Supplementary Table 1. Search strategies used in the systematic review, by database.

PubMed

("genetic testing"[Title/Abstract] OR "genetic test"[Title/Abstract] OR "genetic tests"[Title/Abstract] OR "genetic technology"[Title/Abstract] OR "genetic technologies"[Title/Abstract] OR "genetic panel"[Title/Abstract] OR "genetic panels"[Title/Abstract] OR "gene panel"[Title/Abstract] OR "gene panels"[Title/Abstract] OR "genetic profiling"[Title/Abstract] OR "genetic application"[Title/Abstract] OR "genetic applications"[Title/Abstract] OR "genetic screening"[Title/Abstract] OR "genomic testing"[Title/Abstract] OR "genomic test"[Title/Abstract] OR "genomic tests"[Title/Abstract] OR "genomic technology"[Title/Abstract] OR "genomic technologies"[Title/Abstract] OR "genomic panel"[Title/Abstract] OR "genomic panels"[Title/Abstract] OR "genome panel"[Title/Abstract] OR "genome panels"[Title/Abstract] OR "genomic profiling"[Title/Abstract] OR "genomic application"[Title/Abstract] OR "genomic applications"[Title/Abstract] OR "genomic screening"[Title/Abstract] OR "next generation sequencing testing"[Title/Abstract] OR "NGS testing"[Title/Abstract] OR "next generation sequencing test"[Title/Abstract] OR "next generation sequencing tests"[Title/Abstract] OR "NGS test"[Title/Abstract] OR "NGS tests"[Title/Abstract] OR "next generation sequencing technology"[Title/Abstract] OR "next generation sequencing technologies"[Title/Abstract] OR "NGS technology"[Title/Abstract] OR "NGS technologies"[Title/Abstract] OR "next generation sequencing panel"[Title/Abstract] OR "next generation sequencing panels"[Title/Abstract] OR "NGS panel"[Title/Abstract] OR "NGS panels"[Title/Abstract] OR "next generation sequencing profiling"[Title/Abstract] OR "NGS profiling"[Title/Abstract] OR "next generation sequencing application"[Title/Abstract] OR "next generation sequencing applications"[Title/Abstract] OR "NGS application"[Title/Abstract] OR "NGS applications"[Title/Abstract] OR "next generation sequencing screening"[Title/Abstract] OR "NGS screening"[Title/Abstract] OR "pharmacogenetic testing"[Title/Abstract] OR "pharmacogenetic test"[Title/Abstract] OR "pharmacogenetic tests"[Title/Abstract] OR "pharmacogenetic technology"[Title/Abstract] OR "pharmacogenetic technologies"[Title/Abstract] OR "pharmacogenetic panel"[Title/Abstract] OR "pharmacogenetic panels"[Title/Abstract] OR "pharmacogenetic profiling"[Title/Abstract] OR "pharmacogenetic application"[Title/Abstract] OR "pharmacogenetic applications"[Title/Abstract] OR "pharmacogenetic screening"[Title/Abstract] OR "pharmacogenomic testing"[Title/Abstract] OR "pharmacogenomic test"[Title/Abstract] OR "pharmacogenomic tests"[Title/Abstract] OR "pharmacogenomic technology"[Title/Abstract] OR "pharmacogenomic technologies"[Title/Abstract] OR "pharmacogenomic panel"[Title/Abstract] OR "pharmacogenomic panels"[Title/Abstract] OR "pharmacogenomic profiling"[Title/Abstract] OR "pharmacogenomic application"[Title/Abstract] OR "pharmacogenomic applications"[Title/Abstract] OR "pharmacogenomic screening"[Title/Abstract] OR "molecular testing"[Title/Abstract] OR "molecular test"[Title/Abstract] OR "molecular tests"[Title/Abstract] OR "molecular technology"[Title/Abstract] OR "molecular technologies"[Title/Abstract] OR "molecular panel"[Title/Abstract] OR "molecular panels"[Title/Abstract] OR "molecular profiling"[Title/Abstract] OR "molecular application"[Title/Abstract] OR "molecular applications"[Title/Abstract] OR "molecular screening"[Title/Abstract] OR "genetic predisposition testing"[Title/Abstract] OR "predictive testing"[Title/Abstract]) **AND** ("evaluation"[Title/Abstract] OR "evaluations"[Title/Abstract] OR "evaluate"[Title/Abstract] OR "evaluating"[Title/Abstract] OR "evaluated"[Title/Abstract] OR "assessment"[Title/Abstract] OR "assessments"[Title/Abstract] OR "assess"[Title/Abstract] OR "assessing"[Title/Abstract] OR "assessed"[Title/Abstract] OR "health technology assessment"[Title/Abstract] OR "HTA"[Title/Abstract] OR "evidence review"[Title/Abstract] OR "evidence reviews"[Title/Abstract] OR "appraisal"[Title/Abstract]) **AND** ("report*"[Title/Abstract] OR "summary"[Title/Abstract] OR "summaries" [Title/Abstract] OR "synthesis"[Title/Abstract] OR "guidance*"[Title/Abstract] OR "guideline*"[Title/Abstract] OR "recommendation*"[Title/Abstract] OR "appraisal"[Title/Abstract])

Scopus

TITLE-ABS-KEY("genetic testing" OR "genetic test" OR "genetic tests" OR "genetic technology" OR "genetic technologies" OR "genetic panel" OR "genetic panels" OR "gene panel" OR "gene panels" OR "genetic profiling" OR "genetic application" OR "genetic applications" OR "genetic screening" OR "genomic testing" OR "genomic test" OR "genomic tests" OR "genomic technology" OR "genomic technologies" OR "genomic panel" OR "genomic panels" OR "genome panel" OR "genome panels" OR "genomic profiling" OR "genomic application" OR "genomic applications" OR "genomic screening" OR "next generation sequencing testing" OR "NGS testing" OR "next generation sequencing test" OR "next generation sequencing tests" OR "NGS test" OR "NGS tests" OR "next generation sequencing technology" OR "next generation sequencing technologies" OR "NGS technology" OR "NGS technologies" OR "next generation sequencing panel" OR "next generation sequencing panels" OR "NGS panel" OR "NGS panels" OR "next generation sequencing profiling" OR "NGS profiling" OR "next generation sequencing application" OR "next generation sequencing applications" OR "NGS application" OR "NGS applications" OR "next generation sequencing screening" OR "NGS screening" OR "pharmacogenetic testing" OR "pharmacogenetic test" OR "pharmacogenetic tests" OR "pharmacogenetic technology" OR "pharmacogenetic technologies" OR "pharmacogenetic panel" OR "pharmacogenetic panels" OR "pharmacogenetic profiling" OR "pharmacogenetic application" OR "pharmacogenetic applications" OR "pharmacogenetic screening" OR "pharmacogenomic testing" OR "pharmacogenomic test" OR "pharmacogenomic tests" OR "pharmacogenomic technology" OR "pharmacogenomic technologies" OR "pharmacogenomic panel" OR "pharmacogenomic panels" OR "pharmacogenomic profiling" OR "pharmacogenomic application" OR "pharmacogenomic applications" OR "pharmacogenomic screening" OR "molecular testing" OR "molecular test" OR "molecular tests" OR "molecular technology" OR "molecular technologies" OR "molecular panel" OR "molecular panels" OR "molecular profiling" OR "molecular application" OR "molecular applications" OR "molecular screening" OR "genetic predisposition testing" OR "predictive testing") **AND** TITLE-ABS-KEY ("evaluation" OR "evaluations" OR "evaluate" OR "evaluating" OR "evaluated" OR "assessment" OR "assessments" OR "assess" OR "assessing" OR "assessed" OR "health technology assessment" OR "HTA" OR "evidence review" OR "evidence reviews" OR "appraisal") **AND** TITLE-ABS-KEY ("report*" OR "summary" OR "summaries" OR "synthesis" OR "guidance*" OR "guideline*" OR "recommendation*" OR "appraisal")

Web of Science

TS=("genetic testing" OR "genetic test" OR "genetic tests" OR "genetic technology" OR "genetic technologies" OR "genetic panel" OR "genetic panels" OR "gene panel" OR "gene panels" OR "genetic profiling" OR "genetic application" OR "genetic applications" OR "genetic screening" OR "genomic testing" OR "genomic test" OR "genomic tests" OR "genomic technology" OR "genomic technologies" OR "genomic panel" OR "genomic panels" OR "genome panel" OR "genome panels" OR "genomic profiling" OR "genomic application" OR "genomic applications" OR "genomic screening" OR "next generation sequencing testing" OR "NGS testing" OR "next generation sequencing test" OR "next generation sequencing tests" OR "NGS test" OR "NGS tests" OR "next generation sequencing technology" OR "next generation sequencing technologies" OR "NGS technology" OR "NGS technologies" OR "next generation sequencing panel" OR "next generation sequencing panels" OR "NGS panel" OR "NGS panels" OR "next generation sequencing profiling" OR "NGS profiling" OR "next generation sequencing application" OR "next generation sequencing applications" OR "NGS application" OR "NGS applications" OR "next generation sequencing screening" OR "NGS screening" OR "pharmacogenetic testing" OR "pharmacogenetic test" OR "pharmacogenetic tests" OR "pharmacogenetic technology" OR "pharmacogenetic technologies" OR "pharmacogenetic panel" OR "pharmacogenetic panels" OR "pharmacogenetic profiling" OR "pharmacogenetic application" OR "pharmacogenetic applications" OR "pharmacogenetic screening" OR "pharmacogenomic testing" OR "pharmacogenomic test" OR "pharmacogenomic tests" OR "pharmacogenomic technology" OR "pharmacogenomic technologies" OR "pharmacogenomic panel" OR "pharmacogenomic panels" OR "pharmacogenomic profiling" OR

“pharmacogenomic application” OR “pharmacogenomic applications” OR “pharmacogenomic screening” OR “molecular testing” OR “molecular test” OR “molecular tests” OR “molecular technology” OR “molecular technologies” OR “molecular panel” OR “molecular panels” OR “molecular profiling” OR “molecular application” OR “molecular applications” OR “molecular screening” OR “genetic predisposition testing” OR “predictive testing”) **AND** TS=(“evaluation” OR “evaluations” OR “evaluate” OR “evaluating” OR “evaluated” OR “assessment” OR “assessments” OR “assess” OR “assessing” OR “assessed” OR “health technology assessment” OR “HTA” OR “evidence review” OR “evidence reviews” OR “appraisal”) **AND** TS=(“report*” OR “summary” OR “summaries” OR “synthesis” OR “guidance*” OR “guideline*” OR “recommendation*” OR “appraisal”)

International HTA database

Title((“genetic testing” OR “genetic test” OR “genetic tests” OR “genetic technology” OR “genetic technologies” OR “genetic panel” OR “genetic panels” OR “gene panel” OR “gene panels” OR “genetic profiling” OR “genetic application” OR “genetic applications” OR “genetic screening” OR “genomic testing” OR “genomic test” OR “genomic tests” OR “genomic technology” OR “genomic technologies” OR “genomic panel” OR “genomic panels” OR “genome panel” OR “genome panels” OR “genomic profiling” OR “genomic application” OR “genomic applications” OR “genomic screening” OR “next generation sequencing testing” OR “NGS testing” OR “next generation sequencing test” OR “next generation sequencing tests” OR “NGS test” OR “NGS tests” OR “next generation sequencing technology” OR “next generation sequencing technologies” OR “NGS technology” OR “NGS technologies” OR “next generation sequencing panel” OR “next generation sequencing panels” OR “NGS panel” OR “NGS panels” OR “next generation sequencing profiling” OR “NGS profiling” OR “next generation sequencing application” OR “next generation sequencing applications” OR “NGS application” OR “NGS applications” OR “next generation sequencing screening” OR “NGS screening” OR “pharmacogenetic testing” OR “pharmacogenetic test” OR “pharmacogenetic tests” OR “pharmacogenetic technology” OR “pharmacogenetic technologies” OR “pharmacogenetic panel” OR “pharmacogenetic panels” OR “pharmacogenetic profiling” OR “pharmacogenetic application” OR “pharmacogenetic applications” OR “pharmacogenetic screening” OR “pharmacogenomic testing” OR “pharmacogenomic test” OR “pharmacogenomic tests” OR “pharmacogenomic technology” OR “pharmacogenomic technologies” OR “pharmacogenomic panel” OR “pharmacogenomic panels” OR “pharmacogenomic profiling” OR “pharmacogenomic application” OR “pharmacogenomic applications” OR “pharmacogenomic screening” OR “molecular testing” OR “molecular test” OR “molecular tests” OR “molecular technology” OR “molecular technologies” OR “molecular panel” OR “molecular panels” OR “molecular profiling” OR “molecular application” OR “molecular applications” OR “molecular screening” OR “genetic predisposition testing” OR “predictive testing”) **AND** (“evaluation” OR “evaluations” OR “evaluate” OR “evaluating” OR “evaluated” OR “assessment” OR “assessments” OR “assess” OR “assessing” OR “assessed” OR “health technology assessment” OR “HTA” OR “evidence review” OR “evidence reviews” OR “appraisal”)) **OR Abstracts**((“genetic testing” OR “genetic test” OR “genetic tests” OR “genetic technology” OR “genetic technologies” OR “genetic panel” OR “genetic panels” OR “gene panel” OR “gene panels” OR “genetic profiling” OR “genetic application” OR “genetic applications” OR “genetic screening” OR “genomic testing” OR “genomic test” OR “genomic tests” OR “genomic technology” OR “genomic technologies” OR “genomic panel” OR “genomic panels” OR “genome panel” OR “genome panels” OR “genomic profiling” OR “genomic application” OR “genomic applications” OR “genomic screening” OR “next generation sequencing testing” OR “NGS testing” OR “next generation sequencing test” OR “next generation sequencing tests” OR “NGS test” OR “NGS tests” OR “next generation sequencing technology” OR “next generation sequencing technologies” OR “NGS technology” OR “NGS technologies” OR “next generation sequencing panel” OR “next generation sequencing panels” OR “NGS panel” OR “NGS panels” OR “next generation sequencing profiling” OR “NGS profiling” OR “next generation sequencing application” OR “next generation sequencing applications” OR “NGS application” OR “NGS applications” OR “next generation sequencing screening” OR “NGS screening” OR “pharmacogenetic testing” OR “pharmacogenetic test” OR “pharmacogenetic tests” OR “pharmacogenetic technology” OR “pharmacogenetic technologies” OR “pharmacogenetic panel” OR “pharmacogenetic panels” OR “pharmacogenetic profiling” OR “pharmacogenetic application” OR “pharmacogenetic applications” OR “pharmacogenetic screening” OR “pharmacogenomic testing” OR “pharmacogenomic test” OR “pharmacogenomic tests” OR “pharmacogenomic technology” OR “pharmacogenomic technologies” OR “pharmacogenomic panel” OR “pharmacogenomic panels” OR “pharmacogenomic profiling” OR “pharmacogenomic application” OR “pharmacogenomic applications” OR “pharmacogenomic screening” OR “molecular testing” OR “molecular test” OR “molecular tests” OR “molecular technology” OR “molecular technologies” OR “molecular panel” OR “molecular panels” OR “molecular profiling” OR “molecular application” OR “molecular applications” OR “molecular screening” OR “genetic predisposition testing” OR “predictive testing”) **AND** (“evaluation” OR “evaluations” OR “evaluate” OR “evaluating” OR “evaluated” OR “assessment” OR “assessments” OR “assess” OR “assessing” OR “assessed” OR “health technology assessment” OR “HTA” OR “evidence review” OR “evidence reviews” OR “appraisal”))

Supplementary Table 2. Assessment components of reports included in the systematic review, by domain (N=41).

	N (%)	Reference
Disease/condition of interest		
Epidemiology		
Investigated, evidence found/produced	41 (100.0)	[17–57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	0 (0.0)	–
Etiopathogenesis		
Investigated, evidence found/produced	26 (63.4)	[17–20,22,26,28,29,31,33–36,38,41–44,48,49, 51–55,57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	15 (36.6)	[21,23–25,27,30,32,37,39,40,45–47,50,56]
Natural history		
Investigated, evidence found/produced	37 (90.2)	[17–39,41–45,47–52,54,55,57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	4 (9.8)	[40,46,53,56]
Current management		
Investigated, evidence found/produced	40 (97.6)	[17–29,31–57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	1 (2.4)	[30]
Application		
Analytic accuracy		
Investigated, evidence found/produced	16 (39.0)	[17,22,23,28,30–33,35–37,43,45,52,53,56]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	25 (61.0)	[18–21,24–27,29,34,38–42,44,46–51,54,55,57]
Clinical aspects		
Clinical accuracy		
Investigated, evidence found/produced	22 (53.7)	[17–19,21,23,27,28,32–35,38,40,42,43,46,48,50,51,54,55,57]
Investigated, no evidence found/produced	3 (7.3)	[26,29,52]
Not investigated	16 (39.0)	[20,22,24,25,30,31,36,37,39,41,44,45,47,49,53,56]
Efficacy		
Investigated, evidence found/produced	16 (39.0)	[21,23–25,32,33,38,40,41,44,45,47–50,56]
Investigated, no evidence found/produced	19 (46.3)	[19,20,22,26,27,29,30,35–37,39,43,46,51–55,57]
Not investigated	6 (14.6)	[17,18,28,31,34,42]
Effectiveness		
Investigated, evidence found/produced	26 (63.4)	[22,23,26–30,32,35,37–48,50,54–57]
Investigated, no evidence found/produced	6 (14.6)	[18,19,36,51–53]
Not investigated	9 (22.0)	[17,20,21,24,25,31,33,34,49]
Safety		
Investigated, evidence found/produced	14 (34.1)	[25,26,29,34,35,38,40,43,46,48,49,55–57]
Investigated, no evidence found/produced	9 (22.0)	[19,20,27,30,42,45,47,51,53]
Not investigated	18 (43.9)	[17,18,21–24,28,31–33,36,37,39,41,44,50,52,54]
Patient-reported outcomes		
Investigated, evidence found/produced	9 (22.0)	[17,18,24,25,34,38,41,43,47]
Investigated, no evidence found/produced	5 (12.2)	[20,42,48,53,54]
Not investigated	27 (65.9)	[19,21–23,26–33,35–37,39,40,44–46,49–52, 55–57]
Post-test clinical pathways		
Investigated, evidence found/produced	34 (82.9)	[17,19–27,29,33–36,38–46,48–57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	7 (17.1)	[18,28,30–32,37,47]
Personal and societal aspects		
Non-health-related outcomes		
Investigated, evidence found/produced	21 (51.2)	[17,18,20,22,23,35,37–39,42,43,45–50,53–55,57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	20 (48.8)	[19,21,24–34,36,40,41,44,51,52,56]
Ethical aspects		
Investigated, evidence found/produced	11 (26.8)	[18,23,27,29,35,38,42,46,50,54,57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	30 (73.2)	[17,19–22,24–26,28,30–34,36,37,39–41, 43–45,47–49,51–53,55,56]
Legal aspects		
Investigated, evidence found/produced	2 (4.9)	[35,38]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	39 (95.1)	[17–34,36,37,39–57]
Social aspects		
Investigated, evidence found/produced	9 (22.0)	[18,23,29,38,42,46,47,50,57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	32 (78.0)	[17,19–22,24–28,30–37,39–41,43–45,48,49, 51–56]

Organizational aspects		
Burden of disease		
Investigated, evidence found/produced	37 (90.2)	[17–25,27,29,31–38,40–57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	4 (9.8)	[26,28,30,39]
Professional resources need		
Investigated, evidence found/produced	24 (58.5)	[17–19,21,25,27,29,31,34,37,38,40–43,45, 48–50,52–55,57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	17 (41.5)	[20,22–24,26,28,30,32,33,35,36,39,44,46,47,51,56]
Material resources need		
Investigated, evidence found/produced	32 (78.0)	[17–19,21,22,25,27,29,31–38,40–43,45–50,52–57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	9 (22.0)	[20,23,24,26,28,30,39,44,51]
Financial resources need		
Investigated, evidence found/produced	34 (82.9)	[17–21,23,25,27,29,31–38,40–43,45–57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	7 (17.1)	[22,24,26,28,30,39,44]
Budget Impact Analysis		
Investigated, evidence found/produced	22 (53.7)	[17,19,23–25,27,29,32,37,40–42,45–50,54–57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	19 (46.3)	[18,20–22,26,28,30,31,33–36,38,39,43,44,51–53]
Equity in access		
Investigated, evidence found/produced	12 (29.3)	[19,22,31,35,36,38,44,46,48,55–57]
Investigated, no evidence found/produced	0 (0.0)	–
Not investigated	29 (70.7)	[17,18,20,21,23–30,32–34,37,39–43,45,47,49–54]
Economic evaluations		
Investigated, evidence found/produced	34 (82.9)	[17,18,20,21,23–27,29,31–36,38–51,54–57]
Investigated, no evidence found/produced	3 (7.3)	[19,22,37]
Not investigated	4 (9.8)	[28,30,52,53]

Supplementary Table 3. Gaps reported in component assessment, by domain. The denominator (N) is the overall number of reports that investigated a single component.

Domain	Component	Gap	N (%)	Reference
Disease/condition of interest	Epidemiology (N=41)	Target population potentially under-represented	3 (7.3)	[18,38,44]
	Etiopathogenesis (N=26)	No gap reported	–	–
	Natural history (N=37)	No gap reported	–	–
	Current management (N=40)	Need for updated clinical practice guidelines/lack of standardized clinical pathways	6 (15.0)	[17,18,21,29,46,49]
Application	Analytic accuracy (N=16)	Concerns on quality of evidence	4 (25.0)	[31,34,52,53]
		Invasive test procedure	1 (6.2)	[17]
		Lack of evidence	5 (31.2)	[22,23,28,30,56]
		Limited generalizability/applicability of findings	2 (12.5)	[31,36]
		Need for larger gene panel	2 (12.5)	[33,49]
		Need for reference standard/thresholds	5 (31.2)	[28,31–33,45]
		Poor accuracy of test	1 (6.2)	[45]
Clinical aspects	Clinical accuracy (N=25)	Concerns on quality of evidence	4 (16.0)	[27,34,55,57]
		Lack of evidence	11 (44.0)	[26,28,29,32–34,42,48,51,52,57]
		Limited generalizability/applicability of findings	9 (36.0)	[18,19,26,27,32,33,35,55,57]
		Need for larger gene panel	1 (4.0)	[21]
		Need for reference standard/thresholds	6 (24.0)	[19,27,32,34,42,57]
		Target population potentially under-represented	2 (8.0)	[50,55]
	Efficacy (N=35)	Concerns on quality of evidence	3 (8.6)	[25,37,41]
		Lack of evidence	30 (85.7)	[19,20,22–24,26,27,29,30,32,33,35–37,39–43,45–49, 51–55,57]
		Limited generalizability/applicability of findings	14 (40.0)	[21,23,27,29,30,33,35,38,40,47,49,50,54,56]
	Effectiveness (N=32)	Concerns on quality of evidence	5 (15.6)	[23,28,37,41,46]
		Lack of evidence	20 (62.5)	[18,19,22,26–28,30,32,36,40–42,45,47,48,51–55]
		Limited generalizability/applicability of findings	16 (50.0)	[22,26,27,29,30,35,37–40,43,47,50,54,56,57]
		Target population potentially under-represented	2 (6.2)	[22,26]
	Safety (N=23)	Concerns on safety	2 (8.7)	[40,46]
		Lack of evidence	12 (52.2)	[19,20,27,30,38,42,45,47,48,51–53]
		Limited generalizability/applicability of findings	3 (13.0)	[29,43,56]
	Patient-reported outcomes (N=14)	Lack of evidence	7 (50.0)	[20,42,43,48,52–54]
		Limited generalizability/applicability of findings	1 (7.1)	[18]
	Post-test clinical pathways (N=34)	Lack of evidence	2 (5.9)	[17,22]
		Limited generalizability/applicability of findings	1 (2.9)	[38]
		Need for updated clinical practice guidelines/lack of standardized clinical pathways	3 (8.8)	[17,22,33]
Personal and societal aspects	Non-health-related outcomes (N=21)	Lack of evidence	4 (19.0)	[22,42,45,50]
		Limited generalizability/applicability of findings	9 (42.9)	[17,18,45–47,49,50,54,55]
		Need for updated clinical practice guidelines/lack of standardized clinical pathways	1 (4.8)	[47]
	Ethical aspects (N=11)	No gap reported	–	–
	Legal aspects (N=2)	No gap reported	–	–
	Social aspects (N=9)	Lack of evidence	1 (11.1)	[50]
Organizational aspects	Burden of disease (N=37)	No gap reported	–	–
	Professional resources need (N=24)	Lack of data/difficulty in estimating costs	2 (8.3)	[52,53]
	Material resources need (N=32)	Lack of data/difficulty in estimating costs	7 (21.9)	[26,34,45,47,52,53,57]
	Financial resources need (N=34)	Lack of data/difficulty in estimating costs	2 (5.9)	[46,57]
	Budget Impact Analysis (N=22)	Lack of data/difficulty in estimating costs	2 (9.1)	[54,55]
		Limited generalizability/applicability of findings	1 (4.5)	[50]
	Equity in access (N=12)	No gap reported	–	–
	Economic evaluations (N=37)	Lack of evidence	11 (29.7)	[18,19,22,26,27,29,37,40,42,48,57]
		Limited generalizability/applicability of findings	27 (73.0)	[17,18,20,21,26,27,31–33,36,39–51,54–57]

References

17. Grimshaw G, Szczepura A, Hulten M, MacDonald F, Nevin N. Evaluation of molecular tests for prenatal diagnosis of chromosome abnormalities. Health Technology Assessment [Internet]. 2003 May 19;7(10):156. Available from: http//journalslibrary.nihr.ac.uk/hta/hta7100

18. Bryant J, Cooper K, Picot J, Clegg A, Roderick P, Rosenberg W, et al. Diagnostic strategies using DNA testing for hereditary haemochromatosis in at-risk populations: a systematic review and economic evaluation. Health Technol Assess [Internet]. 2009;13:23. Available from: https://doi.org/10.3310/hta13230

19. Buckley L, Wang S, Merlin T. Molecular testing for myeloproliferative disease [Internet]. Medical Services Advisory Committee; 2009. Available from: msac.gov.au/internet/msac/publishing.nsf/Content/049E5816272060BCCA25801000123B46

20. Simpson E, Stevenson M, Rawdin A, Papaioannou D. Thrombophilia testing in people with venous thromboembolism: systematic review and cost-effectiveness analysis. Health Technol Assess [Internet]. 2009;13:2. Available from: https://doi.org/10.3310/hta13020

21. Donnan JR, Ungar WJ, Mathews M, Hancock-Howard R. Health technology assessment of thiopurine methyltransferase testing for guiding 6-mercaptopurine doses in pediatric patients with acute lymphoblastic leukemia [Internet]. The Hospital for Sick Children Technology Assessment Task Force; 2010. Available from: tspace.library.utoronto.ca/handle/1807/81028

22. Fleeman N, McLeod C, Bagust A, Beale S, Boland A, Dundar Y, et al. The clinical effectiveness and cost-effectiveness of testing for cytochrome P450 polymorphisms in patients with schizophrenia treated with antipsychotics: a systematic review and economic evaluation. Health Technol Assess [Internet]. 2010;14:3. Available from: https://doi.org/10.3310/hta14030

23. Medical Advisory Secretariat. Gene expression profiling for guiding adjuvant chemotherapy decisions in women with early breast cancer: an evidence-based and economic analysis. Ontario health technology assessment series [Internet]. 2010;10(23):1–57. Available from: https://pmc.ncbi.nlm.nih.gov/articles/PMC3382301/

24. Medical Advisory Secretariat. Epidermal Growth Factor Receptor Mutation (EGFR) Testing for Prediction of Response to EGFR-Targeting Tyrosine Kinase Inhibitor (TKI) Drugs in Patients with Advanced Non-Small-Cell Lung Cancer: An Evidence-Based Analysis. Ontario health technology assessment series [Internet]. 2010;10(24):1–48. Available from: https://pmc.ncbi.nlm.nih.gov/articles/PMC3377519/

25. Medical Advisory Secretariat. KRAS Testing for Anti-EGFR Therapy in Advanced Colorectal Cancer: An Evidence-Based and Economic Analysis. Ontario health technology assessment series [Internet]. 2010;10(25):1–49. Available from: https://pmc.ncbi.nlm.nih.gov/articles/PMC3377508/

26. Fleeman N, Saborido CM, Payne K, Boland A, Dickson R, Dundar Y, et al. The clinical effectiveness and cost-effectiveness of genotyping for CYP2D6 for the management of women with breast cancer treated with tamoxifen: a systematic review. Health Technol Assess [Internet]. 2011;15:33. Available from: https://doi.org/10.3310/hta15330

27. Morona JK, Newton S, Wang S, Tamblyn D, Ellery B, Merlin T. Genetic testing for hereditary mutations in the VHL gene that cause von Hippel-Lindau syndrome [Internet]. Medical Services Advisory Committee; 2011. Available from: msac.gov.au/internet/msac/publishing.nsf/Content/3474C4A5A7E262E5CA25801000123B1A

28. Meleth S, Whitehead N, Evans TS, Lux L. Technology Assessment on Genetic Testing or Molecular Pathology Testing of Cancers with Unknown Primary Site to Determine Origin. Rockville (MD); 2013.

29. Newton S, Schubert C, Morona J, Fitzgerald P, Merlin T. Genetic testing for hereditary mutations in the RET gene [Internet]. Medical Services Advisory Committee; 2013. Available from: msac.gov.au/internet/msac/publishing.nsf/Content/F6C5F35C618BE08CCA25801000123B22

30. Meleth S, Reeder-Hayes K, Ashok M, Clark R, Funkhouser W, Wines R, et al. Technology assessment of molecular pathology testing for the estimation of prognosis for common cancers. Agency for Healthcare Research and Quality; 2014.

31. Snowsill T, Huxley N, Hoyle M, Jones-Hughes T, Coelho H, Cooper C, et al. A systematic review and economic evaluation of diagnostic strategies for Lynch syndrome. Health Technol Assess [Internet]. 2014 Sep 22;18(58). Available from: http://journalslibrary.nihr.ac.uk/hta/hta18580

32. Westwood M, Joore M, Whiting P, van Asselt T, Ramaekers B, Armstrong N, et al. Epidermal growth factor receptor tyrosine kinase (EGFR-TK) mutation testing in adults with locally advanced or metastatic non-small cell lung cancer: a systematic review and cost-effectiveness analysis. Health Technol Assess [Internet]. 2014 May 15;18(32). Available from: http://journalslibrary.nihr.ac.uk/hta/hta18320

33. Westwood M, van Asselt T, Ramaekers B, Whiting P, Joore M, Armstrong N, et al. KRAS mutation testing of tumours in adults with metastatic colorectal cancer: a systematic review and cost-effectiveness analysis. Health Technol Assess [Internet]. 2014 Oct 15;18(62). Available from: http://journalslibrary.nihr.ac.uk/hta/hta18620

34. Huxley N, Jones-Hughes T, Coelho H, Snowsill T, Cooper C, Meng Y, et al. A systematic review and economic evaluation of intraoperative tests [RD-100i one-step nucleic acid amplification (OSNA) system and Metasin test] for detecting sentinel lymph node metastases in breast cancer. Health Technol Assess [Internet]. 2015 Jan 14;19(2). Available from: http://journalslibrary.nihr.ac.uk/hta/hta19020

35. Kessels SJM, Morona JK, Mittal R, Vogan A, Newton S, Schubert C, et al. Testing for hereditary mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene [Internet]. Medical Services Advisory Committee; 2015. Available from: msac.gov.au/internet/msac/publishing.nsf/Content/D54C11627FEA9785CA25801000123BDC

36. Snowsill T, Coelho H, Huxley N, Jones-Hughes T, Briscoe S, Frayling I, et al. Molecular testing for Lynch syndrome in people with colorectal cancer: systematic reviews and economic evaluation. Health Technol Assess [Internet]. 2017 Sep 12;21(51):280. Available from: https://doi.org/10.3310/hta21510

37. Zierler A, Mohammed M, Soulodre C, Laing A, Kaulback K, Mitchell A, et al. Prolaris Cell Cycle Progression Test for Localized Prostate Cancer: A Health Technology Assessment. Ontario health technology assessment series. 2017;17(6):1–75.

38. Varela-Lema L, Puñal-Riobóo J, Ballini L. Screening of fetal trisomies 21, 18 and 13 by noninvasive prenatal testing [Internet]. EUnetHTA; 2018. Available from: eunethta.eu/wp4-otca03-screening-fetal-trisomies-21-18-13-non-invasive-prenatal-testing-now-available/

39. Elston L, Hughes S, Prettyjohns M, Washington J. Pharmacogenetic testing to identify the risk of adverse reactions to anti-epileptic medications [Internet]. Health Technology Wales; 2019. Available from: healthtechnology.wales/reports-guidance/pharmacogenetic-testing/

40. Fagerlund BC, Chudasama KK, Brurberg KG, Robberstad B, Rose C, Juvet LK, et al. Prosigna Gene Signature to Assess Expected Benefit from Chemotherapy in Breast Cancer. Assessment of manufacturer's submission [Internet]. Norwegian Institute of Public Health; 2019. Available from: fhi.no/en/publ/2019/Prosigna-Gene-Signature-to-Assess-Expected-Benefit/

41. Harnan S, Tappenden P, Cooper K, Stevens J, Bessey A, Rafia R, et al. Tumour profiling tests to guide adjuvant chemotherapy decisions in early breast cancer: a systematic review and economic analysis. Health Technol Assess [Internet]. 2019;23:30. Available from: https://doi.org/10.3310/hta23300

42. McKane J, Wang M, Xie X, Tu HA, Gilbert J, Higgins C, et al. Noninvasive Prenatal Testing for Trisomies 21, 18, and 13, Sex Chromosome Aneuploidies, and Microdeletions: A Health Technology Assessment. Ontario health technology assessment series. 2019;19(4):1–166.

43. Stürzlinger H, Conrads-Frank A, Eisenmann A, Ivansits S, Jahn B, Janzic A, et al. Stool DNA testing for early detection of colorectal cancer [Internet]. EUnetHTA; 2019. Available from: eunethta.eu/otja10-final-assessment-report-and-related-documents-are-now-available/

44. Berg G, Deogaonkar R, Harbour J, MacGann T, Stewart J. Pre-treatment DPYD genetic testing for patients who are prescribed chemotherapy involving fluoropyrimidines [Internet]. Healthcare Improvement Scotland; 2020. Available from: shtg.scot/our-advice/pre-treatment-dpyd-genetic-testing-for-patients-who-are-prescribed-chemotherapy-involving-fluoropyrimidines/

45. Betsch EJ, Maguire T, Lambrinos A, Falk L, Gajic-Veljanoski O, Cheng L, et al. Cell-Free Circulating Tumour DNA Blood Testing to Detect EGFR T790M Mutation in People With Advanced Non-Small Cell Lung Cancer: A Health Technology Assessment. Ontario health technology assessment series. 2020;20(5):1–176.

46. Betsch E, Herington E, McCormack S, Duthie K, Vandersluis S, Li CM, et al. Genome-Wide Sequencing for Unexplained Developmental Disabilities or Multiple Congenital Anomalies: A Health Technology Assessment. Ontario health technology assessment series. 2020;20(11):1–178.

47. Maguire T, Wang M, Zhang Y, Shafique A, Walter M, Smith A, et al. Gene Expression Profiling Tests for Early-Stage Invasive Breast Cancer: A Health Technology Assessment. Ontario health technology assessment series. 2020;20(10):1–234.

48. Zierler A, Schaink A, Gajic-Veljanoski O, Li C, Guo J, Wells D, et al. Noninvasive Fetal RhD Blood Group Genotyping: A Health Technology Assessment. Ontario health technology assessment series. 2020;20(15):1–160.

49. Betsch E, Jokic M, Vandersluis S, Higgins C, Gajic-Veljanoski O, Xie X, et al. Multi-gene Pharmacogenomic Testing That Includes Decision-Support Tools to Guide Medication Selection for Major Depression: A Health Technology Assessment. Ontario health technology assessment series. 2021;21(13):1–214.

50. Maguire T, McKane J, Turnbull J, Soulodre C, Harrison S, Abdolzahraei S, et al. DPYD Genotyping in Patients Who Have Planned Cancer Treatment With Fluoropyrimidines: A Health Technology Assessment. Ontario health technology assessment series. 2021;21(14):1–186.

51. Stinton C, Jordan M, Fraser H, Auguste P, Court R, Al-Khudairy L, et al. Testing strategies for Lynch syndrome in people with endometrial cancer: systematic reviews and economic evaluation. Health Technol Assess [Internet]. 2021;25:42. Available from: https://doi.org/10.3310/hta25420

52. Flodgren GM, Hamidi V, Meneses Echavez JE, Bidonde J. Molecular tests for detection of PIK3CA mutations in men and postmenopausal women with HR+/HER2–, locally advanced or metastatic breast cancer: a health technology assessment [Internet]. Norwegian Insitutute of Public Health; 2022. Available from: fhi.no/en/publ/2022/Molecular-tests-for-detection-of-PIK3CA-mutations-in-men-and-postmenopausal-women-with-locally-advanced-metastatic-breast-cancer/

53. Hamidi V, Flodgren GM, Meneses-Echavez JF, Bidonde J. Tests for the detection of NTRK gene fusions in patients with locally advanced or metastatic solid tumours. A Health Technology Assessment [Internet]. Norwegian Institute of Public Health; 2022. Report No.: ID2019_119 and ID2019_029. Available from: fhi.no/publ/2022/tester-for-deteksjon-av-ntrk-genfusjoner-hos-pasienter-med-lokalavanserte-e/

54. Maguire T, Kabali C, Lambrinos A, Holubowich C, Zhang Y, Gajic-Veljanoski O, et al. Genetic Testing for Familial Hypercholesterolemia: Health Technology Assessment. Ontario health technology assessment series. 2022;22(3):1–155.

55. McKane J, Vandersluis S, Holubowich C, Li C, Xie X, Wells D, et al. Molecular Testing for Thyroid Nodules of Indeterminate Cytology: A Health Technology Assessment. Ontario health technology assessment series. 2022;22(2):1–111.

56. Newton S, Vogan A, Lett B, Liufu V, Parsons J. Small gene panel testing for non-squamous non-small cell lung cancer [Internet]. Medical Services Advisory Committee; 2022. Available from: msac.gov.au/internet/msac/publishing.nsf/Content/1721-public

57. Comber L, Byrne D, Carty P, Finnegan É, Carrigan M, Weir A, et al. Health technology assessment of the addition of severe combined immunodeficiency (SCID) to the National Newborn Bloodspot Screening Programme [Internet]. Health Information and Quality Authority; 2023. Available from: hiqa.ie/reports-and-publications/health-technology-assessment/hta-addition-severe-combined-immunodeficiency