Barriers and Facilitators to the Implementation of Pre-Symptomatic Genetic Testing in Clinical Practice: An Updated Systematic Review

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Abstract

Background: Pre-symptomatic genetic and genomic testing can identify individuals at increased disease risk before symptoms develop, but implementation depends on more than analytical validity or test availability. This review updates the evidence on barriers and facilitators to implementation after the search period of Shen et al. [2].

Methods: PubMed was searched on 3 August 2026 using three separately constructed search sets covering genetic/genomic testing, presymptomatic or asymptomatic screening, and implementation; the final PubMed History search combined these as #1 AND #2 AND #3. Results were limited to English-language publications from 2020–2026, followed by a publication-boundary audit excluding records published on or before 22 May 2020. Study selection, critical appraisal, extraction, and synthesis were conducted by a single reviewer. Design-appropriate Joanna Briggs Institute (JBI) tools were used [3]. Findings were coded as barriers or facilitators, assigned to implementation levels, and mapped to the 58 themes reported by Shen et al. or to new themes.

Results: A total of 824 PubMed records were identified before the recorded filtering step and 347 remained for title/abstract screening. Of these, 256 were excluded and 91 reports were sought for full-text retrieval. Eleven reports were not retrieved; 80 full texts were assessed and 17 were excluded, leaving 63 studies in the narrative synthesis. The extraction contained 424 barrier/facilitator findings. Thirty-four of the 58 earlier themes were confirmed and 24 were not addressed. Seventeen additional implementation themes were identified. Care-pathway design and follow-up infrastructure, health equity and representativeness, workforce capacity and training, provider inadequate knowledge, family dynamics in cascade testing, logistics of sample collection and access, organisational readiness, and perceived cost were among the most widely represented themes.

Conclusion: Within this PubMed-based update, the post-2020 evidence places substantially greater emphasis on organisational and health-system implementation than the earlier review. Pre-symptomatic testing depends on complete care pathways for recruitment, informed decision-making, sample collection, result communication, counselling, referral, surveillance, prevention, and treatment. Findings should be interpreted in light of the PubMed-only search, English-language restriction, single-reviewer workflow, 11 unretrieved reports, lack of prospective protocol registration, and heterogeneity of the evidence.

Keywords: genetic testing; genomic screening; presymptomatic testing; population screening; implementation science; barriers; facilitators; systematic review

1. Introduction

Genetic and genomic testing increasingly enables identification of individuals at elevated risk of disease before clinical symptoms develop. Testing may be offered through population genomic screening, cascade testing after identification of a familial pathogenic variant, genomic newborn screening, carrier screening, or other predictive pathways. These approaches can move healthcare toward earlier risk identification, surveillance, prevention, reproductive decision-making, and treatment.

The ability to generate a genetic result does not, however, ensure successful clinical implementation. A functioning programme must identify an appropriate target population, reach eligible individuals, support informed decision-making, collect and process samples, interpret and communicate results, provide genetic counselling when appropriate, establish referral pathways, and connect identified individuals to

surveillance, prevention, treatment, or reproductive care. Barriers can therefore arise at patient, clinician, organisational, health-system, policy, and interpersonal levels.

Shen et al. [2] systematically reviewed barriers and facilitators to population genetic screening in healthy populations using literature searched through 22 May 2020. Their synthesis described 58 barrier and facilitator themes and highlighted gaps in provider-focused evidence, macro-level determinants, community facilitators, health equity, education and counselling, ethical/legal/social implications, and long-term outcomes.

Since that search period, the evidence base has increasingly included operational population genomic programmes, genomic newborn screening, cascade-testing pathways, carrier-screening programmes, and genotype-first approaches. An updated review can therefore examine whether earlier themes remain visible and identify determinants that become apparent when testing is implemented within real clinical systems.

The primary research question was: What barriers and facilitators affect the implementation of genetic and genomic pre-symptomatic testing in clinical practice for people without symptoms?

A secondary objective was to compare the updated evidence with the themes and research gaps identified by Shen et al. [2].

2. Methods

2.1 Review Design

This study was conducted as an updated systematic review of barriers and facilitators affecting implementation of pre-symptomatic genetic and genomic testing. Study-selection reporting was structured according to PRISMA 2020 [1]. Because the eligible literature was methodologically heterogeneous, quantitative meta-analysis was not considered appropriate; a narrative and thematic synthesis was used. The review protocol was not prospectively registered.

2.2 Search Strategy

PubMed was the only bibliographic database searched. The search was conducted on 3 August 2026. Three search sets were constructed separately in PubMed and retained in the project documentation:

Concept 1 – genetic/genomic testing: "Genetic Testing"[Mesh] OR "Genomics"[Mesh] OR "genetic testing"[tw] OR "genomic testing"[tw] OR "genetic screening"[tw] OR "genomic screening"[tw] OR "polygenic risk score*"[tw] OR "carrier screening"[tw] OR "cascade screening"[tw]

Concept 2 – pre-symptomatic: "Asymptomatic Diseases"[Mesh] OR "Mass Screening"[Mesh] OR presymptomatic[tw] OR pre-symptomatic[tw] OR asymptomatic[tw] OR "predictive testing"[tw] OR "population screening"[tw]

Concept 3 – implementation: "Implementation Science"[Mesh] OR "Attitude of Health Personnel"[Mesh] OR implementation[tw] OR adoption[tw] OR uptake[tw] OR barrier*[tw] OR facilitator*[tw] OR enabler*[tw]

The three retained search sets were combined through PubMed History as #1 AND #2 AND #3. Filters were applied for English language and publication years 2020–2026. Because the year-based 2020 filter also includes publications from January through 22 May 2020, publication dates were subsequently audited against the prespecified update boundary and records published on or before 22 May 2020 were excluded. The complete retained search sets, final combination, filters, and search date are reported in Appendix B and the supplementary search-strategy file.

2.3 Eligibility Criteria

Studies were eligible if they involved individuals without symptoms who were eligible for pre-symptomatic molecular genetic or genomic testing, or healthcare professionals, health-system stakeholders, or policymakers involved in offering, delivering, integrating, or regulating such testing.

Eligible testing contexts included population genetic/genomic screening, cascade genetic testing, predictive testing, genomic newborn screening, carrier screening, genotype-first screening, and other

molecular pathways directed toward pre-symptomatic risk identification. Studies had to report implementation-relevant evidence such as adoption, uptake, access, workflow, scale-up, recruitment, referral, follow-up, barriers, facilitators, or other implementation determinants.

Studies were excluded if they focused only on diagnosis of symptomatic or already diagnosed patients; evaluated laboratory or analytical validity without a clinical implementation component; presented modelling without observed implementation; were non-empirical publication types without completed implementation results; did not involve molecular genetic/genomic testing; lacked extractable implementation evidence; or were published on or before 22 May 2020.

2.4 Study Selection

The screening dataset contained 824 PubMed records before the recorded date/language filtering step. After filtering, 347 records remained for title and abstract screening. Records were classified as Include, Exclude, or initially unresolved. After final quality control, 256 records were excluded at title/abstract stage and 91 reports were sought for full-text assessment. Eleven reports could not be retrieved and were not excluded on abstract information alone. Eighty full texts were assessed; 17 were excluded and 63 studies were included.

2.5 Publication-Boundary Audit

Because the review was explicitly designed as an update after 22 May 2020, a final audit used the earliest identifiable electronic or publisher publication date. Five studies that had initially been carried forward were found to fall on or before the cutoff and were reclassified as OUTSIDE-DATE exclusions. The correction was retained explicitly in the audit trail.

2.6 Critical Appraisal

Methodological quality was assessed using design-appropriate JBI critical appraisal tools [3]. The tool set included Analytical Cross-Sectional Studies, Qualitative Research, Case Series, Cohort Studies, Quasi-Experimental Studies, Prevalence Studies, Case Reports, and Textual Evidence/Expert Opinion. Each applicable item was recorded as Yes, No, Unclear, or Not Applicable; no numerical quality score was calculated.

Mixed-methods studies contributing qualitative as well as quantitative implementation evidence received component-specific appraisal where necessary. Studies #3, #34, #58, #70, and #81 received an additional qualitative appraisal. Item-level judgments from the first group of appraisals completed during the early phase of the project were retained transparently rather than retrospectively replaced.

2.7 Data Extraction

For each included study, the structured extraction recorded author/year, country, design, population or stakeholder group, testing context, setting, sample size, implementation framework when reported, and JBI tool. Individual implementation findings were extracted separately and coded as barrier or facilitator, implementation level, stakeholder, evidence type, mapping to an existing Shen et al. theme, and consolidated new theme where no suitable earlier theme existed.

Newer extraction included page or section locators where available. The initial ten-study extraction did not contain page locators; these locations were not reconstructed or invented retrospectively.

2.8 Data Synthesis

Each finding was compared with the 58 themes reported by Shen et al. [2]. A theme was classified as confirmed when at least one included study contained a corresponding extracted finding; themes without a supporting finding were classified as not addressed. “Not addressed” does not imply that a theme was disproven. Findings outside the earlier framework were first retained as granular NEW codes and then consolidated into 17 broader implementation themes. Original codes and consolidated themes were both preserved in the analysis dataset.

3. Results

3.1 Study Selection

A total of 824 PubMed records were identified before the recorded filtering step. After date/language filtering, 347 records remained; 256 were excluded during title/abstract screening. Ninety-one reports were sought, 11 were not retrieved, and 80 underwent full-text assessment. Seventeen full-text reports were excluded, leaving 63 studies in the final narrative synthesis.

Table 1. Study-selection flow.

Review stage	n
Records identified in PubMed before date/language filters	824
Records screened after date/language filters	347
Records excluded at title/abstract	256
Reports sought for retrieval	91
Reports not retrieved	11
Reports assessed for eligibility	80
Reports excluded at full text	17
Studies included in review	63

3.2 Study Characteristics

The 63 included studies represented a heterogeneous evidence base spanning population genomic screening, cascade testing, genomic newborn screening, carrier screening, familial hypercholesterolaemia, hereditary cancer, cardiovascular genetics, reproductive genetics, and other pre-symptomatic contexts. The United States contributed the largest number of studies, followed by Australia and the United Kingdom; most evidence came from high-income health systems. Study populations included general-population participants, parents of newborns, relatives eligible for cascade testing, patients invited to screening, primary-care and specialist clinicians, genetic counsellors, policy experts, programme staff, and public-health stakeholders.

Qualitative interviews and focus groups, cross-sectional surveys, programme evaluations, case series, cohort studies, quasi-experimental studies, mixed-methods designs, expert consensus, and other observational implementation designs were represented. This heterogeneity supported a multi-level implementation synthesis but precluded meaningful statistical pooling.

3.3 Critical Appraisal

All 63 included studies underwent design-appropriate JBI appraisal. Five mixed-methods studies contributed an additional qualitative component appraisal, yielding 68 component-level appraisals. Appraisal was used to characterise methodological limitations rather than to generate a numerical quality score or exclusion threshold. The most recurrent issues were incomplete reporting of researcher positioning/reflexivity in qualitative studies and incomplete handling of confounding in analytical cross-sectional studies. Table 2 summarises the appraisal profile by JBI tool.

Table 2. Summary of JBI critical appraisal by appraisal tool.

JBI tool	Component-level appraisals (n)	Main appraisal signal
Analytical cross-sectional	25	Inclusion criteria were unclear in 13/25; confounding strategies were absent in 13/25. Outcome measurement was generally strong.
Qualitative research	25	Researcher positioning was not reported in 19/25 (unclear in 1); reflexivity was not addressed in 17/25 (unclear in 1).
Case series	8	Most criteria were met; isolated concerns involved consecutive/complete inclusion and reporting of clinical information.
Cohort studies	4	Most criteria were met; completeness of follow-up was unclear in 2/4 studies.
Other designs	6	Three quasi-experimental appraisals showed weak control/comparison structures; the single prevalence, case-report, and expert-opinion appraisals had few or no major concerns.

3.4 Overall Distribution of Barriers and Facilitators

The final extraction contained 424 individual barrier/facilitator findings. System-level determinants formed the largest category (141 findings across 47 studies), followed by patient-level (103 across 40 studies), policy-level (69 across 36), clinician-level (48 across 24), organisational (37 across 16), and interpersonal determinants (26 across 15).

Table 3. Distribution of extracted implementation findings by level.

Implementation level	Findings	Studies represented
Patient	103	40
Clinician	48	24
Organisational	37	16
System	141	47
Policy	69	36
Interpersonal	26	15

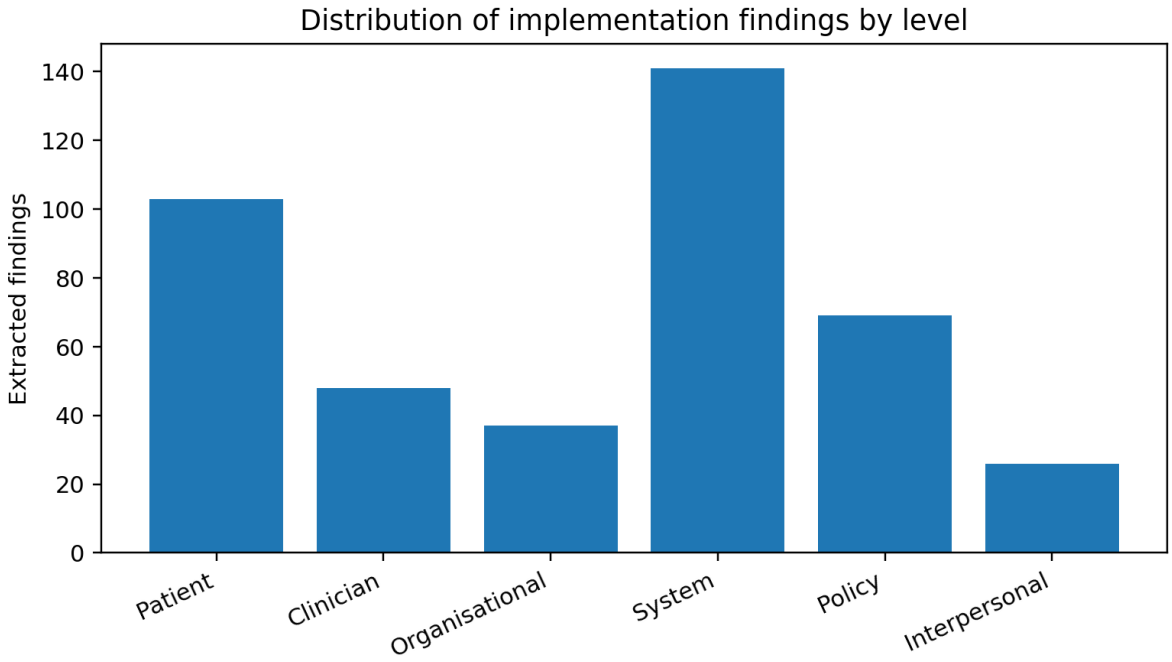


Figure 1. Distribution of the 424 extracted findings across implementation levels.

3.5 Comparison With Themes Reported by Shen et al.

Of the 58 themes described in the earlier review, 34 were confirmed in the updated evidence and 24 were not addressed. Provider inadequate knowledge was represented across 16 included studies and perceived cost of screening across 13. Other repeatedly observed earlier themes included psychological concerns, uncertainty of results, potential insurance effects, confidentiality/privacy concerns, lack of time, and perceived limited clinical utility. These results indicate persistence of multiple patient- and clinician-level barriers despite technological progress.

The complete supporting-study mapping for every confirmed and new theme is retained in the supplementary Theme Summary sheet; therefore, citations in the narrative below are illustrative rather than exhaustive.

3.6 New Implementation Themes

Seventeen consolidated implementation themes were identified beyond the earlier Shen framework. The most widely represented was care-pathway design and follow-up infrastructure (25 studies), followed by health equity and representativeness (20), workforce capacity and training (18), organisational

readiness and inner setting (14), logistics of sample collection and access (14), and family dynamics in cascade testing (14).

Table 4. Consolidated implementation themes identified in the updated evidence base.

Theme	Studies	Implementation meaning
Workforce capacity and training	18	Availability of trained clinicians, genetic counsellors, education, implementation support, and specialist expertise.
Champions and leadership	6	Presence of clinical champions or organisational leadership supporting implementation.
Organisational readiness and inner setting	14	Capacity to absorb testing into routine workflow, staffing, scheduling, and administrative systems.
Planning, coordination and stakeholder engagement	9	Cross-service planning, implementation teams, communication, stakeholder alignment, and coordination.
Care pathway design and follow-up infrastructure	25	Processes linking screening to counselling, confirmatory testing, referral, surveillance, treatment, and long-term care.
Logistics of sample collection and access	14	Testing location, travel, appointments, sample collection, existing specimens, and practical access.
Recruitment channel and reach	9	How eligible individuals are invited, contacted, reminded, and reached.
Health equity and representativeness	20	Differences associated with geography, race/ethnicity, socioeconomic status, language, health literacy, and under-represented groups.
Consent process design	8	Timing, length, complexity, and method of pre-test information and consent.
Post-test support and results communication	8	Return of results, counselling, psychosocial support, interpretation, and communication of implications.
Treatability and actionability	5	Whether identifying genetic risk leads to meaningful preventive, reproductive, surveillance, or therapeutic action.
Funding models and provider incentives	11	Programme financing, reimbursement, provider compensation, and sustainability.
Guidelines, evidence requirements and regulation	10	National guidance, professional standards, regulation, and evidence thresholds.
Legal protection against genetic discrimination	1	Protection against insurance, employment, or other genetic discrimination.
Family dynamics in cascade testing	14	Communication between relatives, family responsibility, privacy, and transmission of risk information.
Competing priorities and inertia	4	Other clinical demands, comorbidities, limited attention, and postponed testing.
Professional role and scope of practice	4	Whether clinicians perceive genomic screening as part of their professional responsibility.

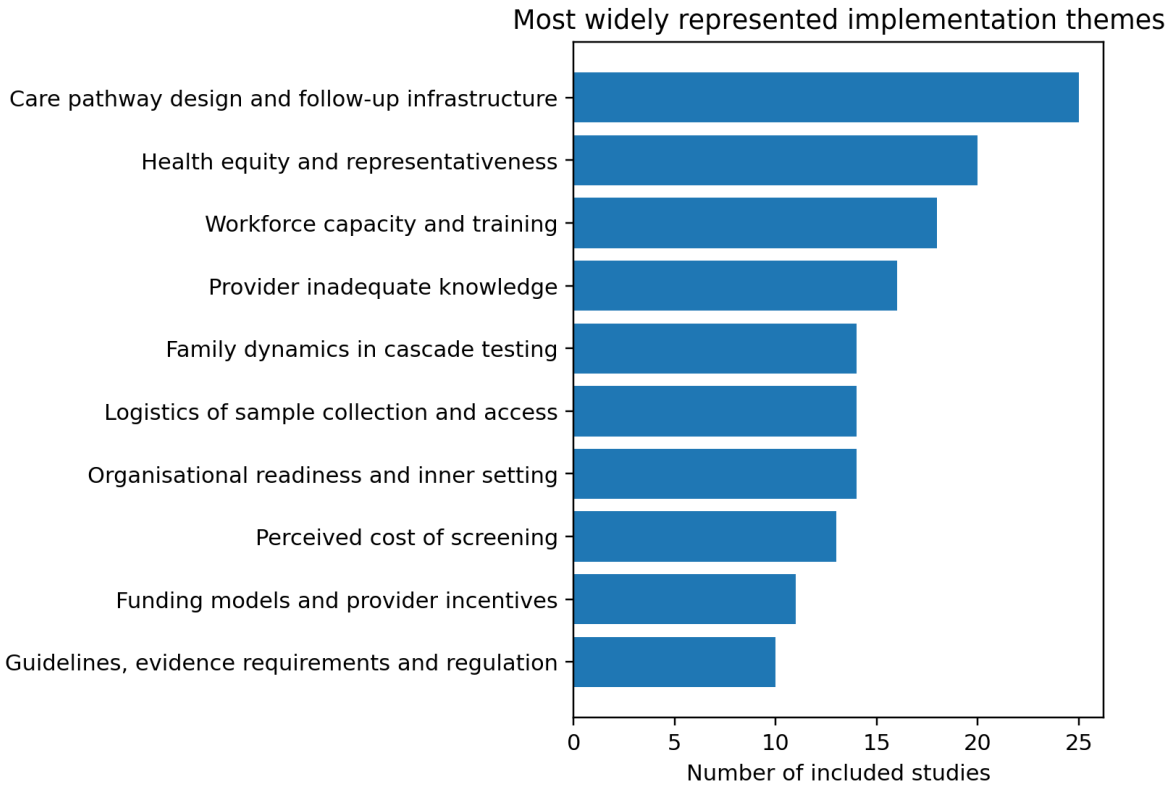


Figure 2. Most widely represented themes by number of included studies.

3.6.1 Care-Pathway Design and Follow-Up Infrastructure

Care-pathway design was the most widely represented new theme. Screening offers depended on functioning links to counselling, confirmatory testing, referral, surveillance, treatment, reproductive services, and family-based follow-up. Barriers arose when downstream responsibility was unclear or fragmented; facilitators included predefined pathways, central coordination, tracking systems, and integration with existing care. Representative examples include [4, 14, 22, 34, 45]; complete supporting-study lists are provided in the supplementary workbook.

3.6.2 Health Equity and Representativeness

Implementation differed across socioeconomic, racial/ethnic, geographic, language, and community contexts. Barriers included limited healthcare access, lower health literacy, language differences, rural residence, digital exclusion, and under-representation. Community partnerships, flexible testing options, and explicit monitoring of who is reached were recurring facilitators. Representative examples include [10, 12, 15, 43, 53]; complete supporting-study lists are provided in the supplementary workbook.

3.6.3 Workforce Capacity and Training

Provider knowledge remained important, but the updated evidence distinguished knowledge from capacity. Even informed clinicians may lack time, genetic counsellors, specialist support, referral pathways, or organisational permission to redesign workflow. Education was therefore most plausibly useful when paired with structural implementation support. Representative examples include [11, 14, 21, 30, 45, 46]; complete supporting-study lists are provided in the supplementary workbook.

3.6.4 Provider Knowledge and Professional Role

Clinicians reported uncertainty about interpretation, patient selection, counselling, clinical utility, and downstream management. Some also viewed genetic screening as belonging primarily to genetics specialists. Role clarification, clinical guidance, decision support, referral infrastructure, and access to

specialist expertise may therefore matter alongside education. Representative examples include [21, 37, 46, 48]; complete supporting-study lists are provided in the supplementary workbook.

3.6.5 Family Dynamics in Cascade Testing

Cascade testing depends on information moving between relatives. Barriers included difficulty contacting relatives, family conflict, privacy concerns, uncertainty over who should disclose risk, and reliance on the index patient. Structured family communication tools and facilitated contact pathways were recurrent facilitators. Representative examples include [8, 20, 33, 44, 54, 65]; complete supporting-study lists are provided in the supplementary workbook.

3.6.6 Logistics of Sample Collection and Access

Testing was easier to implement when collection could be combined with routine care or existing specimens, or offered through local, remote, or community-based options. Additional appointments, travel, complex referrals, and inconvenient collection procedures created barriers even when the underlying test was available. Representative examples include [4, 15, 22, 43, 53]; complete supporting-study lists are provided in the supplementary workbook.

3.6.7 Organisational Readiness

Implementation depended on staffing, administrative support, leadership, information systems, time, and workflow flexibility. Clinical enthusiasm alone was insufficient when organisations lacked the capacity to integrate screening into routine practice. Representative examples include [14, 22, 30, 34, 46]; complete supporting-study lists are provided in the supplementary workbook.

3.6.8 Funding, Consent, Post-Test Support, and Regulation

Cost operated at both individual and system levels. Removing the direct price of a genetic test did not necessarily resolve barriers related to referral, transportation, counselling, follow-up, staffing, or downstream treatment. The updated corpus also contained multiple consent and education models, but did not establish a single best approach across contexts. Limited post-test counselling capacity could itself constrain scale, while guidelines, data governance, insurance/employment concerns, and responsibility for follow-up shaped both participation and clinician confidence.

3.7 Research Gaps

The updated evidence addressed several research gaps identified by Shen et al. but did not eliminate them. Provider-focused research, macro-level determinants, health equity, and community/interpersonal factors were substantially better represented. Prescreen education/counselling and ELSI remained only partly resolved, and long-term clinical/economic outcome evidence remained incomplete.

Table 5. Status of research gaps identified by Shen et al. [2].

Research gap	Status	Interpretation
Evaluate the best manner of prescreen education and counselling for specific contexts	Partly	The updated corpus contains multiple education/counselling and consent models, but the extraction does not establish one best delivery model across contexts.
Examine provider buy-in and expand the definition of clinical utility	Yes	Provider knowledge, role, perceived utility, readiness and implementation support are repeatedly examined.
Investigate the views of healthcare and public-health providers and develop educational resources	Yes	Multiple studies directly sample clinicians/policy stakeholders or evaluate training and implementation support.
Investigate macro-level determinants of population genetic screening	Yes	Organisational, system and policy determinants are prominent in the completed extraction.
Address ethical, legal and social implications (ELSI)	Partly	ELSI issues are frequently reported, but formal ELSI frameworks are not consistently used.

Research gap	Status	Interpretation
Assess long-term outcomes of population genetic screening	Partly	Several studies report programme follow-up or multi-year implementation experience, but the extracted evidence does not establish a mature body of long-term clinical and economic outcome data after presymptomatic screening.
Improve reporting of race/ethnicity and focus on health equity	Yes	Equity, representativeness, access and sociodemographic differences are directly evaluated in multiple studies.
Address the scarcity of provider- and public-health-based studies	Yes	The completed corpus contains numerous clinician, health-system, implementation and policy stakeholder studies.
Identify community-level facilitators and interpersonal factors	Yes	Family communication, community outreach, local partnerships and recruitment channels are represented across the updated evidence base.

4. Discussion

This updated systematic review shows that implementation of pre-symptomatic genetic and genomic testing is shaped by a broader set of determinants than test availability alone. Many earlier barriers remain: uncertainty, fear, mistrust, privacy concerns, perceived cost, questions of clinical utility, and limited provider knowledge. At the same time, the newer corpus contains a much larger body of organisational and system-level implementation evidence.

The apparent shift should be interpreted cautiously. This update used a PubMed-only search and was not an exact re-run of the multi-database search used in the earlier review. Differences between the two syntheses may therefore reflect both maturation of the field and differences in search coverage, eligible study types, and the composition of the included evidence.

4.1 Implementation Is a Care Pathway, Not a Test

The most prominent new theme was care-pathway design. A genetic test does not constitute an implementation programme by itself. Successful implementation requires coordinated identification, recruitment, information provision, consent, sample collection, analysis, interpretation, return of results, counselling, referral, surveillance, prevention, treatment or reproductive care, and—where relevant—communication with relatives.

Failure at any point may reduce the clinical impact of screening. This helps explain why removing one barrier, such as the price of testing, may still leave incomplete uptake or follow-up.

4.2 From Individual Uptake to System Capacity

Earlier genetic-screening literature often framed implementation primarily as an individual decision. The updated evidence suggests that observed uptake is also a property of programme design. Individuals can only accept testing that is offered, accessible, understandable, affordable, and connected to meaningful downstream care.

Low participation may therefore reflect not only preference, but also recruitment strategy, appointment burden, language barriers, fragmented referrals, lack of counselling, digital exclusion, or failure to integrate testing into routine care.

4.3 Health Equity Should Be Treated as an Implementation Outcome

Equity emerged as one of the most prominent new themes. Population-scale genomic screening may widen access beyond specialist clinics, but it can also reproduce existing inequalities through digital recruitment, travel requirements, complex consent, language barriers, and unequal access to downstream services.

Implementation success should therefore be evaluated not only by overall participation but also by who is invited, who responds, who completes testing, who receives results, and who successfully reaches downstream care.

4.4 Provider Education Is Necessary but Not Sufficient

Provider inadequate knowledge remained highly represented, supporting continued investment in genomic education. However, knowledge and capacity were not equivalent. Clinicians may understand the rationale for screening while lacking time, genetic-counselling support, referral options, reimbursement, electronic systems, or authority to redesign workflow.

Education should therefore be paired with system-level support such as decision support, standardised referral criteria, implementation tools, specialist consultation, protected time, and clearly assigned responsibility for follow-up.

4.5 Family Communication Is a Distinct Implementation Layer

Cascade testing creates a distinct interpersonal implementation layer because identification of one familial variant can have implications for multiple relatives. Programmes that rely entirely on the index patient to transmit information may be limited by family conflict, loss of contact, privacy concerns, uncertainty over responsibility, or difficulty explaining genetic risk.

Family letters, structured counselling, digital communication tools, family meetings, and facilitated contact pathways represent plausible implementation supports.

4.6 Actionability, Funding, and Downstream Care

The perceived value of screening is closely linked to whether a result can change management. Clinical utility should therefore be understood as more than the ability to identify a pathogenic variant; it also depends on whether the health system can act on that information.

Likewise, financing discussions should extend beyond the laboratory test itself. Sustainable implementation may require funding for confirmatory testing, counselling, clinician time, programme coordination, information systems, specialist referral, surveillance, prevention, and treatment.

4.7 Comparison With the Earlier Review

The updated review confirms much of the earlier framework while adding substantial organisational, system, policy, and interpersonal evidence. Within this corpus, the implementation question has increasingly moved from whether people would accept testing toward how healthcare systems can deliver testing safely, equitably, and sustainably. Because of differences in search strategy and evidence coverage, this should be interpreted as an observed pattern in the updated corpus rather than definitive proof of a temporal causal shift in the field.

5. Strengths and Limitations

A major strength is the extraction of 424 individual implementation findings across 63 studies rather than reliance only on broad study conclusions. The dataset retains both original Shen-theme mapping and consolidated new themes, enabling the interpretive process to be audited. Critical appraisal used design-appropriate JBI tools, and mixed-methods studies contributing qualitative evidence received component-specific appraisal where necessary. Full-text decisions, date-boundary decisions, retrieval status, appraisal judgments, and supporting-study mappings were retained in the supplementary files.

- PubMed was the only bibliographic database searched; relevant evidence indexed only elsewhere may have been missed.
- Only English-language publications were included.
- The review was conducted by a single reviewer; study selection, full-text eligibility, appraisal, extraction, and thematic synthesis were not independently duplicated.
- Eleven reports could not be retrieved and therefore could not be assessed for eligibility.
- The evidence base was heterogeneous in design, testing context, disease area, population, implementation stage, and outcome measurement.

- Several study designs did not map perfectly to a single appraisal tool; the closest design-appropriate JBI checklist or component-specific approach was used and documented.
- The first ten-study extraction did not record page-level source locators; these were not retrospectively invented.
- Thematic consolidation required interpretive judgment, and study counts indicate representation in the included literature rather than effect size or causal importance.
- Direct comparison with Shen et al. is limited by different database coverage and corpus composition; apparent shifts may therefore reflect both genuine field development and methodological differences.

6. Future Research

Future updates should extend the search to additional bibliographic databases and archive the complete search history, exported results, and any database-specific syntax alongside the review materials. Independent duplicate screening and appraisal should be incorporated where possible.

Long-term clinical and economic outcomes remain especially important. Many implementation studies evaluate feasibility, uptake, or early follow-up, but the current extraction does not establish a mature body of evidence showing whether pre-symptomatic identification improves long-term outcomes.

Direct comparisons of implementation models are also needed, including digital versus clinician-led recruitment, traditional counselling versus streamlined consent, centralised versus primary-care-based testing, patient-mediated versus facilitated cascade testing, and in-person versus remote result disclosure.

Greater representation of lower-resource health systems is required, and equity should increasingly be treated as a primary implementation outcome rather than a secondary demographic analysis.

7. Conclusion

Implementation of pre-symptomatic genetic and genomic testing is shaped by interacting patient-, clinician-, organisational-, system-, policy-, and interpersonal-level determinants. Many barriers described before 2020 remain relevant, including provider knowledge, perceived cost, psychological concerns, mistrust, and uncertainty regarding clinical utility. The updated corpus adds a substantial implementation-system dimension: care-pathway design, downstream follow-up, health equity, workforce capacity, sample access, organisational readiness, family communication, funding, guidelines, and regulation are prominent determinants.

The central practical challenge is therefore not simply whether genetic risk can be identified, but whether healthcare systems can reliably reach eligible people, support informed participation, perform testing, communicate results, and translate genetic information into appropriate clinical action. Successful implementation requires genomic testing to be embedded within complete and sustainable systems of care.

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Conflicts of Interest

The author declares no known competing financial interests or personal relationships that could have influenced the work reported in this manuscript.

Ethics Statement

This systematic review used information from previously published studies and did not involve direct recruitment of human participants, collection of identifiable private data, or animal experimentation. Ethics approval was therefore not required.

Data Availability Statement

The study-level screening decisions, full-text eligibility log, critical appraisal tables, data extraction table, thematic synthesis, research-gap analysis, search strategy, and review audit trail are available in the accompanying supplementary spreadsheets.

AI Use Statement

Generative artificial-intelligence tools were used as assistive tools for review-data organisation, extraction support, critical-appraisal structuring, quality control, language editing, manuscript drafting, and document formatting. The source articles and retained review files—not AI-generated information—were treated as the evidentiary basis for reported data. AI systems were not treated as authors or independent sources of evidence. The author remains responsible for final verification, interpretation, and submission.

Author Contributions

R. Kadylbekov conducted the final study screening, full-text eligibility assessment, critical appraisal, data extraction, thematic synthesis, analysis, manuscript preparation, and final quality-control workflow.

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Appendix A. Full-Text Exclusions

The following 17 reports reached full-text assessment but were excluded. The table preserves the final decision code and the rationale retained in the QC audit.

Table A1. Full-text exclusions.

ID	Report	Reason	Rationale
#5	Genome-based newborn screening for severe childhood genetic diseases has high positive predictive value and sensitivity in a NICU pilot trial (2024)	WRONG-POP	NICU cohort not considered appropriate for genetic testing; study evaluates diagnostic/genome-screening performance in a clinically ill NICU population rather than presymptomatic screening in an asymptomatic population.
#8	Genetic screening for hypertrophic cardiomyopathy in large, asymptomatic military cohorts (2020)	NO-IMPL	Simulation/modeling study of hypothetical military HCM screening and cost-benefit; does not evaluate adoption, uptake, integration or scale-up of an implemented clinical program.
#15	International Survey on Genetic Literacy and Awareness in Patients With Spinal and Bulbar Muscular Atrophy (2026)	WRONG-POP	Participants are patients with symptomatic SBMA; study examines their genetic literacy and views about relatives' testing rather than implementation among asymptomatic people or testing providers.
#16	Public preferences for the value and implementation of genomic newborn screening: Insights from two discrete choice experiments in Australia (2025)	NO-IMPL	Discrete-choice experiment in the Australian public using hypothetical gNBS scenarios; no clinical or health-system implementation/adoption was evaluated.
#20	The implementation of newborn screening for spinal muscular atrophy: the Australian experience (2020)	OUTSIDE-DATE	Publisher publication date 14 October 2019, before the update cutoff of 22 May 2020.
#29	Genetic testing and cascade screening in pediatric long QT syndrome and hypertrophic cardiomyopathy (2020)	OUTSIDE-DATE	Journal publication was January 2020 (with 2019 online indexing/DOI), before the update cutoff of 22 May 2020.
#31	The Provision of Genetic Testing and Related Services in Quebec, Canada (2020)	OUTSIDE-DATE	Published 4 March 2020, before the update cutoff of 22 May 2020.
#42	Managing healthcare for female BRCA carriers in the population screening era: developing a harmonized national policy for surveillance and risk-reduction (2026)	WRONG-TYPE	Excluded as WRONG-TYPE in the retained full-text decision log. The supplied audit record did not preserve a more specific methodological reason; no more specific rationale was invented.
#47	Workplace genetic testing: which employees are likely to participate, what are their concerns with employer sponsorship, and which design features could reduce barriers and increase participation? (2024)	NO-IMPL	Study concerns employer-sponsored workplace genetic testing among a general workforce; the setting is a workplace benefit rather than clinical or health-system practice.
#56	Awareness screening and referral patterns among pediatricians in the United States related to early clinical features of spinal muscular atrophy (SMA) (2021)	WRONG-POP	Focus is awareness of symptomatic SMA and referral for diagnostic genetic testing after developmental signs; not presymptomatic genetic screening implementation.
#65	Making the most of the first prenatal visit: The challenge of expanding prenatal genetic testing options and limited clinical encounter time (2020)	WRONG-POP	Study concerns prenatal fetal screening/testing choices during pregnancy rather than presymptomatic genetic/genomic testing of asymptomatic individuals for future disease risk.
#67	Genomic sequencing in newborn screening: balancing consent with the right of the asymptomatic at-risk child to be found (2025)	WRONG-TYPE	Ethical/policy analysis synthesizing consent issues and existing gNBS programs; no original participant or implementation dataset is reported.

ID	Report	Reason	Rationale
#69	Young Women's Perspectives on Being Screened for Hereditary Breast and Ovarian Cancer Risk During Routine Primary Care (2024)	NO-IMPL	Intervention under discussion is routine family-history/HBOC risk assessment in primary care, not genetic/genomic testing itself.
#73	Perspectives from individuals with familial hypercholesterolemia on direct contact in cascade screening (2020)	OUTSIDE-DATE	Electronic article date 29 March 2020, before the update cutoff of 22 May 2020.
#75	Implementation of the National Collegiate Athletic Association Sickle Cell Trait Screening Policy: Methods and Staff and Athlete Perspectives (2026)	WRONG-TEST	The paper evaluates an NCAA sickle-cell-trait screening policy in which the required direct screening test is a sickle-cell solubility test (or documentation of prior status), not molecular genetic/genomic testing.
#87	Implementation of population-based newborn screening reveals low incidence of spinal muscular atrophy (2020)	OUTSIDE-DATE	Publisher publication date 18 May 2020, four days before the update cutoff of 22 May 2020.
#90	DNA-poli: Design and development of a digital platform for family communication support and predictive genetic counseling on inherited diseases (2025)	NO-IMPL	Design/development paper for a digital predictive-counselling platform; no completed clinical implementation or uptake evaluation of genetic testing is reported.

Appendix B. Retained Search Documentation

The retained search documentation comprised three separately constructed PubMed search sets, which were combined through PubMed History as #1 AND #2 AND #3. The retained sets, filters, and search date are reproduced below.

Database: PubMed

Search date: 3 August 2026

Research question: What barriers and facilitators affect the implementation of genetic and genomic pre-symptomatic testing in clinical practice for people without symptoms?

Concept 1 — genetic/genomic testing: "Genetic Testing"[Mesh] OR "Genomics"[Mesh] OR "genetic testing"[tw] OR "genomic testing"[tw] OR "genetic screening"[tw] OR "genomic screening"[tw] OR "polygenic risk score*"[tw] OR "carrier screening"[tw] OR "cascade screening"[tw]

Concept 2 — pre-symptomatic: "Asymptomatic Diseases"[Mesh] OR "Mass Screening"[Mesh] OR presymptomatic[tw] OR pre-symptomatic[tw] OR asymptomatic[tw] OR "predictive testing"[tw] OR "population screening"[tw]

Concept 3 — implementation: "Implementation Science"[Mesh] OR "Attitude of Health Personnel"[Mesh] OR implementation[tw] OR adoption[tw] OR uptake[tw] OR barrier*[tw] OR facilitator*[tw] OR enabler*[tw]

Final PubMed History combination: #1 AND #2 AND #3
Filters: English language; publication years 2020–2026.
Update boundary: records published on or before 22 May 2020 were excluded during the publication-boundary audit.

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