International Society of Nurses in Genetics

Newsletter

Caring for People´s Genetic and Genomic Health

Jan-Apr 2013

Contents

President´s Message

Greetings ISONG Members,

Happy 25th anniversary! Let me begin by thanking all of you for your diligence and dedication to ISONG. I look forward to celebrating together 25 years of Leadership in Nursing at our Silver Anniversary October 4-6th in Bethesda, Maryland. The conference brochure is online at www.isong.org and registration is open. Janet Williams will serve as our Keynote Speaker to open the conference, and a Gala Anniversary Dinner is planned for Saturday evening, October 5. We had a record 88 abstracts submitted so that educational content of the conference will expose attendees to the best and brightest research, clinical and educational subject material in our field.

Our committees and special interest groups have been busy conducting conference calls and implementing their goals for 2013. Briefly, our webinars series was very successful, in 2013 and will continue into 2013. As we strive to increase our viability discussion are underway to improve the look of our website. If there are any ISONG web-masters out there, we need you! Our bylaws and policy and procedures are being updated, and efforts are under way to increase our global viability and presence. As we strive to preserve our historical footprint in our archives please do let us know if you have any old photos, conference materials, letters or other documents. They can be sent to ISONG headquarters for safekeeping.

As I reflect on the critical role of genetically trained nurses in clinical practice, research, education, scholarship and policy, I am struck by the many contributions of ISONG members over the past 25 years. Long before the completion of the human genome, ISONG pioneers were forging into unchartered areas for nursing and genetics. I am confident this vision and tenacity will continue into the future, and I look forward seeing all of you in Bethesda.

Very Best,

Kathleen

Kathleen T. Hickey EdD, FNP-BC, ANP-BC, FAHA, FAAN

President of ISONG

Columbia University Medical Center

Bylaws Comittee

Submitted by

Ellen Giarelli

Ellen Giarelli, Bylaws Chair
Kathy Sparbel, Member
Janet Williams, Member
Marlena Kern, Member
Marie Twal, Member

In January 2013, the Membership of ISONG voted on and approved updated language for the Vision and Mission Statement of ISONG. The updated version including in the Bylaws is:

Vision and Mission

The International Society of Nurses in Genetics, ISONG, is a global nursing specialty organization dedicated to genomic health care, education, research, and scholarship.

The vision of ISONG is to care for people›s genetic and genomic health throughout the lifespan and across the continuum of health and disease

The mission of ISONG is to serve both the nursing profession and the public. ISONG fosters and advocates for the scientific and professional development of its members and the nursing community, in the discovery, interpretation, application, and management of genomic information, for the promotion of the public's health and wellbeing. ISONG advocates for public understanding of genomic health and use of genomic information.

A. Provide a forum for education and support for nurses providing genetic- and genomics-based healthcare.

B. Promote the integration of genetics and genomic into the delivery of nursing care, and the engagement of the nursing profession in the delivery of genomics-based healthcare services.

C. Lead in the integration of established nursing competencies in genetics and genomics into all levels of nursing education, practice, and research

D. Provide leadership in the development of standards of practice for nurses in human genetics and genomics.

Contribute to the development of public policy in the areas of genetics and genomics healthcare.

F. Lead in the generation and dissemination of the evidence base for genetics and genomics in nursing.

G. Provide a forum for professional, transdisciplinary, and public dialogue, communication, and networking.

H. Promote interprofessional collaboration in the care of people, families, and communities at risk for genetic conditions.

I. Serve as a source of expertise on genetic and genomic nursing for nurses, other healthcare professionals, lay support groups and the public.

Bylaws survey data:

1. ISONG mission statement approved 100% in favor

2. ISONG vision statement approved 98.4% in favor

3. Changes to the ISONG Education Committee Description 96.9% in favor

Education Committee

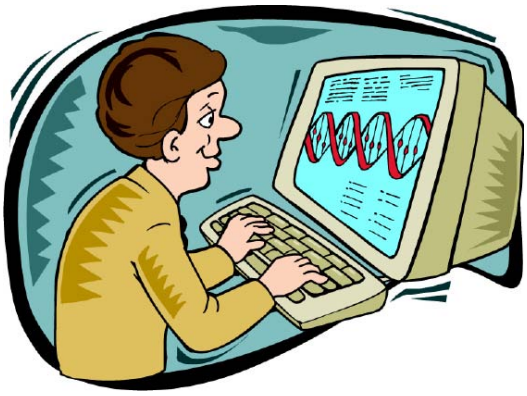
Submitted by

Cathy Read and Sheila Alexander,

Education Committee Co-Chairs

ISONG members plan webinars on genetic/genomic topics.

Many members of ISONG are in the process of preparing webinars that will help disseminate cutting-edge information about genetic and genomic topics. These one to two hour webinars will be offered in a live online format and will also be archived for later viewing through the ISONG website. Webinar participants may earn continuing education credits. ISONG Members will get an email about each upcoming webinar, and we ask that everyone help with marketing by forwarding the message to any relevant groups. More information and details about the procedure for setting up a webinar may be obtained from Cathy Read (readca@bc.edu) or Debbie Zaparoni(isong_admin@msn.com).



Archived ISONG Webinars

The purpose of the International Society Of Nurses in Genetics (ISONG) webinars is to provide ideas for implementing genetic content based on requirements within the Essentials of Master's Education in Nursing (AACN, 2011). Since the completion of genome sequencing in 2003, genomic information has been dramatic in its accumulation need to incorporate into healthcare. Many providers continue to look for methods to learn this dynamic content and how to apply it to clinical practice. These webinars were designed to offer faculty teaching in graduate programs the genetic content for specific "Essentials", resources and some ideas for how to teach this information at the advanced level in nursing. Advanced health care providers may also take away valuable knowledge about genetics/genomics to use in their clinical practice. Visit the ISONG website to view the archived webinars. CE can be provided for a nominal fee and upon completion of the required post-test and evaluation.

Oncology SIG

Submitted by

Lisa B. Aiello-Laws, RN, MSN, AOCNS, APN-C

The Oncology SIG has been reactivated. We met for the first time in October 2012 at the ISONG meeting in Philadelphia. We took a cursory poll of what members wanted from the SIG. Most members stated at this time

they were interested in a mechanism of communication for support and clinical issues. The SIG plans to communicate with the group via email (hopefully by listserv in the future). The emails will state "ISONG Oncology SIG" in the subject line. We will share new research findings, educational opportunities, etc. Any member can also respond to the group to ask for assistance with a clinical case. The group was also interested in sharing resources. The first meeting minutes included the identified resources that members requested at the SIG meeting. The resources included: an educational website for patients, contact information for the Mystery Family Study, information about the Army of Women study, new standard of care regarding BART testing, and language to include on pathology reports sent to practitioners that describes the standard of care for MSI testing. The minutes are located in the ISONG dropbox. A recent announcement also notified members of the new genetics/genomics column in the Oncology Nursing Forum.

If you are a member and have not received any communications, please confirm your contact information with ISONG and the SIG. If you want to be a member, contact ISONG and/or the SIG. Lisa Aiello-Laws has agreed to be the SIG Coordinator. Please contact her at laiell01@villanova.edu. This is the email used for the email communications, so please add it to your address book.

ISONG Newsletter

All ISONG Members are wellcome to contribute in our newsletter:

Please consider sharing reports of your research projects and results

Send us your acoomplishments

Exchange your experience among different cultures by sharing your practice, ethical, social, and legal perspectives

Please contact

Erika M M Santos

at

erikammsantos@gmail.com

Journal of Nursing Scholarship, Genomic Special Issue

Submitted by

Jean jenkins

Ensuring that nurses play a central role in the application of genomics to clinical care is at the core of the 2013 Genomics Special Issue of the *Journal of Nursing Scholarship*. The Issue explores genomic variation and its clinical implications for common diseases in pediatric and adult patients such as cardiovascular diseases, metabolic syndrome and cancer with the implications for nursing practice. Highlights include the genomics of common health conditions, emerging genomic science and technology, and the ethical, legal, social and nursing research issues associated with the translation of genomics into healthcare. All articles included in this special issue are open access and freely available at http://www.genome.gov/27552093.

Genomic Special Issue Webinar Series

As an adjunct to the publication of this special issue, most of the authors have agreed to present more detailed information from their paper and answer questions as part of a series of live open access Webinars.

All webinars are free and were taped for future viewing, accessible at the http://www.genome.gov/27552312.

List of Webminars by Date

February 5, 2013

Relevance of Genomics to Healthcare and Nursing Practice

February 19, 2013

Current and Emerging Technology Approaches in Genomics

March 5, 2013

Cardiovascular Genomics

An Overview of the Genomics of Metabolic Syndrome

March 19, 2013

Implications of Newborn Screening for Nurses and Nursing Faculty

Ethical, Legal, and Social Issues in the Translation of Genomics into Healthcare

April 2, 2013

Integration of Genomics in Cancer Care

Physical, Psychological and Ethical issues in Caring for Individuals with Genetic Skin Disease

April 26, 2013

Genomics and Autism Spectrum Disorder

An Update of Childhood Genetic Disorders

May 7, 2013

A Blueprint for Genomic Nursing Science

Disclosure of Incidental Information Obtained in Exoma and Genoma Sequencing

Submitted by

Erika Santos

The American College of Medical Genetics and Genomics published guidelines about disclosure incidental information obtained in the exoma and genoma sequencing.

The sequencing of the genome and exoma are tools that have become available due to which fallingprice of sequencing and also the advancement of bioinformatics tools. These tools are useful in many clinical situations, such as characterization of rare diseases, individualization of therapy, pharmacogenomics, pre-conception and pre-natal screening, and population screening. However, in all cases there is the possibility of incidental findings (or secondary), which are not related to the indication of the primary sequencing, but it may have utility for the physician or the patient.

Although the literature are discussed ethical aspects, little is said about the practical aspects. The ACMG have established an working group that have met and established a guideline, published in 2013.

The document presents such as clinical, laboratory, patient, primary finding, and incidental finding.

The working group presented a list of clinical conditions whose incidental findings should to be reported in clinical sequencing (which they called exoma and genome sequencing). They selected the most common monogenic diseases. They consider the disease based on clinical validity and utility, and prioritize diseases in which there were preventative measures available, or that patients remain asymptomatic for long periods.

Among the diseases listed are: breast-ovarian cancer hereditary, familial adenomatous polyposis, Marfan syndrome, dilated cardiomyopathy, among others.

An important recommendation is that the findings are contextualized by the professional who requested the examination, considering the data obtained by family history, physical examination, and other sources. It should also be discussed with the patient in the pre-test counseling the possibility of identification of incidental findings, and should be given the opportunity to refuse the patient exam, after the patient evaluates risks and benefits of sequencing.

Regarding the disclosure of incidental findings in children, the working group believes that this is a moment of transition, in which the information about the exoma and genome sequencing still are expensive and not readily available. Thus, in the future, parents will have access to the results of variants adult-onset diseases, and be able to restrict the information to their children. However, currently this is not possible. Therefore, as the potential benefits may be possible, and the identification of incidental findings may be important for the child's parents, the working group recommends that the report of the findings should not be limited by the age of the person whose genome is being sequenced .

In this document they did not address issues related to data maintenance of incidental findings in the patient record. They also did not reported aspects related to incident findings related to newborns screening. Another issue that it was not addressed it was the report the incident findings of research data.

Electronic medical records with Genomic Data

Submitted by

Erika Santos

The use of sequencing exome/ genome is considered a practical reality. However with this technology there is also the obstacle of managing this information.

The electronic medical record based on genomics (genome enable eletronic health record) can receive, store and display genomic information to clinical use, assisting in treatment decision and the individualization of care.

Most EHRs are not prepared to incorporate genetic data, especially from genome sequences and exoma.

This registry is a challenge for practitioners in clinical practice. The interpretation of genomic data is influenced by family history.

The proper integration of genomic information to EHR can reduce the difficulties of professionals in communicating information to patients, which should be considered in the context of risk communication.

Regarding ethical issues: the patient has the right to accept or decline the information itself. All information should be protected and regulated regarding access to confidential information.

The variants included in the medical record must be certified by a valid laboratory with appropriate interpretation. If possible sequencing should provide information with analytical validity necessary to confirm the test.

Once the genome contains a large number of variants that are not in the reference genome, or whose role in disease susceptibility is unknown effort is required to maintain an accurate database with the actionable variants. For the extent that knowledge increases, the data require reinterpretation.

The genome sequence data can be important for clinical decision making available but can be problematic. One solution is to place a minimum value of data in the chart, and the data are reviewed to the extent that new discoveries are identified.

The records can be a source to allow sharing of information, and can also allow the integration of academia with companies that develop software for electronic medical records.

The connection of the genome sequence data with phenotypic information can contribute to the expansion of knowledge. The research questions can be answered more quickly, because patients already have the data recorded. However, ethical issues related to the potential for development of research should be discussed with patients.

Advances in genomics can improve the health of individuals and populations, and the records based on genomics can contribute to better genomic care.

New Loci Associated with Risk of Developing Breast Cancer

Submitted by

Erika Santos

An article published in Nature Genetics presents the identification of new loci associated with risk of breast cancer.

Breast cancer is the most common cancer among women, and there is a significant contribution of inherited susceptibility. Many susceptibility loci contribute to this, including high penetrance gene (BRCA1, BRCA2), moderate penetrance (CHEK2, ATM), and alleles identified in the GWAS.

For this study we performed a meta-analysis of nine GWAS, including 10,052 cases and 12,575 controls. They identified 35,085 SNPs, and drew an Illumina iSelect genotyping array (iCOGs). This array included 211,155 SNPs. After quality control data were obtained from 199,961 SNPs in 52,675 cases and 49,436 controls.

Of the 27 loci that have been previously identified as associated with breast cancer, all but four were associated with risk of breast cancer.

When the results of the GWAS and iCOGs 263 were combined, and 37 SNPs were identified new regions.

Consistent with the pattern seen in other loci, there is strong evidence of specific tumor subtype. Of the 13 loci, the OR was higher for ER-positive than for ER-negative and in most cases, associations were not observed with ER-negative disease.

Four of the new loci identified (rs16857609 at 2q35, rs10759243 at 9q31, rs1195914 at 10q26 and rs2588809 at 14q24) were in regions that were previously associated with cancer risk.

The rs2588809, which is near the RAD18 had association with ER-positive disease, while SNPs (rs999737 and rs104838813) were associated with ER-positive and ER-negative diseases.

They identified two loci near genes of susceptibility. The rs11571833 which is a variant of BRCA2 and rs132390 SNP at 22q13 and which is near the CHEK2.

The results indicate that variants are associated with modest risk of cancer, but research is needed to determine whether this association is associated with risk or linkage desequilibrium.

The other loci were identified intronic or intergenic.

The identification of genes and variants need more refined evaluation, including functional assessment. They identified 53 genes with new associations.

One of the contributions is that the susceptibility loci has a multiplicative combination and they identified 41 new loci explaining 5% of the familial risk of cancer, with a risk of 2 to 3 times higher than the general population. However, excessive significant associations between the SNPs suggest that more susceptibility loci are involved.

Michailidou K et al. Large-scale genotyping Identifies 41 new loci associated with breast cancer risk. Nature Genetics 2013; 43: 353-363.

Genomic Medicine in Clinical Practice

Submitted by

Erika Santos

The potential for application to application of genomic medicine has been recognized, and it is defined as the use of genotypic information for clinical care. This definition is related Mendelian diseases, and also the complex multigenic diseases. While the current focus of genomic medicine has been monogenic Mendelian diseases this emphasis is being modified for the increasing attention to variants.

This article published in Genetics in Medicine summarizes the discussions placed at colloquim from National Human Genome Research Institute that was held on 29 June 2011 that evaluated the opportunities and barriers to the implementation of genomic medicine.

The paper summarizes the applications of genomic medicine. They presented four examples of applications of genomic medicine that has been used in clinical practice.

Many centers have used screening for mutations of high penetrance genes for Lynch Syndromes and Hereditary Breast-Ovarian to identify high-risk individuals. The follow-up of these individuals provides the basis for development broader strategies for follow-up of individuals at risk for cancer.

The self-reported family history was also discussed as an important tool of Genomic Medicine, and strategies of the My Family Health Portrait was discussed. However, the integration of family history to the electronic medical record, which may constitute an important tool for Genomic Medicine, finds barriers to implantation.

The Pharmacogenomics is an application of Genomic Medicine, and available information about changes in response to drugs such as clopidogrel, tamoxifen have been used as examples.

The sequencing of the genome exoma and are in use for the identification of rare diseases, particularly those with onset in childhood. A number of universities and commercial laboratories offer teh service that can be used in clinical practice.

Application of Genomic Medicine finds barriers to its application. The biggest one is the lack of recognition by professionals of the benefits of genomic medicine to improve patient care. Even for strategies with proven utility, such as family history, there is a resistance in its adoption in clinical practice.

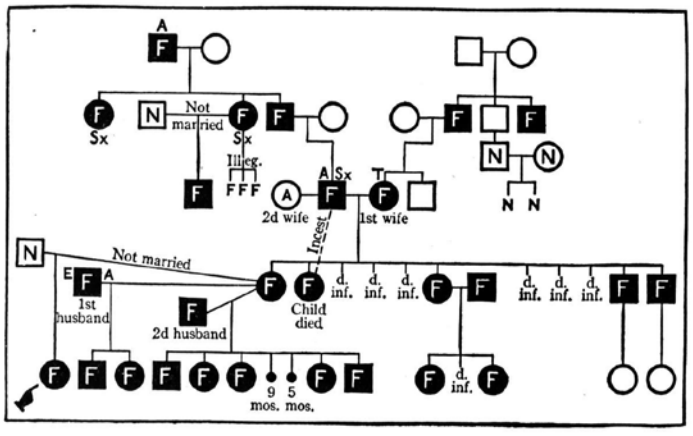


Fig. 499. *Inheritance of Feeble-mindedness* [1]

Another barrier is the use of commission to assess the feasibility of strategies for Genomic Medicine. Institutions use committees that evaluate strategies in isolation. It would be more efficient if the commission acted jointly with a strategy similar to the CDC or EGAPP.

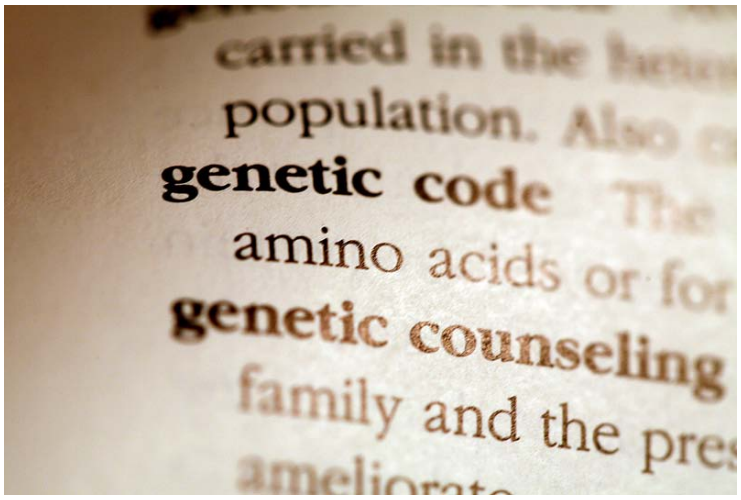
Evaluation of interventions, that are drastic or irreversible, such as prophylactic mastectomy, for example, demand stronger evidence.

The integration of clinical tools to the electronic medical record is required only when the relevant clinical information is indicated. It is necessary to not overload the system with excess genomic information, which may impede the care flow.

The psychological impact has been assessed during the process of informed consent and genetic counseling, but will need expanded evaluation of the impact of genomic medicine.

For the Genomic Medicine is necessary to establish the infrastructure for access information for phenotypic interpretation of genomic data. The research groups work in isolation, and there is the need to establish policies for sharing information. The research groups also have limited access to phenotypic data for the interpretation of results of new variants.

Organizations need to develop guidelines for Genomic Medicine. It is also necessary to establish educational programs directed to clinicians still early in training. It is suggested a fellowship in pharmacogenomics, and given



the relevance of genomic medicine, it is necessary training and certification.

One of the research efforts is the establishment of valid variants with clinical utility. Consent must be obtained in the incorporation of interventions, considering the complexity, cost, risk / benefit comparisons and results.

The implementation of genomic results is regarded as a cultural and political exercise, with scientific recommendations as structural basis. It is necessary to integrate the policy of the institution, and is a work of endurance. The engagement of institutional leadership is critical.

Maniolo et al. Implementing genomic medicine in the clinic: the future is here. Genetics in Medicine 2013, 15:258-267.