CGC Self-Study Guide

ABGC
American Board of Genetic Counseling

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November 2025

Introduction

This study guide was created to support you as you prepare for the ABGC Certification Examination (CGC exam). We understand that preparing for the exam is a significant step, and our hope is that this guide helps you feel more informed, confident, and empowered throughout the process.

Inside, you'll find:

- General information about eligibility for the CGC exam.
- Current ABGC resources that are available for test preparation and understanding of the testing process.
- A summary of the test development and item writing process.
- An overview of the cognitive levels utilized to construct exam questions.
- Suggested strategies for answering questions.
- An overview of the examination content domains and subdomains, highlighting knowledge statements with recommended resources.
- Sample test questions for each domain and cognitive level.
- A suggested syndromes and disorders list.
- A personal reflection readiness checklist.

Feel free to move through this guide at your own pace, and take time to explore the linked and referenced materials. While this resource isn't intended to be your only source of preparation, it's designed to be a helpful companion offering insight into the structure of the exam, tips for studying, and support for your preparation process.

As part of ABGC's commitment to maintaining accreditation through the National Commission for Certifying Agencies (NCCA), we follow standards that require a clear separation between certification and education. This means we don't offer direct instruction or exam prep courses. That said, we've created this guide with care to offer meaningful guidance within those guidelines. Our goal is to support your success by being transparent, encouraging, and informative every step of the way.

Who Is Eligible for the CGC Exam?

Eligibility information can also be found in the **Candidate Guide**. Candidates must meet one of the following requirements at the time of application to take the CGC examination. Examinees who graduated from a training program accredited by the Accreditation Council for Genetic Counseling (ACGC) may apply for eligibility to take the exam in Categories 1–3.

Category 1: Active Candidate Status (ACS)-Eligible First Time Examinees

Graduates of a training program accredited by ACGC are eligible for ACS for five (5) years from the date of graduation (e.g., eligibility period would be 6/1/2020 – 6/1/2025, with 6/1/2020 being date of graduation). Candidates applying under Category 1 must request official transcripts directly from their school to be sent to ABGC at transcript@abgc.net. Official transcripts must state the degree and date the degree was conferred. Please be aware that transcript requests often take an educational institution several weeks to process. Request transcripts as soon as possible so they are received by ABGC well before the application deadline. Unofficial transcripts will not be accepted. The ABGC examination cannot be scheduled without a complete application.

Category 2: ACS-Eligible Repeat Examinees

Graduates of a training program accredited by ACGC are eligible for ACS for five (5) years from the date of graduation (e.g., eligibility period would be 6/1/2020 – 6/1/2025, with 6/1/2020 being date of graduation). ACS expires when the applicant successfully completes the examination or exactly five years from the date of graduation, whichever comes first. Candidates are responsible for being aware of when ACS expires and being prepared to apply for examination administration prior to that date.

Category 3: Non-ACS Attempt Examinees

Candidates who do not obtain certification within the first five years of graduation from an accredited program remain eligible to sit for the examination. However, candidates will not be granted ACS for any examination attempt in Category 3.

Category 4: Internationally Trained Genetic Counselors Who May Be Eligible for the Examination

Internationally trained genetic counselors who hold a current registration/certification from an international body and who have earned a master's degree from a genetic counseling program that has achieved accreditation/recognition from an ABGC-recognized accrediting body may be eligible to apply for the examination. Currently, ABGC only recognizes the international programs accredited through the Human Genetic Society of Australasia.

Individuals applying under the International Pathway must submit the following documentation to the ABGC Executive Office for review:

- Documentation from the certifying/registration body indicating when the individual was initially certified or registered, including expiration or renewal date.
- Official transcript of master's degree in genetic counseling listing the date the degree is conferred – transcripts not in English must be accompanied by an English translation; if the academic records do not include official evidence of the award of the degree, applicants must also submit additional documents that verify the degree conferral.

Please note that through the exam application process, candidates following this path will be prompted to provide this documentation.

ABGC Suggested Resources

The ABGC Certify webpage is a good resource for the first step of your preparation. It is highly recommended that all candidates familiarize themselves with the resources listed below prior to preparing for and taking the CGC exam.

- Candidate Guide
- CGC Exam Content Outline (ECO)
- Examination Reference List
- Exam Advice from CGCs - In these articles you will find advice and inspiration from certified genetic counselors who share how they prepared for the exam; the highs, the lows and everything in between.
- Item Writing 101 – How to Write Questions Like Those on the ABGC Board Exam

CGC Practice Exam

The CGC practice exam consists of 100 single response multiple-choice questions that are mapped to the current CGC Exam Content Outline (ECO) and cognitive levels.

- The practice exam will be delivered on the same test driver as the CGC exam, providing candidates with the opportunity to use the same technology and platform that they will encounter when taking the CGC exam.
- While there is no pass/fail on the practice exam, candidates will receive a breakdown of how they performed per subdomain level.
- Candidates will be allowed 120 minutes to answer the 100 questions, which is exactly half of the items and time allowed on the CGC exam. Candidates will also receive an additional 15 minutes to go back and review any items of their choice.

ABGC Test Development Process

Exam test development is essential to create a fair, reliable, and valid exam. It ensures that test content aligns with learning objectives and accurately measures the intended knowledge or skills.

There are multiple steps involved in the ABGC exam development process. Some of these steps happen annually as part of the form development process, while others occur on a different cadence based on industry standards.

CGC Frequently Asked Questions

Developing the CGC Exam

01 **JOB/PRACTICE ANALYSIS**
Conducting a survey to understand current genetic counseling practices

02 **EXAM CONTENT OUTLINE (ECO)**
Creating a framework for the exam based on survey results

03 **ITEM WRITING**
Subject Matter Experts writing exam questions

04 **ITEM REVIEW**
Reviewing and editing questions for accuracy and fairness

05 **EXAM CONSTRUCTION**
Assembling the final exam with selected questions

06 **STANDARD SETTING PROCESS/CUT SCORE**
Determining the passing score for the exam

Strategies For Answering Questions

The purpose of this section is to familiarize examinees with the format of questions, referred to as items, on the examination. It is not intended to illustrate the scope of the examination or the difficulty of individual questions.

The CGC exam consists of 200 multiple-choice items. Of this total, 170 items are scored, and 30 items are pre-test items that are being evaluated for possible inclusion on future examinations. The scored items and pre-test items are not distinguishable to the examinee. All items should be approached as if they are scored items.

Each item will consist of a statement, question, or scenario, followed by four response options. The options will be lettered A, B, C, D. The examinee is required to select the one best answer to the question. Options other than the one best (correct) answer may be partially correct, but there is only one best answer to each item. Every effort has been made to eliminate unclear or "trick" questions.

Strategies for Answering One Best Answer Single Items

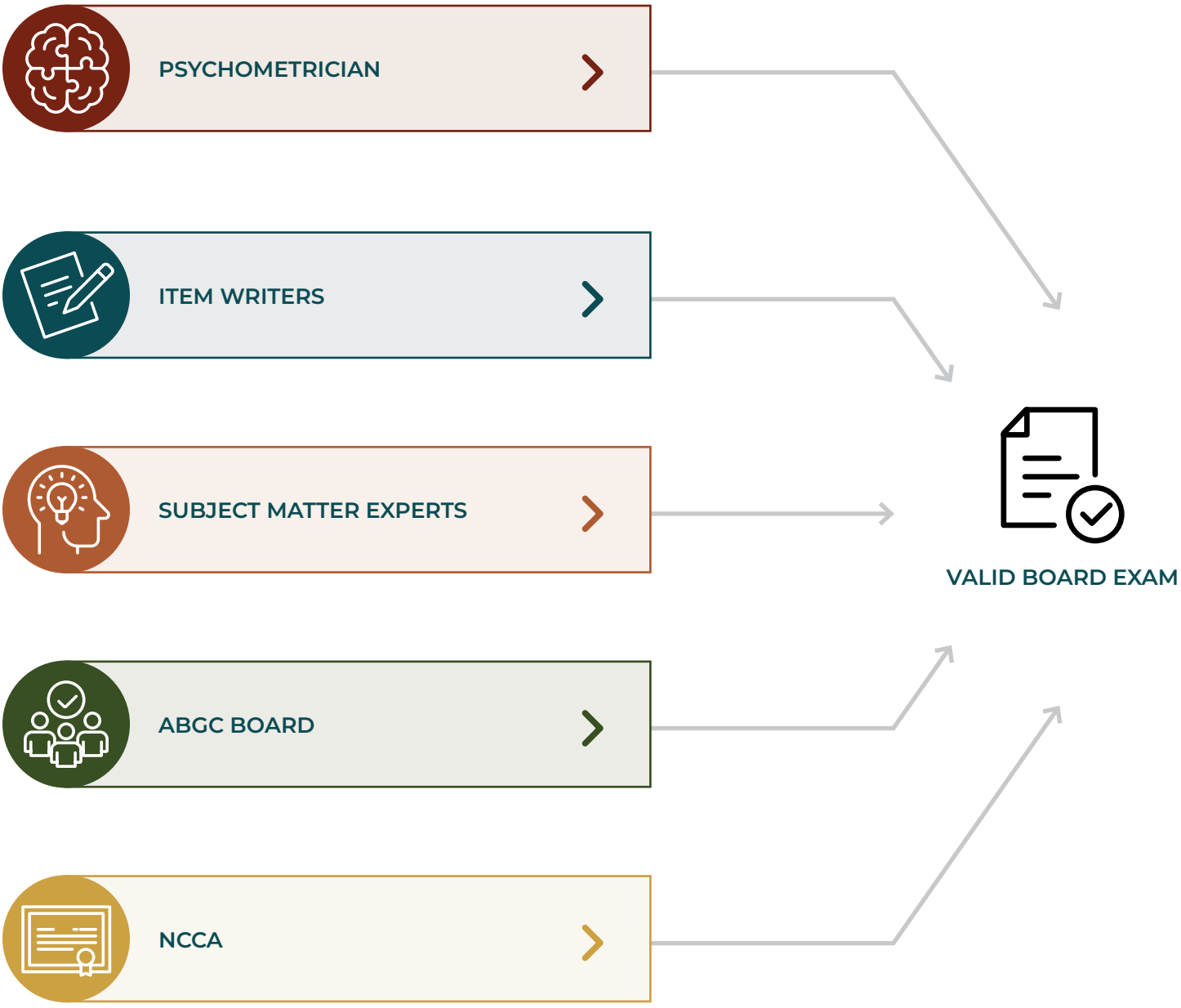
- Read each question or statement carefully. Be certain that you understand what is being asked.
- Read each answer option carefully.
- Look for the best choice and do not be misled by other choices that might be considered possible, but less applicable than, the answer considered correct.
- If an option is only partially correct, tentatively eliminate it as incorrect. Of the remaining options, select the one that you believe to be most correct, and mark that answer.

Exam Advice from CGCs - In these articles you will find advice and inspiration from certified genetic counselors who share how they prepared for the exam; the highs, the lows and everything in between.

ABGC Exam Validity Process

Exam validity ensures that a test accurately measures what it is intended to assess. It is a key to fair and reliable evaluation. Maintaining validity involves collaboration among subject matter experts and testing professionals.

Ensuring Exam Validity



CGC Exam Cognitive Levels

There are three cognitive levels for the CGC exam: **Recall**, **Application**, and **Analysis**. These represent different levels of thinking, understanding, and difficulty. These steps are based on educational taxonomies like **Bloom's Taxonomy** and are used to design and evaluate exam questions. **Recall**, **Application**, and **Analysis** represent increasing levels of difficulty and cognitive demand.

1. **Recall** is the ability to retrieve facts, definitions, formulas, or basic concepts from memory.

- **What it involves:**
  - Memorization
  - Recognition of correct facts
  - Recollection without interpretation
- Recall questions are generally the easiest

2. **Application** involves using learned information in real-life situations or new contexts. It tests whether you can take a known principle or rule and apply it correctly.

- **What it involves:**
  - Solving problems using a known method
  - Applying formulas or procedures to unfamiliar problems
  - Translating theory into practice
- Application questions are more difficult than recall questions

3. **Analysis** is the ability to break down complex information, evaluate it, and draw conclusions or make decisions. It tests critical thinking and judgment.

- **What it involves:**
  - Comparing and contrasting concepts
  - Identifying assumptions, relationships, or flaws
  - Interpreting data and evaluating outcomes
- Analysis questions are the most difficult

CGC Exam Domain, Subdomain, Task and Knowledge Assessed Review

The CGC exam is organized into key domains, each representing a major area of knowledge required for competency in the field. Each domain contains specific tasks, skills, and knowledge areas that candidates are expected to understand.

Domain 1 Overview: Clinical Information, Human Development, and Genetic Conditions

Subdomain	Tasks	Examples of knowledge assessed
1A Clinical Information	1. Medical history, including pregnancy history 2. Teratogens, exposure, and other non-genetic risk factors 3. Differential diagnosis or indication 4. Appropriateness and urgency of referral	• Triage case urgency based on referral indication. • Determine when it is necessary to request additional medical records. • Integrate medical and laboratory findings to develop a differential diagnosis. • Integrate teratogen exposures and non-genetic risk factors, considering timing and duration of exposure. • Utilize family history to develop a differential diagnosis.
1B Human Development	1. Fetal development 2. Development milestones 3. Physical and psychological development	• Demonstrate understanding of the major stages and timelines of human embryological and fetal development. • Determine which fetal anomalies influence the likelihood of a genetic diagnosis. • Differentiate between structural birth defects and their intrinsic or extrinsic causes. • Apply knowledge of pediatric developmental milestones to recognize presence and severity of developmental delays.
1C Genetic Conditions	1. Clinical features 2. Natural history 3. Screening, surveillance, and risk education 4. Treatment options 5. Management options 6. Diagnostic processes including clinical criteria and testing strategy 7. Mode of inheritance 8. Etiology	• Recognize genetic diagnoses based on patterns of clinical features. • Utilize associated clinical features or health risks to identify management or surveillance considerations. • Recognize mode of inheritance based on clinical features, condition name, or a pedigree. • Assess suitability of various specimen types for genetic testing. • Formulate a genetic testing strategy, considering test methodology and clinical urgency. • Recognize treatments available (e.g., gene therapy, enzyme replacement therapy) for various conditions. • Locate management recommendations from published guidelines.

CGC Exam Domain, Subdomain, Task and Knowledge Assessed Review *cont.*

Domain 1 Sample Questions:

QUESTION 1 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Human Development	**Fetal development**	**Recall**

Which of the following isolated ultrasound findings is MOST likely to be associated with fetal aneuploidy?

A. Gastroschisis
B. Duodenal atresia
C. Choroid plexus cyst
D. Holoprosencephaly

QUESTION 2 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Clinical Information	**Differential diagnosis or indication**	**Application**

Which syndrome should be included in the differential diagnosis for a newborn with pulmonary stenosis, coagulopathy and pectus excavatum?

A. Noonan
B. Marfan
C. Loeys-Dietz
D. Williams

QUESTION 3 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Genetic Conditions	**Diagnostic processes including clinical criteria and testing strategy**	**Analysis**

A woman is seen for genetic counseling after her brother was diagnosed with Marfan syndrome. Her past medical history is significant for acute lymphoblastic leukemia at age 12, which was treated with chemotherapy and allogenic bone marrow transplant (BMT) from an unrelated donor. Which of the following is the BEST next step to determine her risk for Marfan syndrome?

A. *FBN1* deletion/duplication using a peripheral blood sample
B. *TGFBR1* sequencing using a peripheral blood sample
C. *FBN1* sequencing using cultured skin fibroblasts
D. *TGFBR1* deletion/duplication using cultured skin fibroblasts

CGC Exam Domain, Subdomain, Task and Knowledge Assessed Review *cont.*

Domain 2 Overview: Risk Assessment and Principles of Human Genetics and Genomics

Subdomain	Tasks	Examples of knowledge assessed
2A Risk Assessment	**1.** Family history, pedigree construction, pedigree analysis **2.** Risk calculations **3.** Residual risk and follow-up medical plan	• Demonstrate appropriate use of pedigree symbols and nomenclature. • Utilize the inclusive pedigree to determine inheritance patterns, differential diagnoses, and risk calculations. • Correctly identify the most appropriate person to test. • Calculate risk using Bayes and Hardy-Weinberg equations. • Calculate recurrence risk in a variety of clinical contexts. • Apply genomics knowledge to determine residual risk.
2B Principles of Human Genetics and Genomics	**1.** Mendelian and non-Mendelian inheritance **2.** Factors that influence phenotype and genotype **3.** Population and quantitative genetics **4.** Human genetic variation and disease susceptibility	• Identify inheritance patterns. • Define and be able to apply genetic concepts such as pleiotropy, variable expressivity, reduced penetrance and anticipation to clinical contexts. • Identify genetic and non-genetic disease modifiers. • Identify factors which influence penetrance. • Apply population genetics concepts to Mendelian disease and multifactorial conditions.

CGC Exam Domain, Subdomain, Task and Knowledge Assessed Review *cont.*

Domain 2 Sample Questions:

QUESTION 1 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Principles of Human Genetics and Genomics	**Human genetic variation and disease susceptibility**	**Recall**

Which term BEST describes when a small group of individuals establishes a new population, carrying only a fraction of the genetic variation from the original population?

A. Genetic drift
B. Founder effect
C. Assortative mating
D. Gene flow

QUESTION 2 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Principles of Human Genetics and Genomics	**Factors that influence phenotype and genotype**	**Application**

Two siblings are seen for genetics evaluation. One sibling has over 50 café-au-lait spots and multiple large neurofibromas requiring surgical removal. The other sibling has severe scoliosis and an optic glioma. The siblings' presentation can BEST be explained by:

A. Variable expressivity
B. Mosaicism
C. Incomplete penetrance
D. Allelic heterogeneity

QUESTION 3 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Risk Assessment	**Risk calculations**	**Analysis**

An unaffected woman receives a negative result on a common cystic fibrosis (CF) variant panel. Assuming the panel detects 90% of pathogenic *CFTR* variants and the carrier frequency is 1/25, what is the chance the woman is a CF carrier?

A. 1/24
B. 1/25
C. 1/241
D. 1/250

CGC Exam Domain, Subdomain, Task and Knowledge Assessed Review *cont.*

Domain 3 Overview: Testing Interpretation, Testing Options, and Reproductive Risk Management

Subdomain	Tasks	Examples of knowledge assessed
3A Testing and Interpretation	**1.** Test methodologies/ technologies and applicable limitations **2.** Variant interpretation, reanalysis, and reclassification **3.** Clinical context **4.** Predictive value of test **5.** Sensitivity and specificity **6.** Analytic validity, clinical validity, and clinical utility **7.** Sample type	• Differentiate between test methodologies and identify their appropriate clinical application based on known benefits, limitations, and clinical context. • Demonstrate understanding of the principles of variant classification, reanalysis, and reclassification and their impact on clinical decision-making and counseling. • Identify situations in which variant reanalysis or reclassification may be appropriate. • Calculate and interpret the predictive value, sensitivity, and specificity of test results considering clinical context. • Apply concepts of analytic validity, clinical validity, and clinical utility to determine the appropriateness of a genetic test. • Select the appropriate sample type based on indication and testing methodology.
3B Testing Options	**1.** Diagnostic **2.** Screening **3.** Predictive/pre-symptomatic **4.** Somatic **5.** Carrier **6.** Research	• Distinguish among types of genetic testing (diagnostic, screening, predictive, carrier, somatic, research, etc.) based on factors such as test purpose, target population, and clinical context. • Recognize the implications of each testing type, including the nature of potential results, psychosocial considerations, and follow-up needs. • Apply knowledge of testing options to clinical contexts.
3C Reproductive Risk Management	**1.** Assisted reproductive technologies (ART) **2.** Family planning and reproductive options **3.** Prenatal screening **4.** Prenatal diagnostic procedures	• Describe the benefits, limitations, and potential risks of prenatal screening and diagnostic options including cell-free DNA (cfDNA) screening, serum AFP screening, chorionic villus sampling (CVS), amniocentesis, and pre-implantation genetic testing (PGT). • Identify the appropriate preconception or prenatal testing option(s) based on indication. • Integrate ultrasound findings and/or other relevant test results to refine reproductive risk assessments or future testing strategy. • Apply knowledge of the clinical context to guide personalized family planning and reproductive options.

CGC Exam Domain, Subdomain, Task and Knowledge Assessed Review *cont.*

Domain 3 Sample Questions:

QUESTION 1 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Testing Options	**Carrier**	**Recall**

A couple presents at 12 weeks gestation to discuss carrier screening. They are both covered by Medicaid and indicate their ancestry is southeast Asian. They state they will continue the pregnancy regardless of the results. What type of screening should you offer first?

A. Expanded carrier screening
B. Nothing, as screening will not be covered by Medicaid
C. Carrier screening based on ethnicity
D. Nothing, as screening will not change medical management

QUESTION 2 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Reproductive Risk Management	**Prenatal diagnostic procedures**	**Application**

What is the negative predictive value of the screening test depicted in the table below?

	Affected	Unaffected
Positive	75	25
Negative	15	65

A. 72%
B. 75%
C. 81%
D. 83%

QUESTION 3 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Testing and Interpretation	**Test methodologies/technologies and applicable limitations**	**Analysis**

A 63-year-old patient presents with a three-year history of ataxia and negative genetic testing through a multigene panel. They report no family history of ataxia, no other medical concerns, and no personal use of tobacco or alcohol. What is the BEST next step to help the patient understand the etiology?

A. Order chromosomal microarray
B. Take a more detailed family history
C. Discuss the unknown etiology of many cases
D. Tell the patient additional testing is not available

CGC Exam Domain, Subdomain, Task and Knowledge Assessed Review *cont.*

Domain 4 Overview: Counseling Skills, Communication, and Education

SUBDOMAIN	TASKS	EXAMPLES OF KNOWLEDGE ASSESSED
4A Counseling Skills	**1.** Contracting **2.** Interviewing techniques **3.** Psychosocial assessment and follow-up **4.** Family dynamics **5.** Client personal identity **6.** Cultural humility **7.** Counseling theories and models **8.** Defense mechanisms and coping strategies **9.** Decision-making styles **10.** Self-awareness and client-counselor dynamics	• Utilize various interviewing techniques to assess client primary concerns and expectations. • Understand how client personal values and beliefs impact comprehension and decision-making. • Understand how culture, class, and language factors can impact effective cross-cultural counseling. • Recognize how family values, structure, and experiences may play a role in client risk perception, coping mechanisms and decision-making. • Describe various client coping styles and defense mechanisms and counselor interventional approaches. • Understand how to respond to client cues with appropriate use of attending, empathy, advanced empathy, and confrontation. • Identify and understand how personal attitudes and beliefs toward cultural differences influence interactions with clients. • Identify strategies for recognizing and managing transference and countertransference.
4B Communication and Patient Education	**1.** Adapting counseling to modes of delivery **2.** Risk communication theories and models **3.** Strategies for working with interpreters **4.** Strategies of results delivery and long-term follow-up **5.** Impact of health literacy **6.** Elements of informed consent **7.** Informed decision making	• Recognize client verbal and non-verbal cues that would indicate misunderstanding of information. • Understand culture-bound and communication style differences among various cultural groups. • Discuss various models of risk communication and how they are best utilized during a genetic counseling session. • Describe best practices using interpreter services. • Adapt counseling approach to various service models such as point-of-care testing, post-test genetic counseling, telemedicine, etc. • Understand how to assess client health literacy and tailor educational approach during a session. • Recognize different strategies for facilitating client decision making through feedback. • Apply the components of informed consent to discussions regarding genetic testing options and outcomes.

CGC Exam Domain, Subdomain, Task and Knowledge Assessed Review *cont.*

Domain 4 Sample Questions:

QUESTION 1 | CLICK HERE FOR THE ANSWER & RATIONALE >		
Subdomain: **Counseling Skills**	Task: **Interviewing techniques**	Cognitive Level: **Recall**
How should a genetic counselor ask questions during a session? **A.** Use "why" questions to allow the patient to explain their perspective. **B.** Combine the use of open and closed questions to focus the response. **C.** Use follow up questions sparingly to not distract from the session goals. **D.** Combine multiple questions together to help the patient understand the end goal.		

QUESTION 2 | CLICK HERE FOR THE ANSWER & RATIONALE >		
Subdomain: **Communication and Patient Education**	Task: **Impact of health literacy**	Cognitive Level: **Application**
During a follow-up session with a 40 year-old patient who has a pathogenic *BRCA1* variant, you learn she has not undergone any of the screening previously recommended. Which of the following would be the BEST next step? **A.** Review the results disclosure letter previously sent. **B.** Discuss the reasons why screening is recommended. **C.** Ask her to describe the types of screening recommended. **D.** Explore her understanding of her personal risk based on the results.		

QUESTION 3 | CLICK HERE FOR THE ANSWER & RATIONALE >		
Subdomain: **Counseling Skills**	Task: **Counseling theories and models**	Cognitive Level: **Analysis**
Which of the following describes the foundation of genetic counseling communication? **A.** Nondirective **B.** Patient-centered **C.** Informational **D.** Empathetic		

CGC Exam Domain, Subdomain, Task and Knowledge Assessed Review *cont.*

Domain 5 Overview: Financial/Reimbursement Issues, Resources and Services for Clients, Legal and Regulatory Requirements, and Professional Frameworks

SUBDOMAIN	TASKS	EXAMPLES OF KNOWLEDGE ASSESSED
5A Financial/ Reimbursement Issues	**1.** Application of insurance policies **2.** Compliance with billing and reimbursement requirements **3.** Letters of medical necessity	• Distinguish between Medicare and Medicaid. • Recognize Medicare coverage policies for hereditary cancer testing. • Demonstrate knowledge of rationale for and effective components of a letter of medical necessity. • Distinguish between relevant numerical identifiers and codes that apply in genetic counseling, including ICD-10, CPT, NPI, Tax ID. • Recognize elements of commercial insurance policies, such as the deductible, out-of-pocket maximum, and coinsurance. • Recognize limitations of prior authorization approval.
5B Resources and Services for Clients	**1.** Financial assistance resources **2.** Appropriateness of resources **3.** Timely delivery of resources	• Consider availability and applicability of financial assistance resources for clients who have financial barriers for genetic testing. • Determine the relevance of a resource for a patient based on their individual circumstance(s). • Determine effective characteristics of a patient financial resource.
5C Legal and Regulatory Requirements	**1.** Genetic non-discrimination legislation **2.** Patient access to information **3.** Privacy and confidentiality regulations **4.** Medical documentation **5.** Standards and regulations for human-subjects research **6.** Genomic data security	• Adhere to medical and legal requirements of clinical documentation. • Demonstrate familiarity with protections offered by the Genetic Information Non-discrimination Act (GINA). • Apply knowledge of HIPAA in clinical contexts. • Differentiate between types of research studies. • Assess when a study qualifies as human subjects research. • Comply with IRB protocols, including appropriate roles and scope of practice, in the setting of a research study. • Recognize characteristics of appropriate storage and security of raw genomic data. • Demonstrate familiarity with the Common Rule.
5D Professional Frameworks	**1.** Code of Ethics **2.** Ethical decision making **3.** Evidence-based practice **4.** Conflict of interest **5.** Clinical supervision of students **6.** Scope of practice **7.** Professional and public education	• Apply ethical principles of the Belmont Report to a given clinical context. • Demonstrate knowledge of models of supervision and tailor supervision approaches to the needs of a student. • Recognize conflicts of interest. • Recognize the scope of practice of a genetic counselor. • Apply knowledge of the NSGC code of ethics to different situations that may arise in genetic counseling practice. • Consider appropriate educational tools and formats for a given target audience. • Recognize and apply evidence-based research in clinical practice.

CGC Exam Domain, Subdomain, Task and Knowledge Assessed Review *cont.*

Domain 5 Sample Questions:

QUESTION 1 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Professional Frameworks	**Code of Ethics**	**Recall**

Which of the following is MOST correct regarding the development of professional codes of ethics?

A. They are created to protect the public from misinformation.
B. They provide a specific ethical framework for decision making.
C. They determine common values and ethical principles for an organization.
D. They create boundaries for professional work to ensure scope is maintained.

QUESTION 2 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Financial/Reimbursement Issues	**Application of insurance policies**	**Application**

Which of the following is MOST accurate about commercial insurance coverage for genetic counseling services?

A. It is not covered until CMS recognizes genetic counselors.
B. It is covered for fully insured plans.
C. It is not covered for self-funded plans.
D. It is covered if insurers and employer groups approve.

QUESTION 3 | CLICK HERE FOR THE ANSWER & RATIONALE >

Subdomain:	Task:	Cognitive Level:
Legal and Regulatory Requirements	**Genomic data security**	**Analysis**

Which of the following is TRUE regarding privacy of stored genomic data?

A. Regulations can vary between and within countries.
B. GINA prevents genomic data from being sold.
C. HIPAA prevents genomic data sharing by DTC companies.
D. Third-party interpretation tools allow users to easily opt-out of data sharing.

CGC Exam Syndromes & Disorders List

The following is a list of syndromes and disorders that may be referenced in the exam. This list is not all-inclusive, and additional conditions may be tested over time. It is intended to serve as a general guide for study and review purposes.

- [ ] **Alagille syndrome**

- [ ] **Ataxias, including:**
  - spinocerebellar ataxias
  - Friedreich ataxia

- [ ] **Bardet-Biedl syndrome**

- [ ] **Bleeding and thrombophilia disorders, including:**
  - factor V Leiden
  - hemophilia

- [ ] **CADASIL**

- [ ] **Canavan disease**

- [ ] **Chromosomal aneuploidies, including:**
  - triploidy
  - trisomies 13, 18, 21
  - X and Y chromosome variations, such as Klinefelter syndrome, Turner syndrome

- [ ] **Chromosomal microdeletion/ microduplication syndromes, including:**
  - 22q11.2 deletion syndrome
  - Angelman syndrome
  - Cri-du-chat syndrome
  - Prader-Willi syndrome
  - Smith-Magenis syndrome
  - Williams syndrome
  - Wolf-Hirschhorn syndrome

- [ ] **Congenital adrenal hyperplasia**

- [ ] **Congenital heart disease, cardiac arrhythmias, and cardiomyopathies, including:**
  - dilated cardiomyopathy
  - hypertrophic cardiomyopathy
  - arrhythmogenic right ventricular cardiomyopathy
  - restrictive cardiomyopathy
  - catecholaminergic polymorphic ventricular tachycardia
  - long QT syndrome
  - sudden cardiac arrest

- [ ] **Connective tissue disorders, including:**
  - Marfan syndrome
  - Loeys-Dietz syndrome
  - hypermobile, classic, and vascular Ehlers-Danlos syndrome

- [ ] **Craniofacial conditions, including:**
  - Treacher Collins syndrome
  - craniosynostosis syndromes
  - holoprosencephaly
  - Pierre Robin sequence
  - cleft lip and palate

- [ ] **Cystic fibrosis**

- [ ] **Epilepsy syndromes, including those caused by variants in:**
  - *DEPDC5*
  - *KCNQ2*
  - *SCN1A*

- [ ] **Familial hypercholesterolemia**

- [ ] **Fanconi anemia**

- [ ] **Fragile X syndrome**

- [ ] **Hearing loss (non-syndromic and syndromic forms), including:**
  - branchio-oto-renal syndrome
  - Stickler syndrome
  - Usher syndrome
  - Pendred syndrome

CGC Exam Syndromes & Disorders List *cont.*

- [ ] **Hereditary cancer syndromes, including those caused by variants in:**
  - *APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CDKN2A, CHEK2, EPCAM, MEN1, MLH1, MSH2, MSH6, MUTYH, PALB2, PTEN, PMS2, RET, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53, VHL*
- [ ] **Hemochromatosis**
- [ ] **Hemoglobinopathies**
- [ ] **L1CAM-associated hydrocephalus**
- [ ] **Meckel-Gruber syndrome**
- [ ] **Mitochondrial disorders, including:**
  - Leigh syndrome
  - Leber hereditary optic neuropathy
  - mitochondrial deletion syndromes
  - MELAS syndrome
- [ ] **Malignant hyperthermia**
- [ ] **Metabolic disorders, including:**
  - fatty acid oxidation disorders such as VLCADD and MCADD
  - adrenoleukodystrophy
  - amino acid disorders such as tyrosinemia, glutaric aciduria, maple syrup urine disease, ornithine transcarbamylase deficiency, phenylketonuria, and galactosemia
  - glycogen storage diseases
  - lysosomal storage diseases such as Fabry, Gaucher, Krabbe, Pompe, Tay-Sachs, and mucopolysaccharidoses
- [ ] **McCune-Albright syndrome**
- [ ] ***MTHFR* gene**
- [ ] **Neurodegenerative diseases, including:**
  - dementia
  - ALS
  - Huntington's disease
  - Alzheimer disease
  - frontotemporal dementia
- [ ] **Neuromuscular disorders, including**
  - myotonic dystrophy
  - spinal muscular atrophy
  - Charcot-Marie-Tooth
  - dystrophinopathies
- [ ] **Neurocutaneous disorders, including:**
  - tuberous sclerosis
  - neurofibromatosis types 1 and 2
- [ ] **RASopathies**
- [ ] **Osteogenesis imperfecta**
- [ ] **Polycystic kidney disease**
- [ ] **Retinoblastoma**
- [ ] **Rett syndrome**
- [ ] **Skeletal dysplasias, including:**
  - achondroplasia
  - thanatophoric dysplasia
- [ ] **Smith-Lemli-Opitz syndrome**
- [ ] **Transthyretin amyloidosis**
- [ ] **Walker-Warburg syndrome**
- [ ] **Wilson disease**

Personal Reflection Readiness Checklist

Before beginning any certification journey, it's important to take a step back and reflect not just on the material to be studied, but on your overall readiness to commit to the process. This Personal Reflection Readiness Checklist is designed to help you assess your current level of preparedness through honest self-evaluation.

Use this checklist as both a starting point and a guide you can revisit throughout your exam preparation. This checklist can help track your understanding of the exam content, key domains, and required knowledge ensuring you're aligned with your goals every step of the way.

Domain & Subdomain	Tasks	Knowledge Readiness Reflection	
Domain 1. Clinical Information, Human Development, and Genetic Conditions **Subdomain 1A Clinical Information**	**1.** Medical history, including pregnancy history **2.** Teratogens, exposure, and other non-genetic risk factors **3.** Differential diagnosis or indication **4.** Appropriateness and urgency of referral	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●
Domain 1. Clinical Information, Human Development, and Genetic Conditions **Subdomain 1B Human Development**	**1.** Fetal development **2.** Development milestones **3.** Physical and psychological development	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●
Domain 1. Clinical Information, Human Development, and Genetic Conditions **Subdomain 1C Genetic Conditions**	**1.** Clinical features **2.** Natural history **3.** Screening, surveillance, and risk education **4.** Treatment options **5.** Management options **6.** Diagnostic processes including clinical criteria and testing strategy **7.** Mode of inheritance **8.** Etiology	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●

Personal Reflection Readiness Checklist *cont.*

Domain & Subdomain	Tasks	Knowledge Readiness Reflection	
Domain 2. Risk Assessment and Principles of Human Genetics and Genomics **Subdomain 2A Risk Assessment**	1. Family history, pedigree construction, pedigree analysis 2. Risk calculations 3. Residual risk and follow-up medical plan	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●
Domain 2. Risk Assessment and Principles of Human Genetics and Genomics **Subdomain 2B Principles of Human Genetics and Genomics**	1. Mendelian and non-Mendelian inheritance 2. Factors that influence phenotype and genotype 3. Population and quantitative genetics 4. Human genetic variation and disease susceptibility	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●
Domain 3. Testing Interpretation, Testing Options, and Reproductive Risk Management **Subdomain 3A Testing and Interpretation**	1. Test methodologies/technologies and applicable limitations 2. Variant interpretation, reanalysis, and reclassification 3. Clinical context 4. Predictive value of test 5. Sensitivity and specificity 6. Analytic validity, clinical validity, and clinical utility 7. Sample type	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●
Domain 3. Testing Interpretation, Testing Options, and Reproductive Risk Management **Subdomain 3B Testing Options**	1. Diagnostic 2. Screening 3. Predictive/pre-symptomatic 4. Somatic 5. Carrier 6. Research	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●

Personal Reflection Readiness Checklist *cont.*

Domain & Subdomain	Tasks	Knowledge Readiness Reflection	
Domain 3. Testing Interpretation, Testing Options, and Reproductive Risk Management **Subdomain 3C Reproductive Risk Management**	**1.** Assisted reproductive technologies (ART) **2.** Family planning and reproductive options **3.** Prenatal screening **4.** Prenatal diagnostic procedures	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●
Domain 4. Counseling Skills, Communication, and Education **Subdomain 4A Counseling Skills**	**1.** Family history, pedigree construction, pedigree analysis **2.** Risk calculations **3.** Residual risk and follow-up medical plan	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●
Domain 4. Counseling Skills, Communication, and Education **Subdomain 4B Communication and Patient Education**	**1.** Mendelian and non-Mendelian inheritance **2.** Factors that influence phenotype and genotype **3.** Population and quantitative genetics **4.** Human genetic variation and disease susceptibility	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●
Domain 5. Financial/ Reimbursement Issues **Subdomain 5A Financial Reimbursement**	**1.** Application of insurance policies **2.** Compliance with billing and reimbursement requirements **3.** Letters of medical necessity	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●

Personal Reflection Readiness Checklist *cont.*

Domain & Subdomain	Tasks	Knowledge Readiness Reflection	
Domain 5. Financial/ Reimbursement Issues **Subdomain 5B Resources and Services for Clients**	**1.** Financial assistance resources **2.** Appropriateness of resources **3.** Timely delivery of resources	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●

Domain 5. Financial/ Reimbursement Issues **Subdomain 5C Legal and Regulatory Requirements**	**1.** Genetic non-discrimination legislation **2.** Patient access to information **3.** Privacy and confidentiality regulations **4.** Medical documentation **5.** Standards and regulations for human-subjects research **6.** Genomic data security	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●

Domain 5. Financial/ Reimbursement Issues **Subdomain 5D Professional Frameworks**	**1.** Code of Ethics **2.** Ethical decision making **3.** Evidence-based practice **4.** Conflict of interest **5.** Clinical supervision of students **6.** Scope of practice **7.** Professional and public education	*Limited to no knowledge*	●
		Foundational understanding of the subject but still seek guidance	●
		Well-prepared and capable of applying my knowledge	●
		Deep understanding of the knowledge and skills needed	●