

Standing Committee on the International System for Human Cytogenomic Nomenclature

Terms of Reference

1. The International System for Human Cytogenomic Nomenclature (ISCN) Standing Committee

The ISCN Standing Committee is a committee of clinical cytogeneticists that reviews and revises the nomenclature that is used to communicate the results of cytogenomic investigations by conventional karyotyping, FISH, molecular cytogenomic assays such as microarray, MLPA, QF-PCR, genome mapping and sequencing. The scope of the work and remit of the group is defined herein.

- 1.1** The ISCN Standing Committee is affiliated with the HUGO Nomenclature Standards Committee.
- 1.2** The ISCN Standing Committee decides which Publisher is engaged (currently Karger) for the publication and online version of the ISCN nomenclature.

2. Committee membership

- 2.1** The size of the Committee should not exceed twelve members, all of whom are voting members.
- 2.2** The Committee should include no more than:
 - Four members from the Americas,
 - Four members from Europe,
 - Two members from Asia, and
 - Two members from Africa/Oceania.
 - These twelve members include those reserved for the ISCN Standing Committee Chair and Vice-Chair.
 - In addition, the Standing Committee includes a representative of the HGVS Nomenclature Committee (who can vote).
- 2.3** The Standing Committee shall have a Chair and he/she may opt to have a Vice-Chair.
 - 2.3.1** The Chair will seek nominations from the SC committee. Where more than one is nominated the committee will vote to elect the vice chair.
- 2.4** Members shall be nominated and elected by their peers through a process organized by the Standing Committee Chair (see Clause 3.8).
- 2.5** The Chair or Vice-Chair will represent the Standing Committee on any other committees (e.g., HGVS Nomenclature Committee, HUGO Nomenclature Standards Committee).
- 2.6** The Standing Committee can recruit advisors for a limited term where specific expertise is required, at the discretion of the Chair (non-voting member).

3. Term of office

- 3.1** The term of office is four years in the first instance.
- 3.2** Members may be re-appointed to the Standing Committee for a further term of office of four years.
- 3.3** A maximum of 50% of the Standing Committee may be re-elected concurrently. In order to maintain 'corporate memory', the Chair and Vice-Chair may ask members to remain on the Committee for more than eight years.

- 3.4 Members may be asked to leave the Committee if they do not contribute to the scope of the Committee (see Clause 5) or do not attend >50% of virtual or face-to-face meetings over a given time period without a valid reason.
- 3.5 Members of the Committee will be removed from the Standing Committee if there is evidence of professional misconduct or a criminal conviction resulting in imprisonment. Regular elections to the Committee take place within six months after every change of Chair or at least every four years. Replacement elections following removal of a member of the Committee are held when required. During regular elections, all voting members of the Committee will need to confirm their willingness to continue on the Standing Committee.
- 3.6 The term of office for Chair and Vice-Chair is four years in the first instance. To elect the new Chair the Standing Chair will call for nominations from the committee. If there is more than one candidate then the committee will vote. If no nominations are received then the Standing Chair will negotiate with committee members outside of the meeting. The appointment of the new Chair will then be ratified by the Committee electronically. The Chair may appoint a Vice-Chair who can replace the Chair when they stand down.
- 3.7 The Chair organises the submission of nominations, ballots for voting and election of members. The Publisher has the right to suggest candidates and to veto nominations where there is a commercial conflict of interest but will not otherwise influence the elections.
- 3.8 The Chair asks the genetics community for nominations for the ISCN Committee giving their name and position. The nomination must be made or seconded by a Head or Section Head of a diagnostic laboratory. Once the nomination period is closed, the ISCN SC will review the applicants for suitable diagnostic experience, expertise in different cytogenomic techniques and evidence of reporting of cytogenomic results including complex cases. The ISCN SC will then vote by ranking the candidates. The individual(s) with the most votes is elected to the Committee. In the event of no clear decision by the ISCN SC, the genetics community are asked to vote for a candidate for each continent (see Clause 2.2). The Chair informs all candidates of the outcome of the vote. The number of votes that an individual receives will not be disclosed. In the event of a tied vote for the same continent, the Chair will have the casting vote.

4. Committee Meetings

- 4.1 The Committee will usually work remotely through digital communication *e.g.* zoom, googlemeet, Microsoft teams. There will be face-to-face meetings at either the European Society of Human Genetics (ESHG) or the American Society for Human Genetics (ASHG) every two years. At the request of the Chair, the Publisher may assist the ISCN Standing Committee with a suitable digital working environment, *e.g.*, Microsoft teams.
- 4.2 When virtual meetings are required, a time will be arranged when all individuals from different time zones can reasonably be expected to attend. The time of the meetings will be given in GMT.
- 4.3 When a revision of the ISCN is required, the Chair will arrange a face-to-face meeting with the Standing Committee.
- 4.4 The Chair can propose face-to-face meetings to the Publisher when required.
- 4.5 No decisions can be made at a meeting unless either the Chair or Vice-Chair is present.

- 4.6 The meeting quorate is >50% attendance. If there is no quorate at the meeting, decisions can be made but must be ratified by the larger group by electronic communication.
- 4.7 At the request of the Chair, individuals who are not Members of the Standing Committee, including the Publisher, may be invited to attend to provide professional expertise, if required. Invitees have no voting rights and may be asked to leave the meeting at any time by the Chair and/or Vice-Chair. Members of the Standing Committee can suggest individuals to nominate.
- 4.8 If there is no unanimous agreement, then a vote is taken and decisions are made on the basis of a majority vote. If there is a split vote, then the Chair has the casting vote.

5. Scope of ISCN Standing Committee

- 5.1 Define a standard for an unambiguous description of normal and abnormal cytogenomic results.
- 5.2 Ensure the ISCN recommendations are published in book and online forms.
- 5.3 Promote the use of the ISCN, revisions and application at conferences, workshops and via webinars where applicable
- 5.4 Promote the use of ISCN by genetics laboratories for banded chromosomes, FISH, microarrays and other molecular assays.
- 5.5 Review proposals to modify or extend the ISCN nomenclature that includes an invitation for suggestions from within the Cytogenomics community via an online portal. The Chair and Vice-Chair will collate the suggestions and the Committee will discuss any amendments needed.
- 5.6 Provide advice to the HUGO Standards Committee.
- 5.7 Communicate with the other committees/working groups in the HUGO Nomenclature Standing Committee where their nomenclature potentially conflicts with ISCN nomenclature principles.
- 5.8 Provide advice to genetic laboratories on the usage of ISCN via email or through the Karger forum.
- 5.9 Evaluate errata and suggested amendments from the genetics community in a timely manner and reject or approve for inclusion in the website version.

6. Publication of ISCN standard

- 6.1 The ISCN standard is published online and as a book by a Publisher (currently Karger).
- 6.2 There is a signed agreement between the Chair of the ISCN Standing Committee and the Publisher that shall continue automatically. The Agreement can be terminated at any time with a notice period of 12 months by either party.
- 6.3 Either party may, by giving a written notice to the other, terminate this Agreement with immediate effect if the other party commits a breach of contract that cannot be remedied; has not been remedied within 30 days of the receipt of notice that identifies the breach and requires its remedy.
- 6.4 The ISCN Editor serves as the Chair *ex officio* of the Committee.
- 6.5 The book will be published in print format. The electronic version will be made available via a portal on the Publisher's website. The book will also be republished as a Special Topic Issue to the journal "Cytogenetic and Genome Research". The Special Topic Issue will be available in print format only and will be released at the same time as the book is published.
- 6.6 The layout of the book will conform with the standard layout of the previous ISCN publications whereas the layout of the Special Topic Issue will conform with the standard layout of the journal "Cytogenetic and Genome Research".

- 6.7** The Publisher's electronic version of ISCN allows for the continuous integration of errata and amendments, creating a dynamic, up-to-date resource complementing the book with version control. The Publisher will make available a means for users of ISCN to point out errata and suggest amendments to which the Standing Committee may respond. The technology, available formats and commercial models used in the Publisher's electronic version are the sole responsibility of the Publisher.
- 6.8** The ISCN Chair and Committee will identify any needs and requirements for the ongoing development of the electronic version of ISCN. Any extension of the website by online working tools catering to the specific needs of cytogeneticists based on the content of the book and other sources will be made in consultation with the Standing Committee and the Publisher.
- 6.9** The Editor and each of the Committee members will receive at least three copies of the book immediately following publication. Committee members are also entitled to free unlimited access to the electronic version of ISCN and the digital version of the journal "Cytogenomic and Genetic Research" during their tenure and, at their request, five years after their term of office.

7. Finance

- 7.1** Committee members give of their time free of charge.
- 7.2** The Publisher will, subject to approval in advance, organize and cover the cost of on-site meetings of the Chair and Committee members as follows:
- ASHG/ESHG registration fees for the Chair and one additional Committee member designated by the Chair
 - Meeting facilities and meals: up to two full days.
 - Accommodation: up to three nights per person attending.
 - Travel and personal costs: five hundred U.S. dollars (\$500) contribution per person attending from the continent of the venue and up to \$1000 contribution per person from other continents.
- 7.3** The Publisher will financially support up to one such event every two years provided such events are being scheduled to take place during the annual meeting of the ASHG or the ESHG.