

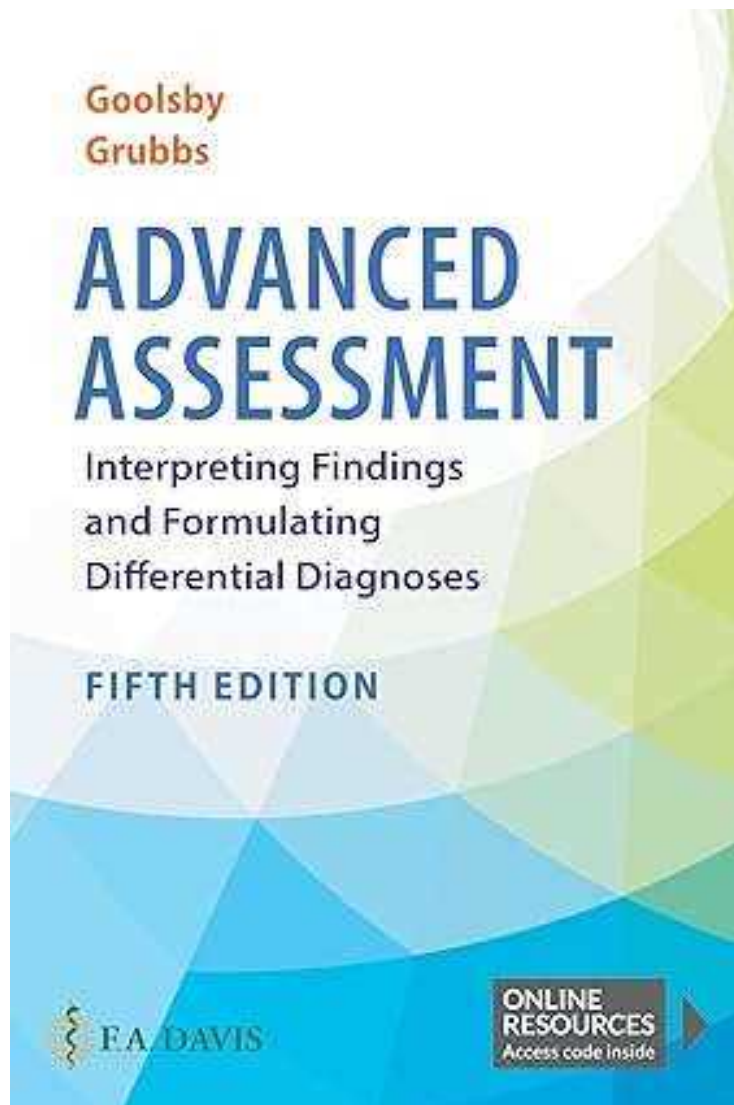
TEST BANK

ADVANCED ASSESSMENT

Interpreting Findings and Formulating
Differential Diagnoses

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5th Edition



Chapter 1. Assessment and Clinical Decision-Making: Overview

Multiple Choice

Identify the choice that best completes the statement or answers the question.

- _____ 1. Which type of clinical decision-making is most reliable?
 - A. Intuitive
 - B. Analytical
 - C. Experiential
 - D. Augenblick

- _____ 2. Which of the following is false? To obtain adequate history, health-care providers must be:
 - A. Methodical and systematic
 - B. Attentive to the patient's verbal and nonverbal language
 - C. Able to accurately interpret the patient's responses
 - D. Adept at reading into the patient's statements

- _____ 3. Essential parts of a health history include all of the following except:
 - A. Chief complaint
 - B. History of the present illness
 - C. Current vital signs
 - D. All of the above are essential history components

- _____ 4. Which of the following is false? While performing the physical examination, the examiner must be able to:
 - A. Differentiate between normal and abnormal findings
 - B. Recall knowledge of a range of conditions and their associated signs and symptoms
 - C. Recognize how certain conditions affect the response to other conditions
 - D. Foresee unpredictable findings

- _____ 5. The following is the least reliable source of information for diagnostic statistics:
 - A. Evidence-based investigations
 - B. Primary reports of research
 - C. Estimation based on a provider's experience
 - D. Published meta-analyses

- _____ 6. The following can be used to assist in sound clinical decision-making:
 - A. Algorithm published in a peer-reviewed journal article
 - B. Clinical practice guidelines
 - C. Evidence-based research
 - D. All of the above

- _____ 7. If a diagnostic study has high sensitivity, this indicates a:
 - A. High percentage of persons with the given condition will have an abnormal result
 - B. Low percentage of persons with the given condition will have an abnormal result
 - C. Low likelihood of normal result in persons without a given condition
 - D. None of the above

- _____ 8. If a diagnostic study has high specificity, this indicates a:
 - A. Low percentage of healthy individuals will show a normal result
 - B. High percentage of healthy individuals will show a normal result
 - C. High percentage of individuals with a disorder will show a normal result
 - D. Low percentage of individuals with a disorder will show an abnormal result

- _____ 9. A likelihood ratio above 1 indicates that a diagnostic test showing a:
 - A. Positive result is strongly associated with the disease
 - B. Negative result is strongly associated with absence of the disease
 - C. Positive result is weakly associated with the disease
 - D. Negative result is weakly associated with absence of the disease

- _____ 10. Which of the following clinical reasoning tools is defined as evidence-based resource based on mathematical modeling to express the likelihood of a condition in select situations, settings, and/or patients?
 - A. Clinical practice guideline
 - B. Clinical decision rule
 - C. Clinical algorithm
 - D. Clinical recommendation

Answer Section

MULTIPLE CHOICE

1. ANS: B

Croskerry (2009) describes two major types of clinical diagnostic decision-making: intuitive and analytical. Intuitive decision-making (similar to Augenblink decision-making) is based on the experience and intuition of the clinician and is less reliable and paired with fairly common errors. In contrast, analytical decision-making is based on careful consideration and has greater reliability with rare errors.

PTS: 1

2. ANS: D

To obtain adequate history, providers must be well organized, attentive to the patient's verbal and nonverbal language, and able to accurately interpret the patient's responses to questions. Rather than reading into the patient's statements, they clarify any areas of uncertainty.

PTS: 1

3. ANS: C

Vital signs are part of the physical examination portion of patient assessment, not part of the health history.

PTS: 1

4. ANS: D

While performing the physical examination, the examiner must be able to differentiate between normal and abnormal findings, recall knowledge of a range of conditions, including their associated signs and symptoms, recognize how certain conditions affect the response to other conditions, and distinguish the relevance of varied abnormal findings.

PTS: 1

5. ANS: C

Sources for diagnostic statistics include textbooks, primary reports of research, and published meta-analyses. Another source of statistics, the one that has been most widely used and available for application to the reasoning process, is the estimation based on a provider's experience, although these are rarely accurate. Over the past decade, the availability of evidence on which to base clinical reasoning is improving, and there is an increasing expectation that clinical reasoning be based on scientific evidence. Evidence-based statistics are also increasingly being used to develop resources to facilitate clinical decision-making.

PTS: 1

6. ANS: D

To assist in clinical decision-making, a number of evidence-based resources have been developed to assist the clinician. Resources, such as algorithms and clinical practice guidelines, assist in clinical reasoning when properly applied.

PTS: 1

7. ANS: A

The sensitivity of a diagnostic study is the percentage of individuals with the target condition who show an abnormal, or positive, result. A high sensitivity indicates that a greater percentage of persons with the given condition will have an abnormal result.

PTS: 1

8. ANS: B

The specificity of a diagnostic study is the percentage of normal, healthy individuals who have a normal result. The greater the specificity, the greater the percentage of individuals who will have negative, or normal, results if they do not have the target condition.

PTS: 1

9. ANS: A

The likelihood ratio is the probability that a positive test result will be associated with a person who has the target condition and a negative result will be associated with a healthy person. A likelihood ratio above 1 indicates that a positive result is associated with the disease; a likelihood ratio less than 1 indicates that a negative result is associated with an absence of the disease.

PTS: 1

10. ANS: B

Clinical decision (or prediction) rules provide another support for clinical reasoning. Clinical decision rules are evidence-based resources that provide probabilistic statements regarding the likelihood that a condition exists if certain variables are met with regard to the prognosis of patients with specific findings. Decision rules use mathematical models and are specific to certain situations, settings, and/or patient characteristics.

PTS: 1

Chapter 2. An Overview of Genetic Assessment

Multiple Choice

Identify the choice that best completes the statement or answers the question.

- _____ 1. The first step in the genomic assessment of a patient is obtaining information regarding:

A.	Family history
B.	Environmental exposures
C.	Lifestyle and behaviors
D.	Current medications

- _____ 2. An affected individual who manifests symptoms of a particular condition through whom a family with a genetic disorder is ascertained is called a(n):

A.	Consultand
B.	Consulband
C.	Index patient
D.	Proband

- _____ 3. An autosomal dominant disorder involves the:

A.	X chromosome
B.	Y chromosome
C.	Mitochondrial DNA
D.	Non-sex chromosomes

- _____ 4. To illustrate a union between two second cousin family members in a pedigree, draw:

A.	Arrows pointing to the male and female
B.	Brackets around the male and female
C.	Double horizontal lines between the male and female
D.	Circles around the male and female

- _____ 5. To illustrate two family members in an adoptive relationship in a pedigree:

A.	Arrows are drawn pointing to the male and female
B.	Brackets are drawn around the male and female
C.	Double horizontal lines are drawn between the male and female

D.	Circles are drawn around the male and female
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6. When analyzing the pedigree for autosomal dominant disorders, it is common to see:

A.	Several generations of affected members
B.	Many consanguineous relationships
C.	More members of the maternal lineage affected than paternal
D.	More members of the paternal lineage affected than maternal

7. In autosomal recessive (AR) disorders, individuals need:

A.	Only one mutated gene on the sex chromosomes to acquire the disease
B.	Only one mutated gene to acquire the disease
C.	Two mutated genes to acquire the disease
D.	Two mutated genes to become carriers

8. In autosomal recessive disorders, carriers have:

A.	Two mutated genes; one from each parent that cause disease
B.	A mutation on a sex chromosome that causes a disease
C.	A single gene mutation that causes the disease
D.	One copy of a gene mutation but not the disease

9. With an autosomal recessive disorder, it is important that parents understand that if they both carry a mutation, the following are the risks to each of their offspring (each pregnancy):

A.	50% chance that offspring will carry the disease
B.	10% chance of offspring affected by disease
C.	25% chance children will carry the disease
D.	10% chance children will be disease free

_____ 10. A woman with an X-linked dominant disorder will:

A.	Not be affected by the disorder herself
B.	Transmit the disorder to 50 % of her offspring (male or female)
C.	Not transmit the disorder to her daughters
D.	Transmit the disorder to only her daughters

_____ 11. In creating your female patient's pedigree, you note that she and both of her sisters were affected by the same genetic disorder. Although neither of her parents had indications of the disorder, her paternal grandmother and her paternal grandmother's two sisters were affected by the same condition. This pattern suggests:

A.	Autosomal dominant disorder
B.	Chromosomal disorder
C.	Mitochondrial DNA disorder
D.	X-linked dominant disorder

_____ 12. A woman affected with an X-linked recessive disorder:

A.	Has one X chromosome affected by the mutation
B.	Will transmit the disorder to all of her children
C.	Will transmit the disorder to all of her sons
D.	Will not transmit the mutation to any of her daughters

_____ 13. Which of the following are found in an individual with aneuploidy?

A.	An abnormal number of chromosomes
B.	An X-linked disorder
C.	Select cells containing abnormal-appearing chromosomes
D.	An autosomal recessive disorder

_____ 14. The pedigree of a family with a mitochondrial DNA disorder is unique in that:

A.	None of the female offspring will have the disease
B.	All offspring from an affected female will have disease
C.	None of the offspring of an

	affected female will have the disease
D.	All the offspring from an affected male will have disease

____ 15. Which population is at highest risk for the occurrence of aneuploidy in offspring?

A.	Mothers younger than 18
B.	Fathers younger than 18
C.	Mothers over age 35
D.	Fathers over age 35

____ 16. Approximately what percentage of cancers is due to a single-gene mutation?

A.	50% to 70%
B.	30% to 40%
C.	20% to 25%
D.	5% to 10%

____ 17. According to the Genetic Information Nondiscrimination Act (GINA):

A.	NPs should keep all genetic information of patients confidential
B.	NPs must obtain informed consent prior to genetic testing of all patients
C.	Employers cannot inquire about an employee's genetic information
D.	All of the above

____ 18. The leading causes of death in the United States are due to:

A.	Multifactorial inheritance
B.	Single gene mutations
C.	X-linked disorders
D.	Aneuploidy

____ 19. Which of the following would be considered a “red flag” that requires more investigation in a patient assessment?

A.	Colon cancer in family member at age 70
B.	Breast cancer in family member at age 75
C.	Myocardial infarction in family member at age 35

D.	All of the above
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- ____ 20. When patients express variable forms of the same hereditary disorder, this is due to:

A.	Penetrance
B.	Aneuploidy
C.	De novo mutation
D.	Sporadic inheritance

- ____ 21. Your 2-year-old patient shows facial features, such as epicanthal folds, up-slanted palpebral fissures, single transverse palmar crease, and a low nasal bridge. These are referred to as:

A.	Variable expressivity related to inherited disease
B.	Dysmorphic features related to genetic disease
C.	De novo mutations of genetic disease
D.	Different penetrant signs of genetic disease

- ____ 22. In order to provide a comprehensive genetic history of a patient, the NP should:

A.	Ask patients to complete a family history worksheet
B.	Seek out pathology reports related to the patient's disorder
C.	Interview family members regarding genetic disorders
D.	All of the above

Chapter 2. An Overview of Genetic Assessment

Answer Section

MULTIPLE CHOICE

1. ANS: A

A critical first step in genomic assessment, including assessment of risk, is the use of family history. Family history is considered the first genetic screen (Berry & Shooner 2004) and is a critical component of care because it reflects shared genetic susceptibilities, shared environment, and common behaviors (Yoon, Scheuner, & Khoury 2003).

PTS: 1

2. ANS: D

A proband is defined as the affected individual who manifests symptoms of a particular condition through whom a family with a genetic disorder is ascertained (Pagon et al. 1993-2013). The proband is the affected individual that brings the family to medical attention.

PTS: 1

3. ANS: D

Autosomal dominant (AD) inheritance is a result of a gene mutation in one of the 22 autosomes.

PTS: 1

4. ANS: C

A consanguineous family is related by descent from a common ancestry and is defined as a “union between two individuals who are related as second cousins or closer” (Hamamy 2012). Consanguinity, if present in the family history, is portrayed using two horizontal lines to establish the relationship between the male and female partners.

PTS: 1

5. ANS: B

For adopted members of the family, use brackets as the appropriate standardized pedigree symbol ([e.g., brackets]).

PTS: 1

6. ANS: A

Pedigrees associated with autosomal dominant (AD) disorders typically reveal multiple affected family members with the disease or syndrome. When analyzing the pedigree for AD disorders or syndromes, it is common to see a “vertical” pattern denoting several generations of affected members.

PTS: 1

7. ANS: C

In autosomal recessive (AR) disorders, the offspring inherits the condition by receiving one copy of the gene mutation from each of the parents. Autosomal recessive disorders must be inherited through both parents (Nussbaum et al. 2007). Individuals who have an AR disorder have two mutated genes, one on each locus of the chromosome. Parents of an affected person are called carriers because each carries one copy of the mutation on one chromosome and a normal gene on the other chromosome. Carriers typically are not affected by the disease.

PTS: 1

8. ANS: D

Individuals who have an AR disorder have two mutated genes, one on each allele of the chromosome. Parents of an affected person are called carriers because each parent carries one copy of the mutation on one chromosome and a normal gene on the other chromosome. Carriers typically are not affected by the disease. In pedigrees with an AR inheritance patterns, males and females will be equally affected because the gene mutation is on an autosome.

PTS: 1

9. ANS: A

It is important that parents understand that if they both carry a mutation, the risk to each of their offspring (each pregnancy) is an independent event: 25% disease free, 25% affected, and 50% carrier.

PTS: 1

10. ANS: B

Everyone born with an X-linked dominant disorder will be affected with the disease. Transmission of the disorder to the next generation varies by gender, however. A woman will transmit the mutation to 50% of all her offspring (male or female).

PTS: 1

11. ANS: D

A man with an X-linked dominant disorder will transmit the mutation to 100% of his daughters (they receive his X chromosome) and none of his sons (they receive his Y chromosome). The pedigree of a family with an X-linked dominant disorder would reveal all the daughters and none of the sons affected with the disorder if the father has an X-linked disorder.

PTS: 1

12. ANS: C

An X-linked recessive disorder means that in a woman, both X chromosomes must have the mutation if she is to be affected. Because males have only one copy of the X chromosome, they will be affected if their X chromosome carries the mutation.

PTS: 1

13. ANS: A

An individual with an abnormal number of chromosomes has a condition called aneuploidy, which is frequently associated with mental problems or physical problems or both (Jorde, Carey, & Bamshad 2010; Nussbaum et al. 2007).

PTS: 1

14. ANS: B

Mitochondrial DNA is inherited from the ovum and, therefore, from the mother. The pedigree of a family with a mitochondrial DNA disorder is unique in that all offspring (regardless of gender) of an affected female will have the disease, and none of the offspring from an affected male will have the disease.

PTS: 1

15. ANS: C

Some individuals or couples have unique identifiable risks that should be discussed prior to conception whenever possible. For example, women who will be 35 years of age or older at delivery (advanced maternal age) are at increased risk for aneuploidy.

PTS: 1

16. ANS: D

The majority of cancers are sporadic or multifactorial due to a combination of genetic and environmental factors; however, approximately 5% to 10% of all cancers are due to a single-gene mutation (Garber & Offit 2005).

PTS: 1

17. ANS: D

On May 21, 2008, President George W. Bush signed the Genetic Information Nondiscrimination Act (GINA) to protect Americans against discrimination based upon their genetic information when it comes to health insurance and employment, paving the way for patient personalized genetic medicine without fear of discrimination (National Human Genome Research Institute 2012).

PTS: 1

18. ANS: A

Most disease-causing conditions are not due to a single-gene disorder but are due to multifactorial inheritance, a result of genomics and environmental or behavioral influences. In fact, the leading causes of mortality in the United States—heart disease, cerebrovascular disease, diabetes, and cancer—are all multifactorial. Most congenital malformation, hypertension, arthritis, asthma, obesity, epilepsy, Alzheimer's, and mental health disorders are also multifactorial.

PTS: 1

19. ANS: C

Early onset cancer syndromes, heart disease, or dementia are red flags that warrant further investigation regarding hereditary disorders.