# 国際プレナリーシンポジウム

### The Evolution of Feathers: Insights from Recent Paleontological and Neontological Data

徐星 (Xing Xu)

Key Laboratory of Vertebrate Evolution and Human Origins of Chinese Academy of Sciences, Institute of Vertebrate Paleontology and Paleoanthropology, Chinese Academy of Sciences, Beijing; Email: xuxing@ivpp.ac.cn 8月20日(木)12:10~12:50 中央大学5号館4・5階 Room 10 (5534)

Feathers are the most complex integumentary structures and their evolution has been one of most attended topics in evolutionary biology and paleontology. Until recently feathers have been considered to be unique to birds, though their presence was inferred in several theropod dinosaurs based on both their close phylogenetic relationships with birds and some osteological correlates, e.g., quill knobs and pygostyles. Since the discovery of the feathered compsognathid theropod Sinosauropteryx in 1996, numerous fossils of many theropod groups and even three ornithischian groups preserving feathers have been recovered from the Jurassic and Cretaceous deposits of northeastern China, Russia, Germany, and Canada. These fossils demonstrate that feathers of various morphotypes have a wide distribution among dinosaurs, and in particular, the presence of monofilamentous integumentary structures in some relatively basal dinosaurs and several pterosaurs led to the hypothesis that the first feathers might have appeared approximately 2.3 million years ago. The phylogenetic distribution of various dinosaur feathers shows an evolutionary trend of increasing complexity closer to birds, concurring with the predictions from a developmental model of living birds, but some of these morphotypes are absent in living birds. Importantly, recent developmental and genomic studies have discovered some feather-specific genes, revealed some molecular mechanisms regulating feather development and regeneration, and provided new comparative data on integumentary structures, which can be combined with fossil data to understand feather evolution. A better understanding of feather evolution requires multifaceted and integrative approaches, yet fossils necessarily provide the final test of any evolutionary model.

### Genetic Basis of Feather Diversity in Birds

李 文雄 (Wen-Hsiung Li)

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8月21日(金) 12:10~12:50 中央大学5号館4・5階 Room 10 (5534)

Feathers have diverse forms with hierarchical branching patterns and are an excellent model for studying the development and evolution of morphological traits. The complex structure of feathers allows for various types of morphological changes to occur. The genetic basis of the structural differences between different parts of a feather and between different types of feather is a fundamental question in the study of feather diversity, yet there is only limited relevant genetic or molecular data on feather development. In the past few years we have conducted (1) studies of the transcriptomes from various parts of feathers, (2) *in situ* hybridization studies of specific keratin genes, (3) ectopic expression of mutant keratin genes, and (4) studies of the effect of knock-down expression of beta keratin genes on feather development. These studies provided abundant data for understanding the genetic basis of feather diversity. In my talk, I shall present a summary of our studies.

#### 分子フェノロジー:植物における遺伝子発現の季節変化

工藤 洋

京都大学生態学研究センター; Email: kudoh@ecology.kyoto-u.ac.jp 8月22日(土)12:10~12:50 中央大学5号館4・5階 Room 10 (5534)

フェノロジーとは、季節に応じて見られる生物現象の研究であり、植物では開花・結実・展葉などが対象と される。例えば、季節的な繁殖同調は個体間の交配に必須であり、実際、どのようなメカニズムで繁殖の同 調がなされているかについての野外研究が欠かせない。私達は、アブラナ科の多年草ハクサンハタザオを対 象に分子フェノロジーの研究を進めている。季節応答を遺伝子発現などの分子遺伝学的手法を用いて研究す るのが分子フェノロジーである。例えば、植物種の多くは、気温の季節変化に応答して一定の時期に花を咲 かせる。ところが、気温の季節変化はあくまで長期的な傾向であり、その実態は大きいノイズを含んだ情報 である。高解像度分子フェノロジー(HMP: high-resolution molecular phenology) データを得ることにより、花 成抑制遺伝子 FLC (FLOWERING LOCUS C) が過去6週間の低温を記憶するかのように調節されることが示 された。さらに、全遺伝子を対象としたトランスクリプトーム、ヒストン修飾解析の結果を紹介する。最後 に、分子フェノロジー研究の展望について議論したい。

# S-01: Sensory genetics, ecology and evolution of primates

8/21 9:00-12:00 会場:Room 1 企画者:Shoji Kawamura (The University of Tokyo), Hiroo Imai (Kyoto University) 使用言語:英語

Primates are generally regarded as visually-oriented mammals, trading a sense of smell for good sight. However, recent studies have questioned this simplistic view and it is not well understood the extent to which senses have evolved interactively, independently or in concert in primates, including humans. For example, the number of olfactory receptor genes is not as clearly differentiated between species with different color vision as once asserted. Among senses, receptors of stimuli for vision, olfaction and bitter/sweet/umami tastes all belong to the G-protein coupled receptor (GPCR) family, for which the genetic mechanism of signal perception is well understood. Thus, it is now possible to explore the evolutionary correlation among different senses in primates by studying these receptor groups. In this symposium, we invite investigators on the cutting edge of the sensory genetics, ecology and evolutionary study on primates to exchange newest findings and discuss the future directions of studies on sensory evolution of humans and other primates.

#### 9:00-9:05 Shoji Kawamura (University of Tokyo): General introduction

9:05-9:30 Shoji Kawamura (University of Tokyo)

#### Significance of color vision diversity in primates inferred from genetic and field studies

9:30-9:55 Amanda D. Melin<sup>1</sup>, Mika Shirasu<sup>2,3</sup>, Yuka Matsushita<sup>4</sup>, Vivek Venkataraman<sup>5</sup>, Jessica, M. Rothman<sup>6</sup>, Kazushige Touhara<sup>2,3</sup> and Shoji Kawamura<sup>4</sup> (<sup>1</sup>Washington University in St. Louis, <sup>2</sup>University of Tokyo, <sup>3</sup>ERATO, JST, <sup>4</sup>University of Tokyo, <sup>5</sup>Dartmouth College, <sup>6</sup>City University of New York)

# Sensory ecology of wild capuchin monkeys (*Cebus capucinus*): Examining the links among fruit signals, nutrition, and primate foraging behavior

9:55-10:20 Yoshihito Niimura (University of Tokyo; ERATO, JST)

#### Evolution of olfactory receptor genes in primates and other mammals

10:20-10:45 Takashi Hayakawa (Kyoto University; Japan Monkey Centre)

#### Dietary adaptation and bitter taste receptor gene evolution in primates

10:45-11:10 Yasuka Toda (Kikkoman Corporation; University of Tokyo)

#### Evolution of the umami taste perception in primates

11:10-11:35 Hiroo Imai (Kyoto University)

# Functional analysis of bitter and sweet receptors of primates by cellular and behavioral experiments

- 11:35-11:55 Discussion (Chaired by Kawamura and Imai)
- 11:55-12:00 Hiroo Imai (Kyoto University): Concluding remarks

# S-03: Genome duplication: integrating comparative genomics, population genetics, and experimentally synthesised polyploids

8/21 9:00-12:00 会場: Room 5

企画者:Kentaro Shimizu (University of Zurich), Jun Sese (AIST)

使用言語:英語

大野乾以来、ゲノム重複は動物・植物・菌類の多様化の主要な原動力であると考えられてきた。過去 150 年 に倍数化によって種分化した新種も知られており、多くの穀物も倍数体である。しかしながら、重複遺伝子 の解析の困難さなどから、倍数体種のゲノム解析は遅れてきた。このシンポジウムでは、別々の手法やタイ ムスケールを用いてすすめられてきたゲノム重複研究を概観し、統合的な研究への展望を目指す。比較ゲノ ム的手法からは、歴史的なゲノム重複と、生物多様化・環境変動との関係が論じられてきた。バイオインフ ォーマティクスの進展により、次世代シークエンサーを用いて重複遺伝子を区別した RNA-seq や多型解析が 可能になり、組み合わさったゲノムのネットワーク進化によって新たな生態学的ニッチェを獲得することが 見えつつある。また、現代に起きた倍数体種分化や実験室での人工倍数体作成は、種分化をリアルタイムで 研究するまたとない機会を提供する。

9:00-9:20 Kentaro Shimizu1, Tim Paape1, Masaomi Hatakeyama1,2, Reiko Akiyama1, Rie Shimizu-Inatsugi1, Satoru Akama3, Jun Sese3, Tanaka Kenta4, Yoshihiko Onda5 (1Univ. Zurich; 2Functional Genomics Center Zurich; 3AIST; 4Univ. Tsukuba; 5RIKEN)
 Introduction to genome duplication and the allopolyploid Arabidopsis kamchatica and Cardamine

spp. as model polyploid species to study ecological speciation

9:20-9:40 OTakashi Makino1, Aoife McLysaght2 (1Tohoku University, 2Trinity College)

Ohnologs in the human genome are dosage balanced and frequently associated with disease 9:40-10:00 Jeffery Fawcett (SOKENDAI)

Significance of polyploidization in the evolution of angiosperms

10:00-10:20 〇小林正樹 1、Kevin Kit Sion Ng 2、Jeffrey A. Fawcett 3、Soon Leong Lee 2、瀬々 潤 1、清水健太郎 4 (1 産総研・創薬基盤、2 FRIM・遺伝、3 総研大・先導科学、 4 チューリヒ大・ 理)

Ancient whole-genome duplication in a tropical tree family, Dipterocarpaceae

10:20-10:40 OHaruki Ochi, Hajime Ogino (Yamagata University)

Genome duplications and evolution of gene expression - What can artificial genome duplicated vertebrate embryos tell us about WGDs? –

10:40-11:00 ○塚谷 裕一1,2、澤田有司3、川出健介2,4、及川彰3,5、平井優美3(1東大・ 院・ 理、2岡崎統合バイオ、3理研・環境資源科学研究セ、4基生研、5山形大・農)

Metabolomics analyses of autopolyploids: Do genome duplications directly affect metabolic pathways?

11:00-11:20 Tetsu Kinoshita (KIBR, Yokohama City Univ.)

Mechanism of hybridization bBarrier in plant endosperm

11:20-11:40 OMasahiro Kanaoka1, Yuri Aoki1, Reiko Akiyama2, RIe Shimizu-Inatsugi2, Kentaro Shimizu2 (1Nagoya University, 2University of Zurich)

Phenotypic plasticity in relation to water environment in allotetraploid Cardamine flexuosa

11:40-12:00 赤間 悟 1,2、清水(稲継) 理恵 3、清水健太郎 3、○瀬々潤 1 (1.産総研・BRD、2. 東 エ 大 ・ ELSI、3. チューリッヒ大・理)

Silencing and ratio changes of homeologs of the allopolyploid *Arabidopsis kamchatica* studied by new bioinformatic workflows HomeoRoq and SIGN

# W-01: Evolution of Modern Humans after "Out of Africa"

8/20 14:10-16:10 会場: Room 1 企画者:斎藤成也(国立遺伝学研究所) 使用言語:英語・日本語

およそ 10 万年前にアフリカから拡散した現代人の進化を、骨と DNA データから論じる。近藤は人骨資料に ついて、木村は肉眼的表現型と DNA の対応について、河合は東北 1000 人ゲノムデータの解析について、 縄 文人の核 DNA ゲノムデータについて、Jinam はネグリトと他の東南アジア人類集団の DNA 解析について、 それぞれ論じる。これらの講演から、主として現代人が東ユーラシアに拡散していった様子を浮かび上がら せることがねらいである。

14:10-14:34 Osamu Kondo (Univ of Tokyo)

#### Evolution of modern humans after "Out of Africa" viewed from hard evidence

14:34-14:58 Ryosuke Kimura (Univ. Ryukyus)

#### Genetic basis of human phenotypic diversity

14:58-15:22 OYosuke Kawai1, Naoki Nariai1, Kaname Kojima1, Yumi Yamaguchi-Kabata1, Yukuto Sato1, Takahiro Mimoro1, Masao Nagasaki1 (1Tohoku Medical Megabank Organization, Tohoku University)

High precession whole genome sequences of 1,070 Japanese individuals

- 15:22-15:46 ○神澤秀明 1、Kirill Kryukov2、Timothy A. Jinam3,4、細道一善 5、佐宗亜衣子 6,7、諏訪元 7、 植田信太郎 6、米田穣 7、田嶋敦 5、井ノ上逸朗 8,4、篠田謙一 1、斎藤成也 3,4(1 科博・人類、2 東海 大・医、3 遺伝研・集団、4 総研大・遺伝、5 金沢大・医、6 東大・理、7 東大・博、8 遺伝研・人遺)
   縄文人核ゲノム分析から見た日本列島人の成立史
- 15:46-16:10 Timothy Jinam, Naruya Saitou (Division of Population Genetics, National Institute of Genetics)
  Tracing human migrations in Southeast Asia using genome-wide Single Nucleotide
  Polymorphisms

# W-09: International activities and collaborations on the evolutionary researches

8/21 14:10-16:10 会場:Room 1 企画者:峯田克彦(KAUST サウジアラビア)・荒木仁志(北海道大学) 使用言語:英語

進化研究のネットワークは世界中に広がっている。本ワークショップでは、海外で活躍中のシニアと若手 PI に講演いただき、海外での進化研究の現状や国際的なネットワークなどについての最新の情報を提供する。 特に、国際的な研究協力の構築を考えている方や、将来海外での研究を考えている若手、学生にとって有用 な情報交換の機会となるようにしたい。

- 14:10-14:15 Introduction
- 14:15-14:40 Masato Yamamichi (1Hakubi Center/Center for Ecological Research, Kyoto Univ., 2Dept. of Ecology and Evolutionary Biology, Cornell Univ.)

#### My postdoc life in Ithaca, or how to survive the long winter

- 14:40-15:05 Kentaro Shimizu (University of Zurich)
- "Internationalization" of the universities, and studying evolution in Japan, USA and Switzerland 15:05-15:30 Takahiro Yonezawa (School of Life Sciences, Fudan University)

#### Looking for the Lost World in China

- 15:30-15:55 Takashi Gojobori, Katsuhiko Mineta (CBRC, KAUST)
  - Arabia Felix!: Expect the unexpected in evolutionary genomics from a scientific community of international heterogeneity

15:55-16:10 General Discussion

## W-13: Endosymbiosis and organellogenesis

8/21 14:10-16:10 会場:Room 5 企画者:中鉢淳(豊橋技術科学大学) 使用言語:日本語・英語

ミトコンドリアや葉緑体といったオルガネラは、太古、真核性単細胞生物に取込まれた細菌の末裔である。 細胞内共生に基づく異系統生物間の融合として究極の例と言えるが、近年、これに匹敵する緊密な関係を築 いた共生系が次々と見つかりつつある。本ワークショップでは、細胞内共生由来オルガネラの進化過程を振 り返るとともに、「新規オルガネラ進化」が進行しつつある、より歴史の浅い共生系について、最新の知見を 共有したい。

14:10-14:40 Kyungtaek Lim (BRD, AIST)

Transitions in rRNA-mRNA interaction during endosymbiosis

14:40-15:10 皿井千裕1、谷藤吾朗2、中山卓郎3、神川龍馬4,5、高橋和也1、石田健一郎2、岩滝光儀6、
 ○稲垣祐司2,3(1山形大・院理工、2筑波大・生命環境、3筑波大・計算科学研究セ、4京大・院人間環境、5京大・院地球環境、6東大・アジア生物資源環境)

## ヌクレオモルフをもつ2種の未記載渦鞭毛藻: 真核藻の細胞内共生を介した葉緑体成立過程を解き明か す新たなモデルとして

15:10-15:40 大熊盛也(理研 BRC JCM)

**シロアリ腸内の原生生物細胞内の細菌の共生と進化** 15:40-16:10 中鉢淳(豊橋技術科学大学・EIIRIS)

動物界のオルガネラ進化

# ポスター発表一覧

### P-01 Heterogeneous characteritics of Conserved noncoding sequences in Eukaryote

Nilmini Hettiarachchi1,3 and Naruya Saitou1,2,3 1School of Life Science, Graduate University for Advance Studies,2 University of Tokyo,3 National Institute of Genetics

#### P-29 Duplication and diversification of amphibian hatching enzyme genes

OMomo H Carlos1, Tatsuki Nagasawa2, Mari Kawaguchi2, Shigeki Yasumasu2 1National University of Sao Carlos, Brazil, 2Sophia University, Japan

# P-37 Genetic diversity of Kazakhstan camel population and its evolutionary relationship with the Arabian camel breed

OXIAOKAITI XIAYIRE1,2; Saitou Naruya1,2,3

1. National Institute of Genetics; 2.SOKENDAI; 3.University of Tokyo

# P-40 Unveiling the relationships and traits evolution within the species of the fern genus Pyrrosia Mirbel (Polypodiaceae)

○VASQUES, D.T. 1、海老原淳2、伊藤元己3

1東京大学大学院・博士課程2年生、2国立科学博物館・植物研究部 陸上植物研究グループ、3 東京大学大学院・教授

### P-57 Cranial shape evolution in adaptive radiation of birds

Masayoshi Tokita1, Wataru Yano2, Helen James3, Arhat Abzhanov1

1 Department of Organismic and Evolutionary Biology, Harvard University, 2 Department of Oral Anatomy, Asahi University School of Dentistry, 3 Department of Vertebrate Zoology, National Museum of Natural History, Smithsonian Institution

# S1-1

Significance of color vision diversity in primates inferred from genetic and field studies

Shoji Kawamura

Department of Integrated Biosciences, Graduate School of Frontier Sciences, The University of Tokyo

Primate color vision is unique, consisting of trichromacy, dichromacy and monochromacy. Trichromacy was generated from ancestral dichromacy via allelic differentiation (e.g. in most New World monkeys) or gene juxtaposition (e.g. in Old World primates) of the L/M opsin gene. The allelic differentiation results in extensive color vision variability in New World monkeys, where trichromats and dichromats are found in the same breeding population. We showed that the ateline New World monkeys renovated the L/M opsin polymorphism by amino acid mutations unique to their lineage so that the spectral separation between the longest-wave and middle-wave alleles was widened, improving chromatic contrast. Contrary to the extensive color vision polymorphism in New World monkeys, Old World primates are less variable. We showed that gibbons (lessor apes) retained uniform trichromacy by the action of natural selection to purge gene conversions between the L and M opsin genes. Overall, our molecular studies reveal the importance of trichromatic vision However, our field behavioral studies do not suggest a uniform advantage of trichromatic color vision. In foraging efficiency for fruits and figs, we have found no apparent superiority of trichromacy over dichromacy in feeding rate. Furthermore, trichromatic capuchin monkeys do not have higher fitness than dichromats based on long-term (26 years) survival and fertility data of a wild population. Together this suggests mixed advantages to dichromats and trichromats. Further interdisciplinary studies on not only opsin genes but also other sensory genes will provide a wealth of data for increasing our understanding of the evolution of primate senses and will generate important advances in the near future.

# S1-3

# Evolution of olfactory receptor genes in primates and other mammals

<sup>O</sup>Yoshihito Niimura

Graduate School of Agricultural and Life Sciences, The University of Tokyo / ERATO Touhara Chemosensory Signal Project, JST  $\,$ 

# S1-2

# Sensory ecology of wild capuchin monkeys (Cebus capucinus): Examining the links among fruit signals, nutrition, and primate foraging behavior

<sup>O</sup>AMANDA D. MELIN<sup>1</sup>, MIKA SHIRASU<sup>2,3</sup>, YUKA MATSUSHITA<sup>4</sup>, VIVEK VENKATARAMAN<sup>5</sup>, JESSICA M. ROTHMAN<sup>6</sup>, KAZUSHIGE TOUHARA<sup>2,3</sup> and SHOJI KAWAMURA<sup>4</sup>

<sup>1</sup>Department of Anthropology, Washington University in St. Louis, <sup>2</sup>Department of Applied Biological Chemistry, University of Tokyo,<sup>3</sup>ERATO Touhara Chemosensory Project, JST, <sup>4</sup>Department of Integrated Biosciences, University of Tokyo, <sup>5</sup>Department of Biological Sciences, Dartmouth College, <sup>6</sup>Department of Anthropology, Hunter College, City University of New York

Through their senses, animals interface with the environment to detect and evaluate food. Yet, our understanding of the sensory ecology of primates is largely limited to the visual system, making us rather blind to the role of the other senses. Although primates are typically viewed as 砺 isual·animals, they routinely sniff and squeeze fruit during assessment before consumption. We present data from a 12-month behavioral study of the foraging behavior of white-faced capuchins (Cebus capucinus), along with olfactory (volatile organic compounds 坊 OCs), haptic (force resistance), visual (spectral reflectance), and nutritional measurements of fruit ripeness of dietary species. We find: 1) differences in the number, diversity and total amount of VOCs in fruits at different stages of mechanical ripeness; 2) that haptic (mechanical) and olfactory (VOC) changes in fruits are significantly related to nutritional ripening (n=7, p=0.0042); and 3) that olfactory and mechanical cues are a better indicator of ripeness than visual (color and size) changes are for some fruit species. Specifically, color changes occur prior to fruit softening, asynchronously with odorant shifts, and before the fruit is nutritionally ripe. These results suggest, for at least some plant species, that color may be used as a long-distance signal to attract frugivores to trees with fruits in stages of advanced ripening, but that foragers must rely on other senses for close-range assessment (touching and sniffing) of individual fruits. This study contributes to our knowledge about the foraging cues available to primates and other frugivores, and how multiple sensory modalities can inform food assessment and selection.

We thank NSERC, Sigma Xi, The Leakey Foundation, The Wenner-Gren Foundation, the Animal Behavior Society, the Japanese Society for the Promotion of Science, ERATO Touhara Chemosensory Project, JST, and Alberta Innovates for funding.

# S1-4

Dietary adaptation and bitter taste receptor gene evolution in primates

<sup>O</sup>Takashi Hayakawa<sup>1,2</sup>

 $^1\mathrm{Primate}$  Research Institute, Kyoto University  $^2\mathrm{Japan}$  Monkey Centre

Olfaction is essential for the survival of mammals. Diverse odor molecules in the environment are detected by olfactory receptors (ORs) expressed in the olfactory epithelium of the nasal cavity. OR genes constitute the largest multigene family in mammals, and the numbers of OR genes vary greatly among species, reflecting the respective species ' lifestyles. For example, higher primates generally have a reduced OR gene repertoires compared with other mammals, reflecting their heavy reliance on vision rather than olfaction. Primates have adapted to various environments, and consequently, structures of their noses, color vision systems, and nocturnal/diurnal habits are diversified. We have previously reported that the reduction of OR gene repertories in primates is not directly linked with the acquisition of trichromatic vision. By comparing OR gene repertories from various primate species, we can infer what factors mainly caused the reduction of OR genes in primate evolution. The OR gene family is also characterized by frequent gene gains and losses during evolution. Recently we have analyzed the diversity in the evolutionary dynamics of individual OR genes using >10,000 OR genes identified from 13 placental mammals. We found that African elephants had a surprisingly large repertoire (~2000) of functional OR genes encoded in enlarged gene clusters. Identification of orthologous gene groups (OGGs) by a newly developed phylogeny-based computational method revealed that some OGGs were highly expanded in a lineage-specific manner, while OGGs showing one-to-one orthology among the 13 species without any gene gains/losses are very rare. These studies provide a basis for inferring OR functions from evolutionary trajectory.

# S1-5

#### Evolution of the umami taste perception in primates

Yasuka Toda

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Among the five basic tastes, umami is sensed by a heteromeric complex of class C GPCRs termed T1R1 and T1R3. T1R1/T1R3 has been considered to be a sensor of protein sources, because the known natural ligands of this receptor are only L-amino acids. T1R1/T1R3 exhibits species-dependent differences in amino acid selectivity. Human T1R1/T1R3 specifically responds to L-Glu, whereas rodent T1R1/T1R3 responds more strongly to other L-amino acids than to L-Glu. We previously revealed the critical mutations that confer the human-type L-Glu specific responses by analyzing the human and mouse chimeric receptors. Among key residues we identified, the electrostatic properties of two residues, which interact with the distal carboxylate side chain of L-Glu, critically affected L-Glu binding. By analyzing the sequences and functional properties of T1R1/T1R3 from three nonhuman primate species, we revealed that macaque and baboon T1R1/T1R3, in which two residues that are critical to L-Glu binding are identical or similar to those in the human receptor, showed high L-Glu activity. On the other hand, squirrel monkey T1R1/T1R3, in which one of two key residues is identical to that in mouse receptor, showed low L-Glu activity. Further analyses of nonhuman primate receptors should reveal when and why the differentiated L-Glu activity was obtained in primates.

Vertebrate bitter taste receptors are encoded by TAS2R genes. TAS2R genes in vertebrate genomes drastically vary in number among species of vertebrates. For example, humans have 26 TAS2R genes and mice have 40 TAS2R genes. Since each bitter taste receptor has the different bitter ligand spectrum, a large number of TAS2R genes allow an individual to recognize many more bitter compounds. Using whole-genome sequences of ~20 species of primates, I showed that duplications of TAS2R genes are particularly evident in the ancestral branches of anthropoids, which may have promoted the adaptive evolution of anthropoid characteristics such as the development of herbivorous characteristics. Population genetics analysis of hominoids (humans, chimpanzees and orangutans) revealed that these anthropoid-specific duplicated TAS2R genes have different forms of natural selection from other TAS2R genes. For example, diversification of TAS2R genes in the western subspecies of chimpanzees may have resulted from balancing selection, whereas purifying selection dominates as the evolutionary form of diversification of the anthropoid-specific TAS2R genes in the eastern subspecies of chimpanzees and the other TAS2R genes were under no obvious selection as a whole. These results suggested that lineage-specific TAS2R repertoire expansions facilitate the recognition of novel toxins in plants during the adaptive radiation of primates.

## S1-6

Functional analysis of bitter and sweet receptors of primates by cellular and behavioral experiments.

Hiroo Imai

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Most animals avoid bitter compounds and prefer sweet compounds. However, some primates ingest food items which are bitter to humans and/or not sweet to humans, suggesting a species-specific sense of taste. To reveal the mechanism of specific taste phenotypes, we conducted functional analysis of taste receptors in cultured cells and behavioral analysis by a two-bottle test. We found species-specific sensitivities of bitter taste receptors, some of which are supported by the behavioral test. For example, Japanese macaques are less sensitive to salicin, a bitter compound in the bark of willow tree, than are humans, due to the change in some amino acid residues that are situated in the putative ligand binding and intracellular regions. On the other hand, the behavioral experiment suggested increased sensitivity of Japanese macaques relative to humans for sweet compounds. These differences in receptor sensitivities highlight the relevant tastes of compounds in the habitat of primates and contribute to their survival and adaptation.

## S3-1

#### Introduction to genome duplication and the allopolyploid Arabidopsis kamchatica and Cardamine spp. as model polyploid species to study ecological speciation

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#### \$3-2

# Ohnologs in the human genome are dosage balanced and frequently associated with disease

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About 30% of protein-coding genes in the human genome are related through two whole genome duplication (WGD) events. Although WGD is often credited with great evolutionary importance, the processes governing the retention of these genes and their biological significance remain unclear. One increasingly popular hypothesis is that dosage balance constraints are a major determinant of duplicate gene retention. We test the dosage balance hypothesis and show that WGD-duplicated genes (ohnologs) have rarely experienced subsequent duplication and are also refractory to copy number variation (CNV) in human populations and are thus likely to be sensitive to relative quantities. This supports the hypothesis of biased retention of dosage-balanced genes after WGD. We also show that ohnologs have a strong association with human disease. In particular, Down Syndrome caused by trisomy 21 is widely assumed to be caused by dosage effects, and 75% of previously reported candidate genes for this syndrome are ohnologs which experienced no other copy number changes. These observations clearly show a persistent resistance to dose changes in genes duplicated by WGD. Dosage balance constraints simultaneously explain gene essentiality and duplicate gene retention after WGD.

Genome duplication, or polyploidization, occurs in animals and fungi, and plants including many crop species. In addition to the critical role of ancient genome duplication in diversification, genome duplication is responsible for up to 15% of the speciation events of flowering plants. Several examples of contemporary polyploid speciation during the past 150 years are described. For example, in Switzerland, a new polyploid Cardamine insueta exploited the man-made meadow habitat with fluctuating water availability. We have developed a novel bioinformatic workflow for polyploid transcriptome named HomeoRoq (Homeolog Ratio and quantification) by using next-generation sequencers. We focus on Arabidopsis kamchatica as an allopolyploid model, which is the allotetraploid derived from A. lyrata and A. halleri, and has a much broader distribution and climatic niche compared to the parental species. We have reported that cold treatment changed the ratio of many homeologous pairs, implying abundant cis-regulatory differences. A. halleri is known as a hyperaccumulator of zinc and other metal ions and survives in contaminated soils. We found that A. kamchatica also accumulated high amount of zinc in the leaves. Many genes known to function in hyperaccumulation showed high expression of halleri-derived homeologs. These data suggest that environmental responses specific for parental species would be safeguarded in the allopolyploid owing to cis-differences, and confer broader environmental responses of allopolyploids.

#### S3-3

# Significance of polyploidization in the evolution of angiosperms

Jeffrey Fawcett

SOKENDAI

Although polyploids were once considered "evolutionary dead-ends", it is now clear that they have played a crucial role in the evolution of plants. Most, if not all angiosperms are derived from a polyploid ancestor, and many lineages have subsequently experienced additional rounds of polyploidization. However, despite the huge contribution of polyploids to plant evolution, it is still highly questionable whether polyploids, in general, have higher adaptability or higher fitness compared to diploids. Indeed, it has been suggested that polyploids are "rarely successful" in evolution. This brings up the following question: why then were certain polyploids successful? What enabled the long-term evolutionary success of certain polyploids? In this talk, I will first give an overview of the history of polyploid research in plants. I will then introduce our previous findings and discuss how periods of unstable environmental and ecological conditions, such as around the Cretaceous-Paleogene extinction event, might have been instrumental in enabling the success of polyploids in the evolution of angiosperms.



**S**3-5

# Ancient whole-genome duplication in a tropical tree family, Dipterocarpaceae

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Dipterocarpaceae, which consists of more than 500 species, is a dominant tree family in the Southeast Asian tropical rainforest. To understand the diversification of this family, we sequenced the genome of one of the ecologically and economically important species, Shorea leprosula. The genome assembly showed that the draft genome contained >40,000 protein-coding genes. Many of these protein-coding genes had similar paralogous genes, and the Ks distribution for the paralogous gene pairs suggested a whole-genome duplication event. Transcriptome data from seven different genera of the same family independently showed the pattern of whole-genome duplication, suggesting that the whole-genome duplication occurred in the ancestor of these dipterocarps. The GO enrichment test showed that a large number of drought response genes retained their paralogous pairs after the duplication event. The large number of drought response genes observed might contribute to the adaptation of dipterocarps in different water availability conditions from aseasonal to seasonal tropics. Our findings suggest that drought has been an important environmental factor for the dipterocarps in the Southeast Asian tropical rainforests where the amount of rainfall is relatively high.

## S3-6

#### 同質倍数体のメタボローム解析:ゲノム倍加は代謝 経路に直接影響するのか

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In plants, autopolyploidy is often seen in natural populations. Some autopolyploid strains are known to exhibit enhanced tolerance against environmental or biotic stresses; this feature has been speculatively attributed to increased amounts of secondary metabolites. Indeed some autopolyploid cultivars have been cherished by their high contents of metabolites.

But there are only few studies that have directly compared the metabolite levels between diploids and autopolyploids. Even in such rare studies, comparisons were often carried out for individuals selected from natural populations; thus it is unclear whether the detected differences are reflecting the direct effects of autopolyploidy or a mere result of additional secondary selections under diverse habitats.

Here we compared the metabolic profiles and amounts between diploids and newly generated autotetraploids of a model species, *Arabidopsis thaliana* to exclude the effects of any secondary selection. As a result, although we could detect some metabolic differences between diploids and autotetraploids, such differences were significantly influenced by genetic background and growth conditions. Thus the formerly reported metabolic differences between diploids and autopolyploids were very likely acquired by secondary selections after establishment of polyploidy.

#### Genome duplications and evolution of gene expression – What can artificial genome duplicated vertebrate embryos tell us about WGDs? –

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It is widely accepted that two rounds of whole-genome duplications (WGDs) occurred in the stem lineage of extant modern vertebrates, followed by a third round in the teleoset lineage. These WGD events have provided many duplicated gene pairs from the set of ancestral genes and also many duplicated cis-regulatory elements (CREs). Many studies have suggested that evolution of genes and CREs after these WGD events has contributed to morphological innovations in vertebrates. Although the significance of WGDs for vertebrate evolution has extensively been recognized, we still do not know what happened immediately after the WGD. Late cold shock gynogenesis in Xenopus tropicalis, a diploid frog, prevents the first mitotic cell division and creates an artificial tetraploid embryo. To investigate what happened after the WGD, we used an artificially genome duplicated vertebrate embryo of X. tropicalis. This provided us with the unique opportunity to understand the initial state of vertebrate evolution. We would like to discuss about features of the genome duplicated vertebrate embryos and the evolution of gene expression following WGDs.

## S3-7

#### Mechanism of Hybridization Barrier in Plant Endosperm

Tetsu Kinoshita

KIBR, Yokohama City Univ.

Species hybridization is a potential source for phenotypic variation and speciation in plants. However, many angiosperm genera have an effective post-zygotic barrier in the endosperm that results in seed abortion. Based on classical genetic analyses and recent studies, it is hypothesized that post-hybridization barrier involves epigenetic mechanisms in the endosperm. To elucidate the molecular basis of this mechanism, we have investigated and compared two types of crosses, i.e. interspecies and interploidy, in Oryza species. We found that those hybridizations induced similar phenotypic abnormalities and altered patterns of the imprinted gene expressions in the endosperm (Plant J. 2011, 2013). Although many characters are similar in each type of cross, an index of rate of nuclear divisions in the syncytial stage of endosperm is different; the nuclear division rates are varied in the interploidy crosses, while change in nuclear division rates are not observed in any interspecific crosses. In addition to this finding, we overcome species hybridization barrier between O. sativa and O. *longistaminata* by manipulation of ploidy level.

# S3-8

# Phenotypic plasticity in relation to water environment in allotetraploid Cardamine flexuosa

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Allotetraploid species possess two sets of genomes from different parental species. It is thought that high plasticity of gene expression in allotetraploid species allows them to show unique and variable phenotypes, however, the relationship between phenotypic plasticity and gene expression pattern is not well understood. In Brassicaceae family, Cardamine flexuosa is a known allotetraploid whose parental species are C. amara and C. hirsuta. In natural environments, C. amara inhabits in a wet environment, whereas C. hirsuta in a dry environment. C. flexuosa inhabits in wide range of environments encompassing wet and dry habitsts. To understand the relationship between adaptation to the environment and gene expression pattern in C. amara, C. flexuosa and C. hirsuta, we investigated phenotypic plasticity of these species grown in different water-related environments, both in field sites and laboratory conditions. We found that stomatal development differed between parental species, and C. floexuosa showed both developmental phenotypes. The plasticity of transpiration rates in C. flexuosa was the biggest among the three species. These data suggest that C. flexuosa dynamically regulates stomatal behavior. In addition, we are analyzing genes expression profiles of three species in different environments.

# **S8−6** がんゲノム進化

<sup>〇</sup>柴田 龍弘 東大・医科研 ゲノム医科学分野

Clonal evolution is the general feature of cancer to explain how cancer cells are generated in a step-wise manner (a.k.a. multi-step carcinogenesis). This concept was originally proposed by the pathological observation of precancerous lesions, early and advanced cancer, but a simple selective sweep model cannot fully explain cancer evolution and would be replaced by more complex ones (e.g. a big bang model or branched evolution model). Recent multi-regional or single cell sequencings have prompted us to regard each tumor not as a simple collection of clonal cells but as a mixture of genetically distinctive sub-populations. By these intrinsic complexities cancer cells can adapt to harsh environmental changes including anti-cancer drugs, which is an inevitable source of therapeutic resistance and should be conquered for better cancer treatment. In this presentation, next -generation sequencing analysis of multi-regional liver cancer tissues will be demonstrated to uncover cancer genome evolution.

# S3-9

# Silencing and ratio changes of homeologs of the allopolyploid Arabidopsis kamchatica studied by new bioinformatic workflows HomeoRoq and SIGN

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Genome duplication with hybridization, or allopolyploidization, occurs commonly in wild and crop plants, and is considered to be a strong force for generating new species. However, genome-wide quantification of homeolog expression ratios was technically difficult because of the high homology between homeologous gene pairs. We have focused on the allotetraploid Arabidopsis kamchatica derived from A. halleri and A. lyrata, which has one of the widest climatic niches in the genus Arabidopsis encompassing lowland hot sandy shores to alpine regions. We first assembled the two diploid parental genomes of A. halleri and A. lyrata, then generated a synthetic allotetraploid, mimicking the natural allopolyploid A. kamchatica. To quantify the homeolog expression ratio using RNA-seq obtained from polyploids, we have developed a new bioinformatic workflow named HomeoRoq. Our new statistical test identified 226 homeologs (1.11% of 20 369 expressed homeologs) with significant ratio changes after cold treatment including RD29B and COR15A. Next, we have developed a method to detect silenced genes based on the distribution of ratio, which does not require arbitrary threshold values. Although polyploidization has been thought to induce silencing, we found that about a quarte of silenced genes in parental diploid were de-silenced in synthetic polyploid A. kamchatica. These methods detected a large number of silenced genes also in polyploid crop species.

Evolution of modern humans after "Out of Africa" viewed from hard evidence

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The sequencing of Neanderthal and Denisovan genomes evidenced that these archaic fossil members genetically contributed at least in part to the origins or formation of modern humans after their "Out of Africa." In contrast to the well-documented dispersals of modern humans into Europe and West Asia, those into eastern Eurasia have remained far from an understanding. This is mainly due to the lack of hard evidence such as archaeological and paleoanthropological records with firm chronological contexts. Recent efforts to excavate new data as well as technical developments for chronological dating enable us to make inferences by gathering and scrutinizing such kind of information. Several lines of inferences, including a more complex population history, possible admixture with local archaic hominins, and variable patterns of modern human behavioral expressions, are reviewed on a few regional units such as South/East Asia, Sahul and Australia, Siberia and Mongolia.

# W1-3

# High precession whole genome sequences of 1,070 Japanese individuals

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## W1-2 Genetic basis of human phenotypic diversity

Ryosuke Kimura

Univ. Ryukyus

We have innate differences between individuals in physical and physiological characteristics. Biological characteristics are also diversified between populations. To understand how the global patterns of biological diversity in humans have generated, it is indispensable to reveal 1) how humans dispersed all over the world, 2) how humans genetically adapted to their environment, and 3) how phenotypes are associated with genotypes. Especially, our research team is working to better understand genetic and phenotypic characteristics of Japanese with special focus on Ryukyu people. We are also tackling studies to identify genetic factors associated with the variation in human visible traits including craniofacial morphology. The final goal of our project is to clarify why and how people acquired their characteristics through their population history. Recent advances in genome technologies and establishments of dense catalogs of the genomic variation have dramatically changed research strategies, enabling us to perform genome-wide association studies (GWAS) and population genomics studies. In this workshop, I will introduce our efforts to understand human phenotypic diversity.

Tohoku Medical Megabank Organization (ToMMo) has conducted the whole genome sequencing of 1,070 participants of the prospective genome cohort study in Miyagi Prefecture. We discovered 12 million novel single nucleotide variants (SNVs) out of 21.2 millions SNVs detected in this study. In addition, 3.4 millions of insertions and deletions and 25 thousands of genic copy number variations were identified. Population genetic analysis of these variants data provide insights into demographic history of Japanese individuals as well as the impact of weak deleterious selection acting on noncoding segments of human genome.

# W1-5

# Tracing human migrations in Southeast Asia using genome-wide Single Nucleotide Polymorphisms

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Anatomically modern humans has been present in the Southeast Asian region (SEA) since at least 40,000 years ago (YBP). Archaeological evidence has suggested that the earliest settlers of the region migrated out of Africa via a southern coastal route. The majority of current SEA populations are thought to have originated from an Out-of-Taiwan expansion approximately 5,000 YBP based on linguistic and archaeological data. Genetic data has been used to support these two migration events. Here we utilized both mitochondrial and autosomal SNP markers to infer demographic events that may have occurred within the long time period between these two migrations and to elucidate the evolutionary forces that has shaped the genetic diversity of current human populations in SEA.

## W8-4

Characteristics of *Triparma laevis* f. *longispina* NIES-3699 with insights to its life cycle

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1国立環境研

Parmales are pico-size siliceous alga mainly found in cold regions, available as culture since 2008. Phylogeny studies revealed their close relationship with *Bolidomonas*, a pico-size naked flagellated alga from warm regions. Both algae have distinctive physiological requirement and the life cycle linking the two is still not elucidated, though highly presumed.

Morphological features of Triparma laevis NIES-2565 and T. laevis f. longispina NIES-3699 grown under different conditions were investigated by light and scanning electron microscopies. Partially silicified cells and naked cells were steadily found in the course of batch culturing of NIES-3699. Non-motile naked cells of about  $3 \mu$  m size could have round shape or displaying unique chloroplast extensions. Large size cells from 6 to  $10 \,\mu$  m often displayed irregular shapes with multi chloroplasts content. Naked flagellated cells similar to Bolidomonas were generated either from the non-motile round cells and the larger cells, with a relative abundance varying according to the culture conditions. Fate and linkage among the different type of NIES-3699 cells were investigated leading to new insights into the Parmales life cycle and opening speculation on the natural population behavior.

## W9-1

# My postdoc life in Ithaca, or how to survive the long winter

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I would like to introduce my personal experience staying in the United States of America as a postdoc, especially focusing how to survive the long winter. After I received a Ph.D. degree in 2012, I worked at Cornell University, Ithaca, New York as a JSPS postdoctoral fellow for research abroad for 2 years. I am interested in eco-evolutionary feedbacks, where ecological and evolutionary processes are interacting dynamically. As Cornell is one of the great research centers on this topic, I enjoyed my stay in Ithaca. However, the only problem was that winter in Ithaca is long and cold: it lasts for 6 months and can be minus 20 degrees Celsius. In this talk, I provide advices to graduate students who are going to stay in such a cold place as a postdoc.

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# W9-2

" Internationalization " of the universities, and studying evolution in Japan, USA and Switzerland

Kentaro Shimizu

University of Zurich

Globalization has influenced basic researches and universities in that universities in many countries are aiming at "internationalization" and are recruiting more and more students and researchers from all over the world. For example, in the University of Zurich in Switzerland, a large proportion of PhD students and PIs came from foreign countries, and English is now the common language in research. I have enjoyed research of Evolution in Japan, USA and Switzerland, and would like to discuss the strength of each. International collaboration is becoming more important and feasible. Human Frontier Science Program originated from Japan has been providing a unique support, and I would like to give an example on the project on genome duplication (see Symp-3: Genome duplication: integrating comparative genomics, population genetics, and experimentally synthesised polyploids).

As a former associate editor of the Newsletter of the Society of Evolutionary Studies, Japan, I started the series of the reports from foreign countries, and I am glad to see that it has continued until 20 times. I hope that the society will continue to serve as an important node of international evolutionary studies.

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# W9-4

Arabia Felix!: Expect the Unexpected in Evolutionary Genomics from a Scientific Community of International Heterogeneity

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W9-3

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Looking for the Lost World in China

Takahiro Yonezawa School of Life Sciences, Fudan University

China is an attractive place for the evolutionary biologist due to following reasons. First, the high level of the biodiversity in China: China is the third largest country in the world (9.6 million km2), spanning from tropical to subarctic zones as well as desert and alpine climate zones. The numerous numbers of the wild species are distributed in such diversified environments. The same holds for the fossil species such as Jehol biota or Chengjiang biota. Second, China is a center of the domestication and cultivation: the domestic animals and cultivated plants are result of the artificial selection, and they are good model of the evolution. During the long history of China, many animals and plants have been domesticated, and many breeds established. It is one of the hot topic in evolutionary biology. Third, the geographical position of China: to understand how Japanese fauna and flora established, the data from Chinese fauna and flora is essential. It also holds on the domestic animals and plants because of the long historical relationships between Japan and China.

However, there are few foreign researchers in China, and the academic life in China is little known to Japanese researchers (see also  $\times$ <sup>2</sup>/<sub>2</sub>, 2011). In this workshop, at first, I want to give a talk about my own evolutionary research in China, and then hope to mention about the academic life style in China.

米澤隆弘(2011)海外研究室だより【第12回】復旦大学・生命科学 学院

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Spending almost one and half years since we joined KAUST (King Abdullah University of Science and Technology) in Saudi Arabia, we have started a comparative study of marine metagenomics between the sea surrounding Japan and the Red Sea in Saudi Arabia. Because of unique characteristics of the Red Sea such as high temperature and salinity in the seawater, we are obtaining very interesting observations of remarkable differences in microorganismic diversities. Since the KAUST community is now composed of different nationalities from more than 100 different countries, our lab members are naturally very international. Here we present our experiences in conducting evolutionary research in such academic environments of heterogeneous ethnic origins, particularly obtaining a full of unexpected insight through intimate discussion. "Arabia Felix" means "Happy Arabia" that describes the southern part of the Arabian Peninsula, which has long enjoyed more productive fields. We enjoy expecting the unexpected in such an international atmosphere, in the hope of attaining a scientific goal of Arabia Felix. We are so glad if we can share some of our experiences with the audience.

## W13-1

Transitions in rRNA-mRNA interaction during endosymbiosis

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By the process of 'endosymbiosis', many bacterial endosymbionts have emerged and further evolved into organelles such mitochondria and plastids (including chloroplasts). An endosymbiotic lifestyle drives unique genomic evolutionary processes such as facilitated gene loss and radical nucleotide compositional bias. Understanding of such endosymbiotic evolution is central to eukaryotic biology and pathology because the most endosymbionts and endosymbiont-derived organelles are essential for eukaryotic metabolism,

As a new perspective on endosymbiotic evolution, here I introduce collective views on evolutionary changes in rRNA-mRNA interaction for translation initiation, called Shine-Dalgarno (SD) interaction. Although SD interaction is considered essential for bacterial genetic system, it has often been lost or altered during endosymbiosis. For example, mitochondria, that have undergone endosymbiosis for approximately 1.5 billion years, do not possess any SD signals except for a clade that features gigantic genome size. Such loss is much less frequent in plastids and endosymbiotic bacteria probably owing to the much shorter endosymbiotic history. Furthermore, the rRNA-side motif for SD interaction has evolved coordinately with mRNA-side SD signals in several plastid lineages, showing unexpected evolutionary plasticity of genetic system during endosymbiotic evolution

# W18-1

The origin of Antarctic terrestrial organisms / 南極 陸上生物の起源

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# W18-2

#### Bioaerosols transported toward the Antarctic/南極へ 輸送されるバイオエアロゾル

Fumio Nakazawa/中澤文男

Transdisciplinary Research Integration Center, Research Organization of Information and Systems (ROIS)/新領域融 合研究センター、情報・システム研究機構

Antarctica is isolated from other continents and is in a closed environment. The Shyowa Station is more than 4000 km away from Africa, being the nearest continent. Some pollen grains have been found in snow and ice collected from the surroundings of the Syowa Station. Also, recent studies on atmospheric aerosol particles have suggested that bioaerosol particles are transported toward the Antarctic away from other continents. This talk will discuss the origin of the airborne pollen grains by analyses using DNA markers.

南極大陸は他の大陸からは隔離された閉鎖的な環境にあ る。日本の観測基地である昭和基地は、最も近いアフリカ大 陸から4,000 km 以上離れている。昭和基地周辺の雪氷中か らは、周辺大陸から飛来したと思われる花粉が見つかってお り、また近年の大気エアロゾル粒子の研究においても、他の 大陸から南極へのバイオエアロゾル粒子の輸送が示唆され てきた。今回、DNA マーカーを用いた解析から、花粉粒子 の起源について考察する。

W18-4

Under the extreme low temperature and water availability, quite poor terrestrial ecosystem has been established on the bare ground in Antarctica. The terrestrial biome is quite simple, with some mosses, lichens, algae as primary producers, and mites, springtails and nematodes as consumers, and bacteria. The origin of these staffs has been discusses and the presence of relict species from the age of Gondwana super continent and the immigrants from other continents during the postglacial period were supposed. In the case of an aquatic moss species composing "moss pillar" in Antarctic lakes, results of molecular systematic and sediment analysis showed that the airborne diaspore of the moss from South America had established in lakes at several thousand years ago.

極度の低温、乾燥のため、夏季には砂漠のような荒野が広 がる南極の露岩地帯では、コケなどの無維管束植物や線虫 などの微小動物が、貧弱な生態系を作っている。南極の陸 上生物の起源については、ゴンドワナ大陸からの遺存種と 後氷期に進入した種があるとされる。南極湖沼底に見られる 「コケ坊主」と呼ばれる特異な植生構造を作るコケの種で は、分子系統学的解析によってその起源は南米であること が示され、数千年前に気流に乗った散布体が南極へ到達し たであろうことが推測された。

## W18-3

#### Bryosphere within an Antarctic moss pillar / 南極の コケ坊主生物圏

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The aquatic moss *Leptobryum wilsonii* forms underwater tower-like structures called "moss pillars" in ultra-oligotrophic Antarctic lakes. Our aim is to understand how such a unique ecosystem succeeds in the extreme environment. Therefore, we used biochemical and molecular methods to elucidate the microflora of aquatic moss pillars, based on fatty acid profile, rDNA genotype, and metagenomic information. The results revealed that eukaryotic organisms, such as algae, fungi, nematodes, and tardigrades were present along with bacterial communities and their functional potential varied in different sections within the pillar. These findings will shed light on the underlying mechanisms involved in maintaining the bryosphere within Antarctic moss pillars.

南極湖底のコケ坊主が現在に至るまでどのようにして維持されてきたのか? 演者らは、微生物学的な見地から、脂肪酸組成、16S/18S rRNA 遺伝子および機能遺伝子群の諸解析を進め、この謎に挑んでいる。結果として、コケ坊主内には、コケ類だけではなく、藻類や菌類、さらにクマムシや線虫のような微小動物が存在することと、その内外上下において細菌叢やその潜在機能が異なることを明らかにしてきた。目には見えない微生物たちがコケ生物圏 (bryosphere)の存立を担う可能性がある。



# Horizontal gene transfer world in an Antarctic lake / 南極湖沼における遺伝子の水平伝播ワールド

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TRIC, ROIS / 融合センター、情報・システム機構

Organisms living on Antarctica have evolved adjusting themselves to the extreme environments and constructed unique biospheres, for example "bio-mat" and "moss pillars" in some of ultraoligotrophic lakes. It has been revealed by our genomics studies of bacteria isolated from an Antarctic lake that a lot of genes were horizontally transferred from distant species. It is suggested that the highest rates of horizontal gene transfers had provided co-evolutions and promoted environmental adaptations of organisms on Antarctica. I will discuss the biospheres of Antarctic lakes as natural laboratories of biological evolutions on the Earth.

南極大陸上の生物は自らを極限環境に適応させながら進化 し、例えば、極貧栄養の湖にバイオマットやコケ坊主といっ た生物圏を成立させることにより生存を可能にしている。 我々が南極湖沼から分離した細菌のゲノム解析からは、多く の遺伝子で遠縁種からの水平伝播が明らかにされた。これ までに無い高い割合での水平伝播が南極における生物の 共進化と環境適応を促してきた結果と考えられる。地球上に おける生物進化の実験室としての南極の湖沼生物圏につい て議論したい。

# W18-6

# Environmental tolerance genes of Antarctic nematode/南極線虫の環境耐性遺伝子

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The Antarctic nematode, Panagrolaimus davidi, tolerates complete water loss and intracellular ice formation. Genome and transcriptome analysis of P. davidi revealed that they have a lot of LEA anti-desiccation genes. It is suggested that LEA genes originate from bacteria, and spread into plants and animals which have desiccation tolerance, by horizontal gene transfer. P. davidi LEAs are classified into 3 types by their subcellular localization, i.e. cytosolic-, secreted- and membrane bound-LEAs. Unusual excess number of the LEA variants in *P. davidi* implies that LEA genes may rapidly evolve to adapt the extreme environment of Antarctica. 南極線虫 P. davidiは、体内の水分の完全な喪失や、細胞内 の凍結に対する耐性を持つ驚くべき生物である。転写産物 解析から、この生物には多様な乾燥耐性関連遺伝子 LEA バリアントが発現していることが判明した。LEA は細菌に起 源を持ち、遺伝子の水平伝播によって乾燥耐性を持つ植 物、動物などに広がったと考えられている。P. davidiの LEA には細胞質型、細胞外分泌型、膜結合型の3種類があり、厳 しい南極環境に適応するために、急速な進化を遂げた可能 性が高い。

# Genome evolution in Arctic and Antarctic bacterial lineages / 北極および南極細菌系統におけるゲノム 進化

Hiroshi Akashi

National Institute of Genetics, SOKENDAI

Polar terrestrial environments are major challenges for low temperature survival. The newly isolated Arctic and Antarctic bacteria, Pseudomonas sp. ArSA and HMP1, are well-adapted to polar environments. Phylogenetic analyses show that cold adaptation has occurred relatively recently and in parallel in these lineages. We compare the genomes of these species with their close, mesophilic relatives to identify lineage-specific genome changes in both nucleotide and amino acid composition that may underlie adaptation to extreme environments. Maximum likelihood methods that account for both nucleotide compositional bias and non-stationary evolution are critical for this analysis.

極域(北極・南極)の極限環境では、低温への適応は生物の生存に必須である。近年新たに単離された2つの Pseudomonas 属細菌 ArSA 株及び HMP1株はそれぞれ北極、南極に固有の系統であり、独立に低温耐性を進化させてきたことが示唆されている。我々は、これら2系統に中温域の近縁種を加えて祖先ゲノムの推定を行なうことで、ゲノムレベルでの低温耐性の進化メカニズムを検証している。非定常的かつ系統特異的な塩基組成の進化を考慮したモデルを用い最尤法による推定を行なうことで、各系統でのゲノム進化を詳細に解析することが可能になる。



## 1D-4

The origin of genes by means of conflicting multilevel evolution

竹内信人

東大·総合文化

In the modern cell, heredity is provided by a small number of template DNA molecules, the gene. How did genes originate? Here, we demonstrate the possibility that gene-like molecules emerge in protocells because of conflicting multilevel evolution. We extended a previous model of protocells that contain replicating catalytic molecules by incorporating strand complementarity. Protocells are selected towards maximizing the catalytic activity of intracellular molecules, whereas molecules tend to evolve towards minimizing it. These conflicting evolutionary tendencies induce spontaneous symmetry breaking between complementary strands of molecules, whereby one strand becomes the majority and serves as catalysts and templates, whereas the other becomes the minority and functions only as templates---like genes. These gene-like molecules substantially increase the evolutionary stability of protocells by reducing an intragenomic conflict. Our results implicates conflicting multilevel evolution in the origin of genetic complexity after the first major evolutionary transition.

### 2A-2

動物多細胞体制の進化に原生生物から迫る (Protists shed light on the evolution of animal multicellularity)

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# 1E-1

Efficient inference of recombination hot regions in bacterial genomes

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<sup>1</sup> Univ. Tokyo (Kurume Univ.), <sup>2</sup> Imperial College London, <sup>3</sup> Univ. Oxford, <sup>4</sup> Swansea Univ., <sup>5</sup> Max Planck Institute In eukaryotes, detailed surveys of recombination rates have shown variation at multiple genomic scales and the presence of "hotspots" of highly elevated recombination. In bacteria, studies of recombination rate variation are less developed, in part because there are few analysis methods. Here we focus in particular on identifying "hot regions" of the genome where DNA is transferred frequently between isolates. We present a computationally efficient algorithm which for the first time enables to explore recombination hot regions by using more than 100 mutually recombining bacterial genomes. We applied our approach to previously analysed Escherichia coli genomes, and revealed that the new method is highly correlated with the number of recombination events affecting each site. We identified three recombination hot regions in Campylobacter jejuni genome, which are enriched for genes related to membrane proteins. Our approach and its implementation, which is downloadable from https://github.com/bioprojects/orderedPainting, will help to develop a new phase of population genomic studies of recombination in prokaryotes

## 2B-1

# Experimental analysis on isochore evolution by using raw mutation data

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Furthermore, we cannot conduct "evolutionary experiments" to validate our models in the conventional framework. An ideal solution would be to observe ongoing evolutionary process by experimentally detecting and collecting raw mutations. Considering the extremely low evolutionary rate, however, this solution is obviously impractical. How metazoans evolved from a unicellular ancestor? This is one of the most important, yet unanswered questions in evolutionary biology. We aim to elucidate the molecular mechanisms that were responsible for the evolution of metazoan multicellularity, both from a perspective of genome/transcriptome informatics and that of molecular biology and genetics. We use the recently-developed protist models, filastereans and ichthyosporeans, the closest relatives to the metazoans after choanoflagellates. The genome of Capsaspora, a filasterean, has revealed that the putative metazoan ancestor before achieving multicellularity had a variety of "multicellularity-relevant" genes that were later co-opted for constructing and maintaining the multicellular system of animals. Similar notion has been obtained from the choanoflagellate genomes. However, choanoflagellates still denv functional analyses including transgenesis despite a decade effort of several groups. We have established the techniques for transforming Capsaspora (although still needs an optimization) and the ichthyosporean Creolimax cells. In the presentation, we first introduce our organisms, and then show our recent advances in functional analysis of the "multicellularity-relevant genes found in protists".

## 2B-2

# Importance of the genomic locations of conserved noncoding sequences in gene regulation

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Functional studies have found the involvement of certain Conserved Noncoding Sequences (CNSs) in the regulation of the closest gene. On the other hand, reported cases of long range enhancer activities and inter-chromosomal regulation suggest that the genomic location might not be important. To test the importance of genomic location, we extracted the CNSs conserved among chicken, human, mouse, dog and cattle. We first show that the CNSs are under purifying selection, and not just mutational cold spots. Next, we show that genomic distribution of CNSs is not random and that the intergenic ones are often found far away from protein-coding genes (gene desert). The distribution pattern, ChIP-Seq and RNA-Seq data exclude the possibility of the CNSs being unannotated genes. The ChIP-Seq data show that the CNSs are more likely to be regulatory element. If the genomic location of the CNSs is important, we would expect to find a pattern in the expression of closest genes. Otherwise, no unique expression pattern would be found. We report a high concordance between the closest gene ontology term and tissue expression level. Gene expression patterns suggest that CNSs regulate nearby genes. Importantly, we show that genes with more CNSs have more conserved expression. Our results therefore suggest that the genomic locations of CNSs are important for their regulatory function.

In the isochore evolution, various analyses were attempted to elucidate its enigmatic mechanism. So far, no clear conclusions were obtained due to contradictory observations against the proposed models.

Many molecular evolutionary studies primarily rely on accumulated mutational and/or substitutional changes in genetic information. This framework is especially powerful to analyze evolutionary characteristics of homologous sequences that independently evolved after their divergence: i.e., their genetic divergence is supposed to directly reflect the real evolutionary process. However, in case of the isochore evolution, this conventional framework often does not work effectively: we have at least two kinds of issues to be considered: (1) the evolutionary signal of interest might be eroded by unknown evolutionary process; (2) we need to handle non-coding regions, to which it is difficult to apply conventional evolutionary models. Especially, (1) is a critical problem because subtle information in non-coding sequences may be easily worn out even during short-term evolution. Furthermore, we cannot conduct "evolutionary experiments" to

Meanwhile, ENU mutagenesis can efficiently produce a rich data set of raw mutations in any species; i.e., in a sense, we can "accelerate" the evolutionary rate and mimic the evolution of the mammalian genome. We analyzed collected raw ENU-induced mutation data in the mouse and found two different mutation pressures, which may explain the isochore evolution in the mammalian genome.

## 2B-3

# Global discovery of Hominidae-specific unique genomic elements in human genome

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Family Hominidae, which includes human beings and great apes, is recognized for unique complex social behavior and intellectual abilities. Despite the increasing genome data, however, the genomic origin of its phenotypic uniqueness has remained elusive. Clade-specific protein coding genes and highly conserved noncoding sequences (HCNSs) are among the high-potential evolutionary candidates being involved in driving clade-specific characters and phenotypes. On this premise, we analyzed whole genome sequences along with gene expression and orthology data retrieved from major DNA databases to find Hominidae-specific (HS) protein coding genes and HCNSs. We discovered that Down syndrome critical region 4 is the only experimentally verified gene uniquely present in Hominidae family. The evolution of DSCR4 which has no structural homology to any known protein is estimated to have happened in several steps through LTR/ERV1, LTR/ERVL retrotransposition, and transversion. Using genomic distance as threshold for neutral evolution, we identified 1,852 HS HCNSs. Polymorphism coverage and derived allele frequency analysis of HS HCNSs showed that these HCNSs are under purifying selection. indicating that they may harbor important functions. In this study, HS HCNSs are shown to be under accelerated evolution in Hominidae common ancestor, overrepresented in promoters, untranslated regions and in close proximity of genes involved in sensory perception of sound and developmental process. They also showed a significantly lower nucleosome occupancy probability. Low HS coding protein-coding gene to HS regulatory HCNS ratio suggests regulation alteration of existing protein-coding genes to have played a more significant role in Hominidae evolution than emergence of novel genes.



### P-1

# Heterogeneous characteritics of Conserved noncoding sequences in Eukaryotes

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Conserved noncoding sequences (CNSs) are enriched in regulatory sequence elements. We conducted a whole genome analysis on plant CNSs and identified them to be GC rich (Hettiarachchi et al. 2014). Babarinde and Saitou (2013) reported mammalian CNSs to be GC poor. This heterogeneity in GC content might be related to varying sequence features of regulatory elements in different lineages. Since animals and fungi are sister groups, in order to determine the evolutionary origin of low GC content of mammalian CNSs we investigated the features of fungi lineage common CNSs. This investigation was further extended to discover the sequence features of lineage common CNSs of invertebrates, non-mammalian vertebrates with the intension to answer varying regulatory features of different lineages, Currently we have identified that plant, fungi, invertebrate lineage CNSs are predominantly GC rich whereas vertebrates are GC poor. We also found that this GC content feature is directly related to their location in the genome. High GC CNSs showed a tendency to be found in heterochromatin regions. whereas low GC CNSs shows a tendency to locate in open chromatin. The transition of high GC content of CNSs from the majority of multicellular eukaryotes to low GC content in vertebrates and the structural architecture of CNSs with its function are some of the questions we intend to answer in the future.

P-29

gEVE, an endogenous viral elements (EVEs) database, facilitates the evolutionary studies of functional EVEs in various mammalian species.

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1東海大・医、2東海大・マイクロナノセンター

In mammals, it is widely accepted that approximately 10% of genome sequences correspond to endogenous viral elements (EVEs) including endogenous retroviruses (ERVs), which are thought to be derived from ancient viral infections of germ cells. A few open reading frames (ORFs) of EVEs are still active and express viral proteins in the hosts. Indeed, several genes derived from EVEs were found to be functional for host species, such as syncytins (envelope proteins of retroviruses) for placenta development in various mammals. However, no databases of EVE ORFs are available, and therefore evolutionary pathways of EVEs have not yet been understood comprehensively. Hereby, we developed EVE ORF databases for 20 genomes of 19 mammalian species. We first identified EVE candidates using RetroTector and Repeat Masker, and then translated the nucleotide sequences. For each EVE sequence (> 80aa), functional viral motif sequences were searched. Then all obtained EVE sequences as well as exogenous and endogenous viral sequences were searched against every genome sequence. Then, comprehensive EVE ORFs identified for each mammalian species are summarized as a database (gEVE,

## P-37

Genetic diversity of Kazakhstan camel population and its evolutionary relationship with the Arabian camel breed

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The genus Camelus contains two species: one-hump camel (Camelus dromedary) which inhabits the Arabia and Africa, and two-hump camel (Camelus bacterianus) which inhabits the Central Asia. However one-hump camels are not only found in Afro-Arabia, but are also inhabitants of Kazakhstan in central Asia. Although it is believed that one-hump camels originated in Arabia, there has been no in-depth study on the comparison of one-hump camel in Arabia and central Asia. To investigate the possible origin of one hump camels as well as the evolutionary relationship between Arabian and Central Asian populations, we determined the sequences of mitochondrial D-loop regions of 17 Kazakhstan camels, including 11 one-hump, 3 two-hump, and 3 hybrid camels. The sequences were analyzed together with the available camel sequences. Our phylogenetic study supports that the Arabian one-hump camels were the ancestral population. Also, the phylogenetic tree shows that the Kazakhstan one-hump camels donot form a single cluster, which probably suggests that Kazakhstan one-hump camel populations are not homogenous. In addition, we confirmed that wild camel (camel bactrianus ferus) and domestic two-hump camel (bactrian camel) are separate lineages. Furthermore, previous studies have demonstrated the usefulness of camelid microsatellite loci as a genetic tool for the study of one-hump and two-hump camels. We would like to further study the genetic diversity and relationships among Kazakhstan camel populations using microsatellite DNA markers.

# Duplication and diversification of amphibian hatching enzyme genes

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The hatching enzymes of vertebrates are conserved molecules that belong to astacin family metallo-proteases. Tetrapod hatching enzymes except mammals are composed of astacin protease domain and CUB domain(s), while fish hatching enzymes are only protease domain. In avians, it was reported that the two hatching enzyme gene homologues were found in their genome. However, at least thirteen hatching enzyme gene homologues were found in Xenopus laevis genome, and two of them were expressed in hatching gland cells. In present study, the expression of four Xenopus hatching enzyme gene homologues was investigated by PCR and in situ hybridization methods. Some of their genes were detected expression at the tissues other than hatching gland cells, such as neural tube and cement gland. The results suggest that duplication followed by functional diversification of hatching enzyme genes have occurred in amphibian lineage.

## P-40

#### Unveiling the relationships and traits evolution within the species of the fern genus Pyrrosia Mirbel (Polypodiaceae)

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Pyrrosia stands for a genus of Paleotropical ferns nested within the Polypodiaceae family and usually with simple and undifferentiated leaves, being found from Central Africa to Southeast Asia, Oceania and New Zealand. Although some represents of Pyrrosia have been explored in other investigations before, a conclusive global phylogeny for the genus is still not available. The main goal of this research was to generate a molecular phylogeny for the genus Pyrrosia, and thus to investigate the evolution of traits within its species. Sequences for three chloroplast genes (rbcL, rps4-trnS intergenic spacer and atpB) from several species of Pyrrosia were analyzed and compiled into a phylogenetic tree. Additionally, morphological data from the different species were gathered through literature and compared to the acquired phylogeny. As a result, infra-generic groupings proposed before by other authors appear to be non-monophyletic. Instead, 3 big lineages emerged in the present analysis, and thus a new infrageneric classification is proposed. Data regarding the morphology of leaves, rhizomes and appendices present in these plants are explored under this phylogenetic hypothesis. Hence, changes in the traits are discussed under the newly proposed classification, some of them being indicated as possible synapomorphies for the discovered groups.

#### Cranial shape evolution in adaptive radiation of birds

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Adaptive radiation is the rapid evolution of morphologically and ecologically diverse species from a single ancestor. The two classic examples of adaptive radiation are Darwin's finches and Hawaiian honeycreepers, which evolved remarkable levels of adaptive cranial morphological variation. To gain new insights into the nature of their diversification, we performed comparative morphometric analyses supplemented with molecular phylogenies on a multivariate morphospace. We show that cranial shapes in both Hawaiian honeycreepers and Coerebinae (Darwin's finches and their relatives) are much more diverse than in their outgroups but Hawaiian honeycreepers as a group display even higher diversity and disparity than coerebins and all other tested bird groups, most likely due to their significantly lower integration of skull modules. These findings help to better understand the nature of adaptive radiations in general and provide a foundation for future investigations on the molecular mechanisms underlying diversification of these morphologically distinguished groups of birds.

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