

The background of the cover features a complex, abstract pattern of elongated, overlapping shapes in various colors including red, orange, yellow, and purple, resembling a microscopic view of biological tissue or a stained histological slide. The pattern is dense and fills most of the cover area.

Advanced Health Assessment and Clinical Diagnosis in Primary Care

5th
EDITION

Joyce E. Dains
Linda Ciofu Baumann
Pamela Scheibel

ELSEVIER

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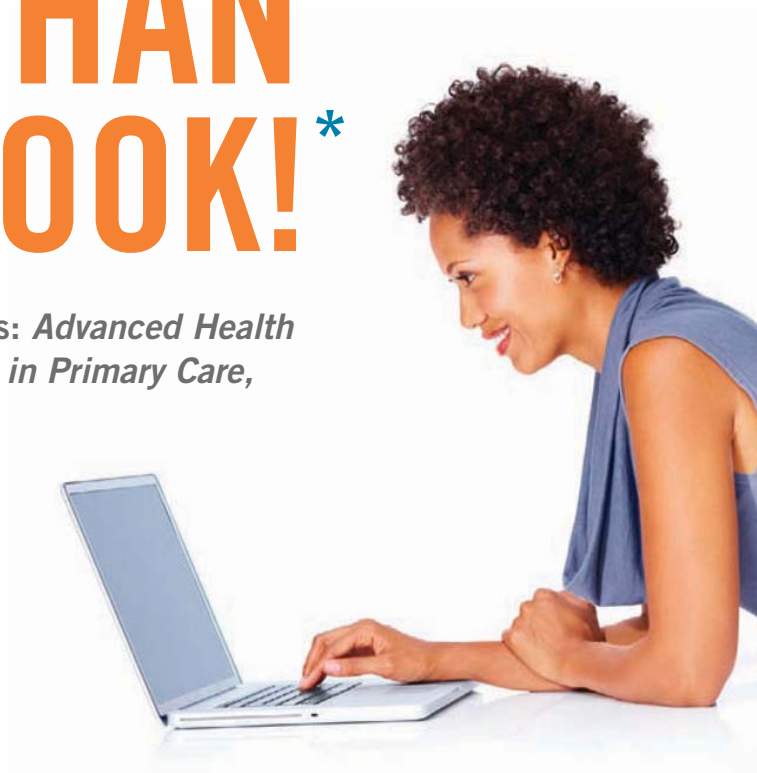
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Advanced Health Assessment and Clinical Diagnosis in Primary Care

5th
EDITION

**Joyce E. Dains, DrPH, JD, RN, FNP-BC,
DPNAP, FAANP**

Associate Professor and Advanced Practice Nursing
Program Director
The University of Texas MD Anderson Cancer Center
Houston, Texas

Linda Ciofu Baumann, PhD, APRN, BC, FAAN

Professor Emerita
University of Wisconsin–Madison
School of Nursing
Madison, Wisconsin

Pamela Scheibel, MSN, RN, PNP

Clinical Professor Emerita
University of Wisconsin–Madison
School of Nursing
Madison, Wisconsin

ELSEVIER

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Executive Content Strategist: Lee Henderson

Traditional Content Development Manager: Billie C. Sharp

Associate Content Development Specialist: Samantha Dalton

Publishing Services Managers: Jeff Patterson and Julie Eddy

Senior Project Manager: Mary Stueck

Design Direction: Ryan Cook

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Reviewers

Nancy F. Altice, DNP, RN, CCNS, ACNS-BC

Cardiology Clinical Nurse Specialist
Carilion Clinic–Roanoke Memorial Hospital
Roanoke, Virginia

Ryan J. Clancy, MSHA, MA, PA-C

Clinical Instructor
Drexel University
College of Nursing and Health Professions
Philadelphia, Pennsylvania

Michelle DiBaise, MPAS, PA-C, DFAAPA

Associate Clinical Professor
Northern Arizona University
Department of Physician Assistant Studies
Phoenix, Arizona

Laura C. Doyle, MSN, BSN, CPNP

Pediatric Nurse Practitioner
Ann & Robert H. Lurie Children’s Hospital of Chicago
Chicago, Illinois

Teresa M. Flores, PA-C

Lecturer
University of Washington–MEDEX
School of Medicine
Seattle, Washington

Drew A. Garcia, PA-C

Faculty
University of Washington–MEDEX
School of Medicine
Seattle, Washington

Stephen D. Heglund, PhD, ARNP

Lecturer
Coordinator RN to BSN/MSN & Concurrent Programs
University of Florida
College of Nursing
Orlando, Florida;
Emergency Department Nurse Practitioner
Sebastian River Medical Center
Sebastian, Florida

Kathleen Sanders Jordan, DNP, MS, RN, FNP-BC, ENP-BC, SANE-P

Lecturer
University of North Carolina–Charlotte
School of Nursing;
Nurse Practitioner
Mid-Atlantic Emergency Medicine Associates
Charlotte, North Carolina

P. Rae Katz, PhD, PA-C

Assistant Professor
Director of Faculty Development
Rosalind Franklin University of Medicine and Science
North Chicago, Illinois

Kari Jean Ksar, RN, MS, CPNP

Pediatric Nurse Practitioner
Pediatric Gastroenterology, Hepatology, and Nutrition
Lucile Packard Children's Hospital
Palo Alto, California

Andrea Mose, RNC, WHNP

Women's Health Nurse Practitioner
University of Missouri–St. Louis
St. Louis, Missouri

Felix Nwamaghinna, MSB, PA-C

Chairman
Assistant Professor
Physician Assistant Program
State University of New York Downstate Medical Center
Brooklyn, New York

Sharon Gallagher Owens, ACNP-BC, PhD

Assistant Director of Nursing-Cardiac Surgery
Johns Hopkins Hospital
Baltimore, Maryland

Jill Harpst Rodgers, DNP, FNP-BC, MSN

Assistant Professor
Carlow University
Graduate School of Nursing
Pittsburgh, Pennsylvania

Megan E. Schneider, MMS, MSPH, PA-C

Clinical Instructor
Drexel University
Philadelphia, Pennsylvania

Michelle Staron, MMS, PA-C

Cardiovascular Surgery Physician Assistant
Ann & Robert H. Lurie Children's Hospital of Chicago
Chicago, Illinois

Laura A. Steadman, EdD, MSN, CRNP, FNP-BC

Assistant Professor
University of Alabama–Birmingham
School of Nursing
Birmingham, Alabama

Andrea Magdalia Trimmingham, MA, RPA-C

Assistant Professor
State University of New York Downstate Medical Center
Brooklyn, New York



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Katharine E. Hohol, MS, APRN, BC, APNP

Sandra K. Roof, MSN, APRN, BC, APNP

Robert W. Vogler, PhD, RN, CS, FNP

Pam Willson, PhD, RN, CS, FNP

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Joyce E. Dains
Linda C. Baumann
Pamela Scheibel

Introduction

This text is designed for beginning clinicians and for students who will be using history and physical examination skills in the clinical setting. Its purpose is to take the student to the “next step” of health assessment, that is, beyond basic history and physical examination to using a diagnostic reasoning process. The book is intended to fill the gap between basic physical examination texts and the medical texts that are aimed primarily at disease management. It is not intended as a substitute for a clinical management text nor does it address management of disorders or diseases. Rather, it is designed specifically to focus on the clinical evaluation of common problems that present in primary care settings, using the tools of history and physical examination to engage in the process of clinical diagnosis.

The fifth edition of this text has several changes to further assist the transition to that “next step” of health assessment. Two new chapters have been added that reflect concerns commonly seen in primary care settings: Evidence-Based Health Screening (Chapter 2) and Heartburn and Indigestion (Chapter 20). The coverage of limb pain has been expanded into two chapters: Lower Extremity Limb Pain (Chapter 22) and Upper Extremity Limb Pain (Chapter 23). We continue to provide a list of selected references at the end of each chapter. These references support the information provided and are suggested for further in-depth learning about the problem. New to this edition, we have included *Evidence-Based Practice* boxes in each chapter. The focus of *Advanced Health Assessment* is primary care patients. Both adults and children are included, with divergence in questions, examination, or interpretation of findings noted where pertinent. This text does not attempt to address all possible patient concerns but rather seeks to focus on the most common concerns as exemplars of the diagnostic reasoning process.

HOW TO USE THIS BOOK

Each chapter is structured in the context of a commonly occurring chief concern rather than a specific diagnosis or disease entity. Patients generally seek care

for relief of symptoms and undiagnosed conditions. The initial challenge for primary care providers is to begin the process of differential diagnosis to determine the cause of a problem, based on history and physical examination and laboratory and other diagnostic tests. However, the steps of the diagnostic reasoning process are seldom articulated in a sequence that reflects the clinician’s thought process. Novice clinicians are often left to their own devices to figure out, for example, which history questions are the most important, which should be asked first and which can be left for later, or to determine which parts of the physical examination must be done as opposed to which will yield little information for a given concern. This text tries to articulate the reasoning process, order the history questions in a meaningful way, and focus the physical examination for a specific chief concern.

The diagnostic or clinical reasoning process is woven into each presenting problem. Each symptom begins with a brief introduction, providing an overview of causative mechanisms and processes. The clinical problem-solving process begins with *Focused History*, which walks through the thinking process involved in obtaining a pertinent, relevant, problem-specific history that will assist with differential diagnosis. The section is designed around questions that experienced clinicians ask themselves to order and organize the questions to be asked of the patient. These “self-questions” are structured according to what information the clinician needs first or most immediately about the presenting complaint, followed by self-questions that help sort through the possible differential diagnoses. The content and order of the self-questions vary, depending on the presenting problem. Sometimes the self-questions are based on what the condition is most likely to be; sometimes they are based on what is too important to miss.

For each of the self-questions there is a list of *Key Questions* to ask of the patient or about the patient if a family member is giving the history. The *Key Questions* are followed by an interpretation or explanation of what the patient responses might signify. For ease of format,

the *Key Questions* are written as though the clinician were addressing the patient. Certainly with young children and sometimes with adults, the clinician will be asking questions of another person about the patient. The intent is to convey what questions to ask, rather than to provide every possible format for each question.

Following these two sections is the *Focused Physical Examination* section. It instructs you in what focused physical examination to perform to assist the diagnostic process. The section is not intended to teach basic physical examination; it assumes you know how, using the techniques of inspection, auscultation, percussion, and palpation. This section, rather, provides focus for the examination, explains how to do more advanced maneuvers, and offers an interpretation of the findings.

Following is the section titled *Laboratory and Diagnostic Studies*. This section provides a brief outline of what kinds of laboratory or diagnostic studies would be appropriate for the chief concern or suspected diagnosis. Because the goal of the text is clinical diagnosis,

the laboratory and diagnostic studies included are those that would be a logical starting point, although perhaps not an ending point.

The final section of each presenting concern is the *Differential Diagnosis*. It contains the most common differential diagnoses for the chief concern and summarizes, in a narrative format, the history and physical examination findings, along with the laboratory and diagnostic studies indicated. The section finishes with one or more *Differential Diagnosis* tables, mirroring the narrative summary, which can be used as a quick reference. An index to the *Differential Diagnosis* tables is provided in the text inside the back cover.

Perfecting advanced health assessment skills is a life-long process. It is our hope that this edition continues to assist you in expanding the diagnostic reasoning process.

Joyce E. Dains
Linda C. Baumann
Pamela Scheibel

CHAPTER

1

Clinical Reasoning, Differential Diagnosis, Evidence-Based Practice, and Symptom Analysis

Basic health assessment involves the application of the practitioner’s knowledge and skills to identify and distinguish normal from abnormal findings. Basic assessment often moves from a general survey of a body system to specific observations or tests of function. Such an approach to assessment and clinical decision-making uses a deductive process of reasoning. For example, a specialist examining a patient with known hyperthyroidism would conduct a physical examination to test for deep tendon reflexes. Brisk or hyperreflexic reflexes would lead the practitioner to conclude that a hyperthyroid state is a likely cause for these findings. This would greatly narrow the choices of diagnostic tests and treatment decisions.

Advanced assessment builds on basic health assessment yet is performed more often using an inductive or inferential process, that is, moving from a specific physical finding or patient concern to a more general diagnosis or possible diagnoses based on history, physical findings, and the results of laboratory and diagnostic tests. The practitioner gathers further evidence and analyzes this evidence to arrive at a hypothesis that will lead to a further narrowing of possibilities. This is known as the process of diagnostic reasoning.

DIAGNOSTIC REASONING

Diagnostic reasoning is a scientific process in which the practitioner suspects the cause of a patient’s symptoms and signs based on previous knowledge. The practitioner gathers relevant information, selects necessary tests, makes an accurate diagnosis, and recommends therapy. The difference between an average and an excellent practitioner is the speed and focus used to arrive at the correct conclusion and initiate the best course of evidence-based treatment with minimum harm, cost, inconvenience, and delay. This expertise of the practitioner is acquired through knowledge and a

skill set developed through experience in clinical practice.

By using diagnostic reasoning, the practitioner is able to accomplish the following:

- Determines and focuses on what needs to be asked, what data need to be obtained, and what needs to be examined
- Performs examinations and diagnostic tests accurately
- Clusters all pertinent findings
- Analyzes and interprets the findings
- Develops a list of likely or differential diagnoses

THE DIAGNOSTIC PROCESS

The Primary Care Context

The process of assessment in the primary care setting begins with the patient stating a reason for the visit or a chief concern. Most visits to primary care providers involve concerns or symptoms presented by the patient, such as an earache, vomiting, or fatigue. The initial evidence is collected through a patient history. Demographic information, such as gender, age, occupation, and place of residence, is obtained to place the patient in a risk category that may rule out certain diagnoses immediately. In most primary care settings, routine vital signs are obtained, which can include height and weight, temperature, pulse, respiratory rate, blood pressure, last menstrual period, and smoking status. While obtaining the history, the practitioner also makes observations of the patient’s appearance, interaction with family members, orientation, and mental and physical condition. The practitioner notes any unusual presentations that could help focus the assessment process.

Symptom Analysis

Presenting symptoms need to be explored with further questions. One useful mnemonic for gathering this information is COLDSPA.

Character: How does it feel, look, smell, sound?

Onset: When did it start?

Location: Be specific. Where is it? Does it radiate?

Duration: How long does it last? Does it recur?

Severity: How do you rate your pain on a scale from 0 (no pain) to 10 (worst pain I've ever had)?

Pattern: What makes it better? What makes it worse? What have you done and did it help?

Associated factors: What other symptoms do you have? How much does it interfere with your usual activities?

Another mnemonic is **OLDCARTS**: **O**nset; **L**ocation; **D**uration; **C**haracter; **A**ggravating/associated factors; **R**elieving factors; **T**emporal factors; **S**everity.

Information can also be gleaned from the review of systems. A final step is to ask about the patient's perception of the meaning of the symptom(s). The practitioner then clusters the information into logical groups based on prior knowledge of symptom clusters associated with specific diagnoses or body systems. At the conclusion, the history of the presenting concerns should give the practitioner a good idea of the most likely differential diagnoses. These hypotheses may be further strengthened during the physical examination.

Performing a Physical Examination

This section may be performed as a complete physical examination or as a focused/localized examination that emphasizes the body or organ systems most likely affected by the patient's presenting symptoms.

Formulating and Testing a Hypothesis

The practitioner then formulates a hypothesis based on expertise and knowledge of probable processes, such as a pathological, physiological, or psychological process. Further interpretation of evidence refines the hypothesis to a working or probable diagnosis. Hypothesis generation, in all likelihood, begins during the assessment of the patient's age, gender, race, appearance, and presenting problem. Age is often the most significant variable in narrowing the probabilities of a problem. Hypothesis generation forms the context in which further data are collected. This context includes the setting in which care is delivered, such as in a hospital, in an outpatient setting, or in another community-based setting where more than a single individual could be affected. Clinical decision-making can be filled with uncertainty and ambiguity. Because available evidence is almost never complete, hypothesis formation involves some element of subjective judgment.

The hypothesis must then be tested and assessed for the following characteristics.

- *Coherence:* Are the physiological links, predisposing factors, and complications for this disease present in the patient?
- *Adequacy:* Does the suspected disease encompass all of the patient's normal and abnormal findings?
- *Parsimony:* Is it the simplest explanation of the patient's findings? The surest way to make this determination is to ask the patient (or the parents) the reason for seeking care and the current understanding of the problem and possible treatment options. This is a crucial step because patients must find the treatment recommendation acceptable.
- *Diagnostic probability:* Is the diagnosis confirmed by radiographic or laboratory tests? A rational diagnostic hypothesis is one that, if confirmed by the select tests, limits the need for additional confirmation.
- *Eliminate a competing hypothesis:* What other diseases could explain the patient's symptoms?

To confirm the hypothesis, the practitioner establishes a working definition of the problem as a basis for a treatment plan and evaluates the outcome. The goal of a clinical decision is to choose an action that is most likely to result in the health outcomes the patient desires. This step of the decision-making process involves personal preference as to whether the benefits outweigh the harms involved, whether the cost is reasonable, and whether the most desired outcomes are short or long term.

Practitioners make extensive use of heuristics, or rules of thumb, to guide the inductive or inferential process of diagnostic reasoning. Heuristics are generally accurate and useful rules to make the task of information gathering more manageable and efficient—rules such as familiarity, salience, and resemblance to a patient who has a known disease. On occasion, however, heuristics can be faulty, particularly if the presentation is atypical or the condition is rare. The practitioner must always be open to a low probability of a serious diagnosis. Heuristics can have negative effects when stereotypes or biases influence judgment. For example, assuming that a patient is heterosexual can lead to errors in clinical reasoning and differential diagnosis when evaluating the symptom of rectal pain.

EXPERT VERSUS NOVICE PRACTITIONERS

Students of advanced assessment have a variety of backgrounds, with many coming from specialized areas of clinical practice. Such students could have difficulty broadening the scope of their observations

and clinical possibilities. In any case, whether they are specialists or not, nonexperts tend to be nonselective in data gathering and in the clinical reasoning strategies they use. Experts, however, are able to focus on a problem, recognize patterns, and gather only relevant data, with a high probability of a correct diagnosis. The goal for a novice practitioner is to aim for competence and expertise.

The competent practitioner will execute the following steps:

1. *Identify the most important cues.* These cues are obtained largely through thorough symptom analysis (e.g., COLSDPA or OLD CARTS), functional assessment, and history to assess the patient's beliefs and understanding or explanatory model of the illness. Research evidence shows that a person's beliefs or explanatory models of an illness or a symptom include a cause, an opinion about the timeline (acute or chronic), consequences of the condition (minor or life threatening), and some type of verbal label used to identify the cluster of symptoms or sensations (e.g., "the flu," "the blues"). Practitioners need to distinguish between the presence of disease, which has a biological basis, and illness, which is the human experience of being sick that could have little correlation with the objective evidence available.
2. *Understand and perform advanced examination techniques.* These techniques can include special maneuvers and closer observation of fine details during the physical examination, more in-depth interviews using valid and reliable instruments to assess the patient's risk for a specific diagnosis, and "gold standard" diagnostic tests for the identification of a specific disorder.
3. *Test differential or competing diagnoses.* A differential diagnosis results from a synthesis of subjective and objective findings, including laboratory and diagnostic tests, with knowledge of known and recognized patterns of signs and symptoms. When using the "rule-out" strategy, the practitioner looks for the absence of findings that are frequently seen with a specific condition; the absence of a sensitive finding is strong evidence against the condition being present. When using the "rule-in" strategy, the practitioner looks for the presence of a finding with high specificity (low false-positive and high true-negative values); the presence of this finding is strong evidence that the condition is present.
4. *See a pattern in the information gathered.* A pattern or cluster of findings can emerge from the

subjective and objective data. This pattern could be evident during one patient encounter or it could depend on a pattern of signs and symptoms that develops over time. Often the expert practitioner can eliminate competing diagnoses only after the initial treatment prescribed is ineffective or after the symptoms either disappear sooner than expected or persist longer than expected.

DEVELOPING CLINICAL REASONING

Clinical reasoning is a situational, practice-based form of reasoning that acknowledges the many variables that are present in an actual clinical situation, such as social relationships or situations involving patient, family, community, and a team of health care providers. Clinical reasoning requires a background of scientific and technological research-based knowledge about general cases and a practical ability to evaluate the relevance of the evidence behind general scientific and technical knowledge and how it applies to a particular patient. In doing so, the clinician considers the patient's particular clinical trajectory; their concerns, values and preferences; and their particular vulnerabilities (e.g., having multiple comorbidities) and sensitivities to care interventions (e.g., known drug allergies and past responses to therapies) when formulating clinical decisions or conclusions.

NEGOTIATING GOALS AND EXPECTATIONS OF A PATIENT ENCOUNTER

It is important, especially in an ambulatory care setting, to identify the patient's goals, expectations, and resources to determine what needs to be achieved during an encounter. A patient who seeks care because of a bothersome symptom could be more interested in having the symptom relieved by a particular date than in knowing the cause or diagnostic explanation for the symptom. Other patients might want reassurance that a symptom or sign is not a serious condition and yet do not expect treatment to alleviate the sensations they are experiencing. An explicit discussion between the practitioner and patient is necessary to establish what the goals and focus of an encounter will be. Goals can be mutually negotiated to assure clinicians that serious conditions can be "ruled out" and to assure patients that their needs and desires are acknowledged.

EVIDENCE-BASED PRACTICE

Evidence-based practice (EBP) is the integration of clinical expertise with the most current, relevant, and sound research evidence to guide clinical practice decisions. Using evidence-based guidelines in practice, informed through research evidence, improves patient outcomes. EBP integrates the best research evidence with clinical expertise and the patient’s values and preferences, and involves the use of simple rules of logic to apply evidence from research to an individual patient. Some of these rules include evaluating the validity, reliability, and generalizability of the evidence. The levels of evidence range from the “gold standard” of the randomized clinical trial to case studies, correlational studies, and expert opinion. Practitioners and patients increasingly gather evidence from web-based sources, such as the Cochrane Library, which includes databases of systematic reviews of a clinical topic, abstracts of reviews of effectiveness, a controlled trial registry, and review methodology. These databases have gathered the “best evidence” related to clinical problems (**Evidence-Based Practice box**). Access to web-based data requires that the practitioner develop skills in health informatics—the application of computer technology to health care delivery—to develop skills in searching for and appraising evidence in the literature to guide care for a specific patient in a specific clinical context.

Evidence-Based Practice Boxes

A feature of the fifth edition of this text is to include evidence-based practice boxes in each chapter. The studies cited represent evidence from epidemiologic studies, meta-analyses, systematic reviews, and randomized clinical trials that informs and guides primary care practitioners in delivering clinical services.

SUMMARY

In the context of primary care practice, the orientation to the patient should be holistic and general and toward the most prevalent or common conditions in a particular population group. This orientation requires that the expert practitioner develop skills in inductive reasoning to arrive at a diagnosis and to develop a treatment plan that is acceptable to the patient. An ongoing relationship with the patient over time greatly enhances the database from which the practitioner works to arrive at the best clinical judgments. Treatment plans in primary care settings rely on low-level technology stress prevention, and encourage self-care behaviors as well as open and effective patient-provider communication.

Practitioners need to be able to search for and evaluate the best evidence to guide assessment, treatment, and evaluation of diagnostic efficacy on health outcomes. A practitioner can progress from novice to expert and become more efficient in exercising clinical

EVIDENCE-BASED PRACTICE *Web Sources*

National Guideline Clearinghouse	www.guideline.gov	Evidence-based practice guidelines and best practices
The Cochrane Collaboration	www.cochrane.org	Cochrane Library of systematic literature reviews about treatments and interventions
Cumulative Index to Nursing and Allied Health Literature	www.cinahl.com	CINAHL database on all aspects of nursing, allied health, alternative health, and community medicine
Medscape (from WebMD)	www.medscape.com	MEDLINE database maintained by the National Library of Medicine for biomedical content for dentistry, veterinary medicine, and nursing
Agency for Healthcare Research and Quality	www.ahrq.gov	A resource for information related to improving quality, safety, efficiency, and effectiveness of care
U.S. Preventive Services Task Force	www.ahrq.gov/clinic/uspstfix.htm	Evidence-based guidelines for screening children and adults in primary care settings
Clinical Evidence	www.clinicalevidence.org	A compendium of resources for informing treatment and patient care decisions
UpToDate	www.uptodate.com	Evidence-based clinical decision support database useful at the point of care

judgment by asking the right questions, seeking pertinent and high quality information from available scientific evidence, and using clinical reasoning to apply the best evidence to clinical practice.

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Evidence-Based Health Screening

Although most primary care visits are related to acute symptoms, the focus of this chapter is health screening conducted in primary care settings on asymptomatic adults and children, and the process of developing evidence-based guidelines for prevention and screening interventions. Primary care practitioners need knowledge of appropriate preventive services to guide screening, counseling, and preventive medication decisions. The benefits of delivering evidence-based screening services include improving the quality of care, achieving desired health outcomes, and reducing health care costs. The ability to analyze and evaluate evidence requires a knowledge base and critical thinking.

In addition to the provision of preventive services, practitioners can educate patients about missed opportunities for screening, as well as the potential harms of inappropriate utilization of these services. For example, screening for colorectal cancer is an underused service even though colorectal cancer is the second leading cause of cancer mortality and it is highly preventable. Overuse of services, such as routine electrocardiograms (ECGs), prostate-specific antigen (PSA) testing, or urinalysis increases health care costs and may potentially harm patients.

STEPS IN EVIDENCE-BASED SCREENING

First, begin with a clinical problem. To examine the suitability of exploring a topic to develop screening guidelines, its public health importance needs to be assessed. Criteria include the burden of suffering a disorder poses to a population, and the anticipated effectiveness of a preventive service or intervention to reduce that burden.

Second, pose a clinical question that focuses on a patient problem and potential preventive service. A useful format that incorporates key components of a well-constructed question is PICO: *problem*, *intervention* or *exposure*, *comparison*, and *outcome*.

An analytic framework can be useful in illustrating the chain of evidence that needs to be evaluated in moving from a screening or preventive intervention to health outcomes, such as improved quality and quantity of life (Figure 2-1). Following the overarching question of “Does screening for X reduce morbidity and/or mortality?” (key question 1) is a series of questions to establish the clinical logic to support the implementation of a preventive service in a primary care setting:

2. Can a group at high risk for X be identified on clinical grounds?
3. Are there accurate (i.e., sensitive and specific) screening tests available?
4. Are treatments available that make a difference in intermediate outcomes when the disease is caught early, or detected by screening?
5. Are treatments available that make a difference in morbidity or mortality when the disease is caught early, or detected by screening?
6. How strong is the association between the intermediate outcomes and patient health outcomes?
7. What are the harms of the screening test?
8. What are the harms of the treatment?

When evaluating the chain of evidence, both certainty and magnitude of evidence for each key question is assessed to address the multiple opportunities for bias.

Third, select appropriate resources and conduct a literature search of each of the key questions that discuss comparisons of interventions and strategies used to examine outcomes of interest. Appraise the evidence for its validity and applicability. To begin a search, large databases such as PubMed or the Cochrane Library will access primary sources. Secondary sources such as the American College of Physicians (ACP) Journal Club, Essential Evidence Plus, and Clinical Evidence, provide assessments of the original study (see Chapter 1). In analyzing the results, consider the following terms:

- *Relative risk (RR)* is the ratio of risk in the experimental group compared to the risk in the control group.

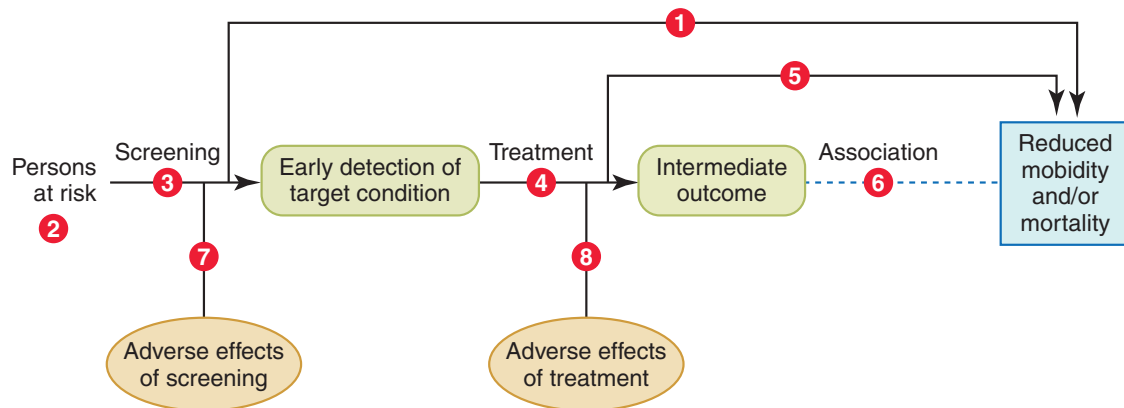


FIGURE 2-1 Template of an analytic framework. See text discussion for the key questions that correspond to the numbers in the template. (From U.S. Preventative Services Task Force: Methods and processes: Procedure manual, Figure 3, n.d. Retrieved from www.uspreventiveservicestaskforce.org/uspstf08/methods/procf3.htm.)

- *Clinical versus statistical significance*—can be a matter of judgment and often depends on the magnitude of the effect being studied. “Is the difference between groups large enough to be worth achieving?”
- *Odds ratio (OR)* is the odds of previous exposure in a case divided by the odds of exposure in a control patient. For example: an OR of 3.0 means that the control cases were three times more likely to have been exposed than were treatment patients.
- *Confidence intervals (CI)* are a measure of the precision of results. Wider CIs indicate lower precision. For example: “36 (CI 27 to 51)” indicates that if the trial was repeated a hundred times 95% of the time the values would fall between 27 and 51.

Finally, apply this knowledge to patients and their preferences. It is important to assess whether the population from which the evidence is gathered matches that of a patient. For example, population-based mammography screening guidelines would not be applied to a woman with a history of breast cancer.

SOURCES OF EVIDENCE

A hierarchy of evidence refers to study designs that allow for less bias or systematic error and may lead to a wrong conclusion. Randomized controlled clinical trials (RCTs) can provide sound evidence of cause and effect, and control for bias. Some limitations of the RCT are threats to the representativeness of the study population and consistency of implementation of the intervention. Meta-analysis examines a number of valid studies on a topic and mathematically combines the results to report them as if they

were one large study. Expert opinion is evidence based on clinical experience, collective experience, and knowledge of professional organizations, such as the American Heart Association or the American College of Obstetrics and Gynecology. Case reports, cohort studies, and qualitative research provide less robust evidence of cause and effect, and have less reliability and validity than higher levels of evidence.

LEVELS OF SCREENING

The three levels of prevention are primary, secondary, and tertiary. Primary prevention involves activities directed at improving general well-being, while also providing specific protection for selected diseases. Interventions can include screening, counseling, or preventive medicines, such as immunizations or dental sealants. Counseling about behavioral risks, such as using seatbelts or bicycle helmets, can reduce injury and death. A common model used to guide behavioral counseling is the 5 *As* model: **A**sk about the behavior; **A**dvice about health risks and benefits of change; **A**gree to set a goal; **A**ssist with identifying and overcoming barriers; **A**rrange for follow up.

The goal of secondary prevention is to identify and detect disease in its earliest stages, before symptoms appear. Screening interventions can identify elevated blood pressure or risk of diabetes with a hemoglobin A1C measurement. With early detection and diagnosis, it may be possible to cure a disease, slow its progression, prevent or minimize complications, and limit disability.

Tertiary prevention programs aim to improve the quality of life for people with various diseases by limiting complications and disabilities, reducing the

severity and progression of disease, and providing rehabilitation (therapy to restore functionality and self-sufficiency). Tertiary prevention can occur over a long period of time, such as optimizing treatment for chronic conditions such as diabetes and hypertension. Unlike primary and secondary prevention, tertiary prevention involves actual treatment for the disease.

POPULATION VERSUS TARGETED SCREENING

Population screening includes all members of a particular population, for example all newborns are screened for congenital hypothyroidism at birth. Targeted screening is more selective and focuses on a population at risk. An example is sexually transmitted infection (STI) screening done in sexually active adolescents and young adults in a specific age group (e.g., those 24 years and younger who have the highest rates of infection) and who have increased risk factors for STI such as new or multiple sexual partners, inconsistent condom use, and sex work.

ETHICAL GUIDELINES FOR SCREENING

Not all diseases or conditions are appropriate for screening. The purposes of screening must be ethically acceptable, information must be used for appropriate purposes, tests must be of high quality, individuals should know what is taking place and informed of their results, counseling must be available to interpret results, and results must be kept confidential. Additionally, genomic medicine has created a new urgency in recognition and application of screening guidelines to assess the value of population screening for genetic susceptibility to diseases and conditions.

Guidelines for determining if a disease or condition warrants screening include the following:

Is the condition significant in the community?

The condition must have a significant impact on the quality or quantity of life, must be measured using morbidity and mortality data, and must be measured by the quality of life. The incidence, or the number of new or undiagnosed cases, of the condition must be sufficient to justify the cost of screening.

Can the condition be screened?

Tests that are acceptable to patients must be available at a reasonable cost to detect the condition in its

asymptomatic period. Measures used to determine acceptability of tests include sensitivity (ability to provide a true positive) and specificity (ability to provide a true negative), as well as measures of reliability (reproducibility) and validity (does it measure what you think it measures?). Other considerations include potential harms of screening, such as labeling and stigma, or morbidity associated with the screening test, as well as patient preference.

Should the condition be screened?

Before screening can be recommended, acceptable treatments must be available. Contextual variables need to be considered such as ethnicity and cultural beliefs and practices, socioeconomic status, and geographic location. The condition must have an asymptomatic period to be considered for screening, and a period in which detection and treatment significantly improve health outcomes compared to a diagnosis obtained based on symptoms.

United States Preventive Services Task Force

The United States Preventive Services Task Force (USPSTF) was established in 1984 as an independent group of experts in prevention and evidence-based medicine. The work of the task force is to make recommendations about clinical preventive services such as screenings, counseling, and preventive medications.

The USPSTF works with Evidence-Based Practice Centers (EPCs) that conduct in-depth systematic reviews of the available evidence, and develop an analytic framework or research plan that includes a set of key questions and outcomes of interest that the review must answer (see [Figure 2-1](#)). After deliberation with input and comments from the public and other experts, a recommendation is reached by calculating the balance between the certainty and magnitude of the net benefit ([Table 2-1](#)) and is then assigned a grade ([Table 2-2](#)). [Box 2-1](#) contains the six questions posed when evaluating evidence. The USPSTF recommendations are considered the gold standard for clinical preventive services.

Electronic Preventive Services Selector (ePSS) is a resource that practitioners can use to electronically access USPSTF recommendations. It is designed to assist primary care practitioners in determining appropriate clinical preventative services for their patients (see <http://epss.ahrq.gov/PDA/index.jsp>).

Table 2-1 Matrix for Arriving at a Grade Recommendation

CERTAINTY OF NET BENEFIT	MAGNITUDE OF NET BENEFIT			
	SUBSTANTIAL	MODERATE	SMALL	ZERO/NEGATIVE
High	A	B	C	D
Moderate	B	B	C	D
Low	Insufficient			

From U.S. Preventive Services Task Force: Procedure manual, n.d. Retrieved from www.uspreventiveservicestaskforce.org.

Table 2-2 What the USPSTF Grades Mean and Suggestions for Practice

GRADE	GRADE DEFINITIONS	SUGGESTIONS FOR PRACTICE
A	The USPSTF recommends the service. There is high certainty that the net benefit is substantial.	Offer or provide this service.
B	The USPSTF recommends the service. There is high certainty that the net benefit is moderate or there is moderate certainty that the net benefit is moderate to substantial.	Offer or provide this service.
C	The USPSTF recommends selectively offering (or providing) this service to individual patients based on professional judgment and patient preferences. There is at least moderate certainty that the net benefit is small.	Offer or provide this service for selected patients depending on individual circumstances.
D	The USPSTF recommends against the service. There is moderate or high certainty that the service has no net benefit or that the harms outweigh the benefits.	Discourage the use of this service.
I Statement	The USPSTF concludes that the current evidence is insufficient to assess the balance of benefits and harms of the service. Evidence is lacking, of poor quality or conflicting, and the balance of benefits and harms cannot be determined.	Read “Clinical Considerations” section of USPSTF Recommendation Statement. If offered, patients should understand the uncertainty about the balance of benefits and harms.

From U.S. Preventive Services Task Force: Grade definitions after July, 2012. Retrieved from www.uspreventiveservicestaskforce.org/uspstf/grades.htm#post.

Current Screening Environment

The Affordable Care Act (ACA) provides coverage for annual wellness visits, which includes the creation of a personalized prevention plan; an individualized health risk assessment, including medical and family history; evaluation of current providers and medications; physical and cognitive assessment; and review of a screening schedule. The ACA provides mandatory coverage of evidence-based practices in prevention services (grade A and B recommendations), for such interventions as smoking cessation, weight control, and the promotion of healthy lifestyles.

Box 2-1 Questions to Consider When Evaluating Evidence

1. Do the studies have the appropriate research design to answer the key question(s)?
2. To what extent are the existing studies of high quality?
3. To what extent are the results of the studies generalizable to the general U.S. primary care population and situation?
4. How many studies have been conducted that address the key question(s)?
5. How consistent are the results of the studies?
6. Are there additional factors that assist with drawing conclusions?