Greetings from the Chair

Time passes all too quickly. Putting together the annual newsletter gives me the chance to pause and review the accomplishments of our faculty, postdoctoral fellows, graduate students, and staff. We have a lot to be proud of this year, and I hope you enjoy this report.

On behalf of our Department, I invite you to join us at our alumni gathering in Philadelphia at the American Society of Human Genetics meeting on November 10, 2008. It will be held in the Marriott, Downtown from 8:00 PM – 10:00 PM.

I encourage all of our alumni and friends to use the enclosed card to share your news with us.

With best regards,

Sally A. Camper, Ph.D.
James V. Neel Professor and Chair

Comings and Goings

David States accepted a position at The University of Texas-Houston, beginning in October. He will continue to be part of our National Center for Biotechnology. We wish him well in this new endeavor.

Anthony Antonellis joined us this fall as an assistant professor with a joint appointment in Neurology. Tony received his Ph.D. from the Joint Program in Genetics at George Washington University and the National Institutes of Health. His thesis work on the role of glycyl-tRNA synthetase mutations in axonal degeneration was performed with Eric Green at the National Human Genome Research Institute (NHGRI). After obtaining his Ph.D. in 2004, Tony stayed at the NHGRI and was co-mentored as a post-doctoral fellow by Eric and Bill Pavan, and used multiple-species comparative sequence analysis to study transcriptional regulation. Here, Tony will be studying the development and diseases of the peripheral nervous system, including the role of aminoacyl-tRNA synthetases in neurodegenerative diseases and identifying loci important for Schwann cell development and function. Tony holds an NIH Pathways to Independence Award (K99/R00) to support this research.

David Kurnit passed away on January 30, 2008 at age 60. He received his M.D. and Ph.D. degrees from Albert Einstein College of Medicine and was an associate professor at Harvard Medical School before joining the University of Michigan. During his career here he was a Howard Hughes investigator...
and professor in the Departments of Pediatrics and Human Genetics. He will be remembered for his scientific contributions, dedication to students and researchers, and his sharp wit.

José Garcia Perez, a research investigator working with John Moran, obtained a faculty position at the Spanish Stem Cell Bank, Center for Biomedical Research at the University of Granada, Spain.

**Faculty Promotions, Awards, Honors**

John Moran was promoted to full professor, Donna Martin to associate professor, and Wendy Uhlmann to clinical assistant professor.

We congratulate Friedhelm Hildebrandt and John Moran for being appointed to the Howard Hughes Medical Institute in January and August, respectively.

Dave Burke received a gift of genetic research funds from Dr. Francis T. Oda. Dave is known for his innovative research bridging molecular biology and bioengineering.

Gil Omenn received the Distinguished Service Award in October 2007 at the 7th World Congress in Seoul, Korea and was elected Vice President of the International Human Proteome Organization (HUPO) for 2009-2010.

The Department faculty, their research activities, and recent publications can be found on the Departmental website at www.hg.med.umich.edu.

**Staff**

Janet Miller, our student services administrator, won the University of Michigan’s Distinguished Service Award. Janet will be retiring next year, so we are thrilled that she was recognized institutionally for her outstanding performance.

**Graduate Education**

We currently have eight postdoctoral research fellows in the labs of primary appointees and twenty-four doctoral students. Three of our students are in the Medical Scientist Training Program and fourteen are pursuing their master’s degrees, thirteen in genetic counseling and one in human genetics. Our Ph.D. students are supported by several different training grants, including Genetics, Biostatistics and Genome Sciences.

We take great pleasure in reporting on recent graduates of our Ph.D. Program.
Lisa Privette (Petty lab) defended her thesis on “CHFR Expression in Breast Cancer Cells: Its Relevance to Tumorigenesis and Genomic Stability,” and is now a post-doc in the laboratory of Susanne Wells at Cincinnati Children’s Hospital Medical Center where she studies the role of the DEK proto-oncogene during breast cancer development.

Clement Chow (Meisler lab) completed his studies on “Mutations in PI(3,5)P2 Signaling and Neurodegeneration in Mouse and Human.” Clement has accepted a joint post-doctoral fellowship with Andy Clark and Marianna Wolfner at Cornell University. He is studying various aspects of evolutionary and population genetics in human and drosophila.

We are very proud of our genetic counseling graduates. Each one received a Rackham grant for their research. All of the graduates are listed below with their current status.

Patty Arscott, Cleveland Clinic, Cleveland, Ohio
Katie Hock is seeking a job in the Great Lakes area.
Michelle Jackson, MD Anderson, Houston, Texas
Jessica McKillop, Eastern Virginia University, Norfolk, Virginia
Jessi Moline, Cleveland Clinic, Cleveland, Ohio

Graduate Student Awards:

The 2008 recipient of the Anita and Howard Cramer Scholarship Award is Martina Kubec (Sekiguchi lab). Martina is working on the regulation of artemis and its relationship to other nucleases in VDJ recombination.

The James V. Neel Fellowship Awards went to Qing Fang (Camper lab), working on the genetics of deafness, and Alyson Krokosky (Branham, Ophthalmology), whose research project is titled “Critical Factors in the Transition to Emerging Adulthood for Adolescents with Visual Impairments.”

Mara Steinkamp (Robins lab) was the inaugural recipient of the Robert and Janet Miller Award for Basic Research on Urologic Cancer. Mara studies the role of the androgen receptor in prostate cancer in humans and mice.

Qing Fang received a Barbour Scholarship. The endowment that funds this was established at the University of Michigan by Levi L. Barbour in 1914 for women of the highest academic and professional caliber from the area formerly known as the Orient to study science, medicine, mathematics and other disciplines critical to the development of their native lands.

Tehmina Masud (Sekiguchi lab) has a Fulbright Scholarship. This program was created following World War II through the efforts of Senator J. William Fulbright with the goal of promoting peace and understanding through educational exchange.

Jennie Mason (Sekiguchi lab) received the award for best poster presentation at the 10th Annual Midwest DNA Repair Symposium.

Department Events

The summer picnic, coordinated by Dave Burke and Janet Miller, was an excellent event at Delhi Park.

The James V. Neel Lectureship was given in May by Dr. Janet Rowley of the University of Chicago. She is well known for her work demonstrating that
chromosome translocations are a key component in the development of leukemia. Dr. Rowley’s findings launched a new era in the field of cancer genetics that paved the way for novel diagnostic, prognostic, and therapeutic options in the management of leukemia and lymphoma.

Our annual fall retreat took place on campus this year at the Michigan Union. Didi Robins, Linda Peasley, and our student and event representatives collaborated to develop an excellent program that included guest speaker, Alex Young, from Zingerman’s. We heard about the value of genetic diversity in heirloom vegetables and got to taste the proof in the form of delicious appetizers made by Alex with produce from the farm that supplies Zingerman’s Roadhouse.

The Tashian family at the Human Genetics picnic

Center for Genetics in Health and Medicine

The recipients of postdoctoral fellowship awards from the Center for Genetics in Health and Medicine were Leon Raskin and Christopher Vlangos. Leon works with Steve Gruber on “Integrated Genetic Approaches to Drug Discovery for Melanoma.” Chris Vlangos and his mentor, Catherine Keegan, are in Pediatrics and Communicable Diseases, and his project is titled “Evaluation of Candidate Genes Involved in Caudal Mesoderm Development in Mice and Humans.”

The Center partnered with the Operating Committee for the Endowment for the Basic Sciences to establish high throughput sequencing and genotyping capabilities at the University of Michigan. Together we also launched the first of three competitions for pilot feasibility studies in the area of genetics and genomics, and we received an enormous interest in these new technologies. Applications from PI’s and Co-PI’s represented twenty different departments and three different schools. Three awards went to Human Genetics faculty: Tom Glover and Tom Wilson (Pathology), Jun Li and John Kim, and Jun Li and Noah Rosenberg. Other awardees were Steve Qin, Goncalo Abecasis and J. T. Elder (Biostatistics, Dermatology), Jessica Schwartz and Steve Qin (Physiology, Biostatistics), and Vincent Young and Gary Huffnagle (Internal Medicine, Microbiology & Immunology).

Alumni Notes

Lt. Gen. Eric B. Schoomaker, M.D., Ph.D. is the commander of Walter Reed Army Medical Center, the U.S. Army Surgeon General, and commander of MEDCOM, which provides medical care for Army service personnel and their families. He completed his M.D. in 1975, and his Ph.D. in Human Genetics in 1979. His thesis work with George Brewer involved “Studies in an animal model of sickle cell disease.” In an interview for the spring 2008 issue of Medicine at Michigan, Schoomaker said “George Brewer kept my interest in medical school. He got me interested in working in red blood cells and recruited me into the M.D./Ph.D. program.” Schoomaker is certified in both hematology and internal medicine.

http://www.medicineatmichigan.org/magazine/2008/spring/rotations/

Dorene Markel, M.S., M.H.S.A. is Director of Clinical and Translational Research and Managing Director of the Michigan Institute for Clinical and Health Research. Her leadership was instrumental in the recent award of $55 million to the University of Michigan by the NIH for clinical and translational research. Markel was one of the first graduates of our genetic counseling program (1983). She also teaches in the Genetic Counseling Program, serves as a research mentor for genetic counseling students, and is an advisor to the CGHM.

Donations and Gifts

Mary Sue Coleman, president of the University of Michigan and a biochemist by training, has shown her support for graduate education by offering a 1 to 2 match for donor gifts for education this year. This donor challenge dovetails with our goal of...
establishing an endowment to support graduate student fellowships. A gift of $50,000 or more will create a named endowment of your choice, honoring your commitment to Michigan and creating a legacy of support for the next generation of leaders and best. The donor challenge will make a one-to-two match of any size gift, up to $1 million, toward graduate and professional student support. By adding one matching dollar to every two dollars you give, the challenge leverages your philanthropy for its greatest impact on graduate education at Michigan. The program continues until $40 million in gifts have been received or December 31, 2008, whichever comes first. To date the Department of Human Genetics has received over $10,000 in support for graduate education through the President’s challenge. We are very grateful to all of you for your support of this important effort.

On June 10th, the Gilbert S. Omenn Atrium and the Flame of Wisdom Sculpture, by renowned Mexican artist Leonardo Nierman, were dedicated in the new Biomedical Science Research Building.

Tom Gelehrter transitioned to professor emeritus, and we honored him and his family at a dinner in May at the Alumni Center. We have created the Thomas D. Gelehter Lectureship in Medical Genetics in acknowledgment of the important roles he has played in teaching, research, and service at the University of Michigan.

How to Get Involved
To make a gift supporting the future of graduate students in scientific research, please visit our website www.hg.med.umich.edu/contribute.php or contact Linda Peasley by email: lpeasley@umich.edu or phone: 734-647-3149.

Department of Human Genetics Donor Honor Roll (2005 - present)

We would like to recognize the following individuals who have given generously to support the Department of Human Genetics during the years beginning in 2005 when Dr. Sally Camper became chair.

Anita and Howard Cramer Fellowship Fund: awarded to a Ph.D. student each year for outstanding academic achievement.
Professor Harvey S. Cramer BS 1965 Pre-Med

Carole McTague Genetic Counseling Student Enrichment: used to offset travel expenses of our genetic counseling students to national meetings.
Elizabeth M. Petty, M.D. Faculty

James V. Neel Fellowship Fund: awarded to a Ph.D. student and an M.S. student in Genetic Counseling each year for outstanding research achievements.
Margit Burmeister, Ph.D., Faculty
Sally A. Camper, Ph.D., Faculty
Ernest H.Y. Chu, Ph.D., Emeritus Faculty
Gretchen Darlington, Ph.D., MS 1966 Human Genetics, PHD 1970 Human Genetics
Julie A. Douglas, Ph.D., MS 1998 Biostatistics, PHD 2001 Biostatistics, Faculty
L. Scott Feiler, M.D., MD 1975
Steven J. Ferrucci, M.D., MS 1980 Human Genetics, MD 1986
Thomas D. Gelehter, M.D., Emeritus Faculty
David Ginsburg, M.D., Faculty
Thomas M. Glaser, M.D., Ph.D., Faculty
Thomas W. Glover, Ph.D., Faculty
Ted Howard Hansen, Ph.D., MS 1972 Human Genetics, PHD 1975 Human Genetics
Sandra Jean Hasstedt, Ph.D., AM 1975 Statistics, PHD 1976 Human Genetics
Denise P. Kalm, MS 1977 Human Genetics
David C. Kohrman, Ph.D., PHD 1991 Microbiology, Research Fellow in Human Genetics, Faculty
Myron Levine, Ph.D., Emeritus Faculty
Jeffrey C. Long, Ph.D., MS 1982 Human Genetics, PHD 1984 Human Genetics, Faculty
Donna M. Martin, M.D., PHD 1992 Neuroscience, MD 1996, Faculty
Melanie K. McDermot, MS 1988 Human Genetics
John V. Moran, Ph.D., Faculty
Mrs. James V. Neel, Friend
Howard C. Passmore, Ph.D., MS 1968 Human Genetics, PHD 1970 Human Genetics
Elizabeth M. Petty, M.D., Faculty
Michael E. Ray, M.D., Ph.D., MS 1993 Human Genetics, PHD 1988 Human Genetics, MD 1998
James V. Neel Lectureship: supports hosting and travel for a lecture by an internationally known geneticist each year.

Georges Brewer is active and publishing his work on the risks of free copper in the body and the development of useful anticopper drugs such as tetrathiomolybdate. His success in obtaining FDA approval for use of zinc to treat Wilson’s disease in 1997 is a paradigm for “orphan” disease research. Orphan diseases are defined as diseases with a population prevalence of less than 200,000 patients in the United States. While Wilson’s disease and others like it are rare, there are over 6,000 orphan diseases in the United States, and they affect one in ten individuals or 25 million people. The clipping below is from our departmental archives, and depicts a group of parents lobbying for research to cure Cooley’s anemia, one of the diseases studied by Jim Neel and others in the beginning of our Department’s history. The children are holding a sign that says “When your child has it - it’s not a rare disease!” Parent groups have always played very active roles in advocating for and raising funds for research on rare diseases. In recognition of the important role academic medical centers play in researching treatments for these diseases, the University of Michigan has launched an Initiative on Rare Disease Research in consultation with faculty, including George, who have expertise in this area. There are now institutional funding competitions for pilot grants and symposia that will stimulate this important area of clinical-translational research.

Dave Burke continues as a curriculum advisor in an NSF-funded science program called “Model Institutions in Excellence” at the Universidad Metropolitana, in San Juan, Puerto Rico. He is collaborating with the science faculty there to develop a new quantitative-based biology curriculum in an effort to enhance educational opportunities for under-represented minorities in science, technology and math.

Margit Burmeister was elected to the Board of the International Society of Psychiatric Genetics and published a review on psychiatric genetics in Nature Reviews Genetics. She received a NARSAD Distinguished Investigator Award for re-sequencing in bipolar disorder. She will become acting director of the Bioinformatics Program next year.

Sally Camper was elected to the Executive Board of the Genetics Society of America, appointed to the Scientific and
Educational Programs Core Committee of the Endocrine Society, and chairs the Genetics Resources Services Advisory Board for the Jackson Laboratory. She gave the McGregor Endowed Lecture at Cornell University and presented her research in endocrine system development in Florence and Rome, Italy.

Ray Chan’s lab has grown to six lab members. Ray welcomes the addition of Sandra Durkin, a new postdoc and HG graduate program alum, and several talented undergraduate researchers. Together, they are making wonderful progress in research.

Julie Douglas was an invited speaker at the University of Texas Health Science Program in Houston. She serves on the Editorial Board of Cancer, Epidemiology, Biomarkers, and Prevention. Her lab recently completed year one of a five-year NIH-funded study of the genetics of breast density and received a competitive renewal of another NIH-funded study of the genetics of prostate cancer.

Eric Fearon’s laboratory continues to emphasize studies of the role of Wnt pathway gene defects in the pathogenesis of human colorectal cancers. The lab published several papers on this topic in the past year and has also developed new research activity on the role of microRNAs in human cancer.

Tom Gelehrter serves as director for the first year medical students course, Patients and Populations, which includes Medical Genetics and is taught in August. He continues to work on preparing the third edition of his popular Principles of Medical Genetics textbook. Given the explosion of changes in our understanding of genetic and environmental risk factors for disease, the third edition will be a completely different text than the first two.

Dave Ginsburg’s lab continues to study the components of the blood clotting system and how disturbances in their function lead to human bleeding and blood clotting disorders. David was a guest speaker this past year in the Department of Genetic Medicine and Development at the University in Geneva, Switzerland, at the 6th Bari International Conference in Foggia, Italy and at the Grifols Symposium at the World Federation of Hemophilia Congress in Istanbul, Turkey. David is chairing a membership committee for the Institute of Medicine.

Tom Glaser has been active in reviewing programs at NIDCD and grants for NSF. His sensory systems research spans the basic aspects of retinal ganglion cell and auditory hindbrain development to identifying genetic risk factors for retinal detachment and anophthalmia.

Tom Glover was an invited speaker at the 38th Annual Meeting of the Environmental Mutagen Society. He served on the Editorial Board of Human Molecular Genetics and scientific advisory boards for the Progeria Research Foundation, the Lymphedema Research Foundation and the Tulane & LSU University COBRE program. He was also an invited participant in a Trans-NIH Lymphatic Research Working Group to establish research directions in the field. He was named co-director of the clinical DNA Microarray Laboratory and continues as co-director of Clinical Cytogenetics.

Steve Gruber is an active advisor for the National Cancer Institute and the American Society of Clinical Oncology. His research focuses on the molecular epidemiology of colorectal cancer, genetic and environmental contributions to melanoma, and a variety of other common malignancies. Together with a large interdisciplinary team, Steve and colleagues published a study in Nature Genetics that used genome-wide association studies to identify several loci that confer risk of colorectal cancer.

Friedhelm Hildebrandt’s laboratory continues their work on positional cloning and functional characterization of novel genes causing kidney diseases. This includes the study of developmental and degenerative defects of the eye and auditory system associated with kidney diseases. Zebrafish models for genetic kidney diseases are being generated for novel genes identified.

Jeff Innis was appointed as a permanent member of the prestigious Genetics of Health and Disease NIH Study Section. Jeff continues as director of the Division of Pediatric Genetics and is the director of the new Michigan Medical Genetics Laboratories. He serves on the editorial boards of Molecular Genetics and Metabolism and Developmental Dynamics.

John Kim was a visiting speaker at Rutgers University, Department of Cell Biology and Neuroscience, and at the University of Chicago. In addition, John gave lectures in the Departments of Pathology and Cell and Developmental Biology at the University of Michigan. John was selected by the Biomedical Research Council to be the University’s nominee for the Pew Scholars Program in Biomedical Sciences.

Dave Kohrman continued a four-year term on the NIH Auditory System Study Section. He is director of the Hearing, Balance, and Chemical Senses Training Grant and the summer Science Mentorship Program for hearing impaired undergraduate students.

Jun Li serves on the Editorial Board of Genome Research. His recent Science paper on human population diversity was featured in Nature News and the popular press. Jun is a co-organizer of an interdisciplinary seminar series on next-generation sequencing funded by the Office of the Vice President for Research.

Jeff Long was an invited speaker at the First Biological Evolution Workshop in Brazil, and gave a keynote address at Indiana University’s symposium on Rethinking Race in the Americas: Anthropology, Politics, and Policy. He serves on
the Federal Advisory Committee for the National Children’s Study and the External Advisory Board for the NIAAA-funded multi-centered project “Collaborative Studies on Genetics of Alcoholism.” He has published on the topics of molecular population genetics and genetic epidemiology.

**Donna Martin** is the genetics PI for the University of Michigan Simons Foundation Autism Simplex Collection and received pilot funding from the Clinical Translational Science Award to perform genetics studies on children with autism. She also served on scientific review panels for the International Rett Syndrome Foundation and the NIH.

**Miriam Meisler** presented talks on mechanisms of epilepsy at the Gordon Conference, the San Raffaele Institute in Milan, the CNR Instituto di Neuroscienze, in Pisa, and the Genetics Department at Washington University in St. Louis. She continues to serve on the Program Committee of the ASHG, as director of the Genetics Training Grant, and as Associate Dean for Research at the University of Michigan.

**John Moran** serves on the editorial boards of Genome Research and Gene. He also serves on the ASHG Awards Committee. He gave invited presentations at the Gordon Conference on Human Genetics and Genomics, the Keystone Symposium on Molecular Evolution as a Driving Force in Infectious Diseases, and at the International Congress on Transposable Elements in Saint Malo, France. He gave lectures at the University of Georgia, the University of Chicago, and the Fred Hutchinson Cancer Research Center. His laboratory continues to study the impact of transposable elements on the human genome.

**Gil Omenn** recently became president-elect of the Harvard Medical Alumni Association and chaired the Alumni Committee on Medical Education. He was appointed to the Center for the Study of the Presidency Study Group on Presidential Science and Technology Advisory Assets. He served as a speaker and on the Advisory Committee of the Aspen Institute World Health Forum. He was appointed to the Scientific Advisory Board of the Battelle/Pacific Northwest National Laboratory Systems Biology and Health Initiative. He is a member of the Advisory Board of Health Promotion Advocates and a member of the Board of the Salzburg Global Seminar.

**Liz Petty** serves our graduate and medical students through her roles as Associate Dean, Office of Student Programs, and as the medical director of our Genetic Counseling Graduate Program in Human Genetics. Liz served as a standing member of the NIH-NCRR Research Review Committee and was an ad-hoc reviewer for the Susan G. Komen Foundation, the Israel Science Foundation, and the Canadian Research Foundation. She is a member of the Programmatic Integration Committee for the Department of Defense Research Program, a question writer for the American Board of Medical Genetics, and editorial board member for the Atlas of Genetics and Cytogenetics for Oncology and Hematology. In addition to teaching residents, medical students, and graduate students, Liz enjoyed teaching various non-science groups about genetics, from members of the Osher Lifelong Learning Institute to medical transcriptionists at the University of Michigan.

**Didi Robins** gave invited talks for the DOD IMPACT Conference, APHMG Workshop, Endocrine Society, and seminars at UT San Antonio and University of Virginia. Didi continues as the director of graduate studies for the Department of Human Genetics and as chair of the ACS Beginning Investigators (UICC) Review Panel. She is a member of the Endocrine Society Annual Meeting Steering Committee and the National Toxicology Program Board of Scientific Counselors.

**Noah Rosenberg** was a keynote speaker at the Isaac Newton Institute for Mathematical Sciences workshop on Future Directions in Phylogenetic Methods and Models. He also gave invited talks at the Universities of Illinois, Pennsylvania, and California (Davis and Irvine), the Santa Fe Institute, the National Center for Biological Sciences in Bangalore, India, and at the Symposium on Trends in Human Genetics in Puri, India. Noah serves on the Editorial Board of the American Journal of Human Genetics.

**JoAnn Sekiguchi** gave presentations at the 10th Annual Midwest DNA Repair Symposium at the University of Pittsburgh and the 2008 Annual Pew Scholars Meeting in Aruba. She was invited to serve as an external grant reviewer for the Telethon Career Award Program in Human Genetics, Rome, Italy.

**Charlie Sing** was chair of the NIH Director’s Pioneer Award Program. He was an invited speaker at The University of Texas MD Anderson Cancer Center. He continues to serve on the Editorial Board of the Journal of Molecular Medicine. He serves as an advisor to the Center for Genome Dynamics, the Jackson Laboratory, and also to the president of Iowa State University.

**Wendy Uhmann** is the NSGC liaison to the National Advisory Council for Human Genome Research. She spoke at the National Society of Genetic Counselors meeting. Wendy is co-editing A Guide to Genetic Counseling 2nd edition with Jane Schuette and Beverly Yashar.

**Bev Yashar** was invited to serve on the Editorial Board of the Journal of Genetic Counseling and elected to the Board of Directors of the Michigan Alliance of Genetic Counselors. Bev was involved in the production of an educational video for Laureate Education, which is a leading international network of accredited campus-based and online universities that provide postsecondary education in over 35 accredited institutions in Asia, Europe, and the Americas.
WE WANT TO HEAR FROM YOU!

Please take a moment to complete the attached card and return it to us. For your convenience you can also keep us informed by emailing kgrahl@umich.edu or visiting our website www.hg.med.umich.edu. We look forward to hearing from you and sending you future updates about the Department, faculty and alumni.

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U of M degree, year received & mentor

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You are invited!

University of Michigan
Department of Human Genetics

Alumni Gathering at the American Society of Human Genetics

Conference Room 414/415
Philadelphia Marriott, Downtown
Philadelphia, Pennsylvania

November 10, 2008
8:00 PM – 10:00 PM

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