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Bridging the Genomic Divide: Translating Research into Clinical Practices in LMICs

Sumreena Mansoor

sumreena.scm@stmu.edu.pk

Shifa Tameer-e-Millat University

Palwasha Arif

Shifa Tameer-e-Millat University

Anoshia Javed

Shifa Tameer-e-Millat University

Warda Gul

Shifa Tameer-e-Millat University

Najmun Nahar

Shifa Tameer-e-Millat University

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Abstract

Background

Pakistan carries one of the highest burdens of recessive genetic disorders in the world, driven by a consanguinity rate exceeding 60% of all marriages. Despite this, access to diagnostic genetics services remains severely limited. Patients face long waits, prohibitive costs, cultural resistance, a shortage of trained professionals and no coordinating national policy. These overlapping challenges cannot be solved in isolation—they require the kind of cross-disciplinary conversation that rarely happens within a conventional conference.

Objective

To evaluate immediate learning outcomes and explore participant perspectives regarding genomics implementation challenges in LMIC settings.

Methods

A 3-hour pre-conference workshop titled “Challenges and Solutions in Genomics Practice in LMICs” was conducted during Biocon 2025 at Shifa College of Medicine, Islamabad, Pakistan. The workshop included lectures, panel discussions, videos, interactive sessions, and role-based small-group discussions. Pre-test and post-test multiple-choice assessments were administered using Socrative. Participants were assigned to thematic discussion groups addressing infrastructure and cost constraints, ethical and cultural barriers, policy and collaboration gaps, workforce limitations, and translational research and education gaps. Quantitative findings were analyzed descriptively using response frequencies and proportions, while qualitative responses were synthesized thematically. Ethical approval was obtained from the Institutional Review Board and Ethics Committee of (IRB #461 – 25).

Results

Sixty-six participants registered for the workshop, comprising clinicians (n = 14), faculty members (n = 19), students (n = 24), and researchers (n = 9). Fifty-seven participants completed the pre-test and 52 completed the post-test. Baseline findings indicated moderate-to-high awareness of core genomics concepts. Post-test results demonstrated an overall increase in correct-response proportions across most domains, particularly in population-specific genomics, data sharing, workforce development, and curriculum integration, suggesting immediate knowledge gain following the intervention. Qualitative thematic analysis identified five key implementation priorities: infrastructure and affordability, ethical and

cultural considerations, policy and collaborative frameworks, workforce development, and integration of genomics into clinical practice and medical education.

Conclusions

Even a brief, well-structured workshop can consolidate knowledge and generate actionable ideas. The findings point to an urgent need for phased diagnostic pathways, community-centered engagement strategies, a national rare disease registry, formal recognition of genetic counseling as a profession, and genomics integrated from day one into medical training. These are not aspirational goals—they are achievable, and this workshop showed that the will to pursue them exists across every level of the health system.

INTRODUCTION

Every year, children in Pakistan are born with conditions that could have been identified, managed, or prevented—if only the right systems were in place. Pakistan has one of the highest rates of consanguineous marriage in the world: more than 60% of unions occur between relatives, and in some communities that figure approaches 85% [1]. This deeply embedded cultural practice significantly elevates the risk of autosomal recessive disorders, including thalassemia, phenylketonuria, congenital adrenal hyperplasia, and a wide range of lysosomal storage diseases [2, 3]. The country's genetic disease burden, by any measure, is severe.

Yet the infrastructure to address it is fragile. Families seeking a genetic diagnosis often find themselves at the start of what is known in the field as a 'diagnostic odyssey'—a prolonged, exhausting, and frequently fruitless search for an answer, in which they move from specialist to specialist, city to city, sometimes over years, without anyone connecting the dots [4]. In many LMIC settings, the average time to a rare disease diagnosis exceeds five years [5]. In Pakistan, where primary care barely exists in rural areas and tertiary genetics services are concentrated in a handful of urban centres, that timeline is often longer still.

When a diagnosis is finally reached, new obstacles appear. Whole exome sequencing (WES) and targeted next-generation sequencing (NGS) panels—now standard tools in high-income country genetics clinics—are financially out of reach for most Pakistani families [6]. Even when sequencing is possible, interpreting the results is treacherous: South Asian populations are substantially under-represented in global variant databases like gnomAD, meaning that a variant found in a Pakistani patient is far more likely to be classified as a 'variant of uncertain significance' (VUS) than an equivalent finding in a European patient would be [7, 8]. The science is there; the infrastructure to apply it equitably is not.

Underneath these technical barriers lie deeper ones. Cultural stigma and religious misconceptions around genetic testing remain widespread, particularly in rural areas [9, 10]. Families fear that a genetic diagnosis will bring shame, disrupt marriage prospects, or contravene their beliefs. There is a chronic

shortage of trained genetic counselors across the country, and the profession is not formally recognised within the Pakistani healthcare system [11, 12]. Research institutions and clinical services rarely communicate with each other, let alone share data. And at the policy level, there is still no national rare disease framework to bring any of this together [13].

Addressing these intertwined problems requires more than a literature review or a policy brief. It requires the people who live and work within the system—clinicians, researchers, students, educators—to sit together, identify what is actually getting in the way, and think creatively about what could realistically be done. Pre-conference workshops, when designed well, can do exactly this. By combining structured knowledge assessment with participatory group activity, they create a rare opportunity to simultaneously measure what stakeholders know, what they are missing, and what they are ready to act on [14].

To our knowledge, no prior study has used this kind of structured workshop format specifically to evaluate stakeholder knowledge and generate practical solutions for medical genetics barriers in Pakistan. This paper fills that gap. It presents the findings of a pre-conference workshop attended by 52–57 participants across five professional groups, reports what participants knew before and after the sessions, and synthesises the recommendations they produced in their own words.

METHODS

Study Design

This was a cross-sectional, mixed-methods educational study embedded within a pre-conference workshop. Quantitative data came from a ten-item pre-test and a parallel ten-item post-test. Qualitative data came from structured group activity sheets completed collaboratively by five groups of participants. The workshop took place in Pakistan and received ethical clearance from the organising institution. Written informed consent was obtained from all participants prior to any data collection.

Participants

Participants were recruited through conference registration and represented a range of professional backgrounds. Five groups were constituted by professional role: Group 1 (researchers), Groups 2 and 3 (students), Group 4 (faculty), and Group 5 (clinicians). Pre-test responses were received from 57 participants; 52 participants completed the post-test. The groups were colour-coded in different colours to represent each group.

The composition of the workshop participants is summarised in Table 1.

Table 1
Participant Composition by Professional Role and Workshop Phase

Category	Subgroup / Description	n
Participant role	Researchers (Group 1)	9
	Students (Groups 2 & 3)	20
	Faculty (Group 4)	14
	Clinicians (Group 5)	14
Pre-test respondents	All participants	57
Post-test respondents	All participants	52
Workshop setting	Pre-Conference BIOCON 25 Workshop, Shifa College of Medicine Islamabad, Pakistan	—

Knowledge Assessment Instruments

A 10-item pre-test and a 10-item post-test were developed by workshop faculty and administered online through Socrative. Each correct response carried one mark.

The ten-item multiple-choice pre-test was developed to assess baseline knowledge across domains relevant to medical genetics in LMICs: the diagnostic utility of genomics, the epidemiology of consanguinity-driven disease, the concept of the diagnostic odyssey, the role of NGS, variant interpretation challenges, standards for variant reporting (ACMG/AMP), and strategies for building sustainable genomics infrastructure. Each item had four response options with one correct answer.

The post-test comprised ten parallel but non-identical items, extending into domains such as endogamy versus consanguinity, system-level versus individual barriers, VUS counseling ethics, global collaboration obstacles, and genomics in medical education. Both instruments were developed by the workshop's scientific committee and independently reviewed for content validity by two senior clinical geneticists.

Group Activity

Five thematic scenarios were designed in advance, each corresponding to a well-documented challenge in LMIC medical genetics [6, 9, 13].

Participants were assigned to five thematic scenarios:

1. Infrastructure and cost constraints
2. Ethical and cultural barriers

3. Policy and collaboration gaps
4. Human resource limitations
5. Translational research and education gaps

One scenario was assigned to each group. Participants worked through a structured activity sheet with five guiding questions per theme, spending 30–40 minutes in deliberation before a comprehensive presentation. Group responses were submitted through Socrative and later compiled for analysis. All responses were transcribed and independently reviewed by two members of the organizing team using conventional content analysis [15].

Data Analysis

Quantitative responses were summarized as frequencies and correct-answer percentages per item. For the qualitative data, recurring themes were identified within and across groups and mapped against the five predefined thematic domains. Illustrative responses are presented as written by participants, with minor edits for legibility only.

Ethical considerations

Ethical approval

was obtained from the Institutional Review Board and Ethics Committee of (IRB# 461 – 25; approval date: 21 October 2025). Written/electronic informed consent was obtained from all participants prior to participation.

RESULTS

What Participants Already Knew: Pre-Test Findings

The pre-test was completed by 57 participants. On the whole, the cohort demonstrated solid foundational knowledge—perhaps unsurprisingly, given that a conference-attending group tends to be self-selected for engagement with the field. The strongest performances were on: the hidden burden of genetic disease in Pakistan (94% correct), the diagnostic utility of genomics as limited by absent locally validated variant databases (93%), and ACMG as the primary body for variant interpretation standards (91%).

The most revealing gap was in variant interpretation. While 72% correctly identified limited representation in global reference databases as the core challenge, that figure also means that more than a quarter of participants could not pinpoint the problem that most directly undermines the usefulness of NGS data for Pakistani patients. Given how consequential that gap is in clinical practice—leading to an excess of VUS classifications that cannot be resolved without better South Asian reference data [7, 8]—it deserved attention.

There was also room for improvement in understanding what drives the prevalence of recessive disorders. Consanguinity and endogamy were correctly identified by 76% of respondents, but 13% attributed the high burden to migration-related admixture—a common but important misconception in the teaching of South Asian population genetics. Correctly understanding the mechanism matters, because it shapes what preventive interventions make sense.

Table 2
Pre-Test and Post-Test Correct Response Rates by Knowledge Domain

Knowledge Domain	Pre-Test % Correct	Post-Test % Correct
Genomic utility limited by absent variant databases	93%	—
Consanguinity/endogamy and recessive disorder prevalence	76%	87%
Critical bottleneck: integrating genomic data into clinical workflows	75%	—
Definition of the diagnostic odyssey	81%	—
Biochemical hallmark of inborn errors of metabolism (IEM)	78%	—
Role of next-generation sequencing (NGS) in LMICs	81%	—
Variant interpretation challenges (limited reference databases)	72%	90%
ACMG as the standard-setter for variant interpretation	91%	—
Sustainable genomics infrastructure: local training	87%	96%
Hidden burden of genetic disease in Pakistan	94%	—
Endogamy versus consanguinity as distinct concepts	—	87%
System-level vs. individual barrier (absent newborn screening)	—	83%
VUS reporting creates ethical dilemmas in counseling	—	87%
Multidisciplinary clinics as most impactful diagnostic intervention	—	69%
Global collaboration barriers: data sharing and absent registries	—	98%
Genomics embedded in undergraduate and residency curricula	—	98%

Note: — indicates the item was not included in that phase of testing. Percentages are calculated from the number of respondents who answered each item.

What Changed: Post-Test Findings

Fifty-two participants completed the post-test. Gains were evident across all directly comparable domains, and several post-test items yielded striking results. Near-universal agreement (98%) that data sharing restrictions and the absence of standardised registries are the primary barrier to global rare disease collaboration suggests the workshop succeeded in grounding a somewhat abstract challenge in something concrete and shared. Similarly, 98% correctly identified embedding case-based genomics learning in undergraduate and residency curricula as the practical first step for integrating genomics into medical education—a recommendation that aligns closely with international consensus [16, 17].

The biggest knowledge gain was in sustainable infrastructure, where 96% post-test respondents correctly identified training local clinicians and bioinformaticians (rather than outsourcing testing or relying on international charity) as the foundation of a durable genomics service. This is not a trivial insight: the temptation in resource-constrained settings is often to look outward for solutions, when the more sustainable path runs through local capacity.

One area that did not score as strongly as might be hoped was the identification of multidisciplinary genomic clinics as the single most impactful intervention for reducing the diagnostic odyssey—only 69% of post-test respondents chose this option. It is possible that participants genuinely found it difficult to prioritise one intervention over others, or that the workshop did not spend enough time on integrated service delivery models. Either way, it points to a topic that warrants more attention in future educational programming.

What Participants Proposed: Group Activity Findings

Each group engaged seriously with their assigned scenario and produced recommendations that were both practical and locally grounded. A synthesis is presented in Table 3, and the substantive findings from each group are described in detail below.

Table 3
Summary of Group Activity Themes, Identified Barriers and Proposed Solutions

Group	Theme	Barriers Identified	Proposed Solutions
1 – Researchers	Infrastructure & Cost	No local sequencing capacity; prohibitive cost of exome sequencing; samples sent abroad	Targeted gene panels; phased diagnostic approaches; regional resource-sharing; NGO and government funding; telemedicine for rural outreach
2 – Students	Ethical & Cultural Barriers	Stigma; religious misconceptions; fear of data misuse; fertility myths; distrust of institutions	Engage religious leaders and LHWs; local-language visual campaigns; corner meetings; confidential, ethical counseling; success stories as advocacy
3 – Students	Policy & Collaboration Gaps	No national rare disease policy; siloed research; no data-sharing incentives; no patient registries	Prioritise newborn screening advocacy; low-cost regional registries; join IRDiRC/RDI; mobilise patient advocacy groups; open-data incentives
4 – Faculty	Human Resource Limitations	No formal genetic counselor cadre; poor post-result follow-up; lack of training in variant interpretation	Online certification; tele-genetics platforms; tertiary hospital-led training hubs; formal career recognition; public–private partnerships
5 – Clinicians	Translational Research & Education Gaps	Research findings not reaching clinical practice; students, researchers, and clinicians work in parallel without collaboration	Genomics as a longitudinal curriculum theme; MDT models; interdisciplinary mentoring networks; professional society grants and guidelines

Group 1 (Researchers): Infrastructure and Cost Constraints

The researchers were given a scenario that will feel familiar to anyone working in a regional hospital in Pakistan: a family needs exome sequencing, but the nearest facility is abroad, the cost is out of reach, and there is no local alternative. The group's central recommendation was that targeted gene panels—rather than broad exome or genome sequencing—represent the most cost-effective starting point for most clinical presentations [6]. This is not a compromise; in many settings, a well-designed gene panel for a specific phenotype will yield a faster and more interpretable result than untargeted whole-exome data.

The group also made a strong case for phased diagnostic approaches. Using congenital adrenal hyperplasia as their worked example, they described how a structured pathway—biochemical screening first, confirmatory molecular testing only where indicated—can dramatically reduce per-patient diagnostic costs without sacrificing clinical utility. For infrastructure sharing, they proposed collaborative models in which institutions with sequencing equipment open access to partner hospitals, sharing both the cost and the research outputs. The roles of NGOs, government subsidies, and the private sector were

seen as essential not just for funding but for raising awareness, creating demand, and sustaining the ecosystem of services. Telemedicine, the group argued, is not a futuristic aspiration—it is an immediately deployable tool for reducing the transportation burden on rural families and cutting overhead costs [18].

Group 2 (Students): Ethical and Cultural Barriers

The student group assigned to this theme took on the genuinely difficult scenario of a rural consanguineous family refusing genetic testing. What struck the group was the breadth of misconceptions they were able to document: beliefs that genetic testing is a form of paranormal interference, that it contravenes religious values, that results will be used to harm the family, that it causes infertility, and—perhaps most significantly—that it will disrupt marriage prospects for other family members. These are not irrational fears. They reflect real experiences of stigma, real gaps in trustworthy information, and real histories of institutions not living up to their promises [9, 10].

The group's response was to think carefully about who communities already trust. Religious leaders, school teachers, lady health workers, community elders, and local NGO workers were all identified as more credible messengers than visiting specialists or institutional campaigns. Importantly, the group recommended that religious leaders be educated first—equipped with accurate, theologically grounded information so they can address concerns from within communities' own frameworks, rather than having outside voices override them. Practical communication tools—local-language short videos, family tree analogies, community corner meetings—were endorsed as complements to the interpersonal approach.

The group drew on thalassemia premarital screening and PKU newborn screening as existing success stories that rural communities could relate to: evidence that genetic knowledge does not inevitably lead to stigma and discrimination, but can enable families to make informed choices and prevent suffering [19]. On confidentiality, the group called for private counseling spaces, coded data storage, full informed consent processes, and strict adherence to bioethics and anti-biopiracy principles—recognising that trust, once broken, is extremely hard to rebuild.

Group 3 (Students): Policy and Collaboration Gaps

The second student group worked through a scenario that sits at the system level: no national rare disease policy, researchers working in silos, and no mechanism for sharing data or coordinating advocacy. Their immediate advocacy priorities were clear: newborn screening, carrier screening before marriage, prenatal diagnosis access, and genetic counseling availability. These are not novel recommendations—they are what every credible international body working on genetic services in LMICs has identified as the starting point [13, 20]. The challenge in Pakistan is not knowing what needs to happen but building the political and institutional will to make it happen.

On registries, the group outlined a four-step initiation process: articulate the registry's purpose, confirm it is the right tool for that purpose, identify stakeholders, and establish feasibility before committing resources. Critically, they recommended linking regional sub-registries to a national platform through existing government agencies and hospitals—avoiding the trap of creating parallel infrastructures that

compete rather than collaborate. For international partnership, they identified the International Rare Diseases Research Consortium (IRDiRC), Rare Diseases International (RDI), and Orphanet as the most relevant networks for Pakistan to engage with [20, 21].

The group's thinking on data sharing incentives was particularly nuanced. They identified both intrinsic motivators—the opportunity for reciprocal data access, contributing to a high-quality evidence base, gaining international collaborators—and extrinsic ones: citation benefits, recognition, funding rewards, and compliance with publisher and funder open-data policies. This multi-layered approach to incentives reflects an understanding that researchers share data not because they are told to, but because it serves their interests to do so.

Group 4 (Faculty): Human Resource Limitations

The faculty group engaged with a scenario that many of them had almost certainly experienced directly: a medical college without trained genetic counselors, leading to pediatricians who struggle to explain results and patients who do not follow up. Their short-term capacity-building recommendations were pragmatic: online certification courses in clinical genetics, tele-genetics training workshops, hands-on sessions focused on variant interpretation and counseling, and webinars on recognising genetic red flags in children. These are all immediately deployable, and each has evidence of effectiveness in other LMIC settings [11, 12].

For the medium term, the group envisioned a tele-genetics model built as a layered hierarchy: locally trained health workers at the base, nationally trained coordinators in the middle, and international genetics experts available for complex cases at the top. This is not a new idea, but it has rarely been articulated as clearly in the Pakistani context. Tertiary care hospitals were proposed as the natural hubs for regional training and exchange programmes, given their existing relationships with peripheral facilities and their ability to host clinical placements.

The group's most structurally significant recommendation was the formal recognition of genetic counseling as a distinct profession within the Pakistani healthcare system. Currently, the role does not officially exist—which means there is no training pathway, no regulatory framework, and no career ladder. Without addressing this foundational gap, every other workforce intervention is building on sand [11]. The group also called for public–private partnerships as a mechanism for expanding infrastructure, with a clear framework that includes needs assessment, policy development, applied research, and population screening programmes as the core components.

Group 5 (Clinicians): Translational Research and Education Gaps

The clinicians tackled what is perhaps the most complex of the five scenarios: the disconnect between what is being discovered in genetics laboratories and what is actually changing at the bedside. Their diagnosis was direct. The problem is not a shortage of research findings—it is that researchers, students, and clinicians work in parallel streams that rarely converge. Sensitizing clinicians to the relevance of

genomic findings, building structured collaboration across these groups, and engaging the pharmaceutical and diagnostics industry as active partners were all identified as necessary steps.

On education, the group called for genetics to be embedded as a longitudinal theme throughout the curriculum at all levels—BS, MS, MBBS, and PhD—rather than confined to a single module in the basic sciences. Interdisciplinary electives, online certificate courses in genomics, and ethics teaching embedded within genetics content were all endorsed [16, 17]. Critically, the group called for this not to be added on top of existing curricula but woven through them, so that students encounter genetic thinking in every clinical context, not just in the genetics ward.

The most powerful structural proposal was the multidisciplinary team (MDT) model, in which geneticists, clinicians, researchers, and students all participate in joint decision-making, joint clinics, and joint research projects. This already works well in oncology—there is no reason it cannot work in genetics. The group also proposed interdisciplinary mentoring networks that pair clinicians with geneticists and extend geographically through online platforms, and called on professional societies and conferences to do more than convene: to offer grants, develop national guidelines, and create the sustained infrastructure for collaboration that no single institution can maintain alone.

DISCUSSION

The findings from this workshop paint a picture that is both familiar and urgent. Familiar because the barriers identified—limited infrastructure, cultural resistance, absent policy, workforce shortages, translational gaps—appear in virtually every LMIC medical genetics literature review. Urgent because the people in this room—the researchers, students, faculty, and clinicians who will actually need to address these barriers—demonstrated both the knowledge and the will to do something about them.

The pre-test results confirmed what the literature would predict: conference-attending professionals have solid baseline knowledge of genomics in their context, but gaps remain precisely where the stakes are highest. The variant interpretation challenge—understanding why South Asian populations are poorly served by existing reference databases, and what that means for a VUS result—was the area where the most improvement was needed [7, 8]. In a clinical context, that gap has direct consequences: a family told their result is 'uncertain' when in fact it is almost certainly pathogenic, or a family reassured when they should not be. Addressing this through better local databases and better education about their limitations must be a priority.

The post-test gains—particularly the near-universal recognition of data sharing and registry gaps, and the strong endorsement of embedding genomics in undergraduate curricula—suggest that even a short, well-designed educational intervention can shift thinking. The fact that the multidisciplinary clinic question scored less strongly (69%) is a useful signal. It may reflect genuine complexity in choosing between system-level interventions, or it may reflect that integrated service delivery is not discussed enough in existing training. Future workshops should build in more time for this.

Several cross-cutting themes from the group activities deserve particular emphasis. The first is trust. Across all five groups, trust emerged as the prerequisite for everything else. Communities need to trust that genetic testing is safe, confidential, and genuinely in their interest. Researchers need to trust that sharing data will benefit rather than disadvantage them. Clinicians need to trust that a positive genetic result will lead to something useful. Policymakers need to trust that investment in rare disease infrastructure will be justified by impact. Without trust at each of these levels, no technical solution will take hold.

The second theme is sequencing—not genomic sequencing, but the sequencing of interventions. Virtually every group endorsed a staged, phased approach: biochemical screening before molecular testing, regional registries before national ones, workshop-based training before formal certification programmes, local gene panels before whole-exome sequencing. This is not a counsel of despair; it is a recognition that durable change is built incrementally, and that each step, if done well, creates the conditions for the next.

The third theme is the enabling role of digital health. Telemedicine, tele-genetics, online certification, and digital reporting were raised by nearly every group as a mechanism for extending the reach of genetics services without proportional increases in physical infrastructure [18]. Pakistan's geography—vast, with enormous variation in the density and quality of health services—makes digital tools not a convenience but a necessity.

Finally, the workshop reinforced what every informed observer of Pakistani genetics already knows: the absence of national rare disease policy is not just a policy gap, it is the root cause of the fragmentation that makes every other barrier harder to address. Without a policy framework that mandates newborn screening, funds registries, recognises genetic counseling as a profession, and incentivises data sharing, individual efforts will remain isolated islands of good practice rather than components of a functioning system.

Limitations

Several limitations should be acknowledged openly. The workshop sample was self-selected—people who register for a genetics conference are, by definition, more engaged with the field than the average clinician or researcher. This means the pre-test scores probably overestimate baseline knowledge across the broader workforce, and the group recommendations probably reflect more optimism about what is possible than would be warranted in a less motivated cohort. The pre- and post-tests were not identical, which limits direct item-level comparison. Group activity responses were collected in writing rather than recorded, which inevitably loses some nuance. And there is no longitudinal follow-up: we do not know whether the knowledge gains persist, or whether any of the recommended actions have been implemented. These are important questions for future studies.

CONCLUSION

Pakistan's medical genetics community faces real, overlapping, and well-understood challenges. What this workshop demonstrated is that the people within that community—across every professional level—are not passive observers. They can diagnose the problems clearly, they can generate creative and locally grounded solutions, and they can learn from each other across the professional divides that too often keep researchers, clinicians, students, and educators in separate conversations.

The recommendations that emerged from this workshop—phased diagnostic pathways, community engagement through trusted intermediaries, low-cost regional registries, formal recognition of genetic counseling, longitudinal genomics curricula, and MDT collaboration models—are not wishful thinking. They are achievable. What is needed now is the institutional and political commitment to pursue them in a coordinated way, and the research infrastructure to evaluate what works.

We hope the approach described here—structured, participatory, and grounded in the experiences of people working in the system—can serve as a model for similar efforts across Pakistan and in comparable LMIC settings. The diagnostic odyssey is not inevitable. With the right investments and the right conversations, it can be shortened.

Declarations

Informed consent:

Written/electronic informed consent was obtained from all participants before participation.

Conflicts of Interest:

The authors declare no conflicts of interest.

ETHICS STATEMENT

Ethical approval was obtained from the Institutional Review Board and Ethics Committee of (IRB #461 – 25; approval date: 21 October 2025).

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Author Contribution

S.M. conceptualized and designed the study, supervised the project, and wrote the main manuscript text. P.A., A.J., and W.G. contributed to data collection, organization, and initial drafting of the manuscript.

N.N. assisted in data analysis and interpretation. S.M. and P.A. performed the formal analysis. All authors contributed to manuscript revision, critically reviewed the content, and approved the final version of the manuscript.

References

1. Zakar R, Zakar MZ, Fischer F (2022) Consanguineous marriages and their association with women's reproductive health and fertility behavior in Pakistan: secondary data analysis from Demographic and Health Surveys, 1990–2018. BMC Womens Health 22(1):118
2. Al-Dewik N, Mohd H, Al-Mureikhi M, Ali R, Al-Mesaifri F, Mahmoud L et al (2019) Clinical, genetic, and genomic findings in a cohort from Qatar with rare hereditary metabolic diseases. Front Genet 10:1024
3. Bittles AH, Black ML (2015) Evolution in health and medicine Sackler colloquium: consanguinity, human evolution, and complex diseases. Proc Natl Acad Sci USA 112(Suppl 1):1779–1786
4. Bauskis A, Strange C, Molster C, Fisher C (2022) The diagnostic odyssey: insights from parents of children living with an undiagnosed condition. Orphanet J Rare Dis 17(1):233
5. Benito-Lozano J, Arias-Merino G, Gomez-Martinez M, Ancochea-Diaz A, Aparicio-Garcia A, De La Paz M et al (2024) Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey. Orphanet J Rare Dis 19(1):310
6. Boycott KM, Rath A, Chong JX, Hartley T, Alkuraya FS, Baynam G et al (2017) International cooperation to enable the diagnosis of all rare genetic diseases. Am J Hum Genet 100(5):695–705
7. Gudmundsson S, Singer-Berk M, Watts NA, Phu W, Goodrich JK, Solomonson M et al (2022) Variant interpretation using population databases: lessons from gnomAD. Hum Mutat 43(8):1012–1030
8. Martin AR, Kanai M, Kamatani Y, Okada Y, Neale BM, Daly MJ (2019) Clinical use of current polygenic risk scores may exacerbate health disparities. Nat Genet 51(4):584–591
9. Rashid Y, Rehman A, Bashir S, Kiani S, Mahmood HZ, Noor MN et al (2024) Genetic testing perspectives in Pakistani population: a survey on knowledge, attitudes, awareness, and concerns. J Community Genet 15(5):489–502
10. Aidhen P, Morales-Soto W, Zaidi SS, Khurshid K, Ali H, Hamid S et al (2024) Stigma associated with genetic testing for rare diseases—causes and recommendations. Front Genet 15:1335768
11. Abacan M, Alsubaie L, Barlow-Stewart K, Caanen B, Cordier C, Courtney E et al (2024) The global status of genetic counselors in 2023: what has changed in the past five years? Genet Med Open 2:100856
12. Tekola-Ayele F, Adeyemo A, Rotimi CN (2015) Translational genomics in low- and middle-income countries: opportunities and challenges. Public Health Genomics 18(4):242–247
13. Chung CC, Chan JC, Chu AT, Chung BH (2022) Rare disease emerging as a global public health priority. Front Public Health 10:1028545
14. Bansal A (2021) Expanding free open-access medical education. Front Med (Lausanne) 8:794667

15. Vaismoradi M, Turunen H, Bondas T (2013) Content analysis and thematic analysis: implications for conducting a qualitative descriptive study. Nurs Health Sci 15(3):398–405
16. Massingham LJ, Rubanovich AV, Robertson ME, Fok W, Henrikson NB, Jarvis JN et al (2022) 2022 APHMG consensus-based update of the core competencies for undergraduate medical education in genetics and genomics. Genet Med 24(10):2167–2179
17. Wonkam A, Noubiap JJ (2019) Strategies for genomic medicine education in low- and middle-income countries. Front Genet 10:1007
18. Brown EG, Watts I, Beales ER, Maudhoo A, Hayward J, Sheridan E et al (2021) Videoconferencing to deliver genetics services: a systematic review of telegenetics in light of the COVID-19 pandemic. Genet Med 23(8):1438–1449
19. Inam SHA, Jamil H, Klair N, Nayyar A, Sheikh N, Arif A (2021) Awareness and acceptance of premarital carrier screening of thalassemia among adults. Pak J Med Health Sci 15(3):521–523
20. Austin CP, Cutillo CM, Lau LP, Jonker AH, Rath A, Julkowska D et al (2018) Future of rare diseases research 2017–2027: an IRDiRC perspective. Clin Transl Sci 11(1):21–27
21. Nguengang Wakap S, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V et al (2020) Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet 28(2):165–173
22. Ashfaq M, Ahmed SA, Aziz-Rizvi R, Hasan Z, Kirmani S, Munim S et al (2023) Identifying the current status and future needs of clinical, educational, and laboratory genetics services in Pakistan: a web-based panel discussion. J Community Genet 14(3):291–302
23. Glinton KE, Potocki L, Dhar SU (2022) An innovative medical school curriculum to enhance exposure to genetics and genomics: updates and outcomes. Genet Med 24(3):722–728

Figures

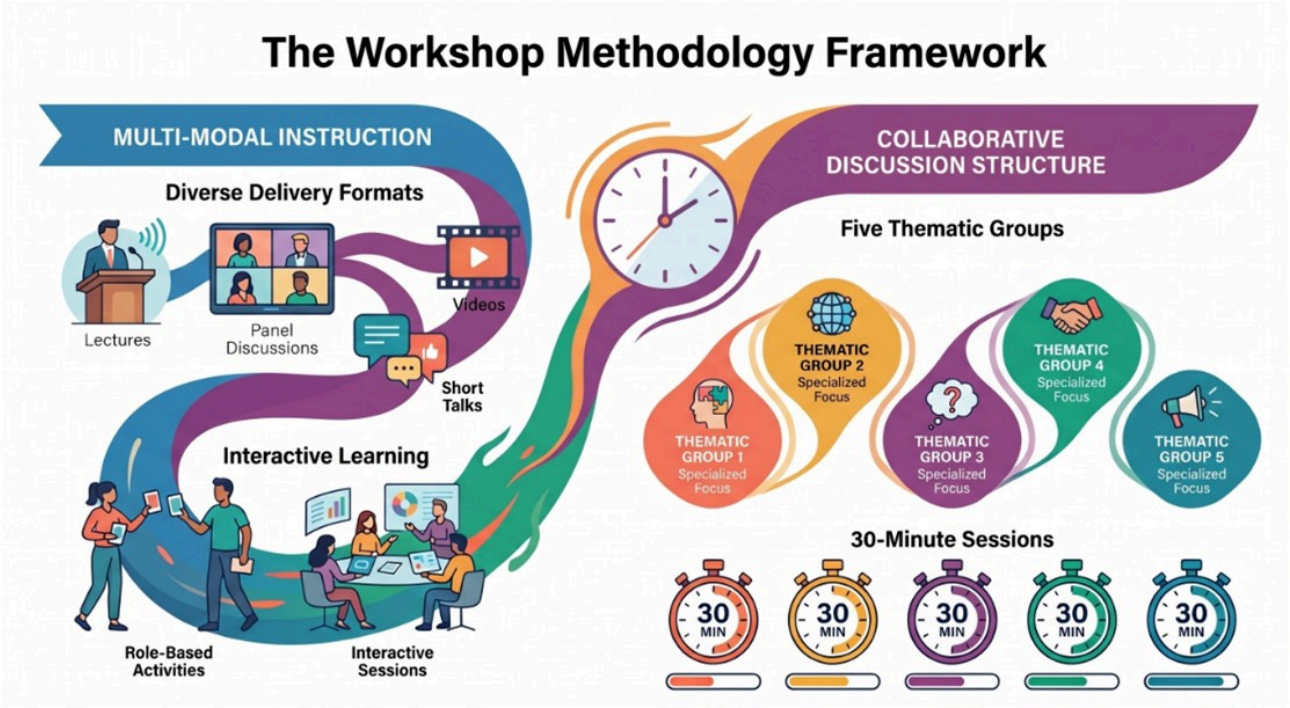


Figure 1

Workshop methodology framework