

Genes, Genomes and Genomics

Abbreviation: Genes Genom. Genomics

Print: ISSN 1749-0383

Scope and target readership: *Genes, Genomes and Genomics* focuses on the analysis of cellular and developmental systems using classical genetics, molecular genetics, genomics, functional genomics – and combinations thereof – in humans, animals, higher plants, yeasts, fungi, algae, protists and cell organelles.

Genes, Genomes and Genomics covers: biochemical and regulatory networks; bioinformatics (computational biology and databases); biomedicine; functional and structural genomics, mapping, organization, expression and evolution of genes/genomes; molecular and cell biology including comparative; molecular enzymology, molecular virology and molecular immunology; physics and physical chemistry of proteins (and proteomics) and nucleic acids; transcriptomics. Other fields of interest are DNA replication, transcription, nucleic acid-protein interaction, RNA processing, intracellular transport, protein biosynthesis. Molecular aspects of genetic variation and evolution, mutation, gene action and regulation, immunogenetics, somatic cell genetics, and nucleic acid function in heredity and development, and biochemical aspects of genetic defects fall into the scope of this journal.

Genes, Genomes and Genomics in particular aims to highlight advances in human genetics: bioinformatics; chemical genomics, cytogenetics and genomic imaging; developmental genetics; ELSI (ethical, legal and social issues); evolutionary genetics; gene structure, organization and expression; genetic epidemiology; genetics of complex diseases and epistatic interactions; genome structure and organisation; genotype-phenotype relationships; identification of genes involved in disease and complex traits, including responses to drugs and other xenobiotics; linkage analysis and genetic mapping; molecular diagnostics; molecular genetics of tumorigenesis; mutation detection and analysis; physical mapping.

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Cover photo: A hypothesized pathway for mitochondrial-nuclear inter-actions inducing cytoplasmic male sterility in higher plants. More details in Yang and Zhang, pp 21-26.

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Printed in Japan on acid-free paper.

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Magda-Viola Hanke, Henryk Flachowsky, Andreas Peil, Conny Hättasch (Germany) No Flower no Fruit – Genetic Potentials to Trigger Flowering in Fruit Trees (pp 1-20)

ABSTRACT

Special Feature: The development of flower buds and a sufficient fruit set is a basic requirement for fruit growers to generate a marketable crop. However, fruit trees remain in a juvenile (nonflowering) phase for years, and after a transition period of getting reproductively competent they enter the adult phase of tree life. The reproductive phase is associated with the ability to alternate between the production of vegetative and reproductive buds. Efficient breeding is limited in fruit trees due to the long period of juvenility. Therefore, it is important to accelerate flowering by reducing the juvenile phase of the tree. In fruit production, precocious flowering of the tree is also favoured to reduce the vegetative phase of tree development after planting in order to obtain the earliest fruit crop. Additional critical aspects of flower development in fruit trees, such as alternate bearing, accentuate the necessity to improve our understanding on genetic factors controlling floral initiation as well as flower and fruit development in perennial fruit trees. Most of what we know about regulating floral development is based on research in annual plants, like *Arabidopsis thaliana*. In this review, we summarize floral transition, meristem development and flower bud formation in *Malus domestica*, one of the most important representatives of temperate fruit trees. We also focus on current findings of the transition from vegetative to reproductive growth obtained in *Arabidopsis* and how this knowledge can be applied to fruit trees, particular to apple. We discuss state-of-the-art and future research to manipulate maturation and flower initiation in apple.

Jing-Hua Yang, Ming-Fang Zhang (China) Molecular Genetics of Mitochondrial Respiratory/ATP-related Genes in Relation to Cytoplasmic Male-Sterility of Higher Plants (pp 21-26)

ABSTRACT

Invited Mini-Review: With the sequencing of the entire mitochondrial genome for some species, including *Arabidopsis thaliana*, *Brassica napus*, *Beta vulgaris*, *Marchantia polymorpha*, *Oryza sativa*, and *Zea mays* it is promising to explore the molecular genetics of mitochondrial respiratory-related genes in relation to some special traits with an emphasis on cytoplasmic male-sterility unique to plant mitochondria. Mitochondrial genomes encode only partial genetic information required for their biogenesis and function, however the vast majority is nuclear-derived, which is encoded by the nucleus and imported into mitochondria through the transported proteins. Consequently, most mitochondrial genetic and biochemical features displayed in plant mitochondria arise from the context of nuclear-mitochondrial co-evolution. Cytoplasmic male sterility (CMS), one useful trait for heterosis in agricultural production, is considered to be the result of the interactions between the mitochondrion and the nucleus in which the former exhibits the role of a tuning fork in nuclear gene expression that induces abnormal reproductive development. Likewise, expression of some mitochondrial genes is also regulated by nuclear genes in which the expression of some CMS-associated genes is influenced by nuclear restorer lines. In this review, we attempt to address the molecular genetics of mitochondrial respiratory/ATP-related genes concomitant with the occurrence of CMS. Furthermore, how the mitochondrion and nucleus interact on a molecular level to influence gene expression and function are also discussed in detail.

Ken-ichiro Yamashita, Masayoshi Shigyo, Shin-ichi Masuzaki, Shigenori Yaguchi (Japan), Tran Thi Minh Hang (Vietnam), Naoki Yamauchi, Sulistyaningsih Endang, Yosuke Tashiro (Japan) The Application of Alien-Chromosome Addition Lines and Cytoplasmic Substitution Lines to Studies on Genetics and Breeding in *Allium cepa* (pp 27-34)

ABSTRACT

Invited Mini-Review: In edible Alliums, shallot (*Allium cepa* L. Aggregatum group, $2n=2x=16$, genomes AA) is an important vegetable crop in South-East Asia and has the highest adaptability to tropical and sub-tropical zones. In diploid *Allium* species, we established eight monosomic additions, representing eight different chromosomes of shallot in a background of Japanese bunching onion (*A. fistulosum* L., $2n=2x=16$, FF). The monosomic additions ($2n=2x+1=17$, FF+nA) were used for genetic analyses of shallot and for improving *A. fistulosum* cultivars. Genetic analyses identified 48 chromosome-specific genetic markers in shallot. The effect of extra shallot chromosomes, on morphology and fertility of *A. fistulosum*, was also identified. Furthermore, onion linkage groups were successfully assigned to *A. cepa* chromosomes by using the *A. fistulosum* – shallot monosomic additions. To develop shallot CMS lines with wild species cytoplasm, we conducted cytoplasmic substitution

between *A. galanthum* and shallot by continuous backcrossing. In a BC₁ generation, male sterile plants appeared, and the male sterility was maintained in a BC₃ generation stably. RFLP analysis of chloroplast DNA confirmed that the lines inherited cytoplasm from *A. galanthum*. Our results demonstrated that the development of shallot CMS lines was possible by this genetic approach. We also induced haploid and doubled haploid plants of F₁ hybrids between the CMS shallot and bulb onion by using unpollinated flower culture. This review paper was prepared, primarily, to introduce the results so far obtained for the gene analyses of *A. cepa* using a complete set of *A. fistulosum* – shallot monosomic additions and for development of shallot CMS lines.

Martin Trick, Ian Bancroft (UK), Yong Pyo Lim (Korea) The *Brassica rapa* Genome Sequencing Initiative (pp 35-39)

ABSTRACT

Invited Mini-Review: *Brassica rapa* ssp. *pekinensis* (Chinese cabbage), a member of the Brassicaceae, is both an economically important crop and a model plant for the study of polyploidization and phenotypic evolution. Its genome (the *Brassica* A-genome) is being sequenced by the Multinational *Brassica rapa* Genome Sequencing Project (MBrGSP), an international consortium of six countries (Korea, Canada, UK, China, USA, and Australia). This consortium has developed a number of genomic resources, including 2 genetic mapping populations, 2 BAC libraries, 22 cDNA libraries, 107,280 BAC-end sequences, and 104,914 ESTs and has constructed a genetic and physical map of *B. rapa* using them. As a strategy for sequencing the genespace, the project has adopted a BAC-by-BAC sequencing approach. In order to implement this, 629 seed BACs mapped genetically throughout the genome have been selected and sequenced. At this preliminary phase of the project, eight of the ten chromosomes have been allocated between the participants; Korea (R3 and R9), Canada (R2 and R10), UK/China (R1 and R8), USA (R6) and Australia (R7) and they are being separately sequenced. In this article, we assess the current status of the *B. rapa* genome sequencing project, including the available and developing computational and bioinformatics infrastructure.

Svetlana B. Gontcharova, Andrey A. Gontcharov (Russia) Molecular Phylogenetics of Crassulaceae (pp 40-46)

ABSTRACT

Invited Mini-Review: Crassulaceae is the most species-rich family (ca. 1400 spp.) of the Saxifragales and the majority of its members are succulents. Great diversity of morphology, cytology and habit complicates systematics of the family and the relationships between species and genera remain poorly understood. Studies using various molecular markers placed Crassulaceae as one of Saxifragales crown groups and showed close relationships between the family and Haloragaceae, lacking any phenotypic background. Earlier molecular data analyses established a number of clades in the family and revealed a disagreement between the traditional taxonomic structure of Crassulaceae and the pattern of phylogenetic relationships between its members. In this paper, we review the major contributions to the phylogeny of Crassulaceae based on molecular data, with emphasis on the major clades established in the family, the clades' structure and polyphyly of some genera. We describe the areas of conflict and agreement between molecular phylogenies and stress that morphological characters provide little evidence for inferring relationships between taxa even at low taxonomic levels.

Ayman A. Diab, Ramesh Kantety, Carlos Mauricio La Rota, Mark E. Sorrells (USA) Comparative Genetics of Stress-Related Genes and Chromosomal Regions Associated with Drought Tolerance in Wheat, Barley and Rice (pp 47-55)

ABSTRACT

Invited Review: An integrated barley consensus map was constructed and used with durum wheat and rice maps to develop comparative genetic maps for durum wheat, barley and rice. Comparative maps were constructed in three stages, each adding a new layer of information. In the first stage, comparative maps were constructed based on common markers present in durum wheat (A and B genomes) and barley genomes compared to rice maps. In the second stage, the marker sequences were matched according to sequence similarity across species. In the third stage, the sequences of drought candidate genes and differentially expressed sequence tags (dESTs) were compared to Bacterial Artificial Chromosome/Phage Artificial Chromosome sequences of the rice genome. The analysis of stress-related genes and dESTs in durum wheat, barley and rice revealed that the genetic response to drought stress is partially conserved among these species. Comparative maps identified conserved genomic regions that are associated with quantitative trait loci (QTLs) for drought tolerance in durum wheat, barley and rice. Some QTLs were unique to only one species, whereas other QTLs for related traits were co-located in all three species.

Shakeel Ahmad Jatoi (Japan/Pakistan), Akira Kikuchi, Kazuo N. Watanabe (Japan) Genetic Diversity, Cytology, and Systematic and Phylogenetic Studies in Zingiberaceae (pp 56-62)

ABSTRACT

Invited Mini-Review: Members of the Zingiberaceae, one of the largest families of the plant kingdom, are major contributors to the undergrowth of the tropical rain and monsoon forests, mostly in Asia. They are also the most commonly used gingers, of which the genera *Alpinia*, *Amomum*, *Curcuma*, and *Zingiber*, followed by *Boesenbergia*, *Kaempferia*, *Elettaria*, *Elettariopsis*, *Etlingera*, and *Hedychium* are the most important. Most species are rhizomatous, and their propagation often occurs through rhizomes. The advent of molecular systematics has aided and accelerated phylogenetic studies in Zingiberaceae, which in turn have led to the proposal of a new classification for this family. The floral and reproductive biology of several species remain poorly understood, and only a few studies have examined the breeding systems and pollination mechanisms. Polyploidy, aneuploidy, and structural changes in chromosomes have played an important role in the evolution of the Zingiberaceae. However, information such as the basic, gametic, and diploid chromosome number is known for only a few species. In this review we highlighted the need to undertake concentrated efforts to address poorly known areas and perform phylogenetic studies to achieve a consensus on the number of genera and species in the family; investigate the pollination biology and breeding system in members of this family; conduct cytological studies to acquire reliable information regarding basic, gametic, and somatic chromosome numbers and ploidy levels in different species; and perform genetic diversity studies to determine the genetic base of the existing genetic resources in the Zingiberaceae.

Yoshinori Kanayama, Kazuhisa Kato, Ryo Moriguchi (Japan) Genetic and Molecular Aspects of *Gypsophila* (pp 63-65)

ABSTRACT

Invited Mini-Review: The genus *Gypsophila* is a member of the family Caryophyllaceae, which also includes carnation (*Dianthus caryophyllus*). Phylogenetic analyses of the Caryophyllaceae have been performed using DNA markers, chloroplast DNA, and rDNA sequences. *Gypsophila* includes more than 100 species, which are distributed mainly in Eurasia. The long-day perennial *G. paniculata* and the annual *G. elegans* are popular in floriculture. In particular, *G. paniculata* is produced in large quantities for use in flower arrangements. The molecular mechanism of flowering has been extensively studied in *Arabidopsis*, a qualitative long-day plant. Many of the genes that regulate flowering time on long-day induction have been characterized. Among them, *CONSTANS* (*CO*) is a key genetic component of the long-day-dependent flowering pathway. Recent studies have suggested that at least four *CO* homologs (*GpCOLs*) are expressed in *G. paniculata*. Each *GpCOL* contains a CCT (*CO*, *CO*-like, *TOC1*) domain near the carboxyl terminus. Phylogenetic analysis of the CCT domain primary sequence indicates that the four *GpCOLs* are Group I *CO*-like proteins. The expression of two of the *GpCOLs* oscillates daily, suggesting a relationship between the *GpCOLs* and flowering in *G. paniculata*. Only a few sequences from *Gypsophila* are available in DNA databases for phylogenetic analyses, including sequences from chloroplast DNA and rDNA, as well as genes involved in anthocyanin formation. Therefore, additional studies are needed at the molecular and genetic levels.

Jaime A. Teixeira da Silva (Japan), Hanna Bolibok, Monika Rakoczy-Trojanowska (Poland) Molecular Markers in Micropropagation, Tissue Culture and *In Vitro* Plant Research (pp 66-72)

ABSTRACT

Review: Molecular marker technologies have become a powerful tool in crop improvement through their use in germplasm characterization, linkage mapping and molecular breeding. *In vitro* plant research has become one of the areas of extensive molecular marker application, as they can be used to monitor the somaclonal variation, verify the genetic fidelity of micropropagated plants and to identify genotypes with desired response to *in vitro* culture conditions. The objective of this paper is to summarize literature concerning application of various molecular markers for genetic fidelity assessment of *in vitro* cultured plants.

Katsuhiro Shiratake (Japan) Genetics of Sucrose Transporter in Plants (pp 73-80)

ABSTRACT

Invited Review: Sucrose is one of the most common and abundant carbon forms in plants. Most plants synthesize sucrose as a major photosynthetic product and use it for long distance carbon transport. Therefore sucrose transport in plants probably is highly regulated and sucrose transporters have indispensable roles in the regulation. In the *Arabidopsis* genome, 69 sugar

transporter homologues have been found, 9 of which are in the sucrose transporter SUC/SUT family. The SUC/SUT family is further divided into three subfamilies based on homology: SUC2/SUT1, SUC3/SUT2 and SUC4 subfamilies. Gene structures, protein structures, kinetics of sucrose transport and subcellular localizations differ between these three subfamilies. Sucrose transporter genes have been isolated from many different plants and their expressions, regulations and physiological roles have been studied. This review summarizes these studies of sucrose transporters.

Antoine X. Deniau, Henk Schat, Mark G.M. Aarts (The Netherlands) Genetics and Genomics of the Heavy Metal Hyperaccumulator Model Species *Thlaspi caerulescens* (pp 81-88)

ABSTRACT

Invited Mini-Review: In the last decade heavy metal hyperaccumulator plants have been increasingly studied, mainly because of their potential use in phytoremediation. *Thlaspi caerulescens* is an attractive model hyperaccumulator plant, because it accommodates a high of intra-specific variation in the degrees and metal-specificity patterns of tolerance and accumulation. In this review we give an overview of recent progress made in the genetics and genomics of heavy metal hyperaccumulation in this species. QTL analysis for zinc and cadmium accumulation in segregating inter-accession crosses demonstrated that these traits are controlled by multiple genes and that there are accession-specific accumulation mechanisms with distinct metal-affinity patterns. Cross-species transcriptome analyses have revealed a large number of genes with differential expression between hyperaccumulators and non-hyperaccumulators. Many of those genes are known to be involved in metal homeostasis, and an even larger number might play a role in this process. However, most of the differentially expressed genes have probably no role in metal homeostasis, owing to the fact that species with different life history and ecology are compared. To confirm the role of candidate genes, mutant research is necessary, but not yet done in hyperaccumulators. In the absence of physical maps and full genome sequences of hyperaccumulators, comparative genomics are indispensable. Co-linearity and micro-synteny analysis should enable the identification of the genes responsible for QTL for accumulation traits in intra- and inter-specific crosses.

Bernard A. Kunz (Australia), Wei Xiao (Canada) DNA Damage Tolerance in Plants via Translesion Synthesis (pp 89-99)

ABSTRACT

Invited Mini-Review: Arrest of replication forks at sites of DNA damage may disrupt the cellular replication machinery leading to cell death. Consequently, cells have evolved damage tolerance mechanisms that do not remove damage but allow replication through or around DNA lesions, which can be repaired subsequently. Damaged templates can be copied by translesion synthesis (TLS), or the damaged segment may be avoided by template switching during replication. Non-essential, low fidelity DNA polymerases catalyse TLS in yeast and mammalian cells. Mechanisms for targeting TLS polymerases to stalled replication forks include interaction with the sliding clamp proliferating cell nuclear antigen (PCNA). Regulation of this interaction and the mode of damage tolerance involves post-translational modification of PCNA, TLS polymerase stability and DNA damage surveillance genes. SUMOylation of PCNA at lysine-164 prevents recombination at blocked forks and so may participate in tolerance pathway selection. Monoubiquitylation of the same residue is necessary for TLS, and polyubiquitylation at lysine-164 promotes damage avoidance. Surprisingly, much less is known about damage tolerance and its importance in plants despite their obligate exposure to a major environmental source of DNA damage, solar ultraviolet (UV) radiation. Recent isolation and functional characterisation of cDNAs encoding *Arabidopsis thaliana* homologues of TLS polymerases or PCNA-modifying enzymes suggest that plants may rely in part on damage tolerance to help combat the onslaught of UV photoproducts.

Jean-Luc Jestin (France), Achim Kempf (Canada) Degeneracy in the Genetic Code: How and Why? (pp 100-103)

ABSTRACT

Invited Opinion Paper: In the genetic code, which is nearly universal among all known organisms, most amino acids are coded for by more than one codon. For example for half of the genetic code's sixty-four codons, the corresponding amino acid is independent of the codon's third base. Interestingly, this degeneracy of the genetic code clearly reduces the deleterious effects of base substitutions at the third codon base. The genetic code possesses a significant number of further degeneracies and the question arises what the structure of the genetic code may be able to tell us about the circumstances of the genetic code's evolutionary origin. Here, we review selected articles in this context, beginning with works on the likely relatively recent origin of the small differences in the present-day genetic codes. We then review work that uncovered characteristic patterns in the genetic code, followed by work that describes the underlying symmetries in terms of mathematical models. Finally, we address

the question what the structure of the genetic code might be able to tell us about the circumstances of the genetic code's evolutionary origin and therefore about the origin of life itself.

Michèle Amouyal (France) Transition from DNA Looping to Simple Binding or DNA Pairing in Gene Regulation and Replication: A Matter of Numbers for the Cell (pp 104-112)

ABSTRACT

Invited Mini-Review: Repression of the *E. coli* lactose operon is achieved though DNA looping and three operators at the physiological repressor concentration. In strains overproducing the repressor or with plasmids with a high copy number, the cooperative mode of repression is masked by other modes. *In vitro*, when several DNA molecules are present, DNA loop formation is replaced by intermolecular associations still mediated by the *lac* repressor. In bacteria, such associations, known as "handcuffing" and mediated by the initiator protein, are observed in replication of the iteron-class of plasmids. When moderate amounts of initiator and its binding to the replication origin (achieved in some instances, by DNA looping) allow replication to proceed, high concentrations prevent replication and lead to handcuffing that controls the number of plasmids. In principle, when DNA looping is feasible, DNA pairing is also possible if more than one DNA molecule is present in the cell. In eukaryotes, the action of the CTCF protein is particularly representative of this situation. This key component of elements that insulate gene expression from the surrounding genomic effects in vertebrates, also acts as an organizer of higher-order chromatin structures at the β -globin and *Igf2/H19* loci. At this latter locus, CTCF controls genomic imprinting by DNA looping. Recent data suggest that genomic imprinting and monoallelic expression might also be controlled through chromosome pairing.

Long-Zheng Chen, Jin-Feng Chen (Russia) Allopolyploid-Induced Sequence Elimination (pp 113-117)

ABSTRACT

Invited Mini-Review: Allopolyploids, which result from the combination of two or more copies of differentiated genomes, have undergone DNA sequence elimination in the early stage after their formation. Recent investigations have illuminated that sequence elimination is a nonrandom and reproducible event. It can be characterized by rapid, nonrandom, and reproducible event, which vary in different plant systems. Many studies have found that the eliminated sequences involve not only low-copy and non-coding DNA sequences but also high-copy and coding sequences, mostly from one of the parental genomes. Although the molecular mechanisms of sequence elimination are not very clear at present, many studies have indicated that it is not affected by the genotype of parental plants, by the cytoplasm, or by the ploidy level, and that it does not result from intergenomic recombination. However, our recent study indicated that sequence elimination might have some relationship with intergenomic recombination in the *Cucumis* group, complicating the mechanisms of the elimination event. It has been speculated that sequence elimination increases the differentiation of homoeologous chromosomes at the polyploid level, thereby providing the physical basis for rapid restoration of diploid-like chromosome pairing pattern in meiosis following polyploidization, and this may have contributed to the successful establishment of newly formed allopolyploids as a new species. Continued application of molecular genetic approaches to study novel genotypes or new allopolyploid species is needed to clarify the issue of allopolyploid-induced sequence elimination, moreover the genome evolution of allopolyploids.

Trees Ubbink-Kok, Juke S. Lolkema (The Netherlands) Heterologous Expression of the Subunits of the Na⁺ V-ATPase of the Thermophile *Caloramator fervidus* (pp 118-122)

ABSTRACT

Original Research Paper: The V-type ATPase of the thermophile *Caloramator fervidus* is an ATP-driven Na⁺ pump. The 9 genes in the *ntpFIKECGABD* operon coding for the 9 subunits of the enzyme complex were expressed separately in *Escherichia coli*. Except for subunit G, all subunits were produced. The main V₁ subunits A and B and central stalk subunit C were produced as soluble proteins, while the main V₀ subunits I and K ended up in the membrane fraction following cell fractionation. Stalk subunit E had a strong tendency to aggregate and was distributed over the soluble and membrane fractions, while subunit D was produced in inclusion bodies. Expression of subunits D and E in the Gram positive host *Lactococcus lactis* reduced aggregation and breakdown significantly, but subunit D was still produced in inclusion bodies.