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Risk-based stratification for genetic and modifiable factors in non-communicable diseases prevention: a scoping review of current guidance

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Research Article

Keywords:

Posted Date: May 26th, 2026

DOI: https://doi.org/10.21203/rs.3.rs-9413274/v1

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Additional Declarations: No competing interests reported.

Abstract

Introduction.

Non-communicable diseases (NCDs) are the leading cause of global morbidity and mortality, resulting from the interaction between genetic susceptibility and modifiable risk factors. Conventional prevention strategies are largely population-based and do not account for inter-individual risk heterogeneity. Risk-based stratified approaches aim to personalize interventions based on genetic and non-genetic factors, potentially improving effectiveness and efficiency. This study aims to map prevention approaches for major NCDs in which preventive interventions are stratified according to genetic risk and/or modifiable risk factors.

Methods.

We conducted a scoping review of international guidelines, recommendations, and consensus statements published between 2019 and June 2025 identified through database searches and grey literature. Documents were included if preventive approaches were explicitly stratified by genetic and/or modifiable risk factors. We categorized extracted data by disease area, level of prevention, stratification criteria, and type of preventive intervention.

Results.

We included 114 documents, of which 74 (64.9%) focused exclusively on genetic factors, 37 (32.5%) on modifiable risk factors, and 3 (2.6%) addressed both, yielding 661 risk-stratified prevention approaches, of which 632 were unique. Overall, approaches were predominantly based on genetic factors (n = 541), whereas 91 addressed modifiable risk factors, mainly tobacco use. Secondary prevention accounted for 51.9% of all approaches. Strategies based on modifiable risk factors more often targeted primary prevention (52.7%) and cardiovascular diseases (76.9%). In contrast, genetically stratified approaches primarily addressed cancer (78.6%) and were more frequent in secondary prevention settings (56.4%). Tertiary prevention approaches were distributed similarly between genetic and modifiable risk factors (n = 165, 26.8% vs. 22.0%).

Conclusions.

Current prevention guidance for major NCDs predominantly relies on genetic risk stratification, particularly in cancer and beyond primary prevention. Modifiable risk stratification remains underrepresented, highlighting a missed opportunity to strengthen risk-stratified prevention. Greater integration of behavioral and lifestyle risk factors may enhance the effectiveness and sustainability of prevention strategies.

Introduction

Non-communicable diseases (NCDs), including cardiovascular diseases (CVD), cancers, chronic respiratory diseases (CRD), diabetes mellitus (DM), and mental health conditions, are responsible for the majority of global morbidity and mortality [1]. In 2021, NCDs accounted for approximately 43 million deaths worldwide, representing about 75% of all non-pandemic-related mortality. The majority of these deaths occur in low- and middle-income countries, which collectively bear over 70% of the global NCD mortality burden [2, 3]. Among these, cardiovascular diseases remain the leading cause of death, followed by cancers, chronic respiratory diseases, and diabetes, together accounting for roughly 80% of premature NCD mortality [4]. Cancer, in particular, contributes substantially to this burden and is highly influenced by modifiable risk factors. Fink et al. estimated that in 2022, 7.1 million of 18.7 million new cancer cases (approximately 38%) were attributable to 30 modifiable exposures (including tobacco use, alcohol consumption, high body mass index, physical inactivity, infections, and environmental exposures) while lung, stomach, and cervical cancers alone accounted for nearly half of all preventable cases [5].

NCDs arise from a complex interplay of genetic, physiological, environmental, and behavioral determinants. Both fixed factors (e.g., genetic susceptibility) and modifiable risk factors (such as tobacco use, harmful alcohol consumption, unhealthy diets, insufficient physical activity, and exposure to air pollution) substantially contribute to disease onset and progression. Despite this multifactorial etiology, prevention strategies have traditionally relied on population-wide approaches and uniform public health messaging. Even when preventive measures involve pharmacological treatment, early clinical management, or structured screening programs, they are frequently implemented using broad and standardized eligibility criteria rather than individualized risk profiles. As a result, such “one-size-fits-all” approaches often fail to account for substantial inter-individual heterogeneity in susceptibility and response, limiting their effectiveness across diverse populations[5–8].In recent years, growing interest has emerged in prevention approaches based on risk stratification, aiming to tailor preventive strategies to individual characteristics through the integration of genetic, behavioral, environmental, and socio-economic data [9].

By explicitly addressing individual variability, these approaches enhance the identification of individuals at higher risk, to optimize resource allocation, and improve both intervention effectiveness and adherence, representing a meaningful advancement beyond uniform interventions [10].

Recently, Farina et al. [11], mapped personalized prevention approaches for NCDs, based on main genetic testing from primary studies and guidelines. In our study, however, we aim to summarise existing preventive guidance adopting a risk-stratified approach that extends beyond genetics.

Guidelines, recommendations, and consensus statements represent the most consolidated and practice-oriented expressions of expert agreement, integrating available evidence with contextual and implementation considerations. Although the publication of such documents does not guarantee their

uptake in practice, they provide a robust and validated benchmark of current prevention strategies and policy orientations [12].

Within this context, the objective of this study is to summarize global guidelines, recommendations, and consensus statements addressing prevention approaches for major NCDs in which preventive interventions are stratified according to genetic risk and/or modifiable risk factors.

Materials and methods

We conducted this scoping review in the context of the Joint Action Prevent Non-Communicable Diseases (JA PreventNCD), funded by EU4Health program [13], supporting strategies and policies to reduce the burden of cancer and other NCDs through comprehensive prevention approaches.

The review followed the five-stage methodological framework proposed by Arksey and O'Malley [14] and adhered to the PRISMA-ScR guidelines [15]. The review protocol was registered and is publicly available on the Open Science Framework (DOI: 10.17605/OSF.IO/8UK96).

Search strategy

We searched PubMed, Web of Science, and Scopus, along with the first 100 results retrieved from Google and Google Scholar and relevant organizational websites, to identify English-language guidelines, recommendations, and consensus statements on the prevention of major NCDs. We included documents published between January 2019 and June 2025 to capture the most recent and updated guidance.

The search focused on primary, secondary, and tertiary prevention approaches for cancer, CVD, DM, CRD, and mental health conditions. These conditions were selected because they represent the leading causes of global NCD mortality according to WHO estimates [16], while mental health conditions were included due to their growing public health impact and their relevance to the consortium's focus [2, 9].

We developed search strings and detailed strategies in consultation with a medical librarian to ensure both accuracy and comprehensiveness (Supplementary Material 1). All identified documents were uploaded into Rayyan software[17] for screening.

Eligibility criteria

We considered documents eligible if they described prevention approaches in which interventions were explicitly stratified according to one or more risk factors, including genetic risk, selected modifiable risk factors (tobacco use, physical inactivity, unhealthy diet, and alcohol consumption), and a corresponding preventive intervention targeted to the risk-stratified group. Genetic risk stratification was defined as prevention approaches based on germline gene variants associated with an increased likelihood of developing NCDs. This included both single high-penetrance pathogenic variants and the combined

effect of multiple low-penetrance variants across different loci, as well as pharmacogenomic (PGx) applications in which germline variants inform preventive strategies, drug response, treatment adjustments, or anticipated clinical outcomes. In this context, genetic variants refer to germline alterations that increase disease susceptibility without necessarily being directly causative (e.g., *BRCA1*, *BRCA2*, *PALB2*), whereas hereditary genetic diseases refer to conditions caused by pathogenic variants that directly result in a defined genetic syndrome, such as homozygous familial hypercholesterolemia (HoFH).

We included all age groups. When we identified multiple documents from the same organization, we retained only the most recent version, provided it met the inclusion criteria.

Exclusion criteria comprised documents not focused on prevention, stratification based only on age and/or sex, those addressing only pharmacotherapy interventions without genetic risk stratification, documents specifically related to COVID-19 or pregnancy, and those documents recommending preventive interventions exclusively on the basis of family membership, without any genetic testing or individual-level risk assessment. Documents focusing on somatic rather than germline gene variants or lacking clear information on the target population or interventions, were also excluded.

Data extraction

From each eligible document, we extracted publication characteristics (including title, doi, year of publication, issuing organization, country of development) and detailed information on the prevention approach, including the target NCD, level of prevention, target population, risk factors used for stratification (germline genetic variants, tobacco use, physical inactivity, unhealthy diet, alcohol consumption and composite risk scores derived from multiple risk factors), stratification details, specific preventive interventions and reported levels of evidence (LoE).

The LoE supporting each approach was categorized into tiers and is summarized in Supplementary Table 1. Given that the included documents adopted different grading systems for rating the evidence underlying their recommendations, we aligned the shared descriptive characteristics across these systems (e.g., highest levels typically based on multiple RCTs) and developed a standardized classification to ensure comparability [18–20].

When a single document included multiple prevention approaches (e.g., different risk factors, genetic variants, or preventive interventions), each approach was extracted and analyzed separately. Identical approaches were counted only once to avoid duplication, and when the same approach was reported across multiple guidelines (e.g., NCCN and ESMO), the most recent reported LoE was retained.

Data synthesis

We presented the results both narratively and quantitatively. We organized extracted prevention approaches by stratification type (genetic vs modifiable risk factors), disease area, and level of prevention, and reported descriptive statistics as absolute and percentage frequencies. We classified all approaches according to the type of preventive intervention recommended following risk stratification. We defined intervention categories *a posteriori*, including counseling and lifestyle advice, intensified surveillance, pharmacotherapy interventions, preventive therapy, preventive surgery, and relapse-prevention strategies. We applied this classification across all risk factors and disease areas. We compared approaches based on genetic with those based on modifiable risk factors using the chi-square test or Fisher's exact test, as appropriate. We conducted all analyses using Microsoft Excel and R software.

Results

The literature search yielded 114 eligible documents (Supplementary Table 2) from multiple database searches and supplementary Google queries (Fig. 1). Among these, 75 were guidelines, followed by 31 recommendations and 8 consensus statements. The distribution of documents by year was as follows: 2019 (11.4%), 2020 (17.5%), 2021 (10.5%), 2022 (15.2%), 2023 (13.2%), 2024 (19.3%), and 2025 (12.3%). A level of evidence was reported in 69 documents (60.5%) (Table 1).

Table 1
Characteristics of the documents included.

n		All documents
		114
Year of publication, n (%)	2019	13 (11.4)
	2020	20 (17.5)
	2021	12 (10.5)
	2022	18 (15.8)
	2023	15 (13.2)
	2024	22 (19.3)
	2025	14 (12.3)
Document type, n (%)	Consensus statement	8 (7.0)
	Guideline	75 (65.8)
	Recommendation	31 (27.2)
Country of origin, n (%)	Multinational	34 (29.8)
	Only European	14 (12.3)
	North America	30 (26.3)
	USA	26 (22.8)
	Canada	4 (3.5)
	European countries	25 (21.4)
	UK	6 (5.3)
	Spain	4 (3.5)
	France, Germany	3 (2.6)
	Netherlands, Poland	2 (1.8)
	Italy, Greece, Slovakia, Switzerland, Turkey	1 (0.9)
	Asian countries	16 (13.7)
	South Korea	4 (3.5)
	China	3 (2.6)

LoE: Level of Evidence

n		All documents
		114
	India, Japan	2 (1.8)
	UAE, Hong Kong, Philippines, Saudi Arabia, Taiwan	1 (0.9)
	South America countries	8 (6.8)
	Brazil	7 (6.1)
	Argentina	1 (0.9)
	African Countries	1 (0.9)
	Egypt	1 (0.9)
Stratification focus	**Genetic factors**	74 (64.9)
	Modifiable factors	37 (32.5)
	Both genetic and modifiable factors	3 (2.6)
With LoE/ consensus, n (%)		69 (60.5)
LoE: Level of Evidence		

Geographically, 34 documents originated from multinational consortia (including 14 European), followed by 30 contributions from North America (26 from the USA), 25 from European countries, and 16 from Asian countries (Table 1).

Among the 114 included documents, 74 (64.9%) focused exclusively on genetic factors, 37 (32.5%) on modifiable risk factors, and 3 (2.6%) addressed both. A total of 661 prevention approaches were identified, of which 632 were unique. 51.9% of these focused on secondary prevention. All identified approaches are reported in Supplementary Material 2.

Overall, most approaches relied on genetic stratification (n = 541), whereas only 91 were based on modifiable risk factors (Table 2).

Table 2
Characteristics of the preventive approaches extracted from the documents.

N	Total approaches	Modifiable factors	Genetic factors	p
	632	**91**	**541**	
Prevention level, n (%)				< 0.001
Primary prevention	139 (22.0)	48 (52.7)	91 (16.8)	
Secondary prevention	328 (51.9)	23 (25.3)	305 (56.4)	
Tertiary prevention	165 (26.1)	20 (22.0)	145 (26.8)	
NCD, n (%)				< 0.001
Cancer	441 (69.8)	16 (17.6)	425 (78.6)	
Cardiovascular disease	162 (25.6)	70 (76.9)	92 (17.0)	
Chronic respiratory disease	4 (0.6)	2 (2.2)	2 (0.4)	
Diabetes	2 (0.3)	2 (2.2)	0 (0.0)	
Mental health	23 (3.6)	1 (1.1)	22 (4.1)	
Modifiable risk factor, n (%)				
Alcohol	-	6 (6.6)	-	
Physical activity	-	2 (2.2)	-	
Tobacco use	-	83 (91.2)	-	
With LoE/ consensus, n (%)	409 (64.7)	70 (76.9)	339 (62.7)	0.008
Tier LoE	389 (95.1)	70 (100.0)	319 (94.1)	0.051
1	94 (24.2)	19 (27.1)	75 (23.5)	
2	166 (42.7)	21 (30.0)	145 (45.5)	
3	129 (33.2)	30 (42.9)	99 (31.0)	
Consensus*	20 (4.9)	0 (0.0)	20 (5.9)	
100%	1 (5.0)	-	1 (5.0)	

LoE: Level of Evidence;

**For consensus statements, consensus was classified as either complete (100% agreement among panel members) or partial (< 100% agreement).*

N	Total approaches	Modifiable factors	Genetic factors	p
	632	**91**	**541**	
< 100%	19 (95.0)	-	18 (95.0)	
LoE: Level of Evidence;				
**For consensus statements, consensus was classified as either complete (100% agreement among panel members) or partial (< 100% agreement).*				

Among the modifiable risk factors, 83 (91.2%) focus on tobacco use, 6 (6.6%) on alcohol consumption, and 2 (2.2%) on physical activity. Most approaches targeted cancer (69.8%), followed by CVDs (25.6%), and LoE or consensus was reported in 64.7% of cases, with a similar distribution across LoE tiers (Tier 1: 24.2%, Tier 2: 42.7%, Tier 3: 33.2%).

Comparison of preventive approaches based on genetic versus modifiable risk stratification revealed marked differences in both the level of prevention addressed and the diseases targeted. Approaches addressing modifiable risk factors showed a greater emphasis on primary prevention compared with those based on genetic factors (52.7% vs. 16.8%). In contrast, genetically stratified approaches were more frequently associated with secondary (56.4% vs. 25.3%) and tertiary prevention (26.8% vs. 22.0%). Regarding disease focus, cardiovascular disease was more frequently addressed in approaches based on modifiable risk factors (76.9% vs. 17.0%), whereas cancer predominated among genetic approaches (78.6% vs. 17.6%). Reporting of LoE or consensus was slightly more frequent in approaches based on modifiable risk factors than in genetic approaches (76.9% vs. 62.7%), although the distribution across evidence tiers was similar, particularly for Tier 1 evidence (27.1% vs. 23.5%) (Table 2). Table 3 reports preventive interventions associated with genetic and modifiable risk stratification criteria. Intensified surveillance was the most frequently reported intervention across all risk factors. Genetic stratification was more often associated with preventive surgery, pharmacotherapy interventions, and structured surveillance, whereas stratification based on modifiable risk factors primarily involved counseling, lifestyle interventions, smoking cessation, and surveillance.

Table 3
Risk stratification criteria and corresponding preventive interventions across all extracted prevention approaches

Risk factor	Stratification	Intervention	Reference
Smoking	Smokers	Counseling and healthy lifestyle	[21], [40], [41]
		Intensified surveillance	[42], [29], [43], [44], [45], [46]
		Cessation	[36], [31], [43], [47], [25], [48]
	Pack-years	Intensified surveillance	[49], [21], [50], [51], [33], [52], [23], [53], [54]
	Score°	Counseling and healthy lifestyle	[55], [56], [57], [58], [59], [60]
		Preventive therapy	[27], [57], [59], [19]
		Assessment	[61], [62]
		Intensified surveillance	[63], [64]
		Pharmacotherapy	[59]
Alcohol	Drinkers	Counseling and healthy lifestyle	[65]
		Intensified surveillance	[66]
		Reduction	[47]
	Drink/day	Counseling and healthy lifestyle	[67]
		Elimination	[31], [68]
Physical activity	Physical inactivity	Intensified surveillance	[69]
		Physical activity increase for relapse prevention	[35]
Genetic factors	Germline variants	Counseling and healthy lifestyle	[70], [69], [43], [71], [72], [73], [74]

Colors indicate levels of prevention: light yellow = primary, yellow = secondary, dark yellow = tertiary prevention.

*° Score: defined according to the risk assessment schemes described in the source documents. Each score represents a well-established risk prediction model in which the corresponding risk factor (e.g. smoking) is explicitly integrated into the calculation as per the document analyzed (e.g. SCORE2, 10-year ASCVD risk, POL score). * Pharmacotherapy may include genetically informed interventions, including PGx-guided approaches*

Risk factor	Stratification	Intervention	Reference
		Preventive surgery	[75], [72], [76], [20], [77], [78], [79], [22], [80], [81], [82], [83], [84], [28], [73], [85], [86]
		Preventive therapy	[87], [22], [70]
		Intensified surveillance	[77], [88], [78], [89], [70], [81], [90], [82], [91], [84], [92], [93], [94], [72], [95], [96], [86], [76], [97], [20], [98], [99], [89], [100], [101], [22], [69], [102], [103]
		Intensified surveillance for relapse prevention	[104], [105], [106]
		Pharmacotherapy*	[107], [108], [109], [32], [110], [37], [111], [112], [73], [113], [114], [115], [116]
		Physical activity increase for relapse prevention	[79]
		Surgery	[77], [80], [83], [114], [97]
	Hereditary genetic disease	Counseling and healthy lifestyle	[117], [74], [40], [118], [20], [119], [120]
		Preventive surgery	[24], [121], [83], [122], [98], [84], [120]
		Preventive therapy	[111], [123], [124], [119], [125], [126]
		Intensified surveillance	[88], [89], [30], [24], [127], [128], [34], [120], [129], [130], [95], [20], [24], [124], [75], [122], [98], [99], [121], [131], [132], [133], [111], [134], [135]
		Pharmacotherapy*	[120], [124]
	Score°	Preventive surgery	[81], [28]
		Intensified surveillance	[101], [81], [93]
		Pharmacotherapy*	[26]

Colors indicate levels of prevention: light yellow = primary, yellow = secondary, dark yellow = tertiary prevention.

*° Score: defined according to the risk assessment schemes described in the source documents. Each score represents a well-established risk prediction model in which the corresponding risk factor (e.g. smoking) is explicitly integrated into the calculation as per the document analyzed (e.g. SCORE2, 10-year ASCVD risk, POL score). * Pharmacotherapy may include genetically informed interventions, including PGx-guided approaches*

Cancer

Four hundred and forty-one risk stratification approaches for cancer prevention were identified, of which 425 (96.4%) were based on genetic factors and 16 (3.6%) targeted modifiable risk factors, primarily tobacco use (n = 13), alcohol consumption (n = 2), and physical activity (n = 1). When classified by level of prevention, no differences were observed between approaches based on fixed genetic versus modifiable risk factors (p = 0.900) (Supplementary Table 3).

Within primary prevention, 72 approaches were recorded, the vast majority of which (70) were genetic-based, while only 2 focused on modifiable risk factors. Modifiable risk stratification includes advising individuals who smoke to quit and offering evidence-based smoking-cessation counseling and pharmacotherapy [21]. In contrast, genetic-based primary prevention includes recommendations such as risk-reducing salpingo-oophorectomy (RRSO) strongly advised for women carrying pathogenic *BRCA* variants [22].

Secondary prevention included 310 approaches, predominantly genetic (298), with 12 addressing modifiable factors. Examples of modifiable risk–based approaches include recommending annual low-dose computed tomography (LDCT) screening for current or former heavy smokers (defined as at least one pack of cigarettes daily for 30 years or two packs for 15 years) [23]. Genetic stratification examples include colonoscopy surveillance every two to three years for carriers of pathogenic *MLH1*, *MSH2*, and *MSH6* variants, in patients with Lynch Syndrome [24].

Tertiary prevention included 59 approaches, the vast majority of which were based on genetic stratification (n = 57), of which 8 involved PGx, with only two approaches addressing modifiable risk factors. Modifiable risk–based approaches in tertiary prevention mainly consisted of comprehensive smoking cessation programs for patients with cancer who smoke [25]. In contrast, genetic-based approaches included PGx such as *DPYD* genotyping prior to the initiation of chemotherapy, followed by dose adjustment in carriers of specific risk variants [26].

Among all approaches, 265 (58.6%) reported LoE. There was no significant difference between genetic and modifiable risk factors in this regard (59.2% vs. 45.5%). The distribution across evidence tiers did not differ substantially between the two categories, with 21.2% classified as Tier 1, 46.1% as Tier 2, and 32.7% as Tier 3. Consensus percentages were reported only for genetic-based approaches, one of which indicated 100% agreement, while 19 genetic-based approaches reported agreement between 75% and 100%.

Cardiovascular Diseases

One hundred and sixty-two CVDs prevention approaches were identified, of which 92 (56.8%) were based on genetic factors and 70 (43.2%) targeted modifiable risk factors, primarily tobacco use (n = 66) and, to a lesser extent, alcohol consumption (n = 4). When classified by level of prevention, the distribution of approaches differed significantly between genetic- and modifiable risk–based strategies (p < 0.001) (Supplementary Table 4).

Regarding primary prevention, 66 approaches (40.7%) were documented, the majority of which (46) focused on modifiable risk factors, whereas 20 were genetically based of which 17 clinically actionable (e.g., familial hypercholesterolemia and pathogenic variants in genes associated with inherited cardiovascular diseases such as *LMNA* and *TMEM43*), while 3 non-actionable (e.g., the *ALDH2*∗2 dysfunctional allele). Modifiable risk–based stratification for primary prevention includes recommendations targeting adults at increased cardiovascular risk; for example, smoking cessation is recommended for all individuals who smoke to reduce the risk of atherosclerotic cardiovascular disease (ASCVD) [27]. In contrast, genetic-based primary prevention includes recommendations such as implantable cardioverter-defibrillator (ICD) implantation in dilated cardiomyopathy/ hypokinetic non-dilated cardiomyopathy (DCM/HNDCM) patients with a pathogenic mutation in LMNA gene, if the estimated 5-year risk of life-threatening ventricular arrhythmia (VA) is ≥ 10% and in the presence of non-sustained ventricular tachycardia (NSVT) or left ventricular ejection fraction (LVEF), 50% or atrioventricular conduction delay [28].

In secondary prevention, 15 approaches (9.3%) were identified, predominantly modifiable factors (9), with six addressing genetics. Modifiable risk examples in this category involve one-time screening for abdominal aortic aneurysm (AAA) with ultrasonography in men aged 65 to 75 years who have ever smoked [29]. Genetic stratification examples include computed tomography coronary angiography (CTCA) every 5 years, or more frequently if clinically indicated, in HoFH individuals [30].

Tertiary prevention accounted for half of all approaches (n = 81; 50.0%), of which 66 were genetically based, including 64 PGx approaches, and 15 considered modifiable risk factors. Examples of modifiable factor approaches in tertiary prevention include multibehavioral, evidence-based interventions for smoking cessation, including avoidance of secondhand smoke in populations with ischemic stroke [31]. On the genetic side, interventions included tailored statin dosing according to *SLCO1B1*, *ABCG2*, and *CYP2C9* mutations [32].

Across all approaches, a higher frequency of LoE reporting was observed for modifiable risk factors compared to genetic factors (85.7% vs. 65.2%). Analysis by evidence tier revealed difference across tiers between genetic and modifiable risk factors (p = 0.002) that Tier 1 included 35 approaches (29.2%; 33.3% genetic, 25.0% modifiable), Tier 2 included 45 approaches (37.5%; 48.3% genetic, 26.7% modifiable), and Tier 3 included 40 approaches (33.3%; 17.5% genetic, 48.3% modifiable).

Chronic respiratory diseases

Four CRDs prevention approaches were identified, of which 2 (50.0%) were based on genetic factors and 2 (50.0%) targeted modifiable risk factors on tobacco use. These approaches were implemented in the context of primary and secondary prevention (Supplementary Table 5).

In secondary prevention examples of modifiable risk–based approaches include recommending spirometry testing for symptomatic individuals with risk factors such as a history of smoking or

exposure to harmful airborne particles [33]. Examples of genetic stratification approaches include offering a baseline low-dose high-resolution computed tomography (HRCT) scan to patients with, or suspected of having, Birt-Hogg-Dubé (BHD) syndrome to detect pulmonary cysts [34].

Among the four approaches, only one in the genetic factors category has a LoE classified as Tier 2.

Diabetes

Two DM prevention approaches targeted modifiable risk factors. These approaches were implemented in the context of tertiary prevention (Supplementary Table 6).

An example of a modifiable risk–based approach involves diabetic patients with low levels of physical activity and cardiorespiratory fitness, who may benefit most from increasing their daily step count. It is recommended that individuals accumulate at least 5,000 steps per day [35].

Among the two approaches, only one has a LoE classified as Tier 3.

Mental health

Twenty-three mental health prevention approaches were identified in the context of schizophrenia, depression, and anxiety disorders, of which 22 (95.7%) were based on genetic factors, all involving PGx, and 1 (4.3%) targeted modifiable risk factors on tobacco use. These approaches were implemented in the context of tertiary prevention (Supplementary Table 7).

Examples of modifiable risk–based approaches include providing smoking cessation advice to all patients who are current smokers at every clinical encounter. Support should be offered through follow-up appointments and behavioural counselling, alongside pharmacological options such as nicotine replacement therapy, varenicline, or bupropion [36]. Examples of genetic stratification approaches include dosing recommendations for serotonin reuptake inhibitor (SRI) antidepressants in patients carrying genetic variants affecting CYP2C19 and/or CYP2B6 drug metabolism, which influence serum concentrations and treatment response [37].

Across all approaches, a LoE was reported in 22 cases (95.7%), all of which were based on genetic approaches. Among these, 7 (31.8%) were classified as Tier 1, 7 (31.8%) as Tier 2, and 8 (36.4%) as Tier 3.

Discussion

This scoping review provides a comprehensive overview of how current global guidelines, recommendations, and consensus statements address risk stratification in the context of prevention for major non-communicable diseases. By mapping prevention approaches according to the risk factors considered for stratification, fixed genetic and modifiable, this review characterizes how risk stratification is conceptualized within existing guidance across disease areas and levels of prevention.

Overall, prevention approaches identified in this review were predominantly based on genetic factors, particularly in the context of cancer prevention or pharmacotherapy, where genetic stratification accounted for the vast majority of approaches. This predominance is consistent with the historical development of personalized prevention within oncological settings, where hereditary syndromes, high-penetrance pathogenic gene variants, and PGx susceptibility variants have well-defined clinical implications and are supported by relatively robust evidence [38].

Genetic stratification was represented across prevention levels, including primary prevention in high-risk individuals through interventions such as risk-reducing surgery, as well as secondary and tertiary prevention, where strategies such as intensified surveillance and genotype-guided treatment modifications can be more readily integrated into established clinical pathways [39].

In contrast, prevention approaches based on modifiable risk factors were less frequently represented overall and were unevenly distributed across disease areas. Where present, they were more commonly associated with CVD prevention and with primary prevention strategies. This pattern suggests that, despite the well-established contribution of lifestyle and behavioral factors to the global burden of NCDs, modifiable risk stratification remains underutilized in guideline-based prevention. More broadly, this distribution may reflect a longstanding health system orientation that prioritizes clinical disease management, while prevention (particularly for modifiable risk factors) is often implemented through universal public health strategies rather than individualized risk-stratified approaches.

Our results further indicate that modifiable risk stratification is largely driven by tobacco use, with minimal incorporation of other behavioral determinants such as physical activity or alcohol consumption. This imbalance mirrors the availability of long-standing evidence and standardized assessment tools for smoking-related risk, while underscoring the persistent challenges in operationalizing more complex or multidimensional modifiable risk factors within guideline-based prevention. Consequently, current stratification frameworks may capture only a limited portion of preventable risk, particularly in primary prevention settings.

Overall, current prevention guidance appears to translate genetic risk stratification into more intensive and clinically actionable preventive strategies, while stratification based on modifiable risk factors is more often linked to lower-intensity, behavior- or surveillance-focused interventions.

Marked heterogeneity was observed across disease areas. For CRDs, DM, and mental health conditions, only a limited number of risk-based prevention approaches were identified. Although this finding should be interpreted cautiously given the small number of available documents, it highlights persistent gaps in guideline-based prevention for these conditions and suggests that formal risk stratification has yet to be systematically incorporated into preventive guidance.

A major challenge emerging from this review concerns the identification and measurement of non-genetic risk factors required for effective stratification, particularly in primary prevention. Unlike genetic factors, which are stable over time and measurable once, modifiable risk factors are, by definition,

dynamic, context-dependent, and often inconsistently captured in routine care. Their assessment frequently requires longitudinal data, standardized behavioral instruments, or access to community-based prevention services, which are not uniformly available across health systems, although these factors are routinely addressed at the population level. As a result, modifiable risk stratification appears more readily applicable in secondary and tertiary prevention settings, where individuals are already engaged in structured healthcare pathways.

Innovative approaches, including population-based cohorts, integrated data systems, primary prevention centres, and community-based “living laboratories”, may facilitate the implementation of modifiable risk stratification in real-world practice. Approaches that integrate prevention into everyday settings may help translate traditional risk factors, as well as socio-economic, psychosocial and contextual determinants, into tailored preventive interventions, while complementing existing population-level strategies. Although socio-economic stratification was not explicitly addressed in the present review, our findings point to its potential relevance as an additional layer of risk stratification in future prevention frameworks.

These challenges in defining, measuring, and applying risk categories also have important implications for the translation of risk-stratified prevention into practice. Even when stratified recommendations are available, their appropriate implementation depends on the ability of healthcare professionals to identify relevant risk factors, accurately interpret risk information and effectively communicate preventive strategies at the individual level, as well as on individuals’ capacity to understand and act upon such information.

Education and training of healthcare professionals, together with citizen engagement and health literacy, therefore represent essential enabling conditions for the successful implementation of risk-based prevention at the population level. Without these elements, stratification frameworks risk remaining largely theoretical, regardless of their methodological sophistication.

This study benefits from a comprehensive search strategy and a focus on consolidated expert guidance rather than individual primary studies. Nevertheless, several limitations should be considered. First, the review was restricted to English-language documents published from 2019 onward, which may have led to the exclusion of relevant guidance developed earlier or published in other languages. Second, we did not evaluate the effectiveness, feasibility, implementation, or real-world impact of the identified prevention approaches. Instead, the review focused on how risk stratification is conceptualized and operationalized within existing guidance documents. Finally, while guidelines, recommendations, and consensus statements provide an important benchmark for current knowledge, their publication does not guarantee real-world implementation, which was beyond the scope of this review.

Abbreviations

AAA: Abdominal Aortic Aneurysm

ASCVD: Atherosclerotic Cardiovascular Disease

BHD: Birt-Hogg-Dubé (syndrome)

BRCA: Breast Cancer gene (BRCA1/BRCA2)

CVD: Cardiovascular Diseases

CRD: Chronic Respiratory Diseases

CTCA: Computed Tomography Coronary Angiography

DM: Diabetes Mellitus

DCM: Dilated Cardiomyopathy

HRCT: High-Resolution Computed Tomography

HoFH: Homozygous Familial Hypercholesterolemia

ICD: Implantable Cardioverter-Defibrillator

LDCT: Low-Dose Computed Tomography

LVEF: Left Ventricular Ejection Fraction

LMICs: Low- and Middle-Income Countries

NCDs: Non-Communicable Diseases

NSVT: Non-Sustained Ventricular Tachycardia

OSF: Open Science Framework

PGx: Pharmacogenomics / Pharmacogenomic

RCTs: Randomized Controlled Trials

RRSO: Risk-Reducing Salpingo-Oophorectomy

SRI: Serotonin Reuptake Inhibitor

VA: Ventricular Arrhythmia

Declarations

Authors' contributions: EG, NL: Conceptualization, Methodology, Validation, Formal analysis, Investigation, Data Curation, Writing - Original Draft, Writing - Review & Editing, Visualization. AV: Conceptualization, Methodology, Investigation, Writing - Original Draft, Writing - Review & Editing,

Visualization. FC, FAC, MrDP, MtDP, MP, DT: Conceptualization, Methodology, Validation, Investigation, Writing - Review & Editing. MR, HAP, FR, MVDB, SB: Conceptualization, Methodology, Validation, Writing - Review & Editing. RP: Conceptualization, Methodology, Validation, Resources, Writing - Original Draft, Writing - Review & Editing, Supervision, funding acquisition, guarantor.

Competing interests

Nothing to declare.

Funding

This project (GA 101128023) is co-funded by the European Union. Views and opinions

expressed are however those of the author(s) only and do not necessarily reflect those of the

European Union or the European Health and Digital Executive Agency (HaDEA). Neither the

European Union nor HaDEA can be held responsible for them.

Acknowledgements

The authors would like to thank Christa Niehot for her valuable support in literature searches and Sophie Denoël for her contribution to the interpretation and discussion of the findings.

Code availability

No code was used or generated in this study.

Data availability

Part of the data are provided in Supplementary Material 2. Other data are available from the authors upon reasonable request.

Ethics approval and consent to participate

This study is a scoping review based on publicly available literature and did not involve human participants, human data, or individual patient-level information. Therefore, ethics approval was not required. Consent to participate was not applicable.

Consent for publication

Not applicable.

Clinical trial number:

Not applicable.

References

1. Freihat O, Sipos D, Aamir M, Kovacs A. Global burden and future projections of non-communicable diseases (2000–2050): Progress toward SDG 3.4 and disparities across regions and risk factors. PLoS One 2025;20:e0336036. https://doi.org/10.1371/journal.pone.0336036.
2. Li J, Pandian V, Davidson PM, Song Y, Chen N, Fong DYT. Burden and attributable risk factors of non-communicable diseases and subtypes in 204 countries and territories, 1990-2021: a systematic analysis for the global burden of disease study 2021. Int J Surg 2025;111:2385–97. https://doi.org/10.1097/JS9.0000000000002260.
3. Naghavi M, Kyu HH, Bhoomadevi A, Aalipour MA, Aalruz H, Ababneh HS, et al. Global burden of 292 causes of death in 204 countries and territories and 660 subnational locations, 1990–2023: a systematic analysis for the Global Burden of Disease Study 2023. The Lancet 2025;406:1811–72. https://doi.org/10.1016/S0140-6736(25)01917-8.
4. Bennett JE, O'Driscoll ON, Stevens GA, Aldea-Ramos N, Bray F, Farzadfar F, et al. Benchmarking progress in non-communicable diseases: a global analysis of cause-specific mortality from 2001 to 2019. The Lancet 2025;406:1255–82. https://doi.org/10.1016/S0140-6736(25)01388-1.
5. Fink H, Langselius O, Vignat J, Rumgay H, Rehm J, Martinez RX, et al. Global and regional cancer burden attributable to modifiable risk factors to inform prevention. Nature Medicine 2026 2026:1–10. https://doi.org/10.1038/s41591-026-04219-7.
6. Leppo K, Ollila E, Pena S, Wismar M, Cook S. Health in All Policies: Seizing opportunities, implementing policies. Health in All Policies 2013:1–357.
7. Allen LN, Wigley S, Holmer H, Barlow P. Non-communicable disease policy implementation from 2014 to 2021: a repeated cross-sectional analysis of global policy data for 194 countries. Lancet Glob Health 2023;11:e525–33. https://doi.org/10.1016/S2214-109X(23)00042-6.
8. Allen LN, Nicholson BD, Yeung BYT, Goiana-da-Silva F. Implementation of non-communicable disease policies: a geopolitical analysis of 151 countries. Lancet Glob Health 2020;8:e50–8. https://doi.org/10.1016/S2214-109X(19)30446-2.
9. Van Den Bulcke M, Rossetti F, A. Poirel H, Denoël S, Hansen TF, Sandgren BL, et al. Personalised risk-stratified approaches in prevention of cancer and other NCDs: The Joint Action PreventNCD project. Scand J Public Health 2025;53:54–60. https://doi.org/10.1177/14034948251384404.
10. Zettler I, Strandsbjerg CF. Personalized interventions. Curr Opin Psychol 2025;66. https://doi.org/10.1016/j.copsyc.2025.102147.
11. Farina S, Osti T, Russo L, Maio A, Scarsi N, Savoia C, et al. The current landscape of personalised preventive approaches for non-communicable diseases: A scoping review. PLoS One 2025;20:e0317379. https://doi.org/10.1371/journal.pone.0317379.
12. Brignardello-Petersen R, Carrasco-Labra A, Guyatt GH. How to Interpret and Use a Clinical Practice Guideline or Recommendation: Users' Guides to the Medical Literature. JAMA 2021;326:1516–23. https://doi.org/10.1001/jama.2021.15319.

13. JA PreventNCD - Reducing Europe's cancer and NCD burden through coordinated strategies on health determinants. Focusing on effective policies, societal and personal risk factors. n.d. https://www.preventncd.eu/ (accessed February 4, 2026).
14. Arksey H, O'Malley L. Scoping studies: Towards a methodological framework. International Journal of Social Research Methodology: Theory and Practice 2005;8:19–32. https://doi.org/10.1080/1364557032000119616.
15. Tricco AC, Lillie E, Zarin W, O'Brien KK, Colquhoun H, Levac D, et al. PRISMA Extension for Scoping Reviews (PRISMA-ScR): Checklist and Explanation. Ann Intern Med 2018;169:467–73. https://doi.org/10.7326/M18-0850.
16. Noncommunicable diseases n.d. https://www.who.int/news-room/fact-sheets/detail/noncommunicable-diseases (accessed February 4, 2026).
17. Rayyan: AI-Powered Systematic Review Management Platform n.d. https://www.rayyan.ai/ (accessed February 4, 2026).
18. ESC Declaration of Interest Report 2021 ESC Guidelines for the diagnosis and treatment of acute and chronic heart failure n.d.
19. Katsiki N, Filippatos T, Vlachopoulos C, Panagiotakos D, Milionis H, Tselepis A, et al. Executive summary of the Hellenic Atherosclerosis Society guidelines for the diagnosis and treatment of dyslipidemias - 2023. Atherosclerosis Plus 2024;55:74. https://doi.org/10.1016/j.athplu.2024.01.004.
20. Daly MB, Pal T, Maxwell KN, Churpek J, Kohlmann W, AlHilli Z, et al. NCCN Guidelines® Insights: Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic, Version 2.2024. J Natl Compr Canc Netw 2023;21:1001–10. https://doi.org/10.6004/jnccn.2023.0051.
21. Wolf AMD, Oeffinger KC, Shih TY, Walter LC, Church TR, Fontham ETH, et al. Screening for lung cancer: 2023 guideline update from the American Cancer Society. CA Cancer J Clin 2024;74:50–81. https://doi.org/10.3322/caac.21811.
22. González-Santiago S, Ramón Y Cajal · T, Aguirre · E, Alés-Martínez · J E, Andrés · R, Balmaña · J, et al. SEOM clinical guidelines in hereditary breast and ovarian cancer (2019). Clinical and Translational Oncology 2019 22:2 2019;22:193–200. https://doi.org/10.1007/s12094-019-02262-0.
23. NCCN. Lung Cancer Screening n.d. https://www.nccn.org/search-result?indexCatalogue=nccn-search-index&searchQuery=Lung%20Cancer%20Screening (accessed February 5, 2026).
24. Hodan R, Gupta S, Weiss JM, Axell L, Burke CA, Chen LM, et al. Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, and Gastric, Version 3.2024, NCCN Clinical Practice Guidelines In Oncology. J Natl Compr Canc Netw 2024;22:695–711. https://doi.org/10.6004/jnccn.2024.0061.
25. Shields PG, Bierut L, Arenberg D, Balis D, Cinciripini PM, Davis J, et al. Smoking Cessation, Version 3.2022, NCCN Clinical Practice Guidelines in Oncology. Journal of the National Comprehensive Cancer Network 2023;21:297–322. https://doi.org/10.6004/jnccn.2023.0013.

26. Lunenburg CATC, van der Wouden CH, Nijenhuis M, Crommentuijn-van Rhenen MH, de Boer-Veger NJ, Buunk AM, et al. Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene–drug interaction of DPYD and fluoropyrimidines. European Journal of Human Genetics 2019 28:4 2019;28:508–17. https://doi.org/10.1038/s41431-019-0540-0.
27. Grundy SM, Stone NJ, Bailey AL, Beam C, Birtcher KK, Blumenthal RS, et al. 2018 AHA/ACC/AACVPR/AAPA/ABC/ACPM/ADA/AGS/APhA/ASPC/NLA/PCNA Guideline on the Management of Blood Cholesterol: A Report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. Circulation 2019;139:E1082–143. https://doi.org/10.1161/CIR.0000000000000625.
28. Zeppenfeld K, Tfelt-Hansen J, De Riva M, Winkel BG, Behr ER, Blom NA, et al. 2022 ESC Guidelines for the management of patients with ventricular arrhythmias and the prevention of sudden cardiac death: Developed by the task force for the management of patients with ventricular arrhythmias and the prevention of sudden cardiac death…. Eur Heart J 2022;43:3997–4126. https://doi.org/10.1093/eurheartj/ehac262.
29. Owens DK, Davidson KW, Krist AH, Barry MJ, Cabana M, Caughey AB, et al. Screening for Abdominal Aortic Aneurysm: US Preventive Services Task Force Recommendation Statement. JAMA 2019;322:2211–8. https://doi.org/10.1001/jama.2019.18928.
30. Tarugi P, Bertolini S, Calandra S, Arca M, Angelico F, Casula M, et al. Consensus document on diagnosis and management of familial hypercholesterolemia from the Italian Society for the Study of Atherosclerosis (SISA). Nutrition, Metabolism and Cardiovascular Diseases 2024;34:1819–36. https://doi.org/10.1016/j.numecd.2024.05.002.
31. Ford B, Peela S, Roberts C. Secondary Prevention of Ischemic Stroke: Updated Guidelines From AHA/ASA. Am Fam Physician 2022;105:99–102.
32. Cooper-DeHoff RM, Niemi M, Ramsey LB, Luzum JA, Tarkiainen EK, Straka RJ, et al. The Clinical Pharmacogenetics Implementation Consortium Guideline for SLCO1B1, ABCG2, and CYP2C9 genotypes and Statin-Associated Musculoskeletal Symptoms. Clin Pharmacol Ther 2022;111:1007–21. https://doi.org/10.1002/cpt.2557.
33. GOLD Report - Global Initiative for Chronic Obstructive Lung Disease - GOLD n.d. https://goldcopd.org/2024-gold-report/ (accessed February 5, 2026).
34. Geilswijk M, Genuardi M, Woodward ER, Nightingale K, Huber J, Madsen MG, et al. ERN GENTURIS clinical practice guidelines for the diagnosis, surveillance and management of people with Birt-Hogg-Dubé syndrome. European Journal of Human Genetics 2024 32:12 2024;32:1542–50. https://doi.org/10.1038/s41431-024-01671-2.
35. Knaga J, Buresh R. Personalizing activity recommendations for individuals with type 2 diabetes. ACSMs Health Fit J 2022;26:18–24. https://doi.org/10.1249/FIT.0000000000000817.
36. Wichniak A, Dudek D, Heitzman J, Kapłon-Cieślicka A, Mamcarz A, Samochowiec J, et al. Metabolic risk reduction in patients with schizophrenia treated with antipsychotics: recommendations of the

Polish Psychiatric Association. Psychiatr Pol 2019;53:1191–218. https://doi.org/10.12740/PP/113222.

37. Bousman CA, Stevenson JM, Ramsey LB, Sangkuhl K, Hicks JK, Strawn JR, et al. Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. Clin Pharmacol Ther 2023;114:51–68. https://doi.org/10.1002/cpt.2903.
38. Zeng C, Bastarache LA, Tao R, Venner E, Hebbring S, Andujar JD, et al. Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases. JAMA Oncol 2022;8:835–44. https://doi.org/10.1001/jamaoncol.2022.0373.
39. Ginsburg O, Ashton-Prolla P, Cantor A, Mariosa D, Brennan P. The role of genomics in global cancer prevention. Nature Reviews Clinical Oncology 2020 18:2 2020;18:116–28. https://doi.org/10.1038/s41571-020-0428-5.
40. Chao TH, Lin TH, Cheng CI, Wu YW, Ueng KC, Wu YJ, et al. 2024 Guidelines of the Taiwan Society of Cardiology on the Primary Prevention of Atherosclerotic Cardiovascular Disease --- Part II. Acta Cardiol Sin 2024;40:669–715. https://doi.org/10.6515/ACS.202411_40(6).20240724B.
41. Feitosa-Filho GS, Peixoto JM, Pinheiro JES, Neto AA, de Albuquerque ALT, Cattani ÁC, et al. Updated Geriatric Cardiology Guidelines of the Brazilian Society of Cardiology - 2019. Arq Bras Cardiol 2019;112:649–705. https://doi.org/10.5935/abc.20190086.
42. Allehebi A, Al-Omair A, Mahboub B, Koegelenberg CF, Mokhtar M, Madkour AM, et al. Recommended approaches for screening and early detection of lung cancer in the Middle East and Africa (MEA) region: a consensus statement. J Thorac Dis 2024;16:2142–58. https://doi.org/10.21037/jtd-23-1568.
43. Isselbacher EM, Preventza O, Hamilton Black J, Augoustides JG, Beck AW, Bolen MA, et al. 2022 ACC/AHA guideline for the diagnosis and management of aortic disease: A report of the American Heart Association/American College of Cardiology Joint Committee on Clinical Practice Guidelines. J Thorac Cardiovasc Surg 2023;166:e182–331. https://doi.org/10.1016/j.jtcvs.2023.04.023.
44. Al-Jahdali H, Al-Lehebi R, Lababidi H, Alhejaili FF, Habis Y, Alsowayan WA, et al. The Saudi Thoracic Society Evidence-based guidelines for the diagnosis and management of chronic obstructive pulmonary disease. Ann Thorac Med 2025;20:1–35. https://doi.org/10.4103/atm.atm_155_24.
45. Abramson BL, Al-Omran M, Anand SS, Albalawi Z, Coutinho T, de Mestral C, et al. Canadian Cardiovascular Society 2022 Guidelines for Peripheral Arterial Disease. Canadian Journal of Cardiology 2022;38:560–87. https://doi.org/10.1016/j.cjca.2022.02.029.
46. Sönmez A, Özdoğan Ö, Arici M, Salman S, Derici Ü, Değer SM, et al. Consensus Statement of Endocrinology, Cardiology, and Nephrology(ENCARNE) Experts on Prevention, Diagnosis, and Management of Cardiovascular and Renal Complications of Diabetes. Turkish Journal of Endocrinology and Metabolism 2021;25:392–411. https://doi.org/10.25179/tjem.2021-85173.
47. Liu L, Li Z, Zhou H, Duan W, Huo X, Xu W, et al. Chinese Stroke Association guidelines for clinical management of ischaemic cerebrovascular diseases: executive summary and 2023 update. Stroke

Vasc Neurol 2023;8. https://doi.org/10.1136/svn-2023-002998.

48. KDIGO 2020 Clinical Practice Guideline for Diabetes Management in Chronic Kidney Disease. Kidney Int 2020;98:S1–115. https://doi.org/10.1016/j.kint.2020.06.019.
49. Juskanic D, Sandor F, Denkova L, Juhos P, Janik M, Hontzas J, et al. Lung Cancer Screening Initiative in Slovakia: Guidelines of screening implementation. Bratisl Lek Listy 2023;124:109–15. https://doi.org/10.4149/BLL_2023_016.
50. Mazzone PJ, Silvestri GA, Souter LH, Caverly TJ, Kanne JP, Katki HA, et al. Screening for Lung Cancer: CHEST Guideline and Expert Panel Report. Chest 2021;160:e427–94. https://doi.org/10.1016/j.chest.2021.06.063.
51. Recommendation: Lung Cancer: Screening | United States Preventive Services Taskforce n.d. https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/lung-cancer-screening (accessed February 5, 2026).
52. Boyeras I, Roberti J, Seijo M, Suárez V, Morero JL, Patané AK, et al. Argentine consensus recommendations for lung cancer screening programmes: a RAND/UCLA-modified Delphi study. BMJ Open 2023;13:e068271. https://doi.org/10.1136/bmjopen-2022-068271.
53. Al-Shamsi HO, Jaffar H, Mahboub B, Khan F, Albastaki U, Hammad S, et al. Early Diagnosis of Lung Cancer in the United Arab Emirates: Challenges and Strategic Recommendations. Clinics and Practice 2021, Vol 11, Pages 671-678 2021;11:671–8. https://doi.org/10.3390/clinpract11030082.
54. Ferreira Pereira LF, Dos Santos RS, Bonomi DO, Franceschini J, Santoro IL, Miotto A, et al. Lung cancer screening in Brazil: recommendations from the Brazilian Society of Thoracic Surgery, Brazilian Thoracic Association, and Brazilian College of Radiology and Diagnostic Imaging. J Bras Pneumol 2024;50. https://doi.org/10.36416/1806-3756/e20230233.
55. Visseren FLJ, Mach F, Smulders YM, Carballo D, Koskinas KC, Bäck M, et al. 2021 ESC Guidelines on cardiovascular disease prevention in clinical practice: Developed by the Task Force for cardiovascular disease prevention in clinical practice with representatives of the European Society of Cardiology and 12 medical societies With…. Eur J Prev Cardiol 2022;29:5–115. https://doi.org/10.1093/eurjpc/zwab154.
56. Krist AH, Davidson KW, Mangione CM, Barry MJ, Cabana M, Caughey AB, et al. Behavioral Counseling Interventions to Promote a Healthy Diet and Physical Activity for Cardiovascular Disease Prevention in Adults With Cardiovascular Risk Factors: US Preventive Services Task Force Recommendation Statement. JAMA 2020;324:2069–75. https://doi.org/10.1001/jama.2020.21749.
57. Sosnowska B, Stepinska J, Mitkowski P, Bielecka-Dabrowa A, Bobrowska B, Budzianowski J, et al. Recommendations of the Experts of the Polish Cardiac Society (PCS) and the Polish Lipid Association (PoLA) on the diagnosis and management of elevated lipoprotein(a) levels. Arch Med Sci 2024;20:8–27. https://doi.org/10.5114/aoms/183522.
58. Yuan Y, Gao C, Yin X, Zhang X, Ji Y, Zheng X, et al. The Guidelines for use and promotion of low sodium salt in China. J Evid Based Med 2024;17:454–67. https://doi.org/10.1111/jebm.12621.

59. Lee HY, Shin J, Kim GH, Park S, Ihm SH, Kim HC, et al. 2018 Korean Society of Hypertension Guidelines for the management of hypertension: part II-diagnosis and treatment of hypertension. Clinical Hypertension 2019 25:1 2019;25:20-. https://doi.org/10.1186/s40885-019-0124-x.
60. Newman CB, Blaha MJ, Boord JB, Cariou B, Chait A, Fein HG, et al. Lipid Management in Patients with Endocrine Disorders: An Endocrine Society Clinical Practice Guideline. J Clin Endocrinol Metab 2020;105. https://doi.org/10.1210/clinem/dgaa674.
61. Lyon AR, López-Fernánde T, Couch LS, Asteggiano R, Aznar MC, Bergler-Klei J, et al. 2022 ESC Guidelines on cardio-oncology developed in collaboration with the European Hematology Association (EHA), the European Society for Therapeutic Radiology and Oncology (ESTRO) and the International Cardio-Oncology Society (IC-OS): Developed by the …. Eur Heart J 2022;43:4229–361. https://doi.org/10.1093/eurheartj/ehac244.
62. Pearson GJ, Thanassoulis G, Anderson TJ, Barry AR, Couture P, Dayan N, et al. 2021 Canadian Cardiovascular Society Guidelines for the Management of Dyslipidemia for the Prevention of Cardiovascular Disease in Adults. Canadian Journal of Cardiology 2021;37:1129–50. https://doi.org/10.1016/j.cjca.2021.03.016.
63. Chen B, Li Q, Hao Q, Tan J, Yan L, Zhu Y, et al. Malignancy risk stratification for solitary pulmonary nodule: A clinical practice guideline. J Evid Based Med 2022;15:142–51. https://doi.org/10.1111/jebm.12476.
64. McDonagh TA, Metra M, Adamo M, Baumbach A, Böhm M, Burri H, et al. 2021 ESC Guidelines for the diagnosis and treatment of acute and chronic heart failure: Developed by the Task Force for the diagnosis and treatment of acute and chronic heart failure of the European Society of Cardiology (ESC) With the special contributi…. Eur Heart J 2021;42:3599–726. https://doi.org/10.1093/eurheartj/ehab368.
65. Alcohol and Cancer Risks A Guide for Health Professionals Alcohol and Cancer Risks: A Guide for Health Professionals Alcohol and Cancer Risks: A Guide for Health Professionals n.d.
66. Kumar A, Acharya SK, Singh SP, Arora A, Dhiman RK, Aggarwal R, et al. 2019 Update of Indian National Association for Study of the Liver Consensus on Prevention, Diagnosis, and Management of Hepatocellular Carcinoma in India: The Puri II Recommendations. J Clin Exp Hepatol 2020;10:43–80. https://doi.org/10.1016/j.jceh.2019.09.007.
67. Rabi DM, McBrien KA, Sapir-Pichhadze R, Nakhla M, Ahmed SB, Dumanski SM, et al. Hypertension Canada's 2020 Comprehensive Guidelines for the Prevention, Diagnosis, Risk Assessment, and Treatment of Hypertension in Adults and Children. Canadian Journal of Cardiology 2020;36:596–624. https://doi.org/10.1016/j.cjca.2020.02.086.
68. Chao TF, Joung B, Takahashi Y, Lim TW, Choi EK, Chan YH, et al. 2021 Focused update of the 2017 consensus guidelines of the Asia Pacific Heart Rhythm Society (APHRS) on stroke prevention in atrial fibrillation. J Arrhythm 2021;37:1389–426. https://doi.org/10.1002/joa3.12652.
69. Tsang THF, Wong KH, Allen K, Chan KKL, Chan MCM, Chao DVK, et al. Update on the Recommendations on Breast Cancer Screening by the Cancer Expert Working Group on Cancer

Prevention and Screening. Hong Kong Med J 2022;28:161–8. https://doi.org/10.12809/hkmj219622.

70. Elghazaly H, Aref AT, Anderson BO, Arun B, Yip CH, Abdelaziz H, et al. The first BGICC consensus and recommendations for breast cancer awareness, early detection and risk reduction in low- and middle-income countries and the MENA region. Int J Cancer 2021;149:505–13. https://doi.org/10.1002/ijc.33506.
71. Feitosa PHR, Castellano MVC de O, da Costa CH, Cardoso A da RO, Pereira LFF, Fernandes FLA, et al. Recommendations for the diagnosis and treatment of alpha-1 antitrypsin deficiency. Jornal Brasileiro de Pneumologia 2024;50. https://doi.org/10.36416/1806-3756/e20240235.
72. Dhawan A, Baitamouni S, Liu D, Yehia L, Anthony K, McCarther A, et al. Cancer and Overgrowth Manifestations of PTEN Hamartoma Tumor Syndrome: Management Recommendations from the International PHTS Consensus Guidelines Working Group. Clin Cancer Res 2025;31:1754–65. https://doi.org/10.1158/1078-0432.CCR-24-3819.
73. Ditsch N, Untch M, Kolberg-Liedtke C, Jackisch C, Krug D, Friedrich M, et al. AGO Recommendations for the Diagnosis and Treatment of Patients with Locally Advanced and Metastatic Breast Cancer: Update 2020. Breast Care (Basel) 2020;15:294–309. https://doi.org/10.1159/000508736.
74. Arbelo E, Protonotarios A, Gimeno JR, Arbustini E, Barriales-Villa R, Basso C, et al. 2023 ESC Guidelines for the management of cardiomyopathies: Developed by the task force on the management of cardiomyopathies of the European Society of Cardiology (ESC). Eur Heart J 2023;44:3503–626. https://doi.org/10.1093/eurheartj/ehad194.
75. Wasserman JD, Schneider KW, Achatz MI, Nakano Y, Zelley K, Gallinger B, et al. Updated Recommendations for Pediatric Surveillance in Hereditary Endocrine Neoplasia Syndromes: Multiple Endocrine Neoplasias, Hyperparathyroidism-Jaw Tumor Syndrome, and Carney Complex. Clin Cancer Res 2025;31:3628–37. https://doi.org/10.1158/1078-0432.CCR-24-3860.
76. Cardoso F, Kyriakides S, Ohno S, Penault-Llorca F, Poortmans P, Rubio IT, et al. Early breast cancer: ESMO Clinical Practice Guidelines for diagnosis, treatment and follow-up. Annals of Oncology 2019;30:1194–220. https://doi.org/10.1093/annonc/mdz173.
77. Tung NM, Boughey JC, Pierce LJ, Robson ME, Bedrosian I, Dietz JR, et al. Management of hereditary breast cancer: American society of clinical oncology, American society for radiation oncology, and society of surgical oncology guideline. Journal of Clinical Oncology 2020;38:2080–106. https://doi.org/10.1200/JCO.20.00299.
78. Dhooge M, Baert-Desurmont S, Corsini C, Caron O, Andrieu N, Berthet P, et al. National recommendations of the French Genetics and Cancer Group - Unicancer on the modalities of multi-genes panel analyses in hereditary predispositions to tumors of the digestive tract. Eur J Med Genet 2020;63. https://doi.org/10.1016/j.ejmg.2020.104080.
79. Sessa C, Balmaña J, Bober SL, Cardoso MJ, Colombo N, Curigliano G, et al. Risk reduction and screening of cancer in hereditary breast-ovarian cancer syndromes: ESMO Clinical Practice Guideline. Ann Oncol 2023;34:33–47. https://doi.org/10.1016/j.annonc.2022.10.004.

80. Kawai M, Ohtani S, Iwasaki M, Yamamoto S, Takamatsu K, Okamura H, et al. The Japanese Breast Cancer Society clinical practice guidelines for epidemiology and prevention of breast cancer, 2022 edition. Breast Cancer 2024;31:166–78. https://doi.org/10.1007/s12282-023-01531-9.
81. Hanson H, Kulkarni A, Loong L, Kavanaugh G, Torr B, Allen S, et al. UK consensus recommendations for clinical management of cancer risk for women with germline pathogenic variants in cancer predisposition genes: RAD51C, RAD51D, BRIP1 and PALB2. J Med Genet 2023;60:417–29. https://doi.org/10.1136/jmg-2022-108898.
82. Hanson H, Brady AF, Crawford G, Eeles RA, Gibson S, Jorgensen M, et al. UKCGG Consensus Group guidelines for the management of patients with constitutional TP53 pathogenic variants. J Med Genet 2020;58:135–9. https://doi.org/10.1136/jmedgenet-2020-106876.
83. Carneiro VCG, Gifoni ACLVC, Mauro Rossi B, Andrade CEM da C, Lima FT de, Galvão H de CR, et al. Cancer risk-reducing surgery: Brazilian society of surgical oncology guideline part 1 (gynecology and breast). J Surg Oncol 2022;126:10–9. https://doi.org/10.1002/jso.26812.
84. Ovarian cancer: identifying and managing familial and genetic risk | Guidance | NICE n.d.
85. Lee B, Chang SJ, Kwon BS, Son JH, Lim MC, Kim YH, et al. Clinical practice guidelines for ovarian cancer: an update to the Korean Society of Gynecologic Oncology guidelines. J Gynecol Oncol 2025;36. https://doi.org/10.3802/jgo.2025.36.e69.
86. Shenoy S. CDH1 (E-Cadherin) Mutation and Gastric Cancer: Genetics, Molecular Mechanisms and Guidelines for Management. Cancer Manag Res 2019;11:10477–86. https://doi.org/10.2147/CMAR.S208818.
87. Ortmann O, Schüler-Toprak S, Kast K, Speiser D, Seitz S, Rhiem K, et al. The risk of endocrine interventions in carriers of a genetic predisposition for breast and gynecologic cancers: recommendations of the German Consortium for Hereditary Breast and Ovarian Cancer. J Cancer Res Clin Oncol 2024;150. https://doi.org/10.1007/s00432-024-05936-7.
88. Goggins M, Overbeek KA, Brand R, Syngal S, Del Chiaro M, Bartsch DK, et al. Management of patients with increased risk for familial pancreatic cancer: updated recommendations from the International Cancer of the Pancreas Screening (CAPS) Consortium. Gut 2020;69:7–17. https://doi.org/10.1136/gutjnl-2019-319352.
89. Das A, MacFarland SP, Meade J, Hansford JR, Schneider KW, Kuiper RP, et al. Clinical Updates and Surveillance Recommendations for DNA Replication Repair Deficiency Syndromes in Children and Young Adults. Clin Cancer Res 2024;30:3378–87. https://doi.org/10.1158/1078-0432.CCR-23-3994.
90. van Leerdam ME, Roos VH, van Hooft JE, Balaguer F, Dekker E, Kaminski MF, et al. Endoscopic management of Lynch syndrome and of familial risk of colorectal cancer: European Society of Gastrointestinal Endoscopy (ESGE) Guideline. Endoscopy 2019;51:1082–93. https://doi.org/10.1055/a-1016-4977.
91. Frebourg T, Bajalica Lagercrantz S, Oliveira C, Magenheim R, Evans DG, Hoogerbrugge N, et al. Guidelines for the Li–Fraumeni and heritable TP53-related cancer syndromes. European Journal of Human Genetics 2020 28:10 2020;28:1379–86. https://doi.org/10.1038/s41431-020-0638-4.

92. Ovarian Cancer Screening Guidelines | Mays Cancer Center n.d. https://cancer.uthscsa.edu/cancer-care/screenings-diagnosis/ovarian-cancer-screening-guidelines (accessed February 5, 2026).
93. Monticciolo DL, Newell MS, Moy L, Lee CS, Destounis S V. Breast Cancer Screening for Women at Higher-Than-Average Risk: Updated Recommendations From the ACR. Journal of the American College of Radiology 2023;20:902–14. https://doi.org/10.1016/j.jacr.2023.04.002.
94. Brzezinski JJ, Becktell KD, Bougeard G, Brodeur GM, Diller LR, Doria AS, et al. Update on Surveillance Guidelines in Emerging Wilms Tumor Predisposition Syndromes. Clin Cancer Res 2025;31:18–24. https://doi.org/10.1158/1078-0432.CCR-24-2488.
95. Sawhney MS, Calderwood AH, Thosani NC, Rebbeck TR, Wani S, Canto MI, et al. ASGE guideline on screening for pancreatic cancer in individuals with genetic susceptibility: summary and recommendations. Gastrointest Endosc 2022;95:817–26. https://doi.org/10.1016/j.gie.2021.12.001.
96. Tsoulaki O, Evans DG, Sinha K, Rajan N, Bakr F, Hatcher H, et al. UK clinical practice guidelines for the management of patients with constitutional POT1 pathogenic variants. J Med Genet 2025;62:559–65. https://doi.org/10.1136/jmg-2025-110638.
97. Paluch-Shimon S, Cardoso F, Partridge AH, Abulkhair O, Azim HA, Bianchi-Micheli G, et al. ESO–ESMO fifth international consensus guidelines for breast cancer in young women (BCY5). Annals of Oncology 2022;33:1097–118. https://doi.org/10.1016/j.annonc.2022.07.007.
98. Motzer RJ, Jonasch E, Boyle S, Carlo MI, Manley B, Agarwal N, et al. Kidney Cancer, Version 1.2021: Featured Updates to the NCCN Guidelines. J Natl Compr Canc Netw 2020;18:1160. https://doi.org/10.6004/jnccn.2020.0043.
99. Gonda TA, Everett JN, Wallace M, Simeone DM, Amundadottir L, Beyer G, et al. Recommendations for a More Organized and Effective Approach to the Early Detection of Pancreatic Cancer From the PRECEDE (Pancreatic Cancer Early Detection) Consortium. Gastroenterology 2021;161:1751–7. https://doi.org/10.1053/j.gastro.2021.08.036.
100. Achatz MI, Caleffi M, Guindalini R, Marques RM, Nogueira-Rodrigues A, Ashton-Prolla P. Recommendations for advancing the diagnosis and management of hereditary breast and ovarian cancer in Brazil. J Glob Oncol 2020;6:439–52. https://doi.org/10.1200/JGO.19.00170.
101. Urban LABD, Chala LF, Paula IB De, Bauab SDP, Schaefer MB, Oliveira ALK, et al. Recommendations for the Screening of Breast Cancer of the Brazilian College of Radiology and Diagnostic Imaging, Brazilian Society of Mastology and Brazilian Federation of Gynecology and Obstetrics Association. Rev Bras Ginecol Obstet 2023;45:E480–8. https://doi.org/10.1055/s-0043-1772498.
102. Family history of breast cancer and inherited genes | Breast cancer | Cancer Research UK n.d. https://www.cancerresearchuk.org/about-cancer/breast-cancer/risks-causes/family-history-and-inherited-genes (accessed February 5, 2026).
103. Ploussard G, Baboudjian M, Barret E, Brureau L, Fiard G, Fromont G, et al. French AFU Cancer Committee Guidelines – Update 2024–2026: Prostate cancer – Diagnosis and management of

localised disease. The French Journal of Urology 2024;34:102717. https://doi.org/10.1016/j.fjurol.2024.102717.

104. Hassett MJ, Somerfield MR, Baker ER, Cardoso F, Kansal KJ, Kwait DC, et al. Management of male breast cancer: ASCO guideline. Journal of Clinical Oncology 2020;38:1849–63. https://doi.org/10.1200/JCO.19.03120.
105. Hansford JR, Das A, McGee RB, Nakano Y, Brzezinski J, Scollon SR, et al. Update on Cancer Predisposition Syndromes and Surveillance Guidelines for Childhood Brain Tumors. Clin Cancer Res 2024;30:2342–50. https://doi.org/10.1158/1078-0432.CCR-23-4033.
106. Brandi ML, Pieterman CRC, English KA, Lines KE, Shariq OA, Marini F, et al. Multiple endocrine neoplasia type 1 (MEN1): recommendations and guidelines for best practice. Lancet Diabetes Endocrinol 2025;13:699–721. https://doi.org/10.1016/S2213-8587(25)00119-6.
107. Conroy T, Pfeiffer P, Vilgrain V, Lamarca A, Seufferlein T, O'Reilly EM, et al. Pancreatic cancer: ESMO Clinical Practice Guideline for diagnosis, treatment and follow-up☐. Annals of Oncology 2023;34:987–1002. https://doi.org/10.1016/j.annonc.2023.08.009.
108. Lee CR, Luzum JA, Sangkuhl K, Gammal RS, Sabatine MS, Stein CM, et al. Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2C19 Genotype and Clopidogrel Therapy: 2022 Update. Clin Pharmacol Ther 2022;112:959–67. https://doi.org/10.1002/cpt.2526.
109. Hamzic S, Aebi S, Joerger M, Montemurro M, Ansari M, Amstutz U, et al. Fluoropyrimidine chemotherapy: recommendations for DPYD genotyping and therapeutic drug monitoring of the Swiss Group of Pharmacogenomics and Personalised Therapy. Swiss Med Wkly 2020;150:w20375. https://doi.org/10.4414/smw.2020.20375.
110. Hulshof EC, Deenen MJ, Nijenhuis M, Soree B, de Boer-Veger NJ, Buunk AM, et al. Dutch pharmacogenetics working group (DPWG) guideline for the gene–drug interaction between UGT1A1 and irinotecan. European Journal of Human Genetics 2022 31:9 2022;31:982–7. https://doi.org/10.1038/s41431-022-01243-2.
111. Rhee EJ, Kim HC, Kim JH, Lee EY, Kim BJ, Kim EM, et al. 2018 Guidelines for the Management of Dyslipidemia in Korea. J Lipid Atheroscler 2019;8:78–131. https://doi.org/10.12997/jla.2019.8.2.78.
112. Malhotra H, Pramanik R, Srinivas S, Kotwal P, Mehra N, Gupta S, et al. Genetic Counselling, Testing, and Management of Hereditary Breast and Ovarian Cancer Syndrome in India: Updated Expert Consensus Recommendations from Indian Society of Medical and Pediatric Oncology. Indian Journal of Medical and Paediatric Oncology 2025;46:233–52. https://doi.org/10.1055/s-0044-1788727.
113. Ditsch N, Wöcke A, Untch M, Jackisch C, Albert US, Banys-Paluchowski M, et al. AGO Recommendations for the Diagnosis and Treatment of Patients with Early Breast Cancer: Update 2022. Breast Care 2022;17:403–20. https://doi.org/10.1159/000524879.
114. Park YH, Lee SC, Singer CF, Balmaña J, Dent RA, Tan VKM, et al. Part II: consensus statements and expert recommendations for BRCA-associated breast cancer in the Asia-Pacific region: clinical management. Front Oncol 2025;15:1507840. https://doi.org/10.3389/fonc.2025.1507840.

115. Lebreton L, Boyer JC, Lafay-Chebassier C, Hennart B, Baklouti S, Cunat S, et al. French-Speaking Network of Pharmacogenetics (RNPGx) Recommendations for Clinical Use of Mavacamten. Clin Pharmacol Ther 2025;117:387–97. https://doi.org/10.1002/cpt.3502.
116. Konstantinopoulos PA, Norquist B, Lacchetti C, Armstrong D, Grisham RN, Goodfellow PJ, et al. Germline and somatic tumor testing in epithelial ovarian cancer: ASCO guideline. Journal of Clinical Oncology 2020;38:1222–45. https://doi.org/10.1200/JCO.19.02960.
117. Lee CJ, Yoon M, Kang HJ, Kim BJ, Choi SH, Jeong IK, et al. 2022 Consensus Statement on the Management of Familial Hypercholesterolemia in Korea. J Lipid Atheroscler 2022;11:213–28. https://doi.org/10.12997/jla.2022.11.3.213.
118. Taha HSED, Kandil H, Farag N, Oraby A, Sharkawy M El, Fawzy F, et al. Egyptian practical guidance in hypertriglyceridemia management 2021. Egypt Heart J 2021;73. https://doi.org/10.1186/s43044-021-00235-9.
119. Watts GF, Gidding SS, Hegele RA, Raal FJ, Sturm AC, Jones LK, et al. International Atherosclerosis Society guidance for implementing best practice in the care of familial hypercholesterolaemia. Nature Reviews Cardiology 2023 20:12 2023;20:845–69. https://doi.org/10.1038/s41569-023-00892-0.
120. Colas C, Guerrini-Rousseau L, Suerink M, Gallon R, Kratz CP, Ayuso É, et al. ERN GENTURIS guidelines on constitutional mismatch repair deficiency diagnosis, genetic counselling, surveillance, quality of life, and clinical management. European Journal of Human Genetics 2024 32:12 2024;32:1526–41. https://doi.org/10.1038/s41431-024-01708-6.
121. Guillén-Ponce C, Lastra E, Lorenzo-Lorenzo I, Martín Gómez T, Morales Chamorro R, Sánchez-Heras AB, et al. SEOM clinical guideline on hereditary colorectal cancer (2019). Clinical and Translational Oncology 2020;22:201–12. https://doi.org/10.1007/s12094-019-02272-y.
122. Tomita N, Ishida H, Tanakaya K, Yamaguchi T, Kumamoto K, Tanaka T, et al. Japanese Society for Cancer of the Colon and Rectum (JSCCR) guidelines 2020 for the Clinical Practice of Hereditary Colorectal Cancer. International Journal of Clinical Oncology 2021 26:8 2021;26:1353–419. https://doi.org/10.1007/s10147-021-01881-4.
123. Gonzalez-Santos LE, Oliva R, Jimeno C, Gonzales E, Balabagno MM, Ona D, et al. Executive Summary of the 2020 Clinical Practice Guidelines for the Management of Dyslipidemia in the Philippines. J ASEAN Fed Endocr Soc 2021;36:5–11. https://doi.org/10.15605/JAFES.036.01.01.
124. Monahan KJ, Bradshaw N, Dolwani S, Desouza B, Dunlop MG, East JE, et al. Guidelines for the management of hereditary colorectal cancer from the British Society of Gastroenterology (BSG)/Association of Coloproctology of Great Britain and Ireland (ACPGBI)/United Kingdom Cancer Genetics Group (UKCGG). Gut 2020;69:411–44. https://doi.org/10.1136/gutjnl-2019-319915.
125. (UK) NGA. Effectiveness of aspirin in the prevention of colorectal cancer in people with Lynch syndrome. Effectiveness of Aspirin in the Prevention of Colorectal Cancer in People with Lynch Syndrome: Colorectal Cancer (Update): Evidence Review A1 2020.

126. Précoma DB, de Oliveira GMM, Simão AF, Dutra OP, Coelho OR, Izar MC de O, et al. Updated Cardiovascular Prevention Guideline of the Brazilian Society of Cardiology - 2019. Arq Bras Cardiol 2019;113:787–891. https://doi.org/10.5935/abc.20190204.
127. Seppälä TT, Latchford A, Negoi I, Sampaio Soares A, Jimenez-Rodriguez R, Evans DG, et al. European guidelines from the EHTG and ESCP for Lynch syndrome: an updated third edition of the Mallorca guidelines based on gene and gender. BJS 2021;108:484–98. https://doi.org/10.1002/bjs.11902.
128. Zaffaroni G, Mannucci A, Koskenvuo L, de Lacy B, Maffioli A, Bisseling T, et al. Updated European guidelines for clinical management of familial adenomatous polyposis (FAP), MUTYH-associated polyposis (MAP), gastric adenocarcinoma, proximal polyposis of the stomach (GAPPS) and other rare adenomatous polyposis syndromes: a joint EHTG-…. BJS 2024;111:42. https://doi.org/10.1093/bjs/znae070.
129. Achatz MI, Villani A, Bertuch AA, Bougeard G, Chang VY, Doria AS, et al. Update on Cancer Screening Recommendations for Individuals with Li–Fraumeni Syndrome. Clinical Cancer Research 2025;31:1831–40. https://doi.org/10.1158/1078-0432.CCR-24-3301.
130. Morgan DR, Corral JE, Li D, Montgomery EA, Riquelme A, Kim JJ, et al. ACG Clinical Guideline: Diagnosis and Management of Gastric Premalignant Conditions. Am J Gastroenterol 2025;120:709–37. https://doi.org/10.14309/ajg.0000000000003350.
131. Sánchez-Heras AB, Ramon y Cajal T, Pineda M, Aguirre E, Graña B, Chirivella I, et al. SEOM clinical guideline on heritable TP53-related cancer syndrome (2022). Clinical and Translational Oncology 2023 25:9 2023;25:2627–33. https://doi.org/10.1007/s12094-023-03202-9.
132. Barretina-Ginesta MP, Quindós M, Alarcón JD, Esteban C, Gaba L, Gómez C, et al. SEOM-GEICO clinical guidelines on endometrial cancer (2021). Clin Transl Oncol 2022;24:625–34. https://doi.org/10.1007/s12094-022-02799-7.
133. Simard J, Kamath S, Kircher S. Survivorship Guidance for Patients with Colorectal Cancer. Curr Treat Options Oncol 2019;20. https://doi.org/10.1007/s11864-019-0635-4.
134. Brandi ML, Pieterman CRC, English KA, Lines KE, Shariq OA, Marini F, et al. Multiple endocrine neoplasia type 1 (MEN1): recommendations and guidelines for best practice. Lancet Diabetes Endocrinol 2025;13:699–721. https://doi.org/10.1016/S2213-8587(25)00119-6.
135. Greenhalf W, Lévy P, Gress T, Rebours V, Brand RE, Pandol S, et al. International consensus guidelines on surveillance for pancreatic cancer in chronic pancreatitis. Recommendations from the working group for the international consensus guidelines for chronic pancreatitis in collaboration with the International Associati…. Pancreatology 2020;20:910–8. https://doi.org/10.1016/j.pan.2020.05.011.

Figures

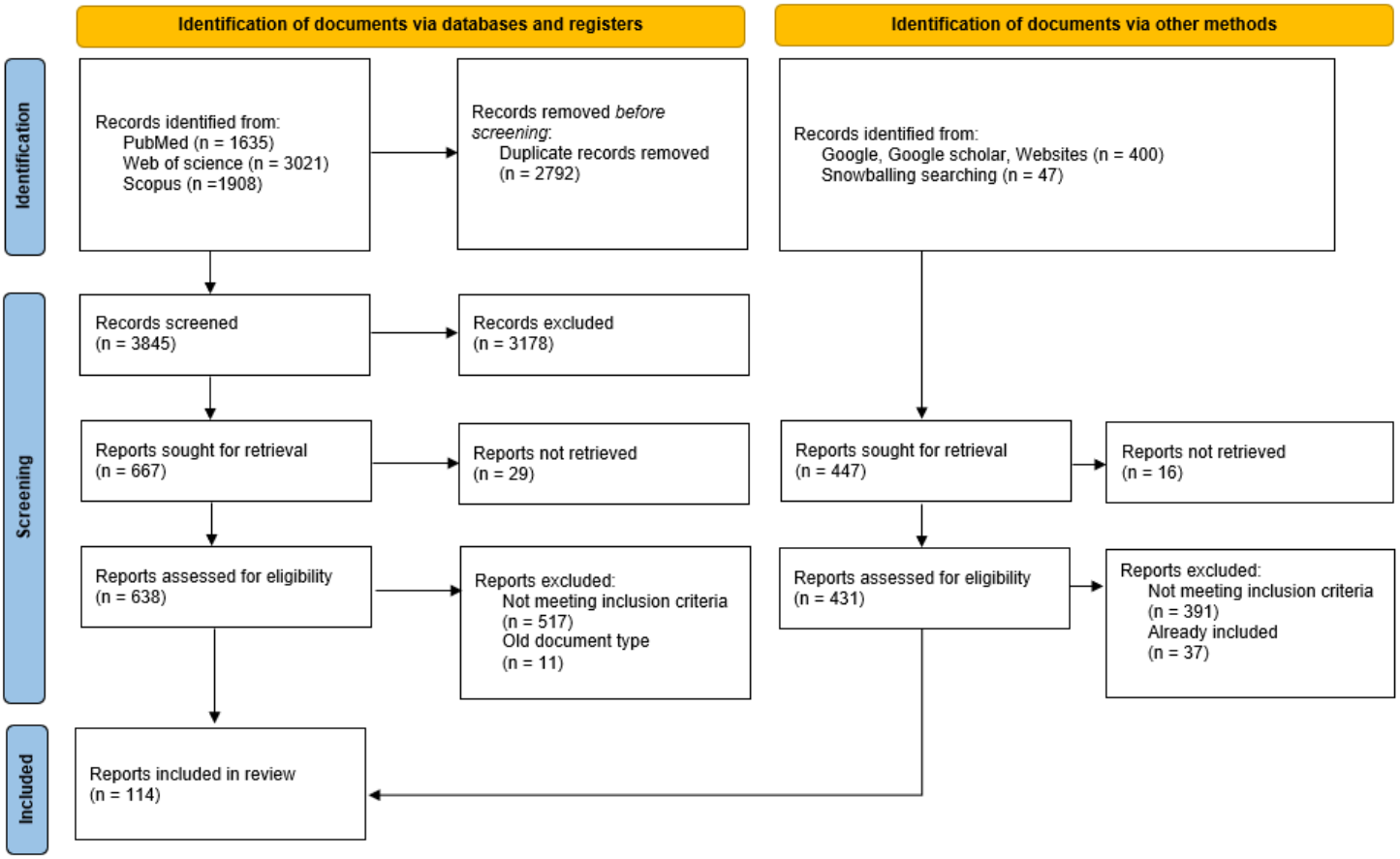


Figure 1

PRISMA flowchart

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