

28TH ANNUAL MEETING

SOCIEDADE PORTUGUESA DE GENÉTICA HUMANA

Secretariado

asic
Associação de Sociologia de Coimbra

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Organização

5 - 7

DECEMBER 2024



SPGH

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Abstract Submission to the 28th Annual Meeting of SPGH

Guidelines

The abstract submission and the registration to attend the 28th annual meeting of SPGH are independent actions. You can submit your abstract before your registration to attend the 28th annual meeting of SPGH.

Abstracts submission deadline: 30th September 2024

Abstract acceptance notification (predicted): 4th November 2024

NOTE: To participate and present your work you must be REGISTERED for the 28th Annual meeting of SPGH.

All abstracts for the **28th Annual SPGH Meeting** must be submitted electronically through the electronic abstract submission system.

Before you submit your abstract, please format it according to the following rules:

1. Abstracts must be written in English.
2. Abstracts should include the following sections:
 - a) Title (**mandatory – Capital Letters, Arial style with font size 14, 400 characters max.** including text spaces)
 - b) Author list (mandatory - e.g. Manuel B. Romeu, Maria C. Duarte - **Arial style with font size 11**)
 - c) Affiliations (mandatory - **Arial style with font size 11**)

Abstract text (mandatory - **Arial style with font size 11, 2200 characters max., including the main text and, if applicable, References, Acknowledgments, and Declaration of interests.**)

Abstracts that exceed 2200 characters including text spaces may be blocked at the Reviewing stage.
3. Abstract text **MUST** be organized into the following sections:
 - a) Introduction
 - b) Methodology
 - c) Results
 - d) Discussion
4. Abstract Author's should ensure that proper grammar and spelling is used as well as that abstracts are well-written, organized, clear, and understandable.
5. **Each author may be co-author of several abstracts, but the presenter of only one work** (oral communication or Poster) submitted to the 28th Annual Meeting of SPGH.
6. When co-author of different abstracts, the authors' names must be written identically in each abstract.

What happens after abstract submission?

1. All abstracts will be evaluated through blind peer review by the SPGH scientific committee.
2. Acceptance of abstracts or their selection for oral communication will be based on the following considerations: Originality, appropriate Methodology used, Quality of scientific results, and relevance of Conclusions.
3. Selected abstracts will be published in the Abstract Book of the 28th Annual SPGH Meeting and in the scientific journal "Medicine®".
4. The presenting author is responsible for notifying all co-authors regarding abstract acceptance or rejection.
5. Specific Guidelines to prepare Oral presentations and Posters will be sent to First/Presenting Authors after acceptance.

Abstract Categories (to be chosen upon abstract submission)

Basic Research – Abstracts submitted in the "Basic research" category are generically those describing research works aimed at elucidating fundamental principles and mechanisms in the area of human genetics. These studies often provide broad insights applicable to many scientific fields, regardless of direct disease relevance. Such research frequently employs cell lines, *in vitro*, *in silico* and/or *in vivo* models, pure bioinformatics approaches, theoretical or *in silico modeling*, population genetics in non-disease cohorts, social or socio-economic approaches, and forensic applications.

Clinical Research – Abstracts submitted in the "Clinical research" category are generically those describing medical - and/or health-related research works aimed at advancing our understanding of human disease, developing diagnostic strategies, or effective prevention and treatment strategies. "Clinical Research" embraces a continuum of studies that involve direct interaction with patients or analysis of clinical data, namely diagnostic clinical materials, clinical-trials, case-control studies or other sorts of patient-related data analysis.

Clinical Case Reports – Abstracts submitted in the "Clinical case reports" category typically describe and analyze the diagnosis and/or the management of **five patients/families or less**. It is the first line of evidence in health care and forms the basis of the evidence-based pyramid. These case reports should serve as primary sources of evidence, and may be accompanied by a literature review of similar cases.

Clinical Cohorts – Abstracts submitted in the "Clinical cohorts" category typically describe research findings from the analysis of a **medium to large cohort of patients/families**. These Clinical cohort studies are expected to represent national and/or international efforts to improve knowledge on genetic disorders, and illustrate the outcome of collaborative networks in the Human Genetics field.

Type of presentation:

- Poster
- Oral communication

The Scientific commission of SPGH has the final decision about the type of presentation (poster or Oral communication).

Please indicate if you are eligible and applying to the two SPGH awards for young investigators (information available below or at <http://spgh.net/regulamento-dos-premios>)

Young Researcher – ESHG National Fellowship award	Young Researcher - Prof. Amândio Tavares	Best Basic Research	Best Clinical Research
Includes ESHG registration, travel grant and hotel accommodation to attend the ESHG 2025 in Milan	Includes ESHG registration, travel grant and hotel accommodation to attend the ESHG 2025 in Milan	500.00€	500.00€

REGULATIONS OF SPGH PRIZES - EN

SPGH Prizes:

B.1 SPGH Young Investigator ESHG National Fellowship Award;

- a) The ESHG National Fellowship is the responsibility of ESHG, and is awarded annually to a young researcher under the age of 35 who is a member of the SPGH (with regularized status before the beginning of the Annual Meeting) and who has shown excellence in the area of human genetics.
- b) This scholarship offers the registration for the ESHG Annual Meeting, as well as travel and accommodation expenses in an amount to be published annually (www.eshg.org).
- c) The selection of the candidate is the responsibility of the SPGH Scientific Committee, which should take into account the following prioritization criteria among candidates who meet the ESHG requirements indicated above: Best Oral Communication/Poster of the current year, preferably in the area of basic research, and in which the presenter meets criterion a).
- d) If the candidate has been selected by work presented at the SPGH Meeting, he/she must submit it to the ESHG meeting.

B.2 SPGH Young Researcher Professor Amândio Tavares Award

- a) The SPGH Professor Amândio Tavares Young Researcher Award is awarded annually and aims to support a young researcher, author of the best research work in clinical genetics or laboratory diagnosis presented at the SPGH Annual Meeting.
- b) The selection for the Prize, among the first authors of oral communications and posters, is the responsibility of the SPGH Scientific Committee. The 1st author must be 30 years of age or younger. In the absence of works of unequivocal merit by authors with this profile, the age may be extended to a maximum of 35 years.
- c) The patron of the award, Prof. Purificação Tavares, following her Father's line of encouragement to young researchers, awards a scholarship to participate in the European Human Genetics Conference of the ESHG the following year, supporting the registration, travel and accommodation expenses of the winner of the award.
- d) At the time of submission of the abstract of the work to the SPGH Annual Meeting, the author must register on the platform that he/she is applying for this award, so that the SPGH Scientific Committee knows which authors meet the age criterion and who wish to have the opportunity to participate in the ESHG European Human Genetics Conference of the following year through this award.

C. SPGH Award for Best Communication (Oral/Poster) in Basic Research

- a) The Award for the Best Communication (Oral/Poster) Basic Research is awarded annually and aims to reward the first author of a basic research work in human genetics, presented at the SPGH Annual Meeting among the selected works.
- b) The selection for the Award among the first authors of communications is the responsibility of the Scientific Committee of SPGH.
- c) This prize has a monetary value to be established annually by the Board of Directors of SPGH and will be delivered at the closing session of the Annual Meeting.

D. Award for the Best Communication (Oral/Poster) in Clinical Research

- a) The Award for the Best Communication (Oral/Poster) Clinical Research is awarded annually and aims to reward the first author of a clinical research work presented at the SPGH Annual Meeting among the selected works.
- b) The selection for the Award among the first authors of communications is the responsibility of the Scientific Committee of SPGH.
- c) This prize has a monetary value to be established annually by the Board of Directors of SPGH and will be delivered at the closing session of the Annual Meeting.

3. General Provisions

- a) The Scientific Committee of SPGH reserves the right to award ex-aequo prizes, honorable mentions (up to a maximum of three for each prize), or not to award prizes.
- b) The four SPGH prizes are not cumulative.
- c) The Scientific Committee of SPGH will deliberate on all aspects omitted from this regulation, and its decision will be sovereign and without the right to complaint.
- d) Each author can only have one presentation as a presenting author submitted to each annual meeting. However, he may be co-author of several submissions.

REGULAMENTO DOS PRÉMIOS - PT

Prémios SPGH:

B.1 Prémio SPGH Jovem Investigador ESHG National Fellowship;

- a) A bolsa ESHG National Fellowship é da responsabilidade da ESHG, sendo atribuída anualmente a um jovem investigador com idade inferior a 35 anos que seja membro da SPGH (com situação regularizada antes do início da Reunião Anual) e que se tenha distinguido por mostrar excelência na área de genética humana.
- b) Esta bolsa oferece a inscrição na Reunião Anual da ESHG, bem como despesas de viagem e alojamento num valor a publicar anualmente (www.eshg.org).
- c) A seleção do candidato é da responsabilidade da Comissão Científica da SPGH, que deverá ter em conta os seguintes critérios de priorização entre candidatos que preencham os requisitos da ESHG acima indicados: Melhor Comunicação Oral/Poster do corrente ano, preferencialmente na área da investigação básica, e em que o apresentador cumpra o critério a).
- d) Caso o candidato tenha sido selecionado por trabalho apresentado na Reunião da SPGH, deverá submetê-lo à reunião da ESHG.

B.2 Prémio SPGH Jovem Investigador Professor Amândio Tavares

- a) O Prémio SPGH Jovem Investigador Professor Amândio Tavares é atribuído anualmente e tem como objetivo apoiar um jovem, autor do melhor trabalho de investigação orientado para a genética clínica ou diagnóstico laboratorial apresentado na Reunião Anual da SPGH.
- b) A seleção para o Prémio, entre os primeiros autores de comunicações orais e posters, é da responsabilidade da Comissão Científica da SPGH. O 1o autor deverá ter idade igual ou inferior a 30 anos. Na ausência de trabalhos de inequívoco mérito de autores com este perfil, a idade poderá ser estendida aos 35 anos no máximo.
- c) O mecenas do prémio, a Prof. Purificação Tavares, seguindo a linha de estímulo do seu Pai a jovens investigadores, atribui uma bolsa para participação na European Human Genetics Conference da ESHG do ano seguinte, suportando as despesas de inscrição, viagem e alojamento do vencedor do prémio.
- d) Na altura da submissão do resumo do trabalho para a Reunião Anual da SPGH, o autor tem obrigatoriamente de registar na plataforma que se candidata a este prémio, de modo a que a Comissão Científica da SPGH saiba quais os autores que cumpram o critério de idade e que desejam ter a oportunidade de participar na European Human Genetics Conference da ESHG do ano seguinte através deste prémio.

C. Prémio SPGH Melhor Comunicação (Oral/Poster) em Investigação Básica

- a) O Prémio para a Melhor Comunicação (Oral/Poster) Investigação Básica é atribuído anualmente e tem como objetivo premiar o primeiro autor de um trabalho de investigação básica em genética humana, apresentado na Reunião Anual da SPGH entre os trabalhos selecionados.
- b) A seleção para o Prémio entre os primeiros autores de comunicações é da responsabilidade da Comissão Científica da SPGH.
- c) Este prémio tem um valor pecuniário a estabelecer anualmente pela Direção da SPGH e será entregue na sessão de encerramento da Reunião Anual.

D. Prémio para a Melhor Comunicação (Oral/Painel) em Investigação Clínica

- a) O Prémio para a Melhor Comunicação (Oral/Poster) Investigação Clínica é atribuído anualmente e tem como objetivo premiar o primeiro autor de um trabalho de investigação clínica apresentado na Reunião Anual da SPGH entre os trabalhos selecionados.
- b) A seleção para o Prémio entre os primeiros autores de comunicações é da responsabilidade da Comissão Científica da SPGH.
- c) Este prémio tem um valor pecuniário a estabelecer anualmente pela Direção da SPGH e será entregue na sessão de encerramento da Reunião Anual.

3. Disposições Gerais

- a) A Comissão Científica da SPGH reserva-se o direito de atribuir prémios ex-aequo, menções honrosas (até um máximo de três por cada prémio), ou de não atribuir prémios.
- b) Os quatro prémios SPGH não são acumuláveis.
- c) A Comissão Científica da SPGH deliberará em todos os aspetos omissos deste regulamento, sendo a sua decisão soberana e sem direito a reclamação.
- d) Cada autor só pode ter um trabalho como autor apresentador submetido a cada reunião anual. Podendo no entanto ser co-autor de vários trabalhos.