



香港醫學遺傳學會  
Hong Kong Society of Medical Genetics

# HKSMG

## Newsletter

Issue no. 5; Dec 2009

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### Message from the Editor

This is the last issue of the HKSMG Newsletter in 2009. In this issue we have a scientific article, contributed by Dr. Nelson Tang, on his achievement in the Joint Metabolic Clinic-Laboratory in Hong Kong for diagnosis of inherited metabolic diseases, also called inborn errors of metabolism. By comparison with data from other parts of the world, Hong Kong seems that only a fraction of the expected cases have been identified. Specialized laboratory facilities for accurate and efficient biochemical and genetic diagnosis are needed to increase the rate of detection. There is also a brief report on two scientific seminars on clinical applications of microarray in disease investigation and diagnosis. Apart from up-coming international conferences, we have new session of members' publications. Last but not least, on behalf of the society we would like to take this opportunity to wish all members a Merry Christmas and Happy New Year.

**Ronald Wang**

Editor

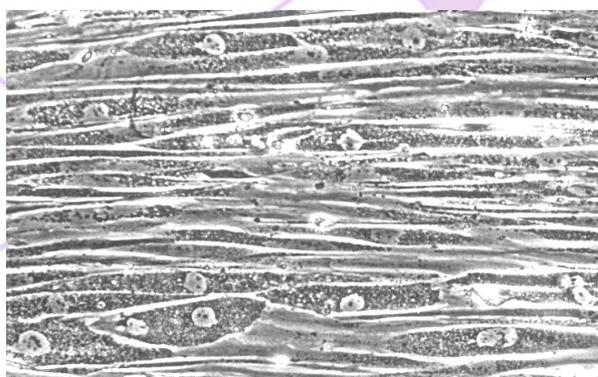
### Scientific Article

**Common genetic defects in the fatty acid  $\beta$ -oxidation pathway in Chinese.  
A review of cases seen in the Joint Metabolic Clinic-Laboratory: a laboratory specialized for inherited metabolic diseases in Hong Kong.**

*Nelson LS Tang, LK Law, Joannie Hui ©*

*Joint Metabolic Clinic, Prince of Wales Hospital/The Chinese University of Hong Kong.*

The Joint Metabolic Clinic (JMC) at Prince of Wales Hospital/The Chinese University of Hong Kong is the first multi-disciplinary Metabolic Clinic in Hong Kong, which has been operating for more than 10 years. The clinic takes care of patients suffered from inherited metabolic diseases (IMD) which is a collective term for a group of hundreds of diseases caused by defects in metabolic pathways. It is also known as inborn errors of metabolism (IEM). The clinic receives input from multiple disciplines; include Pediatrics, Chemical Pathology, Anatomical and Cellular Pathology, Obstetrics and Gynecology, and Bone Marrow Transplantation teams. The JMC-laboratory, led by Dr. Nelson Tang, supports first-line diagnostic investigations, like plasma and urine metabolites (organic acids and carnitine profiling); advanced investigations, like skin fibroblast cultures (Figure 1); enzyme assays and mutation analysis which are essential for making a definitive diagnosis. Samples



**Figure 1.** Skin fibroblast cultures for investigation of inherited metabolic diseases. The Joint Metabolic Clinic - Laboratory pioneered the use of cultured fibroblast for diagnosis of inherited metabolic diseases (IMD) in Hong Kong. This photo shows cultured fibroblast from a patient with Inclusion-cell (I-cell, Mucopolipidosis Type II) disease, which features characteristic cytoplasmic vacuoles. Many definitive diagnosis of various IMDs can be performed on cultured fibroblasts such those with fatty acid  $\beta$ -oxidation defects (FAOD).

for first line investigations came from hospitals both of the New Territories East cluster and other clusters, like Kwong Wah Hospital and private hospitals.

The 10 years' experience of treating more than 100 families with various IMD provided us an unique opportunity to reveal the spectrum of common IMD in the Southern Chinese population, which had little information in the past [1]. Here, I briefly review the spectrum of fatty acid  $\beta$ -oxidation defects (FAOD) found in our population. FAOD commonly causes acute metabolic crisis during infancy and early childhood and is also a common cause of sudden death during infancy.

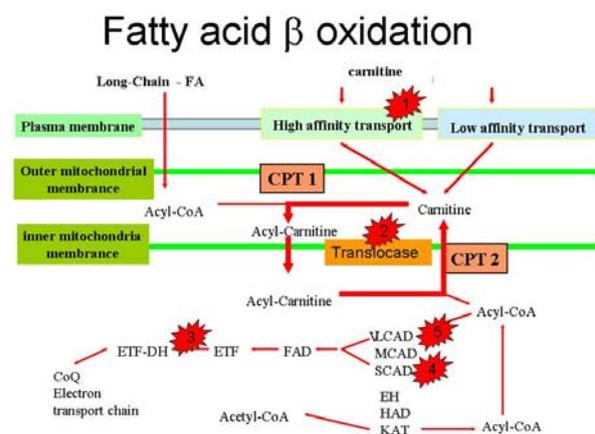
Our cells use a series of more than 20 enzymes to translocate/move fatty acids and its essential co-factor, carnitine, across cellular compartments and consecutively breakdown 2-carbon units per reaction ( $\beta$  oxidation) to generate ketone bodies and ATP as energy currency for cellular activities. As fatty acid serves as both the "emergence" energy source and a key form of storage of energy supply, it is not difficult to understand that any blockage in its energy production reactions would lead to detrimental consequences.

There is a prototype of clinical presentation of FAOD: a previously well infant/child with episode(s) of acute presentation featuring metabolic decompensation, hypoglycemia, liver derangement (sometimes confused with Reye's syndrome, so it has been called Reye-like syndrome), encephalopathy, and even sudden death. The acute attacks are commonly precipitated by other trivial event like mild infection, common cold, etc. The characteristic hypoketotic hypoglycaemia is recognised as a diagnostic indicator for FAOD.

While medium chain acyl-CoA dehydrogenase (MCAD) deficiency is very common in Caucasian, no definitive case has been diagnosed in Chinese. Instead, several other FAOD are common in Southern Chinese (Figure 2): namely primary carnitine deficiency, carnitine-acylcarnitine translocase defect, multiple acyl-CoA dehydrogenase deficiency and short chain acyl-CoA dehydrogenase deficiency.

This laboratory led the discovery of the gene coding for plasma membrane carnitine transporter and mutations in this gene (solute carrier family 22 (organic cation/carnitine transporter), member 5, SLC22A5, also known as Na<sup>+</sup>-dependent organic cation transporter 2, OCTN2) cause primary carnitine deficiency (PCD)[2]. We diagnosed a case of PCD in a family with sudden death of 2 infants and their story was later filmed as a documentary by Radio Television Hong Kong. The fibroblast cultured from patient cannot actively pump carnitine into the cell leading to an intracellular deficiency of carnitine. In-vivo investigation of the parents also demonstrated a renal wastage of carnitine. The JMC-Laboratory has become a regional referral laboratory for this disease and

collaborators are from Macau, Mainland China, and Taiwan. Studies of these samples revealed a common founder mutation of OCTN2 among Southern Chinese [3]. As most Chinese patients carried this mutation, it could be used as screening marker in high-risk patient.



**Figure 2.** Pathway of  $\beta$ -oxidation of fatty acid. Various defects found in Chinese are marked by numbers. 1. Carnitine transporter defect, 2. Carnitine-acylcarnitine translocase defect, 3. multiple acyl-CoA dehydrogenase deficiency; 4. short chain acyl-CoA dehydrogenase deficiency and 5. very long chain acyl-CoA dehydrogenase deficiency. Abbreviations: FA, Fatty Acid; CPT1/2, Carnitine palmitoyltransferase I/II; CoA, Coenzyme A; CAD, acyl-CoA dehydrogenase; FAD, Flavin adenine dinucleotide; ETF, Electron Transfer Flavoprotein; KAT, 3-ketoacyl-CoA thiolase

Furthermore, the mutation is likely descended from a single ancestral founder, its frequency would be high in the population and we determined that carrier frequencies was as high as 1/100 in Southern China [3]. Recently, Dr. Tang was invited by the Taiwan Foundation of Rare Diseases to Taipei to join a Press Interview to explain the importance of this disease in the region (Figure 3). PCD is now widely recognized as 卡尼丁缺乏症.

Carnitine-acylcarnitine translocase deficiency (卡尼丁穿透障礙) frequently presents with severe disease and causes death in neonatal period. This defect was first discovered in a patient born to an American-Chinese parent in 1992 [4]. Interestingly, the same splicing mutation was found in another British-Chinese family [5]. We performed pre-natal diagnosis for a couple carrying the same mutation. Recently, this mutation was also found in another local case of sudden death [6].

Another common FAOD in Chinese is multiple acyl-CoA dehydrogenase deficiency (戊二酸血症二型). It can be caused by mutations in at least 3 different genes (electron transfer flavoprotein alpha-subunit, ETF $\alpha$ ; electron transfer flavoprotein beta-subunit, ETF $\beta$ , and ETF dehydrogenase, ETFDH), all are involved in electron transfer for the fatty acid oxidation reactions. We are looking into the spectrum of mutations and distribution among the 3 genes in our local patients [7].

Short-chain acyl-CoA dehydrogenase deficiency (短鏈

脂肪酸去氫酵素缺乏症) is a defect in the finishing part of the chain of reactions of FAO, breaking down short-chain fatty acid which has already been chopped up by series of enzymes targeted at the fatty acids of longer chain length. Therefore, these patients could

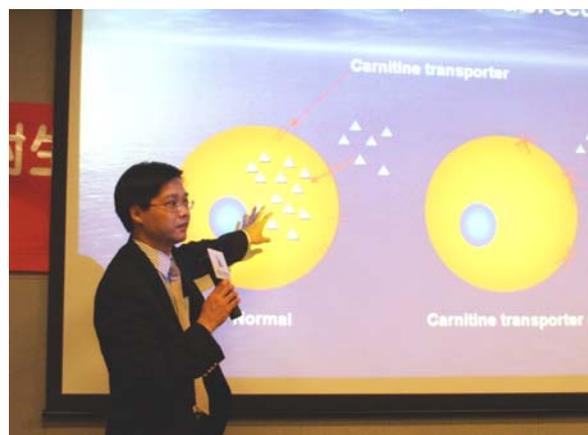


Figure 3. Photo taken during a press conference in Taiwan, and Dr. Tang was describing the defect in primary carnitine deficiency.

have some degree of emergence energy supply from FAO and present with mild disease. However, some patients had more severe disease and died, which might be related to secondary blockage of other mitochondrial reactions due to accumulation of toxic metabolites. Furthermore, we diagnosed the first local case of long-chain acyl-CoA dehydrogenase deficiency (長鏈脂肪酸去氫酵素缺乏症) in an adult patients treated in another hospital [8].

This laboratory is now developing new functional assays to analyze fatty acid oxidation in cultured cells (collected from skin biopsy) [9]. Hopefully, it will become one of the preferred methods for making definitive diagnosis for this group of diseases. In addition, Dr. Tang is active in promoting newborn screening for IMD using the latest tandem mass spectrometry technology in Hong Kong.

## References

Footnote: The official translation of carnitine is “肉鹼”. And the new Chinese translation of “卡尼丁” was first used by Dr. Tang in a press release in 1999 (<http://www.cuhk.edu.hk/ipro/990624c.htm>). He is very pleased to note that 卡尼丁 is now in common use among Chinese colleagues.

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## Report on Scientific Activities in November 2009

Two seminars related to advanced microarray technologies in clinical applications were successfully held at Queen Elizabeth Hospital on 7<sup>th</sup> and 11<sup>th</sup> November 2009, respectively. Here we sincerely thank the speakers and all the members who have attended the seminars.

The first seminar was a scientific seminar in the applications of aCGH technology in autism research and pre-/postnatal diagnosis given by Dr. Jeffrey Gregg from UC Davis and Dr. Richard Choy from CUHK. The seminar was co-organized by Hong Kong Society of Child Neurology and Developmental Paediatrics, Hong Kong Society of Cytogenetics, Hong Kong Society of Medical Genetics and Prenatal Genetic Diagnostic Centre, Department of Obstetrics and Gynaecology, The Chinese University of Hong Kong. This seminar was sponsored by Agilent Technologies. There were over 30 attendees coming from different disciplines including Hospital Authority, Universities, Department of Health, and some clinical specialists in private sectors.

The second seminar was a joint seminar between Hong Kong Society of Medical Genetics & Hong Kong Society of Cytogenetics in clinical cytogenetics and copy number variation given by Dr. Trilochan Sahoo from Signature Genomic Labs and Dr. Peter Matthiesen from Roche NimbleGen. The seminar was sponsored by Roche Diagnostics (HK) Ltd.

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### **Scientific Seminar:**

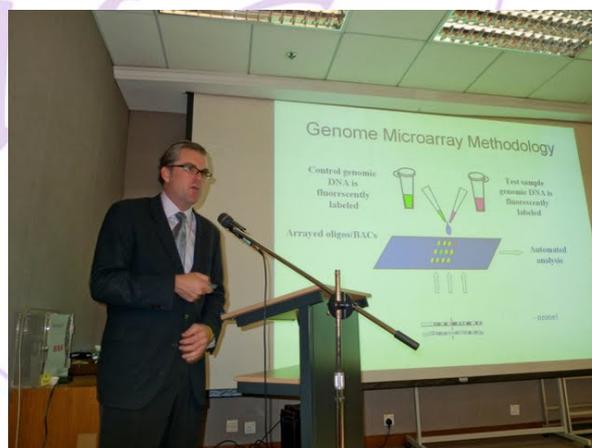
#### **Clinical application of aCGH in diagnosis of autism spectrum disorder/developmental delay**

**Jeffrey P. Gregg, M.D.**

*Associate Professor, Department of Pathology, School of Medicine; Director, Molecular Pathology Shared Resource, UC Davis Cancer Center, and UC Davis M.I.N.D. Institute Genomics Facility, UC Davis Medical Center, USA.*

Dr. Jeffrey Gregg received his M.D. in 1994, from UC Los Angeles. He then entered a Pathology Residency and Genetics Fellowship at UCLA. At UCLA, he focused his research efforts on developing microarray technology. In 1998, he joined the faculty as an Assistant Professor of Pathology and Director of Molecular Diagnostics at UC Davis. Currently, Jeffrey Gregg serves as Associate Professor of Pathology, Director of Molecular Diagnostics, Department of Pathology, and Director of the Gene Expression Shared Resources for the UC Davis Cancer Center and M.I.N.D. Institute. UC Davis M.I.N.D. Institute is a collaborative international research center, committed to the awareness, understanding, prevention, care and cure of neurodevelopmental disorders. His current research focuses on mouse models of mammary carcinoma including a model for ductal carcinoma in-situ. With the DCIS model, Jeffrey Gregg is working on strategies for chemoprevention and nutritional intervention. In addition, his laboratory is currently identifying genes involved or required for progression to invasive carcinoma and metastasis in these models.

Dr. Jeffrey Gregg is an expert in autism. He utilizes genomic technologies to identify biomarkers and



*The lecture delivered by Dr. Jeffrey Gregg and chaired by Dr. Ivan Lo*

genetic aberrations in children with autism. With gene expression profiling, he is utilizing blood genomics, transcriptional profiling of whole blood, to identify genes that are differentially expressed between children with autism and other control populations. With DNA, he is utilizing microarray technology to identify DNA aberrations that are associated with autism. This technology offers the ability to identify aberrations in the kb size range in contrast to the Mb size traditional cytogenetics offers.

## Utilization of aCGH in postnatal and prenatal diagnosis: experience in CUHK

*Richard Choy, Ph.D.*

*Assistant Professor, Deputy Director of Prenatal Genetics Diagnostic Centre, Department of Obstetrics & Gynaecology, The Chinese University of Hong Kong, c/o Prince of Wales Hospital, Shatin, New Territories, Hong Kong.*

At the same event, we also have our local aCGH expert, Dr. Richard Choy from The Chinese University of Hong Kong to share with us his experience of using aCGH technology in prenatal and postnatal genetic diagnosis for our local population in Hong Kong.

In Dr. Richard Choy's laboratory has shown aCGH technologies to be highly accurate for rapid detection of chromosomal aneuploidies and submicroscopic deletions or duplications on fetal samples. Of equal importance, for each fetal sample screened using the high resolution whole-genome coverage CGH array, anywhere between 10 and 150 chromosome Copy Number Variants (CNV) were found.



*Dr. Richard Choy presenting his targeted fetal DNA chip*

Chromosome CNVs are defined as gains and losses of

DNA sequences >1 kb within the human genome. The lacking of knowledge about these genetic variants (CNV) in the general population, including their alleles, frequencies and precise locations, makes differentiation between pathogenic CNVs from polymorphic features difficult and as a result leading to the classification of many CNVs as genomic imbalances of unknown clinical significance and hence, carries limited clinically meaning to the physicians and patients. To eliminate the issue of CNVs, Prenatal Genetic Diagnostic Centre of CUHK (directed by Prof. TK Lau, Dr. Richard Choy and Dr. Ronald Wang) developed a targeted microarray chip which contains 44,000 oligonucleotide probes for prenatal genetic screening for common chromosomal abnormalities.

This Fetal DNA Chip not only targeted on over 100 clinically relevant regions but also give reasonably high resolution coverage of the genome. Pilot study included 21 high risk pregnancy samples including fetus with sonographic anomalies, IUGR, thickened NT but with normal karyotype, and advanced maternal age alone. High-quality array CGH results were obtained in all samples without culture within a week. Although no pathogenic chromosome aberrations was identified among the IUGR and advanced maternal age group, Dr. Richard Choy identified pathogenic submicroscopic deletions/duplications among 4/6 fetus with increased NT with normal karyotyping. It demonstrated the use of array CGH for direct analysis of fetal DNA samples has increased the detection limit at a shorter time compared with conventional karyotyping.

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### **Joint Seminar:**

## **The Use of Microarrays in Clinical Cytogenetics: A New Understanding of the Human Genome in Health and Disease**

***Trilochan Sahoo, M.B.B.S, M.D., FACMG***

*Laboratory Director, Signature Genomics, Spokane, WA 99207, USA*

The evolution of high resolution microarray-based comparative genomic hybridization has revolutionized cytogenetics over the last few years. The introduction of this technology into the molecular cytogenetics laboratory now enables rapid identification of numerous microdeletion-microduplication syndromes, many of which are novel and had hitherto been unidentifiable. The extensive use of this technology has

empowered geneticists in identifying many complex genetic and genomic disorders and often enabled identification of causative genes for certain phenotypes. Importantly, the extensive data generated from array-CGH testing has led to a greater appreciation of the complexities of the human genome, and to better differentiate between benign copy number variations and those with a more adverse impact upon disease

phenotype. The development, validation and implementation of newer array-CGH technologies have greatly advanced our ability to generate useful data. Our current understanding of the technology permits us to predict that extensive use of these methodologies will profoundly impact the identification of novel genomic disorders and a better understanding of the genetic mechanisms involved. The data presented should provide an overview of the design and extensive use of our whole-genome array that interrogates over 3400 genomic loci across the genome and has helped identify most known disorders resulting from copy number alterations with great degree of specificity and sensitivity; and the unraveling a number of novel genetic syndromes.



*The lecture delivered by Dr. Trilochan Sahoo.*

## **High resolution Copy Number Variation Detection Using Roche NimbleGen Microarrays**

***Peter Matthiesen, Ph.D.***

*Global Marketing Director, Roche NimbleGen Inc., Roche Applied Science, Madison, WI 53719, USA*



*The lecture delivered by Dr. Peter Matthiesen.*

Recent advances in high-resolution microarray technology have revealed genomic DNA copy number variation as a significant source of genome variation,

now thought to account for more nucleotide variation than SNPs. Until recently, the recognized contribution of genome structural variation to human disease has been limited to rare genomic disorders (e.g. Trisomy 21, Prader-Willi Syndrome). However, with the emergence of high-resolution maps of genome-wide copy number variants (CNVs), their impact on disease phenotypes has become a key research focus. Roche NimbleGen has developed a suite of high-resolution CGH and CNV arrays with up to 2.1 million probes for comprehensive analysis of DNA copy number variation. In addition to the 2.1M array format, 3x720K and 12x135K multiplex arrays are available for higher throughput and cost-effective analysis. The information presented will provide an overview of the NimbleGen CGH/CNV array portfolio, complete microarray workflow, and application for genetic disease, cancer, and model organism research.

## Members' Publications

This is a new session to report the academic and research publications of our members over the half-year. The purpose is not only to recognize the scientific contributions in medical genetics and genetics, but also to stimulate the research outputs amongst our members in the society. As the beginning of this new session, we list the publications from 1<sup>st</sup> July to 31<sup>st</sup> December 2009 from our council members. Herewith all the publications listed below are linked to the PubMed server with publication ID to show the original abstract of the papers.

Szoeke C, Sills GJ, Kwan P, Petrovski S, Newton M, Hitiris N, **Baum L**, Berkovic SF, Brodie MJ, Sheffield LJ, O'Brien TJ. Multidrug-resistant genotype (ABCB1) and seizure recurrence in newly treated epilepsy: Data from International Pharmacogenetic Cohorts. *Epilepsia* 2009 Jul; 50(7):1689-96. PMID:19453704

Leung CK, Cheung CY, Weinreb RN, Qiu Q, Liu S, Li H, Xu G, Fan N, Huang L, **Pang CP**, Lam DS. Retinal nerve fiber layer imaging with spectral-domain optical coherence tomography: a variability and diagnostic performance study. *Ophthalmology* 2009 Jul;116(7):1257-63, 1263.e1-2. PMID:19464061

Lam AC, Lai KK, Chau AT, **Lo IF**, **Lam ST**. Subtelomeric 1q deletion syndrome causing patella hypoplasia and limb deformities, features overlapping with genitopatellar syndrome. *Clin Genet* 2009 Jul;76(1):102-7. PMID:19500117

**Chung BH**, **Lam ST**, **Tong TM**, Li SY, Lun KS, Chan DH, Fok SF, Or JS, Smith DK, Yang W, Lau YL. Identification of novel FBN1 and TGFBR2 mutations in 65 probands with Marfan syndrome or Marfan-like phenotypes. *Am J Med Genet A* 2009 Jul;149A(7):1452-9. PMID:19533785

Lam DS, Li EY, Chang DF, Zhang MZ, Zhan HK, **Pang CP**. Project vision: a new and sustainable model for eliminating cataract blindness in China. *Clin Experiment Ophthalmol* 2009 Jul;37(5):427-30. PMID:19624336

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Zhang LY, Gong B, Tong JP, Fan DS, Chiang SW, Lou D, Lam DS, Yam GH, **Pang CP**. A novel gammaD-crystallin mutation causes mild changes in protein properties but leads to congenital coralliform cataract. *Mol Vis* 2009 Aug 6;15:1521-9. PMID:19668596

Leung WC, Choi H, Lau WL, Ng LK, Lau ET, Lo FM, **Choy KW**, Lau TK, **Tang MH**, Chin R. Monozygotic dichorionic twins heterokaryotypic for duplication chromosome 2q13-q23.3. *Fetal Diagn Ther*. 2009 Sept; 25(4):397-9. PMID:19786786

Qiu K, Leung CK, Weinreb RN, Liu S, Chueng CY, Li H, Zhang MZ, **Pang CP**, Lam DS. Predictors of atypical birefringence pattern in scanning laser polarimetry. *Br J Ophthalmol* 2009 Sep;93(9):1191-4. PMID:19416934

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Zhao C, Bellur DL, Lu S, Zhao F, Grassi MA, Bowne SJ, Sullivan LS, Daiger SP, Chen LJ, **Pang CP**, Zhao K, Staley JP, Larsson C. Autosomal-dominant retinitis pigmentosa caused by a mutation in SNRNP200, a gene

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Ng TK, Lam CY, Lam DS, Chiang SW, Tam PO, Wang DY, Fan BJ, Yam GH, Fan DS, **Pang CP**. AC and AG dinucleotide repeats in the PAX6 P1 promoter are associated with high myopia. Mol Vis 2009 Nov 5;15:2239-48. PMID:19907666

Lan C, Tam PO, Chiang SW, Chan CK, Luk FO, Lee GK, Ngai JW, Law JS, Lam DS, **Pang CP**, Lai TY. Manganese superoxide dismutase and chemokine genes polymorphisms in Chinese patients with anterior uveitis. Invest Ophthalmol Vis Scii 2009 Dec;50(12):5596-600. PMID: 19628738

*Since this is the very first time, comments and suggestions to improve the session are most welcome. For the next half-year, on behalf of the society we would like to invite all members to provide their publications to our office.*



## Up-coming International Conferences



The banner for the Human Genome Meeting 2010 features a stethoscope on the left, a DNA double helix in the background, and a collage of images on the right showing the Le Corum building, a night view of the building, a swan, a red field, and a group of people. The text reads: "Human Genome Organisation Human Genome Meeting 2010 LE CORUM, MONTPELLIER, FRANCE, 18-21 MAY 2010" and includes the "hgm 2010" logo.

- Date:** 18-21 May 2010  
**Venue:** Le Corum, Montpellier, France  
**Organizer:** Human Genome Organisation (HUGO) International, Singapore  
Inserm, Laboratoire de Génétique Moléculaire, France  
**Registration:** 31 January 2010 (early bird)  
**Abstract:** 31 January 2010 (abstract book)  
**Website:** [www.hgm2010.org](http://www.hgm2010.org)  
**Enquiries:** HGM 2010 Secretariat. Tel: +65 6478 8192, Fax: +65 6478 9057,  
Email: [hugoadmin@qis.a-star.edu.sg](mailto:hugoadmin@qis.a-star.edu.sg)  
Inserm. Tel: 04 67 41 53 60, Fax: 04 67 41 53 65  
Email: [karine.deletang@inserm.fr](mailto:karine.deletang@inserm.fr)



The banner for the Taiwan ACGA 2010 International Conference on Genetic and Genomic Medicine features a yellow circle with the text "TAIWAN ACGA 2010" and a DNA double helix. The text reads: "ACADEMIA SINICA, TAIPEI, TAIWAN May 2-5, 2010, Sunday-Wednesday International Conference on Genetic and Genomic Medicine". It includes logos for the Association of Chinese Geneticists in America, Academia Sinica, and the Taiwan Genomics and Genetics Society.

- Date:** 2-5 May 2010  
**Venue:** 3F, Humanities and Social Sciences Building, Academia Sinica, Taipei, Taiwan  
**Organizer:** Institute of Biomedical Sciences, Academia Sinica, Taiwan  
Taiwan Genomics and Genetics Society  
Taiwan Human Genetics Society  
Association of Chinese Geneticists in America  
**Registration:** 28 February 2010 (early bird)  
**Abstract:** 31 January 2010  
**Website:** <http://taiwan-acga2010.ibms.sinica.edu.tw/>  
**Enquiries:** Email: [ACGA2010@mail.tcm-mice.com.tw](mailto:ACGA2010@mail.tcm-mice.com.tw)