genetics as technology solutions evolve [19, 20].

Case Studies

In October 2013, a 22-year-old female presented with severe intellectual disability, developmental delays, and neuromotor impairment. Abnormal facial features included drooping eyelids and a flat occiput. Blood work was unremarkable. Patient was evaluated at another institution and whole genome sequencing was performed but did not provide a diagnostic result. The patient was referred for further evaluation. The consultation with the genetics team suggested possible deletion of chromosome 1p, Smith–Magenis syndrome, nonsyndromic holoprosencephaly or a possible pathogenic variant in PGBD5. Whole exome sequencing was performed on the proband and parents and revealed a likely pathogenic variant in PGBD5. Clinicians submitted a novel variant for re-classification which was included in the updated information. The operator received a 150X average coverage exome sequencing data from a female patient. The analysis plan included AGVP but the clinic team rerouted the analysis to integrated pipeline, and the issue was solved by using good reference mapping. One candidate deletion was independently detected by screening approach and later confirmed by ad hoc follow-up NGS and Sanger sequencing. The proposed variant of DMD gene was classified pathogenic and declined due to lack of submission support from the treating clinician. The operator received a 154X average coverage exome sequencing data from a 2-year-old male with poor growth and global developmental delay. The analysis plan included AGVP as the clinical team submitted readable log and all phenotypic evidence on interactive web page. With the implementation of AGVP, biallelic pathogenicity of the GRIN2A variant was intriguingly supported by genetic evidence. The proposed variant was classified as pathogenic and later submitted [21, 22].

Future Directions in Genetic Testing

Over the next 2–5 years, advancements in genetic testing will likely arise from increased competition and new sequencing technologies. Improvements in information transfer, organization, interpretation, and communication will enhance return-of-value efforts. Genetic testing and predictive genotyping will expand due to more rigorous evaluations supporting regulatory decisions. The field of epigenetics is developing tests for genetic predisposition to diseases, influencing insurance coverage as well. Competition among companies testing genes linked to cancer and other diseases will press for improved quality, value, and reduced costs. Government and state analyses, as well as input from the insurance industry and academia, will significantly impact the availability, regulation, and delivery of genetic testing. These changes will alter the current genetic testing landscape for companies, patients, and

physicians. Further advancements will be driven by technology that improves genomic laboratory tests. New methods will enable rapid interrogation of the complete human genome, reducing analysis time from days to hours at costs nearing $100. This will empower patients and physicians to derive actionable insights from extensive genomic data in a clinically relevant manner. Health systems' Electronic Health Records (EHRs) will evolve into interactive communication tools, delivering interpretive narrative reports and actionable predictions. Meeting emerging demands for service, organization, and interpretation will be essential for integrating genomic tests into patient care. Advances in clinical information collection and storage will create context for genomic data, making epigenomic information more accessible. Most tested individuals will receive normative information about their genome's 3 billion letters, often including variants of uncertain significance or those conferring modest risk. However, very few will encounter clinically actionable risks that exceed the uncertainty and potential unintended consequences associated with such tests [23, 24].

Impact of Genetic Testing on Healthcare

The ongoing and accelerating revolution of genome testing will have a deep impact on healthcare services, patients, and doctors. This review summarises how genetic tests differ in nature, cost, and complexity, and how the electronic health record (EHR) might facilitate communication between doctors, patients, and institutions necessary to advance genomic medicine. There are four EHR functions that can potentially lead to improvements in the actual clinical use of genetic tests: 1) Data Integration; 2) Clinical Decision Support; 3) Workflow Adjustment; and 4) Performance Tracking. This rapid expansion in genome testing will have a significant effect on the care provision landscape and may offer new services to patients and doctors alike. Genetic tests are a diverse group of medical procedures, requiring different expertise, technology development, implementation, and use. Genetic tests differ greatly in the number of genes tested. A test could be medically equivalent to a single base pair change in a few genes, or to sequencing hundreds of genes at once. The utmost complexity test is, of course, whole genome sequencing. Not only do tests differ greatly in the genes tested, they may also differ in the variation type screened. Thus, one could simply check for a known genome nucleotide substitution, or screen for gene deletions/duplications, large genomic rearrangements, or epigenetic alterations. Tests differ substantially in the nature of the target(s). Target(s) can consist of nucleic acids, proteins, or even classical histological or enzymatic examination, revealing no molecular aberration but an altered pattern of activity [25, 26].

Patient Perspectives

The Rapid Genetic Tool was provided for free to the first set of patients who participated in the pilot study. Patient participants were alerted that they could access the tool via a secure email. After participants accessed this website and connected to RapidGenetic through a secure portal, they were presented with a disclaimer about the approximate time commitment to use the tool, the fact that the tool had been designed specifically for its intended use with genomic test results, and that only testing results presented in the specific format generated by the study platform laboratory would be accepted. Once this disclaimer was acknowledged, patients were presented with instructions for how to submit their genetic test results. Patients were instructed to download their genetic test results as a text file, open this file, select and copy the text beginning with "Result," and paste that text into a designated text entry box in RapidGenetic. The tool was then activated by pressing a button labeled "Enter Genetic Test Results." Patients were informed to expect a waiting period while their results were processed and that they would subsequently be shown options for viewing test results in full or in brief. After explaining these processes, the assessment stage of the tool was demonstrated with a de-identified example of a BRCA1/2 test report. Patients were then given the opportunity to engage with the RapidGenetic Tool on their own. The researcher remained in the same room and answered questions as needed, but did not provide active assistance in using the tool. Independent engagement with the tool lasted approximately 20-45 minutes for each patient participant, and afterward, patients described their experiences using the tool in an interview. Similar to the prior visits, these follow-up interviews were audio-recorded, transcribed, and coded for thematic analysis. Everyone reported being able to use the tool on their own without much difficulty. Many patients also made general positive comments about their experiences, such as feeling that the tool is "cutting-edge," "really neat," and "awesome." One patient, who had worked for many years in hospital IT, specifically said, "I could tell that there was a lot of thought put into software—I think it is well-designed," and another patient remarked, "I think it's amazing that this exists" [27, 28].

Collaboration In Genetic Research

Genetic research is a collaborative multistep process involving various stakeholders. It begins with generating a genetic variant. Bioinformaticians analyze data from high-throughput sequencing, while researchers utilize DNA, transcriptomics, and proteomics data to identify novel genes, establish phylogenetic trees, and uncover interactions between coding and non-coding genes. Statistical geneticists conduct marital, logistic, Bayesian, and combinatorial tests to produce candidate variant lists and filtering pipelines. Compliance can be developed in-house or purchased from other research groups. Geneticists and biochemists perform Sanger screenings, categorizing candidates by disease associations before creating constructs for functional studies. Candidates are further evaluated via in-silico analysis to identify the best options. Experimental biologists create patient and wild-type cell lines in 2D and 3D for cellular perturbations. The Eco-Sciences platform conducts proteomics-level tests like ELISA, western blot, and mass spectrometry while developing necessary parsing software. Computational biologists analyze data and conduct mutational protein structure runs, validating variants across populations and pedigrees. A document summarizing findings is prepared, reviewed by co-authors and non-scientific personnel, and subjected to a verification protocol before submission to journals. Authors respond to reviewer comments post-submission, converting replies to journal formats and providing raw data as needed. Revisions are usually approved on the first submission. Genetic testing results are efficiently transmitted to patients. Genetic stakeholders play vital roles in this process, starting from collecting family history and patient information through intake forms. Genetic education materials are prepared by clinics for patients and physicians. Testing candidates are discussed by geneticists before proposals are sent to larger committees. Data platforms for genetic counseling are established, and inheritance patterns are analyzed for families. Group software engineers design informative region panels for testing family members, converting them to printable formats [29, 30].

CONCLUSION

Genetic testing is poised to reshape healthcare through early detection, tailored treatments, and predictive insights, yet its full potential hinges on two critical factors: accuracy and accessibility. Technological advances have significantly reduced sequencing costs and enhanced test capabilities, but disparities in access—particularly in low-resource settings—remain a pressing issue. Moreover, the complexity of variant interpretation and inconsistent regulatory oversight challenge test reliability and clinical utility. Ethical practices, informed consent, and standardized quality control must be prioritized to build public trust. Enhanced telehealth delivery, improved data sharing, and international collaboration can bridge existing gaps, making genetic testing not just a privilege for the few but a powerful diagnostic tool for all. As we continue engineering the future of genetic testing, the focus must remain on equity, efficacy, and informed application.

REFERENCES

1. Horton RH, Lucassen AM. Recent developments in genetic/genomic medicine. Clinical Science. 2019 Mar;133(5):697-708.
2. Kotze MJ. Application of advanced molecular technology in the diagnosis and management of genetic disorders in South Africa: the new millennium. South African Medical Journal. 2016 Jun 1;106(sup-1):114-8.
3. Marwaha S, Knowles JW, Ashley EA. A guide for the diagnosis of rare and undiagnosed disease: beyond the exome. Genome medicine. 2022 Feb 28;14(1):23.
4. Lopes KD, Snijders GJ, Humphrey J, Allan A, Sneeboer MA, Navarro E, Schilder BM, Vialle RA, Parks M, Missall R, van Zuiden W. Genetic analysis of the human microglial transcriptome across brain regions, aging and disease pathologies. Nature genetics. 2022 Jan;54(1):4-17. nih.gov
5. Liu W, Li L, Jiang J, Wu M, Lin P. Applications and challenges of CRISPR-Cas gene-editing to disease treatment in clinics. Precision clinical medicine. 2021 Sep;4(3):179-91.
6. Anstee QM, Castera L, Loomba R. Impact of non-invasive biomarkers on hepatology practice: Past, present and future. Journal of hepatology. 2022 Jun 1;76(6):1362-78.\
7. Chen E, Facio FM, Aradhya KW, Rojahn S, Hatchell KE, Aguilar S, Ouyang K, Saitta S, Hanson-Kwan AK, Capurro NN, Takamine E. Rates and classification of variants of uncertain significance in hereditary disease genetic testing. JAMA network open. 2023 Oct 2;6(10):e2339571-. jamanetwork.com

8. Roberts GH, Partha R, Rhead B, Knight SC, Park DS, Coignet MV, Zhang M, Berkowitz N, Turrisini DA, Gaddis M, McCurdy SR. Expanded COVID-19 phenotype definitions reveal distinct patterns of genetic association and protective effects. Nature genetics. 2022 Apr;54(4):374-81. nature.com
9. Naito Y, Aburatani H, Amano T, Baba E, Furukawa T, Hayashida T, Hiyama E, Ikeda S, Kanai M, Kato M, Kinoshita I. Clinical practice guidance for next-generation sequencing in cancer diagnosis and treatment (edition 2.1). International Journal of Clinical Oncology. 2021 Feb;26:233-83. springer.com
10. Akacin I, Ersoy Ş, Doluca O, Güngörmüşler M. Comparing the significance of the utilization of next generation and third generation sequencing technologies in microbial metagenomics. Microbiological Research. 2022 Nov 1;264:127154.
11. Javitt G, Hudson K. Public Health at Risk: Failures in Oversight of Genetic Testing Laboratories. Genetics and Public Policy Center; 2006.
12. Chin HL, Goh DL. Pitfalls in clinical genetics. Singapore medical journal. 2023 Jan 1;64(1):53-8.
13. Emmerich CH, Gamboa LM, Hofmann MC, Bonin-Andresen M, Arbach O, Schendel P, Gerlach B, Hempel K, Bespalov A, Dirnagl U, Parnham MJ. Improving target assessment in biomedical research: the GOT-IT recommendations. Nature reviews Drug discovery. 2021 Jan;20(1):64-81. nature.com
14. Vandereyken K, Sifrim A, Thienpont B, Voet T. Methods and applications for single-cell and spatial multi-omics. Nature Reviews Genetics. 2023 Aug;24(8):494-515. nih.gov
15. Hoban S, Archer FI, Bertola LD, Bragg JG, Breed MF, Bruford MW, Coleman MA, Ekblom R, Funk WC, Grueber CE, Hand BK. Global genetic diversity status and trends: towards a suite of Essential Biodiversity Variables (EBVs) for genetic composition. Biological Reviews. 2022 Aug;97(4):1511-38. wiley.com
16. Wang JH, Gessler DJ, Zhan W, Gallagher TL, Gao G. Adeno-associated virus as a delivery vector for gene therapy of human diseases. Signal transduction and targeted therapy. 2024 Apr 3;9(1):78. nature.com
17. Jenkins BD, Fischer CG, Polito CA, Maiese DR, Keehn AS, Lyon M, Edick MJ, Taylor MR, Andersson HC, Bodurtha JN, Blitzer MG. The 2019 US medical genetics workforce: a focus on clinical genetics. Genetics in Medicine. 2021 Aug 1;23(8):1458-64. sciencedirect.com
18. Filip R, Gheorghita Puscaselu R, Anchidin-Norocel L, Dimian M, Savage WK. Global challenges to public health care systems during the COVID-19 pandemic: a review of pandemic measures and problems. Journal of personalized medicine. 2022 Aug 7;12(8):1295. mdpi.com
19. Di Resta C, Pipitone GB, Carrera P, Ferrari M. Current scenario of the genetic testing for rare neurological disorders exploiting next generation sequencing. Neural Regeneration Research. 2021 Mar 1;16(3):475-81. lww.com
20. Doricchi A, Platnich CM, Gimpel A, Horn F, Earle M, Lanzavecchia G, Cortajarena AL, Liz-Marzán LM, Liu N, Heckel R, Grass RN. Emerging approaches to DNA data storage: challenges and prospects. ACS nano. 2022 Oct 18;16(11):17552-71. acs.org
21. Li SS, Zhai XH, Liu HL, Liu TZ, Cao TY, Chen DM, Xiao LX, Gan XQ, Cheng K, Hong WJ, Huang Y. Whole-exome sequencing analysis identifies distinct mutational profile and novel prognostic biomarkers in primary gastrointestinal diffuse large B-cell lymphoma. Experimental Hematology & Oncology. 2022 Oct 15;11(1):71. springer.com
22. Yamada M, Keller RR, Gutierrez RL, Cameron D, Suzuki H, Sanghrajka R, Vaynshteyn J, Gerwin J, Maura F, Hooper W, Shah M. Childhood cancer mutagenesis caused by transposase-derived PGBD5. Science Advances. 2024 Mar 22;10(12):eadn4649. science.org
23. Fang W, Wu J, Cheng M, Zhu X, Du M, Chen C, Liao W, Zhi K, Pan W. Diagnosis of invasive fungal infections: challenges and recent developments. Journal of biomedical science. 2023 Jun 19;30(1):42. springer.com
24. Aragona M, Haegi A, Valente MT, Riccioni L, Orzali L, Vitale S, Luongo L, Infantino A. New-generation sequencing technology in diagnosis of fungal plant pathogens: a dream comes true?. Journal of Fungi. 2022 Jul 16;8(7):737. mdpi.com
25. Ingber DE. Human organs-on-chips for disease modelling, drug development and personalized medicine. Nature Reviews Genetics. 2022 Aug;23(8):467-91.

26. Ji P, Ye J, Mu Y, Lin W, Tian Y, Hens C, Perc M, Tang Y, Sun J, Kurths J. Signal propagation in complex networks. Physics reports. 2023 May 18;1017:1-96. [HTML]
27. Denommé-Pichon AS, Vitobello A, Olaso R, Ziegler A, Jeanne M, Tran Mau-Them F, Couturier V, Racine C, Isidor B, Poë C, Jouan T. Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. European Journal of Human Genetics. 2022 May;30(5):567-76. nih.gov
28. Imafidon ME, Sikkema-Raddatz B, Abbott KM, Meems-Veldhuis MT, Swertz MA, Van der Velde KJ, Beunders G, Bos DK, Knoers NV, Kerstjens-Frederikse WS, van Diemen CC. Strategies in rapid genetic diagnostics of critically ill children: experiences from a Dutch university hospital. Frontiers in Pediatrics. 2021 May 31;9:600556. frontiersin.org
29. Xiong E, Jiang L, Tian T, Hu M, Yue H, Huang M, Lin W, Jiang Y, Zhu D, Zhou X. Simultaneous dual-gene diagnosis of SARS-CoV-2 based on CRISPR/Cas9-mediated lateral flow assay. Angewandte Chemie International Edition. 2021 Mar 1;60(10):5307-15. [HTML]
30. Yen HL, Sit TH, Brackman CJ, Chuk SS, Gu H, Tam KW, Law PY, Leung GM, Peiris M, Poon LL, Cheng SM. Transmission of SARS-CoV-2 delta variant (AY. 127) from pet hamsters to humans, leading to onward human-to-human transmission: a case study. The Lancet. 2022 Mar 12;399(10329):1070-8. thelancet.com

CITE AS: Kabiga Chelule Kwemoi (2025). Engineering Genetic Testing: Improving Accessibility and Accuracy. EURASIAN EXPERIMENT JOURNAL OF SCIENTIFIC AND APPLIED RESEARCH, 7(2):8-16