Connecting and Advancing Health Sciences

Media Release

Basel, Switzerland, 21 Jan.2021

Interactive, Digital, and Innovative: Karger's ISCN 2020

With the new edition of ISCN 2020, Karger offers the first online and interactive version of the International System for Human Cytogenomic Nomenclature (ISCN).

The ISCN Committee has created an updated version of *ISCN 2020* with an extended version of the Human Genome Variation Society (HGVS) nomenclature. Both nomenclatures are now also available online. "We seek to standardize the way the abnormalities in chromosomes are described", says Dr. Jean McGowan-Jordan, co-editor of *ISCN 2020* and chair of the ISCN Committee. This standardization is valid for all technologies.

The online platform also offers access to HGVS, that together with *ISCN 2020* forms a comprehensive nomenclature. Tools that help practitioners in their daily work will increasingly supplement the online platform. For example, the karyotyping tool helps users to generate the correct nomenclature by gradually indicating the abnormalities observed on their karyogram. Next to the latest edition, the platform offers the possibility to view earlier PDF versions of the *ISCN* book, as well.

The further development of *ISCN* has already begun: In 2021, Karger Publishers is planning on offering their users a forum to get in touch with each other and discuss problems, changes, and updates. A Mutalyzer tool, used to verify the HGVS nomenclature, will also be integrated. In addition, the publishing house will offer educational courses on ISCN, HGVS, and Sequencing Quality Assurance based on *Fast Facts-Medical Handbooks* in preparation. Laurenz Baltzer, Strategic Competence Lead, Digital Health at Karger Publishers, said "This is creating a new exciting perspective to the users of ISCN.karger.com. It represents a further step in Karger's ambition of Connecting and Advancing Health Sciences".

The following standardizations can be found in ISCN 2020:

- Chromosomes are now described starting from pter to qter and no longer from the centromere position.
- The sex chromosome abnormalities are always described first for all technologies.
- New abbreviations have been established to clarify the inheritance of rearrangements and to describe polar bodies.

About Karger Publishers

Karger Publishers is a worldwide publisher of scientific and medical content based in Basel, Switzerland. It is independent and family-led in the fourth generation by Chairwoman and Publisher Gabriella Karger. Connecting and advancing health sciences since 1890, Karger has been continuously evolving, keeping pace with the current developments and shifts in research and publishing. The publishing house is dedicated to serving the information needs of the scientific community, clinicians, and patients with publications of high-quality content and services in health sciences. Karger Publishers has 240 employees and is present in 15 countries around the globe.

For more information, please visit karger.com

