Human Genome Center

Department of Public Policy
公共政策研究分野

Professor	Kaori Muto, Ph.D.	教　授	博士(保健学)	武　藤　香　織
Associate Professor	Izen Ri, Ph.D.	准教授	博士(学際情報学)	李　　怡　然
Assistant Professor	Yukitaka Kiya, Ph.D.	助　教	博士(社会学)	木　矢　幸　孝

The Department of Public Policy contributes to the accomplishment of the following major missions: research ethics consultation to help scientists to comply with ethical guidelines and build public trust; public policy science studies of translational research and its societal impact; and promotion of patient and public involvement/ engagement in research and health care. Through qualitative and quantitative social science studies and policy analysis, we facilitate discussion of the challenges posed by advances in medical science.

1. Symposium report—ethical and social considerations regarding return of secondary findings in atomic bomb survivors

The Radiation Effects Research Foundation (RERF) conducts research on the health effects of atomic bomb (A-bomb) radiation, supported by the long-term cooperation of survivors and their children. In response to concerns from A-bomb survivors, their offspring, and the global community, RERF has investigated transgenerational radiation effects for many years. Currently, RERF is planning the Trio Genome Study (TGS), which will utilize advanced genome sequencing to examine potential genetic effects on the offspring of survivors. This study could yield significant findings but also presents ethical challenges, particularly in ensuring that results are returned responsibly to the community. RERF first held an international workshop to address ethical, legal, and social issues related to genomic analysis in 2020, followed by the formation of stakeholder committees to align the study with community perspectives. Most recently, in December 2024, an international symposium further explored frameworks important to the TGS, including risk communication, benefit-sharing, community involvement, and return of secondary findings. In genomic analysis, secondary findings comprise a deliberate search for pathogenic variants in an established list of genes in disorders associated with life-threatening manifestations. Returning these secondary findings to research participants is crucial from the perspective of benefit-sharing. Discussions emphasized the need for transparency in the return of secondary findings, especially those with actionable health implications, including sharing the rationale behind providing information about genetic variants to research participants. These frameworks will guide not only future RERF studies but also genomic research worldwide, ensuring that the findings benefit participants and communities.

2. What does “appropriate scientific justification” mean for the review of human pluripotent stem cell, embryo, and related research?

Rapid advances in human pluripotent stem cell (PSC), embryo, and stem cell–based embryo model research have enabled unprecedented exploration of early human development, while also raising complex ethical and regulatory questions. The 2021 International Society for Stem Cell Research (ISSCR) Guidelines require that such research be supported by “adequate and appropriate scientific justification,” yet this concept has not been clearly operationalized

for investigators and reviewers. In this article, members of the ISSCR Ethics Committee examine what constitutes scientifically justifiable research involving human PSCs, embryos, and related models. Drawing on multidisciplinary expertise and discussions from dedicated workshops, the authors identify seven key characteristics relevant to scientific justification, including scientific rigor, importance of the research question, comparison with alternative approaches, stepwise study design, assessment of research risks, adequacy of the research environment, and investigator qualifications. The paper also discusses how ethical considerations, jurisdictional differences, and evolving societal values intersect with scientific review. By providing practical guidance for oversight committees, regulators, and researchers, this work aims to promote responsible innovation, transparent decision-making, and sustained public trust in sensitive areas of stem cell and embryo research worldwide.

3. International Survey on Genetic Literacy and Awareness in Patients with Spinal and Bulbar Muscular Atrophy

Genetic literacy among patients is important for presymptomatic diagnosis and intervention, family communication, and future therapeutic strategies for inherited neuromuscular diseases. This international survey examined genetic literacy and related attitudes among patients with spinal and bulbar muscular atrophy (SBMA) across Japan, the Americas and Europe, and Asia-Oceania (excluding Japan). Questionnaires were developed in collaboration with patient advocacy groups and distributed to patients with SBMA to assess knowledge of inheritance, genetic testing, information sharing, reproductive technologies, and genetic counseling. Responses from 628 patients revealed that only around one-third demonstrated high genetic literacy across all regions, defined by correct answers regarding inheritance risk. While overall literacy levels were similar, substantial regional differences were observed in willingness to share genetic information with family members and to recommend presymptomatic genetic testing, with patients in Japan and the Asia-Oceania group being more hesitant. In Japan, higher genetic literacy was associated with younger age and greater willingness to participate in clinical research, but such associations were not observed in other regions.

4. Exploring Patient and Public Involvement and Engagement in Allergy Research: Cross-Disease and Cross-Stakeholder Perspectives in Japan

Patient and public involvement and engagement (PPIE) is increasingly recognized as a core ethical and practical component of medical research, yet its implementation varies across disease areas and stakeholder groups. This study investigated the current status of PPIE in allergy research in Japan through a survey of principal investigators and patient advocacy group representatives, with comparisons to rare disease and cancer research. The survey assessed perceptions of the necessity of PPIE, existing practices, unmet needs, and the use of digital tools for engagement. Results showed that patient advocacy groups in allergy research strongly endorsed the importance of PPIE and reported frequent interactions with researchers, whereas many researchers demonstrated lower awareness and limited implementation of formal PPIE frameworks. Key challenges included the need for training, dedicated coordinators, and practical guidance to facilitate effective collaboration. Despite active use of digital platforms by patient groups, researcher utilization remained limited. These findings reveal structural and awareness gaps between stakeholders and emphasize the importance of education, standardized guidelines, and supportive infrastructure to achieve meaningful and sustainable PPIE in allergy research.

Publication list

1. Noriaki Yoshida, Douglas R Stewart, Arikuni Uchimura, Ken-ichi Arita, Yvonne Bombard, Megan Frone, Takao Hinoi, Kenji Kamiya, Shigeru Katamine, Junji Kayukawa, Kazunori Kodama, David T Miller, Kiyonori Miura, Kaori Muto, Fuji Nagami, Hidewaki Nakagawa, Asao Noda, Waka Ohishi, Osamu Tanabe, Chikashi Terao, Preetha Rajaraman, Kazuto Kato, Leslie G Biesecker. Symposium report—ethical and social considerations regarding return of secondary findings in atomic bomb survivors. Carcinogenesis. 46(3): bgaf059. 2025. doi: 10.1093/carcin/bgaf059.
2. Erica C. Jonlin, Misao Fujita, Rosario Isasi, Kazuto Kato, Megan Munsie, Kaori Muto, Kathy Niakan, Krishanu Saha, Jeremy Sugarman, Leigh Turner, Insoo Hyun. What does "appropriate scientific justification" mean for the review of human pluripotent stem cell, embryo, and related research? Stem Cell Reports. 20:10249-102479. 2025. doi: 10.1016/j.stemcr.2025.102479.
3. Shinichiro Yamada, Atsushi Hashizume, Daisuke Ito, Yoshiyuki Kishimoto, Shota Komori, Takahiro Kawase, Ayano Kondo, Yukitaka Kiya, Hideaki Nakatsuji, Yukihiko Suzuki, Yasuhiro Tako, Yukihiko Hamada, Kim Slowe, Kathleen A. Thompson, Terry Thompson, Masahisa Katsuno.International Survey on Genetic Literacy and Awareness in Patients with Spinal and Bulbar Muscular Atro-

phy. Neurology Genetics. 12 (1) : e200336. in Press. Doi: 10.1212/NXG.0000000000200336.
4. Takeya Adachi, Saori Watanabe, Yu Kuwabara, Yuki Abe, Masaki Futamura, Takenori Inomata, Keima Ito, Meiko Kimura, Keiko Kan-o, Hanako Koguchi-Yoshioka, Yosuke Kurashima, Katsunori Masaki, Mayumi Matsunaga, Haruka Miki, Saeko Nakajima, Yuumi Nakamura, Yasushi Ogawa, Aiko Oka, Masafumi Sakashita, Sakura Sato, Kyohei Takahashi, Masato Tamari, Takeshi Tsuda, Satoru Yonekura, Mayumi Tamari, Kaori Muto, Hideaki Morita. Exploring Patient and Public Involvement and Engagement in Allergy Research: Cross-Disease and Cross-Stakeholder Perspectives in Japan. Allergy. 0:1-4. 2025. doi:10.1111/all.70064.
5. 河合香織，島﨑美空，武藤香織．遺伝性疾患における結婚・出産に関する助言のあり方の検討―難病情報センターを手掛かりに―．日本難病医療ネットワーク学会機関誌．12(2): 44-53. 2025.
6. 武藤香織，社会調査と倫理審査委員会：日本社会学会会員への調査を手掛かりに．理論と方法．40 (1): 56-69. 2025.
7. 木矢幸孝．遺伝・ゲノム医療の倫理．三重野雄太郎・秋葉峻介編『概説　生命倫理学』第12章．大学教育出版．2025.
8. 渡部沙織，武藤香織．社会の視点から考える診療ガイドライン：患者・市民参画(PPI)を診療ガイドライン策定にどう取り入れるか．小児科診療．88(1): 46-50. 2025.
9. 渡部沙織，美馬達也，秋葉峻介，杉原正子．患者・市民参画(PPI)とは何か：その現状と課題．医学哲学医学倫理．43: 119-122. 2025.
10. 安藤道人，河田純一．高額療養費改革案はどう見送られたのか：2024年度案の政策形成・修正過程と患者運動．医療経済研究．37(1): 29-43. 2025.
11. 西 千尋，三村恭子，李 怡然，武藤香織．感染症危機事態における病原体研究のELSI．生体の科学．76(5): 420-421. 2025.