



2025 Case Study Reports, and Diagnostic Dilemmas/Challenge Cases (Unknown & Rare Knowns) Abstract Submission Guidelines

All abstract submissions are due by 11:59 p.m. PT on **Friday, November 15, 2024**. Accepted abstracts are required to be presented in person at the 2025 Annual Meeting in Los Angeles, CA.

All abstracts accepted for presentation at the 2025 ACMG Annual Clinical Genetics Meeting must be presented in person. Presenters who are unable to travel to Los Angeles to present their work in person will have the opportunity to designate a co-author to serve as the presenter.

Publication: All accepted abstracts will be published in *Genetics in Medicine Open*.

Submission Requirements:

- Submitters must select the appropriate submission type during submission for their abstract to be considered for acceptance.
- The choice of submission type is final and cannot be changed after the review committee has reached its decision.

Case Study Reports and Diagnostic Dilemmas/Challenge Cases (Unknown & Rare Knowns)

Case Study Reports and Diagnostic Dilemmas: Three or more cases should be considered a scientific abstract. Case Study abstracts are not eligible for platform presentation or for awards.

Overview:

- A case study report abstract provides an overview of the symptoms, diagnosis, treatment, and follow-up of an individual patient, especially for unique or rare cases.
- Diagnostic Dilemmas/Challenge Cases involve presenting rare knowns and unknowns in accredited interactive sessions, offering a platform to share best practices.

Diagnostic Dilemmas/Challenge Cases (Unknown & Rare Knowns) Concurrent Session Options:

- **Laboratory Diagnostic Challenges:** Focuses on constitutional and neoplastic cases in molecular, cytogenomic, and biochemical genetics.
- **Medical Diagnostic Challenges:** Includes adult and cancer cases.
- **Challenge the Experts - Pediatric and Prenatal Diagnostic Dilemmas:** Addresses rare knowns and unknowns.

Submission Options:

1. Case Study Report Poster Presentation Only
2. Diagnostic Dilemmas/Challenge Cases (Unknown & Rare Known) Presentation Only

3. Case Study Report or Diagnostic Dilemmas/Challenge Cases (Unknown & Rare Known) Presentation with Reflex to Poster

- If not accepted for presentation, the abstract may be accepted as a poster, depending on the score.

Abstract Structure:

- **Introduction:** Emphasizes the significance or rarity of the case.
- **Case Presentation:** Details the patient's symptoms, history, and any relevant factors.
- **Diagnostic Workup:** Describes the diagnostic process, tests performed, and any unusual findings.
- **Treatment and Management:** Discusses the care provided, including any experimental treatments.
- **Outcome and Follow-up:** Outlines the patient's response and any long-term follow-up actions.
- **Discussion:** Explores the implications of the case, comparing it with existing literature or similar cases.
- **Conclusion:** Summarizes key insights and suggests areas for future research.

Submission Guidelines:

- Abstracts must be structured and are allotted 600 words.
- Use italic type where necessary; bold and underline formatting are not allowed.
- Text may be in multiple paragraphs.
- Special Greek and mathematical symbols are available in a character map within the submission system.
- Use numerals to indicate numbers, except when beginning sentences.
- Do not include figures, diagrams, images, references, or tables.
- Proofreading is the responsibility of the author.

Important Dates

Tuesday, October 1, 2024	Call for Abstracts and Registration Opens
Friday, November 15, 2024	Abstract Submission Deadline
November 19 – December 6	Abstract Review Process
Wednesday, December 11, 2024	Abstract Acceptance/Rejection Notices emailed
Wednesday, December 18, 2024	Early Bird Registration Deadline
Friday, January 3, 2025	Platform & Diagnostic Dilemmas Presenters complete Participation Forms in Presenter Portal
Friday, February 7, 2025	Abstract Withdrawal Deadline
Friday, February 28, 2025	Final PowerPoint Presentations Uploaded Test Questions Submitted for Accredited

	Sessions ePosters with audio tour uploaded
February 3 – March 7, 2025	Slide Review and Peer Review Process
March 19, 2025	Posters – Embargoed until Wednesday, March 19, at 5:00 pm PT Oral (Platform) Scientific Presentations & Diagnostic Dilemmas – Presentations Embargoed until start of live session
March 18 - 22, 2025	See you in Los Angeles, CA

Topic Categories

ACMG 2025 Abstract Categories

1. ASOs (antisense oligonucleotides)
2. Basic Research
3. Cancer
4. Clinical - Adult
5. Clinical - Adult and Pediatric
6. Clinical - Pediatric
7. Clinical Genetics
8. Enzyme Replacement Therapy
9. Ethical, Legal and Social Issues
10. Gene Therapy
11. General Education
12. Genetic Counseling
13. Genomic Medicine
14. Health Care Inequities and Health Disparities
15. Health Services and Implementation
16. Laboratory Genetics
17. Metabolic Genetics
18. Non-Clinical
19. Other Therapies
20. Population Genetics
21. Prenatal Genetics
22. Public Health Genetics
23. RNA Therapy
24. Substrate Reduction Therapy

ACMG 2025 Abstract Key Topic Categories

1. Biochemical/Metabolic and Therapeutics
2. Cancer genetics and therapeutics
3. Clinical Genetics and Therapeutics
4. Health Services and Implementation
5. Laboratory Genetics and Genomics
6. Prenatal Genetics
7. Research Methods & Resources
8. Ethical, Legal, Social Issues (ELSI), Public Health and Policy

Responsibilities of the Submitter (First Author)/Co-author/Presenting Author

- The **submitting author must be the first author and** is responsible for the following:
 - The first author serves as point of contact for all correspondence regarding the abstract.
 - Ensuring that all authors have read the abstract and agreed to be co-authors. Failure to get approval from all authors will result in rejection of the abstract.
 - Notifying all co-authors of any additions, deletions, and changes to the program, as may be communicated by ACMG.
 - Ensuring that all authors approve of submitting this work for presentation and publication.
 - Abiding by all formatting
- The author(s) agree(s) to materially confine the presentation to information in the abstract, if accepted for presentation. If an author has more than one abstract accepted, each presentation will be materially confined to the information in the abstract selected for the specific session.
- The presenting author will be available to present the abstract if selected for the program. The author(s) will immediately notify ACMG if the first author is not the presenting author.
- The **presenting author** must be one of the co-authors listed on the submitted abstract. The presenting author must be confirmed prior to abstract submission.
- An individual may be first author (i.e., presenting author) of more than one abstract. If the first author should have multiple accepted Posters and has a conflict with the Poster presentation schedule, then it is acceptable to designate one of the co- authors to present one of the Posters.

Authors' Consent and Waiver of Claims

Completion of all required information in the online abstract submission system serves as an agreement. It certifies the ACMG abstract submitter's understanding of the rules for participation contained in the online abstract submission program and affirms that:

- Each abstract author agrees and certifies that they:
 - **have read all the rules and agrees to be bound by them,**
 - is responsible for submission of the abstract in accordance with the rules, and

- waives any and all claims against ACMG and any reviewer arising out of or relating to the abstract submission and review process, including but not limited to peer review and the grading of abstracts.

Abstract Submission Requirements:

- The abstract must address scientific questions, detail clinical observations, and/or contain primary scientific data. We encourage authors to highlight why their abstract is novel or how it provides a unique contribution or perspective.
- Authors assign copyright of the abstract to ACMG upon submission, unless one of the authors is a U.S. Federal employee (in such case, ACMG does not hold copyright). This means that the identical abstract may not be republished or submitted to another meeting.
- All research and studies reported in submitted abstracts that involve human and animal subjects must comply with the guiding principles for experimental procedures found in the [Declaration of Helsinki](#) of the World Medical Association.
- Interim analysis of a prospective randomized clinical trial will be considered only if it is performed as planned in the original protocol and is statistically valid. If your abstract involves interim analysis, use the Interim Analysis of a Clinical Trial section of the abstract form to explain the details of your study. The reviewers will have this information available during their evaluation.
- No revisions can be made after the abstract submission deadline (November 15, 2024).
- The presentation at the annual meeting must reflect the submitted abstract. In particular, the abstract title, authorship, and scientific content of the presentation at the annual meeting must reflect the submitted abstract.
- Abstracts should be written in clear and concise English to ensure that reviewers can focus solely on the scientific merits of the submission. For authors whose primary language is not English, we encourage having abstracts checked for grammar and spelling before submission.
- It is expected that the presenting author will have a sufficient command of English to deliver the presentation and respond to questions. If this is not the case, authors are encouraged to choose a poster presentation as their preference.
- Abstracts can be submitted if:
 - It is unpublished and original
 - Accepted for publication but not yet published
 - Encore presentations are allowed, only if material was previously presented at non-genetics meeting within the last 6 – 12 months.

Case Study Reports and Diagnostic Dilemmas/Challenge Cases (Unknown & Rare Knowns) Abstracts Review and Selection Process

- After the submission deadline, all completed and eligible abstracts will be made available to the ACMG Abstract Reviewers for review and scoring.
- Abstracts will be evaluated and scored based on the clarity and significance of the case, its contribution to understanding rare or challenging clinical scenarios, and its potential to inform or challenge existing clinical guidelines and best practices.
- Incomplete abstracts will not be reviewed.

- Abstracts based on the same study with the same content reworded must not be submitted as multiple abstracts. Abstracts that are simply different versions of a single study will be rejected.
- Abstracts will be peer reviewed according to the subject categories. Authors must indicate during online submission the appropriate review category (one only). **All category selections will be final. There will be NO re-classification of abstracts after the abstract submission site has been closed.** Abstracts submitted to an inappropriate category are scored in that category and may receive a lower score as a result.
- All abstracts submitted will be considered eligible for one presentation format as explained above: either a platform or a Poster.
 - **Diagnostic Dilemmas/Challenge Cases (Unknown & Rare Known) Presentation** are formal oral presentations ranging from a 5-minute case setup with a 10 minute interactive panel discussion and audience participation. Pre-recorded presentations or remote presentations are NOT allowed. Additionally, presenters will need to follow standard presentation protocols with completion of HIPAA documentation, recording release, and financial disclosure.
 - **Poster Sessions** In addition to the display of your traditional printed posters, you are required to upload an electronic poster for online viewing. Instructions will be provided in January 2025.

Acceptance/Rejection Notification

- Notification regarding acceptance or rejection of abstracts will be sent to the submitting author on December 11, 2024, by email; consequently, an accurate personal email address is critical. If your abstract is accepted, the email will specify whether it is accepted as Platform or Poster presentation.
- Once the abstracts are accepted, they will be formatted for publication in an online supplement to *Genetics in Medicine Open* (<https://www.gimopen.org/>). Therefore, abstract authors should carefully review their abstract before submission to ensure that all authors and their affiliations are listed correctly.
- The decision of the ACMG Program Committee regarding acceptance and presentation of abstracts is final.

Abstract Withdrawal

- Once an abstract is accepted, a written request to withdraw must be submitted no later than February 7, 2025, to Education@ACMG.net if the first author decides to withdraw the abstract for any reason. Abstract withdrawal requests received after the deadline will be considered on a case-by-case basis.
- ACMG reserves the right to withdraw abstracts that are in violation of the College's policies and guidelines, or *GIM's* formatting guidelines, have been deemed scientifically unsound, or have been found to include inaccurate data, etc.
- For any abstract submitters with non-withdrawn abstracts who do not fulfill their professional commitment of presenting at the meeting, ACMG reserves the right to consider this information in the selection of abstracts for presentation in the future.

ACMG Annual Meeting Embargo Policy

All meeting attendees, presenters, authors, the media, exhibitors, institutions, industry and all other organizations and individuals are required to abide by the ACMG Annual Meeting Embargo Policy.

All abstracts, posters and oral (platform) scientific presentations are under strict embargo once ACMG has notified a presenter of their abstract's acceptance.

An embargo means that information from any abstract, poster or presentation is confidential and may not be announced, publicized, or disseminated before the embargo lift dates and times, as detailed below. The purpose of the ACMG Embargo Policy below is to protect the scientific integrity of all content presented at the ACMG Annual Clinical Genetics meeting and to ensure publicity is not released prematurely.

All ACMG Annual Clinical Genetics Meeting content to be presented or published is embargoed until the time of an actual conference presentation, ACMG has updated its guidelines to reflect the current e-publishing and online environment. The Embargo Policy below provides more details regarding the types of content that are embargoed.

The ACMG Embargo Policy is based on trust. Violation of the ACMG Embargo Policy may result in the abstract being withdrawn from the meeting and other measures deemed appropriate may be taken. Media who violate the Embargo Policy may lose their ACMG press credentials and be removed from the list to receive embargoed copies of ACMG news releases.

Specific Embargo Dates and Times:

Abstracts – Embargoed until posted on the ACMG Meeting website

Accepted abstracts will be made available online in advance of the ACMG Annual Clinical Genetics Meeting and will also be published in a special online supplement to our official open access journal, *Genetics in Medicine Open*. The information contained in abstracts is embargoed **until the abstracts are available and published online**. Information contained in abstracts may not be released in any format, including preprints, until the abstracts appear online.

ACMG continues to require that any information that goes beyond that contained in the submitted abstract is under embargo until the times specified below.

Posters – Embargoed until Wednesday, March 19, 2025, at 5:00 pm PT

For abstracts selected for posters, the embargo will lift **when the eposters are published via ACMG**.

Oral (Platform) Scientific Presentations – Presentations Embargoed until start of live session

The embargo for all scientific presentations will lift at the start time of the session in which the live presentation is being made. Interviews with presenters as well as any information to be presented for the first time at the ACMG Annual Meeting, are embargoed **until the time of the live presentation in which the paper will be given**.

For questions about the ACMG embargo policy, email education@acmg.net.

Abstract Submission Policies

Financial Disclosure Policy (Will be completed by **Platform Presenter or Case Challenge Presenter**)

The American College of Medical Genetics and Genomics (ACMG) is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide Accredited Continuing Education (ACE) for physicians. ACMG is an organization committed to improvement of patient care and general health by the incorporation of genetics and genomics into clinical practice.

ACMG has implemented the following procedures to ensure the independence of ACE activities from commercial influence/promotional bias, the Accreditation Council for Continuing Medical Education (ACCME) requires that providers (ACMG) must be able to demonstrate that: 1) everyone in a position to control the content of an ACE activity has disclosed all financial relationships that they have had in the past 24 months with ineligible* companies; 2) ACMG has implemented a mechanism to mitigate relevant financial relationships; and 3) all relevant financial relationships with ineligible companies are disclosed to the learners before the beginning of the educational activity. The learners must also be informed if no relevant financial relationships exist.

**Ineligible companies are defined as those whose primary business is producing, marketing, selling, re-selling, or distributing healthcare products used by or on patients.*

Standard to Prevent Commercial Bias and Marketing in Accredited Continuing Education

ACCME requirements designed to ensure that CME activities are independent and free of commercial bias.

Accredited continuing education must protect learners from commercial bias and marketing.

The accredited provider must ensure that all decisions related to the planning, faculty selection, delivery, and evaluation of accredited education are made without any influence or involvement from the owners and employees of an ineligible company.

Accredited education must be free of marketing or sales of products or services. Faculty must not actively promote or sell products or services that serve their professional or financial interests during accredited education.

The accredited provider must not share the names or contact information of learners with any ineligible company or its agents without the explicit consent of the individual learner.

Platform or Case Challenge Presenters Responsibility Regarding Financial Disclosure (ACCME Compliance)

- The presenting author for a platform presentation will be contacted and must complete the online financial disclosure form and additional items pertaining to their presentation and participation.
- Employees of ACCME defined ineligible company may not present an accredited platform presentation.

- The content of the information with which you are involved will promote quality in health care or advances in science and will not promote a specific proprietary or commercial interest. Content for this publication will be well-balanced and unbiased.

ACMG Education Policies

Please review the policies below regarding the ACMG Education program

- [Financial Disclosures and Mitigation Procedure](#)
- [ACMG Content Validation Policy](#)
- [ACMG Content Branding Policy](#)

How to Submit an Abstract

The abstract submission site opens on October 1, 2024.

All **abstracts must be submitted by Friday, November 15, 2024, 11:59 pm PT**. Submissions that are incomplete by the deadline will be rejected.

- Abstracts must be submitted online through the official online abstract **submission system**. Emails and word processing files not submitted through the site will not be accepted.
- Once you have submitted the title page information, a draft of your abstract will be saved, and you will be able to return to edit and update it at any time until **November 15, 2024**, at 11:59 p.m., PT. You can log back into the abstract portal at www.acmgmeeting.net.
- For technical support please click on the "Feedback and Support" tab to the right of your screen. Technical support for Administrative Application users provided by email during the hours of 9:00 am to 5:00 pm Central Time on regular working days (excluding weekends and holidays)

Preparing an Abstract for Submission

Please read the formatting guidelines before submission. Abstracts that are incorrectly formatted may be returned prior to review.

Contact Information

- Your name, degree, institution, address, phone number, and email address must be provided. As the corresponding author, you will receive all future correspondence from ACMG.
- The submitting author should be the first author (presenter) of the abstract, unless otherwise noted during submission.

Co-Authors

- Names of co-authors and institutions must be provided. **Changes will not be made to the spelling of authors' names after the submission deadline**; please proof your co-authors' names carefully.

Copyright Policy

- All authors must assign copyright of the abstract to ACMG, unless one of the authors is a U.S. Federal employee (in such case, ACMG does not hold copyright).

Abstract Title

- The abstract title should be brief and clearly indicate the nature of the abstract. You may use up to 20 words.
- The abstract title must be in title case. Capitalize all nouns, pronouns, adjectives, verbs, adverbs, and subordinate conjunctions (i.e., as, because, although). Except for the first word of the title, lowercase all articles, coordinate conjunctions (i.e., and, or, nor), and prepositions, regardless of length. Also, lowercase “to” when used as an infinitive. See examples [here](#).
- Additionally, keep letters lowercase if the lowercase letters have a specific meaning, such as aCGH.
- Do not put a period at the end of the title.
- For example: Cancer Genetics: Translation of New Concepts to Clinical Care

Use of Product Names

- Non-proprietary (generic/scientific) names should be used and should be lowercase.
- If necessary, you may include a proprietary name in parentheses directly following the generic name after its first mention in the body of the abstract; the first letter of the name of a proprietary drug should be capitalized. ACMG reserves the right to replace proprietary names with generic names to adhere to this policy.

Abbreviations

- Use standard abbreviations. Place abbreviations in parentheses immediately after the first mention of a term or phrase; the abbreviation can then be used throughout the abstract.

Terminology

Please refer to [Terminology in the *Genetics in Medicine* instructions to authors for detailed information](#).

ACMG has preferred **terminology** that **must** be followed. We also require that all sequence variants follow **HGVS nomenclature** and that variants are classified as per the **ACMG/AMP Guidelines**.

Terminology Check

Whole genome sequencing or whole-exome sequencing

- The preferred terminology is Genome sequencing (GS) or Exome sequencing (ES)

Invasive/NIPS/NIPT

We note the use of the terminology "noninvasive prenatal screening" and/or its acronym. In accordance with the ACMG, please use the term "prenatal cell-free DNA screening" or prenatal screening by cell free DNA to describe this screening test. The term "cell-free DNA" can be abbreviated as cfDNA; no other abbreviated forms of this term are permitted. When using the term

"cell-free DNA" to describe its application in settings other than prenatal (such as in oncology), after being defined in the manuscript as cell-free DNA, the abbreviation cfDNA should be used.

Incidental

- Incidental findings when results are not being intentionally sought though they may be anticipated (known to be potentially associated with the test) or unanticipated (not typically associated)
- Secondary findings when results are actively sought but not the primary reason for testing

Mutation and Carrier

GIM has preferred usage for certain terms

(<https://www.gimjournal.org/content/authorinfo#terminology>). We ask you to check your article to ensure those terms are used correctly. In particular, please note the following preferred terminology:

Mutation

- variant instead of mutation
- pathogenic variant to denote a disease-causing variant
(Use of frameshift mutation, mutational, mutation hotspot, mutation rate is permitted)

Carrier

- heterozygote (rather than carrier) when referring to a person's genotype/genetic test result. Explanation: The usage is intended to deal with the issue that "heterozygote" describes the presence of a variant, regardless of classification, but "carrier" typically implies that the variant is disease-causing. Preferred terminology is "heterozygous for a pathogenic variant". Carrier status, Carrier frequency, Carrier testing, Carrier result, and carrier screening are all permissible.

Caucasian, etc.

We note the use of the term "Caucasian" to describe one of the studied populations. For race, ethnicity, and genetic ancestry terms, GIM Open follows the "[Updated Guidance on the Reporting of Race and Ethnicity in Medical and Science Journals published in JAMA](https://jamanetwork.com/journals/jama/fullarticle/2783090)" (<https://jamanetwork.com/journals/jama/fullarticle/2783090>) and the copy editing guidelines in the AMA Style Guide (<https://doi.org/10.1093/jama/9780190246556.003.0011>). Specifically, we do not allow the use of the term "Caucasian" unless it refers to people from the Caucasus region.

VUSs

Please use the acronym VUS (not VUSs) as VUS can be plural or singular (variants of uncertain significance or variant of uncertain significance)

Unknown vs Uncertain

- Use the term "variant(s) of uncertain significance" instead of "variant(s) of unknown significance"

Subject

- When referring to persons included in research studies, 'participant' is preferred. A research 'participant' (preferred to subject'; see below) is a person who undergoes an intervention as part of a scientific investigation. A 'patient' is a person under medical care and 'patient' is not expected to be widely used in GIM articles; rather 'participant' or 'person' are generally

preferred. 'Subject' (as in study/research subject) is impersonal, perhaps derogatory, suggesting a subservient role and should not be used.

Primacy

- Claims of primacy and novelty are discouraged, e.g., *for the first time*.

Electronic Signature

Completion of all required information in the online abstract submission system serves as an agreement. It certifies the ACMG abstract submitter's understanding of the rules for participation contained in the online abstract submission program and affirms that:

1. All authors approve of submitting this work for presentation and publication;
2. The author(s) transfer(s) all copyright ownership of the named abstract to the American College of Medical Genetics and Genomics (except when one or more authors are U.S. Government employees);
3. Submitter is acting on behalf of authors and has read the **ACMG Conflict-of-Interest Policy** and has acted in accordance with that policy;
4. The author(s) agree(s) to materially confine the presentation to information in the abstract, if accepted for presentation. If an author has more than one abstract accepted, each presentation will be materially confined to the information in the abstract selected for the specific session;
5. The presenting author will be available to present the abstract if selected for the program. The author(s) will immediately notify ACMG if the first author is not the presenting author. Presenting author will be required to complete presenter documents including financial disclosure.

Contact Information

Send related correspondence and questions regarding abstract submissions or notifications to Education@ACMG.net.

ACMG Annual Meeting Registration and Housing www.acmgmeeting.net

- **Please note that submitting an abstract does not register you for the ACMG annual meeting.**
- Platform moderators, co-moderators, and presenters are not eligible to receive entitlements.

To register, you must complete and return an Attendee Registration Form, or register online through the ACMG Meeting website (www.acmgmeeting.net).