東アジア人類遺伝学会連合 第14回大会シンポジウム / The 14th Annual Meeting of the East Asian Union of Human Genetics Societies

東アジア人類遺伝学会連合 第 14 回大会 / The 14th Annual Meeting of the East Asian Union of Human Genetics Societies

日　時：11 月 20 日（木）9:30 ～ 10:50、11:00 ～ 12:20、14:45 ～ 16:25　第 2 会場（小ホール）
Date : Nov. 20 (Thu.) 9:30-10:50, 11:00-12:20, 14:45-16:25 Room 2 (Small Hall 5F)

Chairs : Xue Zhang (Chinese Society of Human Genetics)
Masao Nagasaki (Tohoku University)

EAUHGS-1 9:30
Korean Reference Genome Variation Project (KRGVP): Contents and Implication
○ Seong Beom Cho
Division of Biomedical Informatics, Center for Genome Science, National Institute of Health, KCDC

EAUHGS-2 9:50
One thousand Japanese whole genome project in ToMMo
○ Masao Nagasaki
a Department of Integrative Genomics, Tohoku Medical Megabank Organization, Sendai, 980-8575, Japan

EAUHGS-3 10:10
Molecular diagnosis of hereditary hearing loss and its related translational medicine
○ Guangxin Xiang
Translational Medicine Institute, CapitalBio Corporation & National Engineering Research Center for Beijing Biochip Technology, Beijing, China

EAUHGS-4 10:30
The role of *de novo* variants in rare disease pathogenesis
○ Murim Choi
Department of Biomedical Sciences, Seoul National University College of Medicine, Seoul, Korea.

10:50-11:00 break

Chairs : Jin-Sung Lee (Korean Society of Medical Genetics)
Yoko Aoki (Tohoku University)

EAUHGS-5 11:00
MeCP2 in autism － displays allelic expression imbalance and affects both nerve system and heart development by regulating Notch pathway
Yufang Zheng、Yumeng Wang、Yahui Liu、Zhangmin Yang、Yanqin He、Keping Hu、○ Hongyan Wang
State Key Laboratory of Genetic Engineering, School of Life Sciences Fudan University

EAUHGS-6 11:20
Brain Somatic Mutations in Neurodevelopmental Disorders: Lessons from Epilepsy Patients
○ Jeong Ho Lee
Graduate School of Medical Science and Engineering, KAIST (Korea Advanced Institute of Science and Technology), Daejeon 305-701, Korea

EAUHGS-7 11:40
Next-generation sequencing applications in pre- and postnatal genetic diagnostics
○ Desheng Liang[1,2]、Weigang Lv[1]、David S Cram[3]、Lingqian Wu[1,2]
1）State Key Laboratory of Medical Genetics, Central South University, Changsha, China
2）Hunan Jiahui Genetics Hospital, Changsha, Hunan, China
3）Berry Genomics C., Limited

EAUHGS-8 12:00 **次世代シークエンサーを用いた RASopathies の遺伝子解析 / Molecular analysis of RASopathies using next generation sequencer**

○青木 洋子[1)]（Yoko Aoki）、新堀 哲也[1)]（Tetsuya Niihori）、井上 晋一[1)]（Shin-ichi Inoue）、松原 洋一[1,2)]（Yoichi Matsubara）

1）東北大学大学院　医学系研究科　遺伝病学分野
（Department of Medical Genetics, Tohoku University School of Medicine, Sendai, Japan）
2）国立成育医療研究センター研究所
（National Research Institute for Child Health and Development, Tokyo, Japan）

12:20-14:45 Lunch break

Chairs : Kyuyoung Song (University of Ulsan College of Medicine)
Taisei Mushiroda (RIKEN)

EAUHGS-9 14:45 **Genome-wide association analyses in Han Chinese identify two new susceptibility loci for amyotrophic lateral sclerosis**

○ Min Deng[1)]、Ling Wei[2)]、Xianbo Zuo[3)]、Xiaodong Ju[4)]、Kai Wang[2)]

1）Medical Research Center, Peking University Third Hospital, Beijing, 100191, China.
2）Department of Neurology, No.1 Hospital, Anhui Medical University, Hefei, Anhui, 230022, China.
3）State Key Laboratory Incubation Base of Dermatology, Ministry of National Science and Technology, Hefei, Anhui, 230032, China.
4）Institute of Sports Medicine, Peking University Third Hospital, Beijing, 100191, China.

EAUHGS-10 15:05 **Esophageal squamous cell carcinoma in China: interaction of environmental and genetic factors on esophageal carcinogenesis**

○ Li Dong Wang

Henan Key Laboratory for Esophageal Cancer Research The First Affiliated Hospital of Zhengzhou University, Zhengzhou, Henan Province 450052

EAUHGS-11 15:25 **大規模ヒト疾患ゲノム解析を通じた疾患病態解明・新規創薬への展望 / Comprehensive strategies to utilize large scale human genetic studies for elucidation of novel disease biology and drug discovery**

○岡田 随象[1,2)]（Yukinori Okada）

1）東京医科歯科大学　大学院医歯学総合研究科　疾患多様性遺伝学分野
（Department of Human Genetics and Disease Diversity, Tokyo Medical and Dental University Graduate School of Medical and Dental Sciences, Tokyo, Japan.）
2）理化学研究所　統合生命医科学研究センター　統計解析研究チーム
（Laboratory for Statistical Analysis, Center for Integrative Medical Sciences, RIKEN, Yokohama, Japan.）

EAUHGS-12 15:45 ***NUDT15* R139C is a risk factor for thiopurine-induced leukopenia**

○ Kyuyoung Song

Department of Biochemistry and Molecular Biology, University of Ulsan College of Medicine, Seoul, Korea

EAUHGS-13 16:05 **薬疹関連ゲノムバイオマーカーの同定と医学的有用性の検証 / Identification of genomic biomarkers associated with cutaneous adverse drug reactions and validation of medical utility of genetic testing**

○莖田 泰誠（Taisei Mushiroda）

理化学研究所 統合生命医科学研究センター
（RIKEN Center for Integrative Medical Sciences）