Greetings to ISONG Members!

As you read this newsletter, you will be pleased to see that ISONG continues to be strong and led by a dedicated and hard working group of volunteers who serve on the Board of Directors, as chairs and co-chairs, and as committee members. It is an honor to work with this group of professionals who promote the vision and mission of ISONG.

The 2009 Annual Conference Program Committee is busy organizing for the October 16-19, 2009 preconference and annual conference to be held in San Diego, CA. In the preconference you can learn about implications of genomics for nursing practice, and curriculum integration and faculty resources. The annual conference is a great opportunity to catch up on recent genetic and genomic initiatives as well as network with others.

I want to highlight a few happenings since our fall Newsletter.

- As part of our collaborative efforts, our Japanese members and President-elect Karen Greco working with the American Nurses Association have completed a Japanese translation of ISONG’s *Genetics and Genomics Nursing: Scope and Standards of Practice*;

- ISONG and the American Nurses Association co-published the book, *Genetics and Ethics in Health Care: New Questions in the Age of Genomic Health*, edited by Rita Monson. If you are interested in purchasing a book, please notify ISONG Headquarters;
President's Message

• ISONG will be coordinating the 2010 annual conference in Dallas, TX with the National Society of Genetic Counselors.

Through these collaborations, we will facilitate and contribute to genetic and genomic health worldwide.

We hope that you enjoy receiving the electronic quarterly Newsletter and monthly Update. Let us know your ideas, and consider becoming more active in ISONG’s committees.

Agatha Gallo
ISONG President

ISONG Late Submission Policy

ISONG

Late Submission Policy
For Abstract or Grant Proposal

Late submissions will be considered for reasons such as: death of an immediate family member of the Principal Investigator/First Author, sudden acute severe illness of the Principal Investigator or immediate family member, or natural disasters or storms that result in inability to submit on time.

A letter identifying the reason for request of a late application must be submitted electronically or by mail. While the reasons are sometimes personal in nature, an objective evaluation of their merit requires that some details be provided. Specific information about the timing and nature of the cause of the delay is necessary so that a decision can be made. Only the explanatory letter is needed; no other documentation is expected.

An application for late submission of an abstract for presentation at the annual meeting will be considered by the conference abstract subcommittee. An application for late submission of a grant application will be considered by the research committee. The reasons for the delay and the allowable length of delay will be carefully considered by the appropriate committee. To provide the committee the opportunity to plan appropriately, it is expected that the request for late submission be submitted as soon as possible but no later than seven days after the original due date.
GNCC Report

Submitted by
Jeanine Seguin Santelli, PhD, ANP-BC/GNP-BC
GNCC Executive Director

APNG and GCN Portfolio application order forms are available on our website homepage http://www.geneticnurse.org/. The next portfolio deadline is March 1, 2010.

Credentialing Class of 2004, its time to start putting the pieces together for your renewal! Renewals are due September 1, 2009. The guidelines can be found on our website www.geneticnurse.org. Just click on either the APNG or GCN button to access the file.

Online Genetics Courses for Nurses at The University of Iowa

Submitted by
Janet Williams

For the past three years, The University of Iowa College of Nursing has offered online genetics courses for nurses in undergraduate or graduate degree programs as well as nurses or other health care professionals taking courses for professional improvement. Three courses are in the online program. The first, Introduction to Human Genetics course [96:116] is a 3 credit hour course, offered in the fall semester each year. This course provides an introduction to the organization of the human genome and basic principles of inheritance in humans. Course content includes an overview of cells and development, organization of the human genome, chromosome structure and function, gene structure and function, genes in pedigrees and populations, and the implications of genetic variation on health. This course was designed for upper level undergraduate or graduate students. Students with backgrounds in nursing and other healthcare professions as well as students preparing for roles in a healthcare profession – have taken this course.

Starting in 2009, nurses, and other health care professionals, will have more flexibility in registering for the content in the clinical practice courses by selecting content modules that meet their individual education needs. Students have said “I have never learned as much information in one class” and “Dr Daack-Hirsch was a terrific on line instructor. She was organized, provided appropriate and informative feedback, and was available during her office hours for telephone contact. Thank you for making this wonderful learning opportunity available to those of us across the country who do not have such access otherwise!”

The Advanced Practice in Genetic Nursing I (96:228) and Advanced Practice in Genetic Nursing II (96:230) courses are offered in the fall semester odd years [next 96:228 course in fall 2009] and spring semester even years [next 96:230 course 2010]. Students can now select up to 3 content modules from 96:228 and up to 3 modules from 96:230 Advanced Practice in Genetic Nursing II. Each module is equivalent to one credit hour. Topics for the modules include:

• Module 1 - Basic Risk Assessment (pedigree construction and interpretation)
• Module 2 - Advance Risk Assessment (Bayesian analysis, nontraditional inheritance and dysmorphology)
• Module 3 - Molecular Genetics-implications for testing and ELSI
• Module 4 - Genomics and Healthcare delivery (primary care and public health)
• Module 5 - Common Genetic Disorders (Childhood onset)
• Module 6 - Common Genetic Disorders (Adult onset)
To register for these courses, offered through The University of Iowa’s Center for Credit Programs, call 1-800-272-6430, or complete the online registration form at http://www.continuetolearn.uiowa.edu/ccp/de/regform.htm. To learn more about the courses, please contact Dr. Sandra Daack-Hirsch, at 319-335-9967 [sandra-daack-hirsch@uiowa.edu]; to learn more about undergraduate, genetics education opportunities, and the T32 postdoctoral clinical genetics research fellowship at The University of Iowa, please contact Dr. Janet Williams, at 319-335-7046 [janet-williams@uiowa.edu].

**Genetics and Rare Diseases Information Center (GARD)**

*Submitted by Dale Lea*

The Genetic and Rare Diseases Information Center (GARD) was created in 2002 by the National Human Genome Research Institute (NHGRI) and the Office of Rare Diseases Research (ORDR) - two agencies at the National Institutes of Health (NIH) - to help people find useful information about genetic and rare diseases. GARD provides immediate, virtually round-the-clock access to experienced information specialists who can furnish current and accurate information - in both English and Spanish - about genetic and rare diseases. So far, GARD has responded to over 15,500 inquiries on rare and genetic diseases. Requests come not only from patients and their families, but also from physicians, nurses and other health-care professionals.

Contact information for GARD can be found at http://www.genome.gov/10000409. GARD has also launched a web site where nurses, patients and other providers and members of the general public can go to seek information. The GARD web site is at http://rarediseases.info.nih.gov/GARD/

**DNA Day April 25, 2009**

*Submitted by Virginia Minichiello, MS, RN,C*

A month ago, Dale Lea sent out a memo to all of our ISONG members, informing them that again this year, the NHGRI will be sponsoring an on-line chat. As the date of the DNA DAY is on a Saturday, the on-line chat will be held on Friday April 24th. Also mentioned was the fact that NHGRI had launched a National DNA DAY Facebook page as a means to reach out to more students. As a participant with NHGRI, our organization is also linked on the Facebook DNA Day page (http://www.facebook.com/pages/Bethesda-MD/National-DNA-DAY/47309007669)

If you have not had an opportunity to visit the website, I urge you to do so and to share it with everyone you know that might be interested in the DNA DAY activities. By providing the link to our organization, it is hoped that it will generate interest in our mission. Think of the youth you know, either personally or through your professional contacts and send them the link via email. We will be able to track the number of hits coming from the facebook link and that will help to demonstrate the value of this outreach effort.

As last year, we hope you will promote, discuss, and inform others of the wonderful work being done through the NHGRI and the many organizations affiliated with them for DNA DAY.
Genetics and Genomics Nursing: Scope and Standards of Practice Translated into Japanese

Submitted by Karen Greco

ISONG is pleased to announce that the *Genetics and Genomics Nursing: Scope and Standards of Practice*, that ISONG co-published with the American Nurses Association (ANA) in 2007, is now being translated into Japanese. After several months of negotiations, ANA, ISONG, and the Japanese Society of Genetic Nurses (JSGN) have signed a contract in which ANA and ISONG grant JSGN rights to translate the book into Japanese and distribute a translated copy in Japan, without charge (except postage), to those who purchase an English copy of the book. This is the first time an official ISONG publication has been translated into another language, making this a historic event for ISONG. Beth Kassalen, ANA CEO Linda Stierle signed contract on behalf of ANA, ISONG executive director, signed the contract on behalf of ISONG and Dr. Michiko Mizoguchi, JSGN president, signed the contract on behalf of JSGN.

ANA is pleased that the scope and standards are being translated into Japanese and were generous with the contract terms. According to Rosanne Roe at ANA, “ANA is excited to have this contract signed and it is receiving recognition within ANA at the corporate level. We are hoping that other translations will follow.”

According to ANA, over 850 copies of *Genetics and Genomics Nursing: Scope and Standards of Practice* have been sold to date. If you would like more information about the book, a table of contents and purchasing information can be found on the ISONG webpage or you can call ANA at 1-800-637-0323. ISONG members living in Japan interested in purchasing a copy of the English version of the scope and standards, along with the Japanese translation, can contact the Japanese Society of Genetic Nurses by emailing Dr. Mizoguchi, JSGN President, at mizomichi@is.icc.u-tokai.ac.jp.
Committee Reports

Awards and By-laws Committees

Submitted by
Susan Tinley

As past-president I chair two committees, the Awards Committee and the By-Laws Committee. There is nothing to report at this time for either committee because there has not been a need for any activity yet. However, I am recruiting members for both committees. The Awards Committee will request nominations for the Founders’ Awards for Service, Education and Research this summer. The committee will then select the recipients from those who are nominated and the awards are presented at the annual meeting. I am looking for members who are willing to assist with this process, an important yet not terribly time-consuming contribution to the organization.

The By-Laws committee is charged with drafting any changes to the By-Laws that are requested and then putting those potential changes up for a vote of the membership. There may or may not be a need for any changes in the By-Laws but I am recruiting members for the committee to help me review the By-Laws for areas where change should be considered by the Board. We would draft any changes that the Board deems necessary.

If you can help with either of these committees please send an e-mail to:
tinley@creighton.edu

Ethics and Public Policy Committee

Submitted by
Ellen Giarelli

Committee Purpose: This Committee evaluates ethical and policy issues related to scientific advances and political changes. The committee weighs the need to prepare a response on behalf of the Society. Members recommend and establish criteria, with approval by the Board of Directors, for such evaluations that reflects the Vision, Mission and goals of the Society. Specifically, the committee writes position statements and commentaries.

The Ethics and Public Policy Committee has established the following goals for the committee.

1. Update membership and explore formation of subgroups.
2. Identify and get contact information for International organizations that address ethical, social and policy issues related to genetics in general and genetic nursing in particular.
3. Prepare a POSITION STATMENT on the issue of Direct to Consumer Marketing of Genetic tests.
4. Identify issues for which ISONG may wish to prepare statements of position.
5. The committee will hold at least 2 teleconferences

Since the last board meeting, I am reporting on the following:

• Goal: Update membership and explore formation of subgroups.

Action taken: Reviewed and updated members list, twice.

Status report: New email addresses are needed for Elizabeth Twiner and Kathleen Weil. Michelle Bishop
Committee Reports

and Heather Skirton are no longer on the committee. 20 members on roster as of Feb 1, 2009. Core group of 6 active members.

• Goal: Identify and get contact information for International organizations that address ethical, social and policy issues related to genetics in general and genetic nursing in particular.

   Action taken: Martha Turner is working on this goal and has prepared a letter to be sent to the list she is compiling. She will prepare collect information from the orgs as it arrived and report on the status of this goal at the next meeting.

   Status report: In progress. Martha will send letter to the organization on behalf of EPPC.

• Goal: Prepare a POSITION STATEMENT on the issue of Direct to Consumer Marketing of Genetic tests.

   Action taken: The subcommittee developed a draft of a position statement. The subcommittee is composed of Cheedy, Pam, and Marie. They met earlier in the month of February and have been working on background information since the November meeting in Philadelphia; Cheedy prepared a summary document of the issues related to DCT marking of GT; Marie Twal prepared a summary statement on background information; Information has been solicited from international members of ISONG to assure that our position incorporates all relevant perspectives; See list below of concerns.

   Status report: The next step is to prepare a preliminary draft. The subcommittee will work through the month of March to prepare a preliminary draft with a target date of March 31st. At that time the subcommittee will circulate the draft to EPPC for the first review and comment.; Review of the drafts of the position on DTC will be conducted via email. A teleconference will be planned for late Spring.

• Goal: Identify issues for which ISONG may wish to prepare statements of position

   Action taken: The existing ISONG POLICY: Definitions, Process and Format of Position Statement needs review and revision; Additional issues: Biobanking and public genetic databases. Pam Williams will start to collect some background information. Assistive Reproductive Technology. Pharmacogenomics. Personalized medicine.

   Status report: To be worked on.

• Goal: Small Project: to evaluate the availability of audio-visual instructional materials related to ethical issues in genetics/genomics for nurses

   Status report: No action on this goal.

• Goal: The committee will hold at least 2 teleconferences

   Action taken: The DTC subcommittee has met on two occasions. EPPC had a conference call on February 11, 2009, 12-1PM. Attended by : Ellen Giarelli, Pam Williams, Cheedy Yaya, Marie Twal, Dale Lea, Lynn Woods.

   Status report: The committee will meet at the end of March or early April to discuss goals.
The following is a partial list of concerns related to DTC marketing of genetic technology:

1. **Exaggerated claims regarding genetic information** – Encouraging in the public’s mind the notion that genes play a deciding role in many diseases at the expense of addressing issues of poverty, pollutants, and life-style behaviors that have been shown to have a far greater impact on the development of many diseases. Unrealistic expectations should not be a reason to submit to genetic testing.

2. **Psychological effect** – While one of the stated objectives of genetic testing is to change lifestyle behaviors if a genetic risk is indicated data suggests that some people are unable to make such changes (personal will-power, economic factors, employment imperatives, etc.) and instead become depressed or fatalistic. Who will be watching out for them?

3. **Varying risks in different populations** – As data from the BRCA1 and BRCA2 genetic testing studies reveals, the risks of developing breast cancer for those with the gene and a family history differs from the risk for those with the gene but no family history, and the risk for those without the gene. These risks projections continue to be revised as our knowledge of the genetic. A similar scenario may be expected for the many other genes that have been associated with specific diseases in populations suspected to be at risk for the disease due to family history.

4. **Allocation of research grants and the impact of patents** – It has been estimated that 90% of research focuses on 10% of the global disease burden, primarily chronic diseases affecting those in the Western world. The genetic testing industry has the possibility of encouraging genetic research which will be used by those in the Western world. Could medical research put more resources in addressing concerns of “rich healthy people” at the expense of the actual disease-related burden of those in the third world?

5. **Genetic discrimination** – GINA was recently passed in the United States. We need to monitor this law carefully to be sure it accomplishes its intended goal of preventing genetic discrimination. We also need to be aware of the personal and/or economic risk individuals in other countries face.

6. **Social Justice** – Few national health care systems can we afford mass treatment of individuals possessing a specific gene to prevent a small percentage of those individuals from getting ill. Would we be taking money from other health care programs or needed social programs to pay for speculative illness?

Follow-up plans:
1. Review of the drafts of the position on DTC will be conducted via email. A teleconference will be planned for late Spring
2. Martha T. will send a letter to the International organizations on behalf of EPPC soliciting information on how they deal with EPP issues.
3. The committee will hold a teleconference at the end of March or early April to discuss goals and progress on position statement.
Global Membership Committee

Co-chairs
Ida Spruill (Domestic)
Sivia Barnoy (International)

In our role as Co-Chairs of the Global Membership Committee for ISONG, we have established the following goals for the committee.

1. Recruitment of new members.
2. Retention of members.
3. Activate ISONG BUDDY SYSTEM.
4. Respond to needs of membership in timely manner and
5. Diversify the membership (International and Domestic)

Since the last board meeting, we are reporting on the following:

• Goal: Recruitment
  Action taken: New revised membership application available
  Status report: Posted on Web-Page
  Action taken: Designed a welcome letter for new members
  Status report: Need to work with Beth and Debbie to send welcome letter to new members

• Goal: Retention
  Action taken: Developed survey; and it was mailed by Beth to 135 members who did not renew 11-28-08. I followed up with 35 and got 2 responses.
  Status report: Only 35 responded - 69.2% ISONG did not help professionally; 57.1% misplaced renewal; 83.3% never received a renewal notice; 83.3 request multiple renewal notices; 47.6% desire to apply on-line; 55.6% would like a renewal notice;

• Goal: BUDDY System
  Action taken: Created profile of buddies on a spreadsheet, and sent update on who need a buddy and who want a buddy from Debbie’s 12-08 updated list; sent second request for Buddies to entire membership re Buddy System; On Super Bowl Sunday.
  Status report - 12-08: 12 members requested Buddies; 7 members Volunteered to be Buddies; over 75 e-mails sent out from Global Committee regarding the Buddy System to members interested in the Buddy System
  Status report - 2-1-09: match over 20 Buddies/Matched list will be provided later; 1 member still not matched because of interest area in Lab;

• Goal: Member needs
  Action taken: Sent e-mail to Global membership committee re the new membership application
  Status report: Need to plan a conference call
  Action taken: Received new member list from Debbie Zaparoni 10-08
  Status report: Received new member list from Debbie Zaparoni 10-08
Committee Reports

• Goal: Diversity

*Action taken:* Need informational display at NCEMNA conference in March, 2009, or flyers for registration packets. I plan to attend.

*Status report:* Current pictures on web-page are not inclusive, and do not include international nurses nor ethnic minority nurses. Will submit photos to Web-manager

Follow-up plans:
1. Continue to work out logistics for recruiting and matching Buddies
2. Send renewal reminders to members
3. Send welcome letters to new members
4. Develop plans for information sharing at professional conferences
5. Revisit standardized membership renewal dates

Nominations Committee Report

*Co-Chairs*
*Ann Cashion*
*Heather Skirton*

The following goals for Nominations Committee include:
1. Establishing a 3-4 member Nominations Committee.
2. Providing a slate of officers to include president-elect, secretary and member at large, for the 2009 ISONG Elections.
3. Overseeing an electronic voting process.
4. Reporting the voting results via listserv and newsletter.

Since the last board meeting, we are reporting on the following:

• Goal: providing a slate of officers to include president-elect, secretary and member at large, for the 2009 ISONG Elections.

*Action taken:* Nominations are being sought. One person has called since last BOD meeting to self-nominate.

*Status report:* Announcement will be placed in newsletter.
Meeting Report

Submitted by
Sharon J Olsen, MS, RN, AOCN
Assistant Professor
CNS Track Coordinator
Johns Hopkins University, School of Nursing

The National Advisory Council for Human Genome Research (NACHGR) met on February 9, 2009. You can download the agenda at www.genome.gov/27530008 and Dr. Alan Guttmacher’s Acting Director’s Report (which has links to several pertinent articles and press releases at www.genome.gov/27530011).

Some highlights:

• The NHGRI, as part of its long-range planning process, has issued 4 white papers (just 4-5 pages each in length) for comment by February 27, 2009: 1) Applying Genomics to Clinical Problems—Diagnostics, Preventive Medicine, Pharmacogenomics 2) Applying Genomics to Clinical Problems – Therapeutics 3) A Vision for the Future of Genomics: Education and Community Engagement and 4) The Future of Genome Sequencing. Specifically, NHGRI seeks input on whether these are the right questions to ask and whether other questions should be considered. You can view comments and add your own at: www.genome.gov/10001307

• Family History Activities: In January 2009, a new family history tool was released by the Surgeon General, “My Family Health Portrait,” that is compatible with electronic health records/personal health records and more comprehensive. There is also a help desk for consumers. NHGRI’s Genomic Healthcare Branch played a lead role in creating this tool and developing national standards for family history in electronic health records/personal health records. https://familyhistory.hhs.gov/

• Large Cohort Study: Kathy Hudson, PhD, Director Genetics & Public Policy Center presented study results on “Public Perspectives on a Proposed Large Cohort Study of Genes, Environment and Health.” Most participants were supportive of a large cohort study and creation of national biobank. Of note, were views on the researcher-participant relationship (we say “consent”; they say “contract”) and the fact that the return of research results to participants had the most impact on willingness to participate. www.dnapolicy.org/pub.reports.php?action=detail&report_id=27; www.dnapolicy.org/pub.bib.html

• NIH Peer Review Process. Changes to enhance review process and transparency include: shorter R01 applications (12 pages); scoring – 9-point scale instead of 5-point scale; “impact score” instead of “priority score,” more emphasis on research significance than methodological weakness; engage best reviewers by having service over a 6-year period for increased flexibility, “virtual reviews” to reduce travel; revised review criteria and templates for written critiques. http://enhancing-peer-review.nih.gov/index.html; http://grants.nih.gov/grants/guide/notice-files/NOT-OD-09-023.html
• Race, Ethnicity and Gender Issues: The Biennial Report on Inclusion of Women and Minorities in NHGRI-Supported Research was presented. NHGRI partnered with NCI and NCMHD and held a workshop in September 2008 on Ethnicity and Genetics: Understanding the Role of Genomics in Racial and Health Disparities.

• ELSI held a Natural Selection Workshop in Oct. 2008 to identify future directions and best practices for research regarding natural selection.

• ENCODE and modENCODE data release policy finalized in October 2008. Human Microbiome Project (HMP) has begun sequencing samples from 5 body sites and initiated research to explore how microbes interact with the human body to affect health and disease. http://genome.gov/pfv.cfm?pageID=27528386

• Updates were presented on NHGRI’s Program Portfolio, GTEX (Genotype-Tissue Expression), 1000 Genomes, PhenX (Phenotypes and eXposures) and MGC (Mammalian Gene Collection). The MGC project has been completed. NHGRI’s Advanced DNA Sequencing Technology Program: www.genome.gov/Pages/News/Documents/GenomesatOneTen-ThousandthCost-Schloss.pdf

Publications/Findings of Interest

• Science magazine (12/19/08) cited Cancer Genes as the number 3 Breakthrough of the year and Genome Technologies as number 10.

• Nature – News (12/18–25/08) called 2008 as the year of personal genomics and cited the 1000 genomes project.

• New England Journal of Medicine will feature a series of articles on Genomics in Medicine, starting in 2010, edited by Dr. Gregory Feero, Dr. Alan Guttmacher and Dr. Francis Collins. Planned topics include disease risk assessment/GWAS, type 2 diabetes/obesity, mental retardation and autism, genomics and eye disorders, new therapeutic approaches to Mendelian disorders, cardiovascular disease and stroke, pharmacogenomics, ELSI issues.

• Cleft lip Gene Variant Identified: A one-nucleotide difference in a gene involved in facial development may account for ~20% of isolated cleft lip cases (SNP rs642961 in IRF-6 gene, G >A). www.genome.gov/27528380

• Adult and childhood obesity: six new genetic variants were identified, two variants confirmed. www.genome.gov/27529231

• Lung cancer: The Tumor Sequencing Project (TSP) has identified 26 genes that are frequently mutated in lung cancer (including tumor suppressor genes NF1, ATM, RB1, APC), detailed key pathways and described patterns of genetic mutations in smokers and non-smokers with lung cancer. www.genome.gov/27528559
  • Progeria: experimental anti-cancer drug, tipifarnib, prevents, reverses cardiovascular damage in mouse model. www.genome.gov/27528377

• Long-lasting Immunity: protein, SAP, identified that enables T and B cells to interact in crucial way for establishing long-lasting immunity after an infection. www.genome.gov/27528397

Policy Updates and Outreach

• Regulations are being written by the Department of Labor, Department of Health and Human Services and the EEOC for GINA (Genetic Information Nondiscrimination Act), which was passed in May 2008. Implementation for health insurance will be in May and for employment in November 2009.

• There was an Appropriations Update – American Recovery and Reinvestment Act of 2009 and a comparison of the Senate and House versions in regards to NIH funding.

• Darwin Day (2/12/09), the 200th anniversary of his birth: NHGRI and the National Museum of Natural History sponsored a program for high school students and the public at the Smithsonian. www.genome.gov/27529500

• DNA Day: April 25, 2009
Copy Number Variation and Human Disease

Submitted by
Erika Santos, BSN, PhD

In the article “The Year in Human and Medical Genetics – Highlights of 2007-2008” Moyra Smith (2009) has selected eight topics that were considered the highlights of the year. The topics selected for the review were: structural and copy number variants (CNVs) in the human genome; progress in defining genetic factors in the etiology of schizophrenia; Micro RNAs in central nervous system development and function; progress in elucidation of risk factors for complex common disorders through large-scale association studies; epigenetics and the epigenomic era; reprogramming of somatic cell nuclei to generate pluripotent stem cells; new concepts regarding factors involved in sexual differentiation; and new discoveries on the role of Duffy blood group antigens in malaria and HIV-AIDS. In the research update section in this and the next issues we will summarize some of the findings regarding these topics. In this issue we will present some data regarding CNVs.

The structural variation of the genome has gained much attention in the field of human genetics. Genetic variation can range from large, chromosome abnormalities to single-nucleotide variations. Genomic rearrangements are the first known form of mutations. Until now it has been believed that the most abundant source of genetic variation in humans were single nucleotide polymorphisms (SNPs). However, the importance of copy number variants (CNVs) has been revised, and it has been considered that CNVs are known to be a frequent form of common genetic variation and represent a substantial proportion of total genetic variability. (Beckmann et al., 2008; Redon et al.,2006)

CNVs can be defined as a DNA segment ranging from one kilobase to several megabases and present at variable copy number in comparison with a reference genome. (Redon et al.,2006; Sebat, 2007)

The first evidence that copy-number variation can influence human phenotype came from diseases named genomic disorders. These disorders are consequence of abnormal dosage or dysregulation of one or more genes resulting from rearrangement of the genome. (Lupski e Stankiewicz, 2005). Although the idea of CNV is not new, the attention of the scientific community regarding the idea of CNVs as source of genetic variation that affects all chromosomal domains is relatively recent.

In 2006 Redon et al (2006) published an article that evaluated CNVs in the human genome. They evaluated 270 individuals from four populations (the HapMap collection), and a total of 1,447 copy number variable regions (CNVRs), covering 12% (approximately 360Mb), were identified. At this time, they found that 285 out 1,961 (14.5%) gene in the OMIM morbid map overlapped with CNVs.
CNVs may be neutral with no effect in phenotype, but can be pathogenic through gene disruption or fusion, modification of gene dosage or regulation of expression. There are Mendelian inherited entities affected by CNVs (Idiopathic chronic pancreatitis, Prader-Willi/Angelman syndrome, Neurofibromatosis type 1, Charcot-Marie-Tooth type 1A, Rett like syndrome).

There are several conditions with neuropsychiatric alterations that are associated to CNVs. For example, approximately 25% of individuals with the 22q11.2 deletion syndrome have psychiatric manifestations such as schizophrenia, attention-deficit hyper activity disorder or autism spectrum disorders. (Cook Jr and Scherer, 2008)

Several studies reporting CNV in schizophrenia has been reported by St Clair (2009). Copy number variations at 1q21, 15q11.2, 15q13.3, 16p11.2, 22q12, and Neurexin 1 loci have been related with schizophrenia. According to St Clair these findings have several implications: carriers of some the novo and inherited CNVs (especially when they disrupt gene function) are at high risk of developing schizophrenia; these alterations are related to additional psychiatric disorders; it is also possible to explain why some cases of schizophrenia appear to be familiar and other cases appear to be sporadic; so far, CNVs account for 2% to 4% of schizophrenia´s cases, but according to more optimistic opinions, CNVs may account for 20% of cases; it will be possible to evaluate the environmental factors that influence penetrance and expressivity; finally, there are implications in genetic counseling, once the risk of schizophrenia is lower in siblings of individuals with de novo CNVs.

It should be noticed that the research regarding CNV and gene expression is in the early stages.

Oetting (2008) point out that the results from genetic variants studies must to be analyzed carefully, because it seems that we are entering the age of “recreomics” – a premature commercialization of genetic testing without adequate interpretation provided by health care professionals.

References

St Clair D. Copy number variation and schizophrenia. Schizophr Bull 2009;35:9-12.
Genomic Pipeline: From Bench to Practice

22nd Annual ISONG Conference
The Catamaran Resort and Spa
San Diego, California USA

Pre-conference Workshops:
October 16, 2009

ISONG Conference:
October 17-19, 2009