Dear Colleagues and Friends,

The start of a new year has different meanings. For the GC Program it revolves around students-both current and prospective. Our current students are back from winter break. The first years are still focused on classwork and introductory clinical experiences and looking forward to full immersion in their second year clinicals. Second years are thinking about everything they need to complete (rotations, that little thing called a "research project") and wondering where they will be come summer (hopefully employed in their dream job!). The admissions committee is busy reading applications with the difficult task of choosing, from over 120 prospective students, 35 applicants to interview and then further decide the 5 to whom we will extend an offer. Daunting, all of it. This year the offer of admission coincides with the end of the semester; we will be welcoming new GC interns and wishing the Class of 2013 well. And then wow, the year is almost over! I better get moving; there’s a lot to do!

Enjoy!

- Casey

• We were re-accredited by the ABGC for the maximum time allowed! Reviewers commented on the strength of our curriculum, didactic and clinical. A big thank you to faculty, instructors and clinical supervisors!!

• Our students performed excellently on the national certification exam in 2012 with a 100% pass rate! The average scores were well above the national average. Congratulations to all.

• We continue to collaborate with other Midwest training programs. We hosted a summer student from the University of Michigan for a second year clinical rotation; we held the 4th biennial Adam Rennebohm Memorial Bereavement Program attended by all WI, MN and NW GC students; we held a joint lecture with MN GC students for the Cognitive Behavioral Techniques: Implications for Genetic Counseling using distance education as our format.

• We were again able to award scholarships to all of our incoming students due to the generosity of alumni donors and friends.

• We continue successful placement of our graduates; the class of 2012 were all gainfully employed (with benefits!) by July 1, 2012.

• Congratulations to Dr. David Wargowski for his promotion to full professor and to Dr. Greg Rice for receiving the Dean’s Teaching Award.
empowering them with knowledge and resources. Volunteer experiences with people of many different abilities and bioethics courses taught by experts at UW shaped my drive to promote informed decision-making and advocate for genetic counseling as part of mainstream medicine. I am thrilled to begin the next chapter of my life in the UW Genetic Counseling graduate program, and excited to meet my classmates!

**Rosy Ebel – Recipient of the Dr. Raymond Kessel Scholarship in Outreach**

I grew up in Houston, TX and relocated to Cleveland, OH to attend Case Western Reserve University. I graduated this past May with a BA in Biology and a minor in Psychology. I learned about genetic counseling in a Behavioral Genetics course my sophomore year in college and was excited about the prospect of a career that involves scientific and psychosocial aspects of genetics. In order to prepare for entry into the field, I spent time shadowing genetic counselors, as well as volunteering in HIV intervention at the Free Health Clinic of Cleveland. I feel honored to be receiving the Dr. Raymond Kessel Scholarship in Outreach. Dr. Kessel has been responsible for establishing needed genetics services, such as clinics and screening programs, throughout Wisconsin and has shown dedication to providing resources for genetics education to members of all communities. Having spent time working with individuals with limited healthcare options, I hope to combine my experiences and training as a genetic counselor into a passion that mirrors Dr. Kessel’s work in order to continue his mission of empowering all members of the community with important genetic resources.

**Katie Gallagher – Recipient of the Dr. Renata Laxova Scholarship in Patient Advocacy**

I am originally from Appleton, WI and moved to Minneapolis, MN to attend the University of St. Thomas where I graduated in 2011 with a major in Exercise Science and a double minor in Psychology and Biology. I knew that genetic counseling was perfect for me once I started observations in the clinic and spending a summer interning with a genetic counselor in the oncology field. I have really enjoyed volunteering through college and working on a crisis hotline after graduation as preparation for graduate school. I was thankful to have been awarded The Dr. Renata Laxova Scholarship in Patient Advocacy. I believe this is because I am people and patient-oriented. I enjoy the science that the field of Genetic Counseling gives me, but the greatest reward for me is building a relationship with the patients and helping them through potentially difficult times. I am looking forward to moving back to the wonderful state of Wisconsin and becoming a Badger in the Genetic Counseling program.

**Meghan Kraus – Recipient of the Genetic Counselor Training Program Alumni Scholarship for Leadership**

I grew up in St. Louis, MO and graduated from Mizzou in December 2011 with a major in Biology and minors in Spanish and Psychology. I first became interested in genetic counseling after I shadowed a counselor my junior year of college and have been working with her ever since on various clinical trials for patients with metabolic disorders. During my undergraduate career I also enjoyed volunteering at the Mid-Missouri Crisis Hotline, True North Domestic Abuse Shelter and Special Olympics. I am very grateful to have received the
Alumni Scholarship for Leadership to help me in becoming a genetic counselor. The most important person in my journey to becoming a GC was my undergraduate GC mentor and I look forward to the day I can “pay it forward” and serve as a mentor to educate and assist others interested in this field. I am excited to join the accomplished group of UW-Madison genetic counseling alumni in two years and to help others achieve this goal like I was helped. I’ve heard nothing but great things about Madison and I’m thrilled to be moving there and joining the UW genetic counseling program this fall!

Tara Maga – Recipient of the Dr. Richard Pauli Scholarship in Clinical Research

Originally from Michigan, I attended The George Washington University in Washington, D.C. graduating with a B.S. in Biology in 2005. At GW I volunteered for a program called Asthma Swim where I taught children’s swimming lessons and preventative care. As an undergraduate I had the opportunity to work in a genetics lab. This experience, along with working as an IRTA fellow at the National Cancer Institute (NIH), led to my passion for genetic research. This past year I completed my PhD in Genetics at the University of Iowa. My thesis involved the development of a targeted sequence capture platform paired with next generation sequencing to identify novel genes involved in atypical hemolytic uremic syndrome. While working with genetic counselors during my training, I found the profession to be the perfect combination of what I love most about genetics and the opportunity to help patients in a more direct way. I am very thankful to have been awarded the Dr. Richard M. Pauli Scholarship in Clinical Research. I agree that there is a need for collaboration and understanding between genetic counselors, scientists, medical doctors, and other health care professionals. My experiences during my PhD have shown me that this is especially true now with the new technologies that are just beginning to be used in the clinic. This is why I am excited to be a student once again and start the GC program this fall!

Farewell to the Class of 2012

The Class of 2012, like others before and after, was a great class and much missed. Listed below is a statement from each student about what they appreciated about their training here at UW-Madison as well as their Research Project Abstracts. In keeping with our mission of training students to think critically and participate in research, all students enroll in four research credits during the second year of the program. They work under the direction of a research professor toward the goal of producing a publishable research or other project that contributes to the body of knowledge of the discipline.

Hannah Baker: During her time at UW, Hannah enjoyed the opportunity to work closely with supportive supervisors, talented physicians, and a devoted Program Director. She was especially appreciative of the unique opportunities that UW-Madison offers including the MCH-Lend Mayo Clinic Internship, diverse clinical rotations, and comprehensive coursework. Her research experience gained as a project assistant in the School of Nursing has been priceless. As an out-of-state student, Hannah would like to take this opportunity to express her deep gratitude for the scholarship she received, and thank donors for their helpful contribution in allowing her to reach her professional goals. Hannah Baker currently works as a Pediatric Genetic Counselor at the University of Iowa, in her homeland of the Hawkeye State to work.

Development and Validation of Cystic Fibrosis Genetic Knowledge Questionnaire (CFGKQ)

Presenter: Hannah Baker
Project Advisor: Audrey Tluczek, PhD, RN

Background: Currently, there are no empirically validated instruments to evaluate cystic fibrosis (CF) genetic knowledge in individuals of child-bearing age within the general population. Standardized, valid, and reliable instruments are essential to evaluate the effectiveness of clinical practices and to develop new evidence-based genetic educational interventions for parents of infants with abnormal newborn screening (NBS) results for cystic fibrosis (CF). This study was designed to develop and establish the validity and reliability of a CF genetic knowledge questionnaire.

Methods: The 16-item Cystic Fibrosis Genetic Knowledge Questionnaire (CFGKQ) was completed by 84 members of the general population (GP group) and 441 members of the National Society of Genetic Counselors (NSGC group) in the United States. Participants also rated the importance of genetic content.

Results: Significant differences were found in correct responses provided by participants in the GP group as compared with participants in NSGC group. Results showed consistency between the initial and two week follow-up administrations of the questionnaire for the GP group and
NSGC groups. Two items (#10 and #16) lacking empirical support were removed from the questionnaire. Thus, the final questionnaire consisted of 14 items: five multiple choice and nine true-false. Members of the general population placed greater importance on recurrence risks and implications of a CF diagnosis for family members as compared to the genetic counselors who tended to view genetic mechanisms of CF as highly important.

**Conclusions:** These findings suggest that the 14-item version of the CFGKQ is a valid and reliable instrument that can be used to assess knowledge of CF genetics in adult members of the United States general population. This tool may be particularly valuable in the development of evidence-based practices in CF genetic counseling, particularly in the context of NBS.

**Lior Borovik:** Lior’s decision to apply to the UW-Madison program was based on its excellent reputation and its more than 30 years of experience, but what solidified his decision in his opinion was the friendly and tight-knit community he found. He appreciated his time training as part of an interdisciplinary team member and advocate through the MCH LEND program and the Consumer Health Advocacy Program; both have helped him better understand the healthcare system from the perspective of the patient. He is thankful for the scholarships provided to him as it allowed him to concentrate on learning and ultimately helping him help his patients. He values such philanthropy and hopes to one day “pay it forward.” Lior currently works as a Cancer genetic counselor at Sanford Health, in South Dakota.

**Pelger-Huet Anomaly and a Mild Skeletal Phenotype Secondary to Mutation in LBR**

**Presenter:** Lior Borovik  
**Project Advisors:** Richard M. Pauli, MD, PhD & Peggy Modaff, MS, CGC

The Lamin B receptor (*LBR*) gene has been described as encoding a bifunctional protein. Mutations in that gene can affect neutrophil segmentation, and also may have an effect on sterol reductase activity. Mutations on this gene are responsible for two different recognized conditions, Pelger-Huet anomaly (PHA) and Greenberg skeletal dysplasia. PHA is an autosomal co-dominant laminopathy resulting in bilobed neutrophil nuclei in heterozygotes, and unsegmented (ovoid) neutrophil nuclei in homozygotes. Some putative PHA homozygotes have been reported to have minor skeletal malformations. Greenberg skeletal dysplasia is an autosomal recessive, perinatal dwarfing disorder in which heterozygous carriers are usually without clinical manifestations. We report a girl who has bilobed neutrophil nuclei and a mild skeletal dysplasia. Mutation analysis showed two novel mutations in the *LBR* gene: c.653_655 delinsTGATGAGAAA (p.Ile218MetfsX19) and c.1757G>A (p. Arg586His). Sterol analysis found trace amount of cholesta-8,14-dien-3beta-ol, which is normally undetected in healthy individuals. This case and others previously reported suggest that mutations in LBR can result in a continuum of phenotypic manifestations.

**Anne Heun:** Anne chose the University of Wisconsin-Madison because of its excellent reputation, the diverse opportunities, and the support and knowledge of faculty, supervisors, and the Program Director. She is grateful for the guidance and support she received during her time in the Program and enjoyed the unique opportunities that set UW-Madison apart, such as an internship at the Mayo Clinic, Teaching Assistantships, and a graduate certificate in Consumer Health Advocacy. She is appreciative of the scholarship she received through the Genetic Counselor Training Program Scholarship Fund as it allowed her to focus on becoming the best counselor she can be without having to worry about financial concerns. Anne currently works at the Iowa Health system in Des Moines, at Blank Children’s Hospital and John Stoddard Cancer Center.

**Psychosocial Functioning of Adolescents and Young Adults with Cystic Fibrosis Diagnosed through Newborn Screening or Other Methods**

**Presenter:** Anne Heun  
**Project Advisor:** Audrey Tluczek, PhD, RN

**Objective:** Examine factors contributing to psychosocial function in adolescents diagnosed with cystic fibrosis (CF) through newborn screening (NBS) versus standard practice.

**Methods:** This cross-sectional pathway analysis examined factors associated with psychosocial function of three groups of adolescents ages 16 to 22 years: those diagnosed with CF through NBS (n=13), those diagnosed with CF through standard practice methods at that time (n=26), and healthy peers as a reference (n=42). Data were collected between 2006-2011 from 62 mothers and 8 fathers of

**Results:** Significant results included more depression in parents of adolescents diagnosed through NBS (p=.006-.008); parent-child attachment was related to communication (p=.000). Attachment and communication were associated with adolescent internalizing problems (p=.037, p=.009), emotional symptoms (p=0.018, p=0.022), and personal adjustment (communication only, p=.009). Parent depression was related to adolescent personal adjustment (p=0.022).

**Conclusions:** Regardless of diagnostic group, adolescent with CF report psychosocial function very similar to peers without chronic conditions. Parents of children diagnosed with CF through NBS may be at risk for depressive symptoms when their children reach adolescence and are likely to experience worsening symptoms. Parent depression can adversely affect children’s personal adjustment. Depression screening for parents of adolescents with CF and parent-child relationship assessment is warranted.

**Jenni Mancuso:** Jenni is proud to have completed her graduate studies at the University of Wisconsin-Madison and is especially appreciative of scholarships she received to help fund her education. In addition to working with wonderful genetic counselors and physicians, she enjoyed taking part in unique experiences the training program offers, such as the summer internship at the Mayo Clinic in Rochester, MN, the NSGC annual education conference, and opportunity to work as a teaching assistant and co-instructor for courses in zoology and genetics. She currently works as a Genetic Counselor at the Mayo Clinic in Minnesota.

**Trends, Guideline Usage, and Decision Making in Genetic Testings**

**Presenter:** Jennifer Mancuso

**Project Advisor:** Jennifer Laffin, PhD, FACMG

**Purpose:** To examine trends in genetic testing, utilization of practice guidelines and the decision making process in clinicians ordering genetic tests for patient care.

**Methods:** Physicians and genetic counselors from the University of Wisconsin were asked to complete a survey for each patient they ordered or recommended genetic testing for over a 3 month time period. The survey assessed what type of genetic testing was ordered, other testing that was considered, the use of published guidelines, and factors impacting the clinician’s choice.

**Results:** A total of 117 surveys from 5 departments were collected, representing 220 genetic tests. The most commonly ordered test categories included gene sequencing (51% of tests), and copy number analysis (27% of tests). 78 surveys (67%) indicated that published testing guidelines were used to aid in decision making, 27 (23%) indicated no guidelines were used, and 12 (10%) were unspecified. Practice guidelines were available for 108 of the 117 patient visits surveyed (92%). The most common factors impacting the clinician’s choice were that all likely tests were ordered at once (cited 56 times) and the use of a tiered approach (cited 27 times).

**Conclusions:** Comprehensive genetic testing methods, such as gene sequencing, are being used more frequently in clinical practice than mutation-specific techniques. Practice guidelines are available for the vast majority of patients seen in a genetics clinic, although they are not always utilized to aid in decision making. With the goal of increasing consistency in patient care, there should be a focus on the development or expansion of tools, like Gene Tests, to help clinicians access practice guidelines more efficiently.

**Max Wilson:** Max was honored to have continued his graduate endeavors at the University of Wisconsin. He was drawn to the “hands on” curriculum that allowed him to gain clinical experience while simultaneously building competency through his coursework. He appreciated the challenging clinical rotations, the support of a compassionate Program Director and faculty, the opportunity to delve into a historical research project with Walton O. Schalick III, MD, PhD, as well as participate in several teaching assistantships that furthered his professional development. Max was fortunate to be a recipient of graduate scholarships from the University of Wisconsin Athletic Department, University of Wisconsin Genetic Counselor Training Program, as well as the University Center for Excellence in Developmental Disabilities Graduate Student Award, all of which have deepened his appreciation for philanthropic work; he looks forward to contributing to the education of future
Risk as a Concept for U.S. Clinicians and Patients, 1970-2010

Presenter: Max Wilson
Project Advisor: Walton O. Schalick III, MD, PhD

In its core elements, genetic counseling is about translating the meaning of risk from the clinical and scientific realms into the lay realm for the patient. The purpose of this study was to contribute to the body of knowledge of risk communication to empower genetic counselors to be more effective patient advocates. The study involved a qualitative analysis that consisted of investigations of publications in medical, bioscientific and genetic counseling literatures as well as in lay publications as primary sources offering data into the understanding of risk from approximately 1970-2010. A chronology was created by decade to organize the risk-related primary literature in order to generate an interpretive framework for how risk evolved as a concept in both professional and lay traditions, and was subsequently analyzed against a backdrop of secondary literature regarding risk communication.

Historically, in the lay press, genetic risk initially carried predominantly (and exaggerated) eugenic consequences. Distrust of genetics mirrored a tendency to oversimplify risk as either uniformly correct or incorrect. Risk in the professional literature was originally viewed as an all of none type of phenomenon, but later, was compartmentalized into discrete categories of magnitude (high vs. low risk), origin (somatic vs. genetic), or who is affected by risk (proband, parents, society, humanity). More recently, professional analysis redefines risk as a fantastically complicated construct that is more a reflection of sociocultural boundaries than of empiric data. As clinicians, awareness of the complexity and societal constraints of “risk” is crucial to the efficacy of its communication to a lay audience, as the patient and their families are ultimately responsible for decisions regarding their medical management and reproductive planning.

ALUMNI UPDATES

We love to hear updates from UW-Madison alumni. Please consider sharing any professional updates (e.g. new jobs, honors, memberships, publications, etc.), any personal updates (weddings, births, etc.) as well as any current photos of alumni by emailing Laura Birkeland at lebirkeland@pediatrics.wisc.edu.

- Thanks to Quinn Stein (2000) for his role as Co-Chair of the 2012 AEC in Boston and best wishes to McKinsey Goodenberger (2008) as she embarks on this path for next year’s conference!
- Great work to the following speakers at the 2012 AEC in Boston, MA:
  - Cecelia Bellcross, PhD (1990) Emory University, “Epigenetics: Why DNA Sequence isn’t Everything”
  - Regan Veith (2005) and Amy White (2002), Children’s Hospital of Wisconsin, “The New Landscape of Genetic Testing: how to approach testing minors for adult-onset conditions in the era of large-scale genomic testing”


- Susan (Zucker) Berg (1983) joined the Center for Shared Decision Making at Dartmouth-Hitchcock Medical Center in 2006 where she is the current Interim Program Director. She utilizes her genetic counseling skills while working with patients facing difficult medical decisions and helping clinicians incorporate tools and processes into their practices to increase patients’ involvement in medical decision making, as well as participating in related research and presenting this research both nationally and internationally. She continues to work with genetic counseling colleagues in activities such as creating a patient decision aid about prenatal screening.

- Peggy Modaff (1995) continues to be a wonderful and valued genetic counselor.

- Beth Wood Denne (2000) has left her role as a prenatal genetic counselor at Johns Hopkins and taken a position with Counsyl. Also of note, congratulations are in order as she was elected to be on the ABGC Board of Directors
for a 5-year term starting January 2013, and she and her husband are expecting a baby girl in March of this year.

- **Congratulations to Kristin Rasmussen (2004)** for her promotion to Lead Genetic Counselor of the Marshfield Clinic Medical Genetics Department. Also of significance, she and her family have taken in two fainting goats, apparently as she stated, “genetic interesting in their own right.”

- **Megan Nelson (2010)** has taken a new position working in the Cancer Center at Appleton Medical Center in Appleton, WI.

- **Sara (Knavel) Fisher (2004)** welcomed her second son, Caleb James on August 16, 2012.

- **Laura Birkeland (2007)** welcomed her first son, Oneal, on July 2, 2012.

- **Congratulations to Maureen Flynn (2002).** She and Erik married in September 2011 and honeymooned in January 2012 to Cambodia and Thailand.

- **Emily (Windsor) Decker (2008)** has acquired a husband, Mathew, this this past year and has joined the ranks of Mrs. not Miss.

- **Jay Flanagan (2005)** was awarded the Clinical Employee of the Year for Sanford Health. Quite an accomplishment given that this is a 20,000 employee organization.

- **Three UW alumni Quinn Stein (2000), Lior Borovik (2012) and Jay Flanagan (2005)** are representing UW well at Sanford Health, and all male, too!

### 2012 Alumni Publications

We are certain that this list is an under representation of the work that UW alumni actually completed, so please be certain to send publication updates our way to be included in the next Newsletter by emailing Laura Birkeland at lebirkeland@pediatrics.wisc.edu.


### Annual Alumni Gathering at the 2012 NSGC Annual Education Meeting

Thanks to Maureen Flynn, hostess extraordinaire, for helping to organize the alumni gathering at AEC this past year. It was great to take a break from the talks and get a chance to catch up. Find out where our next AEC UW-Madison Alumni Gathering will be located on our Facebook page (UW Genetic Counselor Training Program Alumni) or the old-fashioned way of looking for flyers on the Message Board at AEC.
Stay in touch and Happy New Year!

We would love to hear from you. Please stay in touch via:

**Facebook:** UW Genetic Counselor Training Program Alumni

**Website:** www.med.wisc.edu/gc

**Email:** reiser@pediatrics.wisc.edu or lebirkeland@pediatrics.wisc.edu

**Mail:** Waisman Center, 1500 Highland Ave, Madison, WI 53705

**Phone:** 608-262-9722

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Your gift to the Genetic Counseling Masters Program Award Fund will help us fulfill our mission of training competent and compassionate genetic counselors who will be lifelong learners and leaders in the field.

This fund was created to support the genetic counseling master's program. It will provide tuition assistance that will allow students to choose the University of Wisconsin-Madison for their graduate training in genetic counseling.

Genetic counselors help individuals and families sort through critical health issues. In a rapidly expanding field, they play an increasing role in research and public health. The University of Wisconsin-Madison Genetic Counseling Master's Program is committed to preparing genetic counselors who will help families and help shape the future of genomic medicine.

Gifts to the Genetic Counseling Masters Program Award Fund are administered through the University of Wisconsin Foundation, the official fundraising and gift-receiving organization for the University of Wisconsin-Madison.

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