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INVESTIGATIONS AND AUTHOR SUMMARIES

- 1–14 **Specific Cooperation Between Imp- α 2 and Imp- β /Ketel in Spindle Assembly During *Drosophila* Early Nuclear Divisions**
Erika Virágh, Mátyás Gorjánác, István Török, Tolga Eichhorn, Sowjanya Kallakuri, Tamás Szlanka, István Kiss, and Bernard M. Mechler
- Importins and RanGTP mediate nuclear protein import and have a critical role in the mitotic process. In *Drosophila*, both *imp- α 2* and *imp- β /Ketel* genes were identified by their mutations causing female sterility. However, their roles in spindle formation during early embryonic mitoses were masked by their strong phenotype in earlier developmental stages. By analyzing genetic interaction between specific mutant alleles, we have demonstrated their critical function in the preblastoderm embryo in mitotic spindle, centrosome, and nuclear envelope organization. *imp- α 2* plays specific role in these processes which could not be substituted by *imp- α 1* or *imp- α 3*.
- 15–22 **Whole-Genome Sequencing to Identify Mutants and Polymorphisms in *Chlamydomonas reinhardtii***
Susan K. Dutcher, Linya Li, Huawen Lin, Leslie Meyer, Thomas H. Giddings, Jr., Alan L. Kwan, and Brian L. Lewis
- Chlamydomonas reinhardtii* is a unicellular, green alga that has served an important model for studying human diseases caused by ciliary defects. Human ciliopathies range from chronic respiratory infections to obesity to learning disabilities. We have used whole-genome sequencing to identify the causative mutants in two previously isolated mutants with flagellar assembly defects. The mutation in the IFT80 genes causes a novel defect in the basal body that may represent a new intermediate in ciliary/flagellar assembly. Whole-genome sequencing in combination with limited mapping allows fast and accurate identification of point mutations in *Chlamydomonas*.
- 23–28 **Genetic Analysis of Fibroblast Growth Factor Signaling in the *Drosophila* Eye**
T. Mukherjee, I. Choi, and Utpal Banerjee
- We report an uncharacterized function of *Drosophila* FGF ligand, Branchless and its receptor, Breathless, during morphogenetic movements in the eye and during retinal basal glial cell migration. The results presented here establish a dual function of FGF at a single point of time, one at the neuronal level, where it promotes proper formation of ommatidial clusters by regulating junctional proteins and second at the level of glial migration. For both functions, ligand is secreted from the same tissue but the receptors on distinct cell types interpret it differently. This new insight makes our work relevant to the general readership of G3.
- 29–34 **Natural Genetic Variation for Growth and Development Revealed by High-Throughput Phenotyping in *Arabidopsis thaliana***
Xu Zhang, Ronald J. Hause, Jr., and Justin O. Borevitz
- In the post genomic era, genetic dissection of complex traits, such as growth and development, is now largely dependent on the accurate quantification of many component sub traits. High-throughput phenotyping approaches can identify heritability and enable genome wide mapping of hundreds or thousands of correlated traits. This study presents a simple time-lapse image analysis approach for real-time phenotyping of plant growth under dynamic conditions that promises to uncover the genetic basis of response to environmental change.

- 35–42 **Linkage and Physical Mapping of Sex Region on LG23 of Nile Tilapia (*Oreochromis niloticus*)**
O. Eshel, A. Shirak, J. I. Weller, G. Hulata, and M. Ron
- There is a variety of evidence that sex determination (SD) in tilapias is controlled by major genetic factors. Quantitative trait loci (QTL) for SD in *Oreochromis niloticus* were previously detected on linkage groups (LG) 1 and 23. Using 33 genetic markers in a segregating family, the QTL peak for SD was mapped to a region on LG23 ($P < 7.6 \times 10^{-14}$). Twelve adjacent markers found in this region were homozygous in females and either homozygous for the alternative allele or heterozygous in males, thus determining the sex region. This region encompasses 1.5 Mbp harboring 51 annotated genes.
- 43–58 **A High-Density Simple Sequence Repeat and Single Nucleotide Polymorphism Genetic Map of the Tetraploid Cotton Genome**
John Z. Yu, Russell J. Kohel, David D. Fang, Jaemin Cho, Allen Van Deynze, Mauricio Ulloa, Steven M. Hoffman, Alan E. Pepper, David M. Stelly, Johnnie N. Jenkins, Sukumar Saha, Siva P. Kumpatla, Manali R. Shah, William V. Hugie, and Richard G. Percy
- A high-density cotton genetic map was developed by mapping 2072 SSR and SNP loci using 186 RILs derived from an interspecific cross between two cotton reference species: *Gossypium hirsutum* and *G. barbadense*. This map covered 3380 cM of the tetraploid cotton genome (AD) with an average interval of 1.63 cM, and equivalent recombination frequencies were observed for each of the two subgenomes (At and Dt). Duplicate loci within and between homeologous chromosomes provided evidence for structural relationships and translocation events. This map represents a useful tool for future work on polyploid plants and cotton crop improvement.
- 59–69 **Genetic Analysis in *Drosophila* Reveals a Role for the Mitochondrial Protein P32 in Synaptic Transmission**
Andrew Lutas, Christopher J. Wahlmark, Shaona Acharjee, and Fumiko Kawasaki
- Mitochondria serve important functions at chemical synapses, however much remains to be learned about the underlying mechanisms at the molecular level. In a forward genetic screen for synaptic transmission mutants, we identified a new mutation in a *Drosophila* gene encoding a conserved mitochondrial protein, P32, which represents the first P32 mutation in a multicellular organism. Our analysis revealed a role for dP32 in regulation of presynaptic calcium and neurotransmitter release, and thus defined a molecular component in an established aspect of mitochondrial signaling in synaptic transmission. Future studies promise to define P32 function in a wide range of biological processes.
- 71–78 **Genome-Wide Survey of Large Rare Copy Number Variants in Alzheimer's Disease Among Caribbean Hispanics**
Mahdi Ghani, Dalila Pinto, Joseph H. Lee, Yakov Grinberg, Christine Sato, Danielle Moreno, Stephen W. Scherer, Richard Mayeux, Peter St. George-Hyslop, and Ekaterina Rogava
- We conducted a genome-wide scan for copy number variations (CNVs) in an Alzheimer's disease case-control dataset of Caribbean Hispanic origin. To generate results with high confidence we focused on rare, large CNVs (>100 kb) that were identified by at least two algorithms, and detected 1774 CNVs. Our results did not reveal significant differences between cases and controls in CNV rate or size. However, we did observe a nominal association between the disease and a duplication on chromosome 15q11.2 encompassing up to five genes. The reported results will be useful for identifying CNVs for validation in follow-up studies. For instance, the pathological significance of several rare CNVs that were found in patients with Alzheimer's disease but not in controls requires further assessment in large cohorts.
- 79–82 ***Drosophila* Reporter Vectors Compatible with Φ C31 Integrase Transgenesis Techniques and Their Use to Generate New Notch Reporter Fly Lines**
Ben E. Housden, Kat Millen, and Sarah J. Bray
- One major challenge in biology is to decipher the roles of non-coding DNA in controlling temporal and spatial patterns of gene activity. Fluorescent reporters are powerful tools for visualizing the activity conferred by non-coding DNA in transgenic animals, including *Drosophila*. However, most existing reporter vectors are incompatible with the new Φ C31 integration system that has revolutionized transgenic approaches in *Drosophila*. We have therefore designed a suite of new reporter vectors for use with this system. We demonstrate their efficacy using a DNA-element that reports activity of the developmentally important Notch signaling pathway. Besides confirming the functionality of the vectors, the transgenic lines will be valuable for investigating Notch function.

- 83–97 **Genetic Diversity and Genomic Plasticity of *Cryptococcus neoformans* AD Hybrid Strains**
Wenjun Li, Anna Floyd Averette, Marie Desnos-Ollivier, Min Ni, Françoise Dromer, and Joseph Heitman
- Hybridization which occurs in almost all sexually reproducing organisms may result in novel genotypes adapted to the changed environment. Compared to haploid serotype A and D isolates, most *Cryptococcus neoformans* AD hybrids exhibit unique multilocus sequence typing (MLST) genotypes, and phylogenetic analyses suggest that multiple independent hybridization events punctuated the origin and evolutionary trajectory of AD hybrids. The *C. neoformans* AD hybrid genome is highly dynamic, with continuous chromosome loss and duplication, which may be a facile route for pathogen evolution through which hybrid vigor is generated.
- 99–102 **The C57BL/6J Mouse Strain Background Modifies the Effect of a Mutation in *Bcl2l2***
Stefanie J. Navarro, Tuyen Trinh, Charlotte A. Lucas, Andrea J. Ross, Katrina G. Waymire, and Grant R. MacGregor
- The C57BL/6J mouse strain is widely used in contemporary biological and biomedical research. Many researchers are unaware of the known mutations in this inbred strain and the impact these can have in modifying the effect of independent mutations. We show another example of this, *i.e.* modification of the phenotype of mice mutant for *Bcl2l2*, which encodes BCL-W, a death-protecting member of the BCL-2 family, by the C57BL/6J strain.
- 103–114 **Strain Variation in the Transcriptome of the Dengue Fever Vector, *Aedes aegypti***
Mariangela Bonizzoni, W. Augustine Dunn, Corey L. Campbell, Ken E. Olson, Osvaldo Marinotti, and Anthony A. James
- This study describes for the first time variation in the transcriptomes of three different strains of the dengue vector mosquito, *Aedes aegypti*. Transcriptome variation is analyzed by RNA-seq across strains at two important physiological conditions, which are a sugar-diet or after blood-feeding. This study has important implications for understanding the well-documented geographic variation of *Aedes aegypti* populations in various traits, primarily vector competence. Additionally, knowledge of transcriptome variability across strains is fundamental for genetic-based strategies of vector control applicable at the species level.
- 115–121 **Chromosome Y Regulates Survival Following Murine Coxsackievirus B3 Infection**
Laure K. Case, Leon Toussaint, Mohamad Moussawi, Brian Roberts, Naresha Saligrama, Laurent Brossay, Sally A. Huber, and Cory Teuscher
- The use of consomic strains of mice presents the opportunity to reveal the ChrY genetic variations that impact biological functions unrelated to male reproduction. A comparison of the available sequence data indicates that there are no single nucleotide polymorphisms within ChrY genes that segregate with susceptibility to CVB3-induced mortality. New reports on *Drosophila* show that ChrY structural polymorphism can cause phenotypic differences in males by changing the way genes on other Chrs function. Our report provides evidence suggesting that epigenetic regulation by ChrY also occurs in mammals.
- 123–130 **Use of Cumulative Poisson Probability Distribution as an Estimator of the Recombination Rate in an Expanding Population: Example of the *Macaca fascicularis* Major Histocompatibility Complex**
Antoine Blancher, Alice Aarnink, Nicolas Savy, and Naoyuki Takahata
- We describe a new method to estimate the rate of recombination per generation (c) from the genotypes of a large individual sample of a expanding population for which the founding event is dated. Under these assumptions, the frequencies of the classes of haplotypes defined by the number of recombination “h” they have experienced (intact founding haplotypes $H_{\text{rec}(0)}$ and various types of recombinant $H_{\text{rec}(h)}$) follow a Poisson distribution defined by a mean which equals “ c ” multiplied by the number of generations from the founding event. The approach is illustrated with an application to estimating the major histocompatibility complex (MHC) recombination rate in the Mauritian macaque population.

- 131–141 **Many *Saccharomyces cerevisiae* Cell Wall Protein Encoding Genes Are Coregulated by Mss11, but Cellular Adhesion Phenotypes Appear Only Flo Protein Dependent**
Michael C. Bester, Dan Jacobson, and Florian F. Bauer
- Adhesion properties in microorganisms are primarily responsible for many phenotypes of scientific and biotechnological relevance such as cell-cell and cell-substrate adhesion, biofilm formation, invasive growth, and pathogenic behavior. *Saccharomyces cerevisiae* has been used as a model system to better understand processes involved in cellular adhesion. Our data support the idea that these phenotypes are primarily, if not entirely, controlled by a limited number of cell wall proteins of the Flo family, and that other cell wall proteins that are co-regulated with the *FLO* genes appear not to affect such phenotypes in any significant way.
- 143–150 **Multiplex Chromosomal Exome Sequencing Accelerates Identification of ENU-Induced Mutations in the Mouse**
Miao Sun, Kajari Mondal, Viren Patel, Vanessa L. Horner, Alyssa B. Long, David J. Cutler, Tamara Caspary, and Michael E. Zwick
- Forward genetic screens in *Mus musculus* are slowed by the challenge of efficiently identifying causative mutations. We describe a multiplex chromosomal exome sequencing protocol that overcomes this critical barrier. We demonstrate the approach by identifying the most likely causative molecular lesions in four ENU-induced mouse lines. Our multiplex protocol allows individual investigators to first systematically identify the molecular lesions in all lines, and then integrate that information in choosing which ones to pursue.