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

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Message from the Editor

In the past three decades, the world of genetics has witnessed tremendous changes in scientific knowledge and the provision of medical genetic services. With the advances in our understanding of the human genome, and corresponding developments in technology, genetics has gained attention not only at the professional and scientific fronts, but also among the lay public and media. New discoveries in genes and the genome are heralded as keys to better ways of prevention of diseases and promotion of health. Some of these are no more than hypes, but most may ultimately be realized. What needs to be fulfilled between the upstream discoveries and downstream utility is a process of translation. This flow of development is applicable to both clinical genetics and at the public health programmes. For example, at the individuals' level, the development and application of genetic testing should be subjected to scrutiny by processes such as gene dossier. Alternatively, for application of genetics and genomics at a population level, such as genetic screening for various conditions, the concepts of clinical and analytical validity and utility should be applied. These are the guiding principles for development of our genetic and genomic services. These kinds of services have been implemented in Hong Kong for the past three decades. It is time for us to take stalk of what had been achieved, assess the present and future needs of our community, identify the gaps for us to meet these needs, draw up a strategy for further development and strive to make it work. Towards this aim, various parties, including Department of Health, Hospital Authority, the Universities and Academy of Medicine have joined hands to work out a scheme of action. Hopefully, it will meet with the support of the public and the funding bodies. The Hong Kong Society of Medical Genetics has always been a platform for our medical genetics community to discuss and explore ways to improve our services. Let's continue to contribute towards this objective and help improve the provision of genetic and genomic services in Hong Kong.

Stephen TS Lam
Editor

Annual General Meeting

The Annual General Meeting (AGM) was successfully held in the Lecture Theater of the Centre for Health Protection (CHP) on 21st November 2012. Apart from reports from the Chairman and the Treasurer, new council members were elected. Dr. Ivan Lo was elected to be the Chairman for the next two years. Two new faces were also elected into the council, including Dr. Edgar Hau and Dr. H. M. Luk.

Clinical Genetic Showcase Series –1st Episode

In the evening of the AGM on 21st November 2012, we have also proudly held the 1st Episode of Clinical Genetics Showcase Series. This is intended to be a regular, at least quarterly, scientific activity of HKSMG. Despite the paucity of clinical geneticists in Hong Kong, interesting genetic cases are not scarce. Through this regular scientific activity, we hope to promote interest in clinical genetics and foster collaboration among specialists from other medical disciplines. This evening, two young and brilliant local clinical geneticists, Dr. Brian Chung and Dr. H. M. Luk, presented two interesting local cases of TGF-beta signaling defects. It was also our great honor to have invited Dr. Carlos Bacino from Baylor College of Medicine to be our expert discussant. For those who were not present, the synopses below give a glance of the presentations.

Aneurysms-osteoarthritis syndrome

Dr. Brian H. Y. Chung

Associate Professor, Department of Paediatrics and Adolescent Medicine, University of Hong Kong

Our proband is a 59 y.o. Chinese woman who was found to have multiple visceral aneurysms and hemoperitoneum, presenting with sudden-onset acute left-sided abdominal pain. Whole body CT angiogram showed multiple visceral aneurysms in the superior mesenteric artery (SMA) territory (jejunal branch, right colic branch), bilateral renal arteries, right hepatic artery, right colic branch of SMA, left anterior descending artery (coronary artery), and left vertebral artery. Other significant clinical findings include joint laxity, painful joints, multiple joint degeneration and deformities, and vaginal prolapse. She had a 38 y.o. sister who suffered from ruptured intracranial aneurysm 1 week after delivery. A p.Arg268His mutation is identified in the SMAD3 gene in our proband and her sister, with no other mutations identified in COL3A1, TGFBR1, TGFBR2 and ACTA2 genes. SMAD3 mutations are associated with an aggressive, "Loeys-Dietz syndrome-like" form of aortic aneurysms and dissections with early-onset osteoarthritis (known as aneurysms-osteoarthritis syndrome). Multidisciplinary care is recommended due to the multi-system involvement. Since this entity is first described only in 2011, experience in the cardiovascular management is limited. We know that aortic dissections can occur at relatively smaller aortic diameters and early elective surgical intervention is indicated. However risk of rupture of intracranial and peripheral aneurysms is not known. Since the individual size, rate of growth and the location of aneurysms determine the treatment strategy, close monitoring by echocardiogram, CT angiogram and/or MRI (from head to pelvis) for carriers of SMAD3 mutation is recommended.

Loeys-Dietz Syndrome

Dr. Luk Ho Ming

Medical & Health Officer, Clinical Genetic Service, Department of Health, HKSAR

The proband is 26 years old lady with strong family history of dissecting thoracic aortic aneurysm. Physical examination showed hypertelorism (inter-pupillary distance >+3SD) and cardiac assessment

showed dilated aortic root (Z score=+3.8). Based on the clinical features, LDS was suspected and confirmed by TGFBR2 gene mutation. LDS is a kind of familial aortopathy syndrome that characterized by hypertelorism, bifid uvula and/or cleft palate, and generalized arterial tortuosity with ascending aortic aneurysm/dissection. It is caused by dysregulation of TGFβ signaling pathway result from mutation in TGFBR1 or TGFBR2 gene. When compared with other familial aortopathy like Marfan syndrome(MFS), they have some common clinical and histopathological features like aortic root dilation, dysfunctional smooth muscle cells within the tunica media, fragmentation of elastic fibers and excessive elaboration of extracellular matrix, therefore the medical (like beta blocker, ACE inhibitor) and surgical therapy for LDS is extrapolated from the treatment model of MFS. However, the cardiovascular course is more aggressive and extensive in LDS. Aortic rupture and dissection occur at younger age and smaller aortic root diameter as compared with MFS. Aneurysms are not confined to the aortic root but occurred throughout the entire arterial tree. Therefore close cardiac and vascular surveillance is warranted for all LDS patients, so early surgical intervention is recommended to reduce the risk of sudden cardiac death.

Reports on The 10th Asia Pacific Conference on Human Genetics 2012

The Society had sponsored two of our members, Dr. Brian Chung and Patrick Au, to attend "The 10th Asia-Pacific Conference on Human Genetics" held in Kuala Lumpur, Malaysia from 5- 8 December, 2012. They have shared their valuable experience with us in the brief reports below.

Dr. Brian HY Chung
Associate Professor, Department of Paediatrics and Adolescent Medicine, University of Hong Kong

Firstly I would like to thank the chairman and the selection committee of the HKSMG for supporting me to attend the 10th Asia Pacific Conference on Human Genetics. It was organized in Kuala Lumpur from 5-8th December 2012 and due to my other commitments, I could only attend the first day of the conference. The main theme of this conference was "Genetic and Genomic Medicine: Working together towards Health for All". Genomic medicine is a new structured approach to disease diagnosis and management that prominently features genome sequence information (E.D. Green. Nature 2011; 470:204-213). The recent breakthrough in elucidating the diagnoses of genetic conditions relies on the rapid expansion and applications of affordable genomic technologies.

The opening plenary session was delivered by David Valle from Johns Hopkins University School of Medicine in which he discussed about the various aspects of Individualized Medicine and how the human genome project, whole genome sequencing biology, evolutionary thinking in medicine, individual genome sequences and the progress in disease gene identification have enabled its rapid development. To practice individualized medicine as "a science of the individuals", Dr Valle stressed the importance of asking (Why this patient has this illness at this time?) and how we can use rigorous basic/translational/clinical researches to answer these questions. He also described the progress as well as current challenges facing efforts to accelerate the application of next-generation technologies to Mendelian disorders, e.g. the U.S.-based Mendelian Genome Centers as well as his effort to integrate genetics and genomics in medical student education in a project called "Genes to Society". This discussion is nicely followed by the presidential address made by Dr Stephen Lam from Hong Kong, in his capacity as the President of both the Asia Pacific Society of Human Genetics and the International Federation of Human Genetics Societies. Using a public health approach, Dr Lam discussed how we could evaluate the impact of novel technologies on genetic healthcare in emerging economies in the Asia-Pacific region.

Interesting poster presentations were delivered from various Hong Kong delegates, including Dr Hui Pui Wah on "Homozygous Haemoglobin Constant Spring presenting as fetal anaemia", Dr Anita Kan on "A fetus with haemoglobin Bart's disease due to maternal uniparental disomy for chromosome 16", as well as our society member Mr Patrick Au on "Characterization of a novel mutation in F8 gene causing severe hemophilia A".

I was invited to attend the APCHG by Professor Thong Meow-Keong (Chairman of the Organizing Committee) as a speaker in the Genetic Counselling Workshop chaired by Prof Srikant Sarangi from the Cardiff University, United Kingdom and Dr Olga Zayts from the School of English, the University of Hong Kong. This interactive workshop focused to reflect on existing culture- and context-specific practices of genetic counselling in South Asia vis-à-vis what is known about genetic counselling communication in the Western world. The workshop was structured in 3 parts. In the

first part, Prof Sarangi gave a presentation to describe the activity of genetic counselling within the broader context of healthcare delivery, and used real-life genetic consultation/counselling data to tease out, at the interactional level, what constitutes 'counselling' as distinct from 'consultation' activity.

This is followed the talk by Dr Zayts who reported insights from recent studies in genetic consultation in Hong Kong concerning prenatal clinics and telephone mediated encounters.

It was followed by sharing of the professional practitioners focusing on the specific communicative challenges that need further reflection. Dr Ivan Lo, representing the Clinical Genetic Service, Department of Health, Dr Anita Kan representing the Prenatal Diagnosis Service, Tsan Yuk Hospital and myself, representing the newly established clinical genetics program in the University of Hong Kong all gave presentations about the Hong Kong scenario of genetic consultation and counselling. The challenges and opportunities we are facing in Hong Kong were shared with other speakers and the audience attending the workshop. Dr Zayts is working towards putting together a short communication article/report summarising the discussions in the workshop to be submitted to the Journal of Genetic Counselling or other peer-reviewed journals in medical/clinical genetics. It is also anticipated that the discussion will lead to formulating a genetic counselling communication protocol as well training opportunities which are sensitive to the cultural and organisational context in the South Asian region.

Overall I think this is a very well organized conference with timely discussions on new technologies and their translation into clinical practices. Just as Prof Thong, Chairman of the Organizing Committee, has said in his welcome message, despite the economic constraints, the wide geographic locations of our peoples and diverse stages of medical genetics development in this region, geneticists and genetic counselors have come together to share, discover and celebrate the knowledge and its potential impact and benefits in medicine. The discussions on the development of genomic medicine in Asia and the culture-specific issues in genetic counseling are unique to this conference and not found in other international human genetics conferences.

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Patrick Au
Prenatal Diagnostic Laboratory, Tsan Yuk Hospital, Hong Kong

The 10th Asia-Pacific Conference on Human Genetics was held in Kuala Lumpur, Malaysia from 5th Dec. to 8th Dec., 2012. I had the opportunity to attend this conference and gave a poster presentation on haemophilia A.

There were speakers and delegates from all over the world including Australia, Canada, China, France, Hong Kong, India, Indonesia, Japan, Malaysia, Philippines, Saudi Arabia, Singapore, Spain, Taiwan, Thailand, The Netherlands, United Kingdom, United States of America and Vietnam. The first day of the conference was a Human Variome Project workshop while the rest of the programme consisted of plenary sessions and symposia on genetics and genomic medicine. There were talks on individualized medicine; thalassaemia; epigenetics; recent advances in diagnosis and treatment of genetic

disorders; new born screening of inborn errors of metabolism and their treatment; genetic counselling and prenatal diagnosis; cancer genetics and genomics; and genetic service in developing countries.

I arrived at the hotel in the evening of 5th Dec. after a 3 1/2 hour flight from Hong Kong and a trip from the airport by KLIA express and monorail. After a good sleep overnight, I got up refreshingly in the morning and walked across the street to the conference venue, the Crowne Plaza Hotel for registration and poster setup.

The Opening Ceremony

The opening ceremony was featured with music and singing performed by Malaysian children and young adults with rare genetic disorders. It was good to see that genetically disadvantaged people could lead positive lives with the help of support groups. As a laboratory personnel who works on the prenatal diagnosis of thalassaemia, the highlight of the conference would be plenary session, symposium and free paper presentations (oral and poster) on thalassaemia.

Symposium on thalassaemia

SNP variations on phenotypic variability in patients with thalassaemia (Vip Viprakasit, Thailand) focussed on the GWAS studies on HbE-β thalassaemia to identify genetic factors that modulate the severity of HbE-β thalassaemia.

Genetics of Thalassaemia in the Northeastern Population of Peninsula Malaysia (Rosaline Hassan, Malaysia) presented the spectrum of thalassaemia mutations in the heterogeneous population in Malaysia and their relevance to the diagnosis and prevention of thalassaemia.

Interacting alpha and beta thalassaemia (Ivy Ng, Singapore) described the difficulty to detect the concurrent alpha and beta thalassaemia in routine screening and the National Thalassaemia Registry established in Singapore in 1992 to facilitate counselling and screening for families. The carrier incidence in Singapore: 3.0% α thalassaemia, 0.9% β thalassaemia and 0.55% Hb E.

Plenary session on thalassaemia

Prevention and control of thalassaemia in Asia (Suthat Fucharoen, Thailand) gave a presentation on the functions of Asia Network for Thalassaemia Control (ANTC) in promoting and coordinating the use of thalassaemia screening techniques; education and screening programmes; prenatal diagnosis as an interim methods for the control of thalassaemia and the development of adequate approaches to treatment in Asian countries. National thalassaemia control programmes were launched in Malaysia, Maldives, Singapore and Thailand as a result.

Genotypes of Thalassaemia and Thalassaemia Haemoglobinopathies in Malaysia (Mary Anne Jin-Ai Tan, Malaysia) presented the outcome of prenatal diagnosis of α thalassaemia in Malaysia from 1996 - 2012: 34% with --SEA/--SEA, 25% with αα/αα and 38% with --SEA/αα. About 3.5% - 4% of the population are carriers of α thalassaemia or β thalassaemia with heterogeneous mutations among Malays, Chinese, Indians and Indigenous group.

Free paper presentations on thalassaemia that won prize awards

Specific molecular characterization of β-globin gene mutations using quantitative real-time PCR. A rapid and effective molecular screening technique for thalassaemia in Malaysia. (Young investigator award)

Non-invasive prenatal diagnosis using informative CpG sites: A possibility for confirmation of β thalassaemia mutations in foetal DNA. (Oral)

Spectrum of beta thalassaemia mutations, a step in prevention. Experience in Egypt. (Oral)

Genetic counselling regarding antenatal diagnosis and abortion for thalassaemia: A survey among Malaysian health care workers. (Poster)

There were other plenary sessions that I found interesting.

Plenary session on Genetic and Genomic Medicine

Individualized Medicine (David Valle, USA) gave a summary on the impact of sequencing technology, availability of individual genome sequences, disease genes identification and evolutionary thinking on medicine that led to the paradigm shift from medical treatment of an average patient to individualized medicine.

Plenary session on Genetic and Genomic Counselling

Responsible Genetics and Genomic Counselling (Judith Hall, Canada) presented the challenges in counselling with the increasing genomic information gained from array CGH and exome sequencing together with the advances in our understanding of genetic and epigenetic mechanisms. The importance of availability of appropriate genetic counselling and therapies for disorders identified was also emphasized.

Plenary session on Recent Advances in Diagnosis and Treatment

Overview of Skeletal Dysplasia (Ravi Savarirayan, Australia) talked about the diagnosis, natural history and treatment of skeletal dysplasia; giving the advances in treatments for osteogenesis imperfecta, hypophospatasia and achondroplasia as examples.

Malaysian food and landmark building in KL

Apart from the busy conference schedule during daytime, I could find time to sample delicious Malaysian food with other delegates from Hong Kong in the evening.

We had tried barbecued chicken wings, satay, fried eggs with oysters, barbecued stingray and curry noodles. And last but not least, the famous Malaysian durian (貓山王).

I even had time to get a close look at the Petronas Towers with the light on at night.

Other Scientific Activities

An evening scientific seminar was successfully organized on 23 January 2013 at Queen Elizabeth Hospital. We were honored to have invited Prof. David Amor (Clinical Geneticist and Director, Victorian Clinical Genetics Service, Royal Children's Hospital, Parkville Vic, Australia and Associate Professor, Department of Paediatrics, University of Melbourne), a very experienced clinical geneticist, to give a talk on "***The application of chromosome microarrays for preimplantation, prenatal and postnatal diagnosis in Victoria, Australia***". Prof. Amor was an excellent speaker and had a great deal to share with the audience. Right before the talk, we also had Dr. H. M Luk and Dr. Connie Fung from Clinical Genetic Service to present several interesting cases with diagnostic problem to share with our overseas expert.

International Conferences

1. 8 – 11 June 2013 – **European Human Genetics Conference 2013** at Paris, France. Organized by: the European Society of Human Genetics. Website: https://www.eshg.org/programme2013.0.html
2. 14 June 2013 – **12th European Symposium on Congenital Anomalies** at Zagreb, Croatia. Organised by European network of congenital anomalies registries. Website: http://www.eurocat2013.com/public/
3. 24 – 30 July 2013 – **Human Genome Analysis: Genetic Analysis of Multifactorial Diseases** at Hinxton, UK. Organised by Wellcome Trust. Website: http://www.wellcome.ac.uk/Education-resources/Courses-and-conferences/Advanced-Courses-and-Scientific-Conferences/Advanced-Courses/WTX026851.htm
4. 4 – 7 August 2013 – **37th Annual Scientific Meeting of the Human Genetics Society of Australasia** at Queenstown, New Zealand. Organized by: the Human Genetics Society of Australasia. Website: http://www.nzconference.co.nz/HGSA2013/index.asp
5. 16 – 18 September 2013 – **British Genetic Medicine Conference 2013** at Liverpool, UK. Organised by the British Society of Genetic Medicine. Website: http://www.clingensoc.org/news-events/news/british-genetic-medicine-conference-2013/
6. 22 – 26 October 2013 – **63rd Annual Meeting of the American Society of Human Genetics** at Boston, Massachusetts. Organized by: the American Society of Human Genetics. Website: http://www.ashg.org/2013meeting/

Members' Publications

1. Cheng, Y. K., C. Wong, H. K. Wong, K. O. Leung, Y. K. Kwok, A. Suen, C. C. Wang, T. Y. Leung and K. W. Choy (2013). **"The detection of mosaicism by prenatal BoBs."** Prenat Diagn **33**(1): 42-49.

2. Chung, B. H., H. M. Luk, I. F. Lo, S. T. Lam and R. H. Li (2013). **"A second report of p.Pro986Leu variant in COL2A1-phenotypic overlap with SEDC and other forms of type II collagenopathies."** Am J Med Genet A **161**(4): 918-920.

3. Ci, H. B., Z. J. Ou, F. J. Chang, D. H. Liu, G. W. He, Z. Xu, H. Y. Yuan, Z. P. Wang, X. Zhang and J. S. Ou (2013). **"Endothelial microparticles increase in mitral valve disease and impair mitral valve endothelial function."** Am J Physiol Endocrinol Metab **304**(7): E695-702.

4. Haerian, B. S. and L. Baum (2013). **"GABRG2 rs211037 polymorphism and epilepsy: a systematic review and meta-analysis."** Seizure **22**(1): 53-58.

5. Haerian, B. S., L. Baum, H. J. Tan, P. Kwan, A. A. Raymond, J. Saruwatari, K. Nakagawa and Z. Mohamed (2012). **"SCN1A IVS5N+5 polymorphism and response to sodium valproate: a multicenter study."** Pharmacogenomics **13**(13): 1477-1485.

6. Ho, A. C., A. P. Liu, K. S. Lun, W. F. Tang, K. Y. Chan, E. Y. Lau, M. H. Tang, T. Y. Tan and B. H. Chung (2012). **"A newborn with a 790 kb chromosome 17p13.3 microduplication presenting with aortic stenosis, microcephaly and dysmorphic facial features - is cardiac assessment necessary for all patients with 17p13.3 microduplication?"** Eur J Med Genet **55**(12): 758-762.

7. HM Luk, IFM Lo, Y Aoki, TMF Tong, DHC Chan, STS Lam. **"Clinical and molecular characteristics of Cardio-facio-cutaneous syndrome in Hong Kong Chinese."** HK J Paediatr (new series) 2013;18:31-36.

8. Luk HM, Lo IF, Lai CW, Ma LC, Tong TM, Chan DH, Lam STS. **"Congenital fibrosis of extraocular muscle type 1A due to KIF21A mutation: first case report from Hong Kong."** Hong Kong Med J 2013 Apr;19(2):182-5.

9. Sen, P., Y. Yang, C. Navarro, I. Silva, P. Szafranski, K. E. Kolodziejska, A. V. Dharmadhikari, H. Mostafa, H. Kozakewich, D. Kearney, J. B. Cahill, M. Whitt, M. Bilic, L. Margraf, A. Charles, J. Goldblatt, K. Gibson, P. Lantz, J. Garvin, J. Petty, Z. Kiblawi, C. Zuppan, A. McConkie-Rosell, M. T. McDonald, S. L. Peterson-Carmichael, J. T. Gaede, B. Shivanna, D. Schady, P. S. Friedlich, S. R. Hays, I. V. Palafoll, U. Siebers-Renelt, A. Bohring, L. S. Finn, J. R. Siebert, C. Galambos, L. Nguyen, M. Riley, N. Chassaing, A. Vigouroux, G. Rocha, S. Fernandes, J. Brumbaugh, K. Roberts, L. Ho-Ming, I. Lo, S. Lam, R. Gerychova, M. Jezova, I. Valaskova, F. Fellmann, K. Afshar, E. Giannoni, V. Muhlethaler, J. Liang, J. S. Beckmann, J. Lioy, H. Deshmukh, L. Srinivasan, D. T. Swarr, M. Sloman, C. Shaw-Smith, R. L. van Loon, C. Hagman, Y. Sznajer, C. Barrea, C. Galant, T. Detaille, J. A. Wambach, F. S. Cole, A. Hamvas, L. S. Prince, K. E. Diderich, A. S. Brooks, R. M. Verdijk, H. Ravindranathan, E. Sugo, D. Mowat, M. L. Baker, C. Langston, S. Welty and P. Stankiewicz (2013). **"Novel FOXF1 Mutations in Sporadic and Familial Cases of Alveolar Capillary Dysplasia with Misaligned Pulmonary Veins Imply a Role for its DNA Binding Domain."** Hum Mutat.

10. Tsang, J. P., W. L. Poon, H. M. Luk, C. W. Fung, C. K. Ching, C. M. Mak, C. W. Lam, T. S. Siu, S. Tam and V. C. Wong (2012). **"Arginase deficiency with new phenotype and a novel mutation: contemporary summary."** Pediatr Neurol **47**(4): 263-269.

11. Verhoeven, V. J., P. G. Hysi, R. Wojciechowski, Q. Fan, J. A. Guggenheim, R. Hohn, S. MacGregor, A. W. Hewitt, A. Nag, C. Y. Cheng, E. Yonova-Doing, X. Zhou, M. K. Ikram, G. H. Buitendijk, G. McMahon, J. P. Kemp, B. S. Pourcain, C. L. Simpson, K. M. Makela, T. Lehtimaki, M. Kahonen, A. D. Paterson, S. M. Hosseini, H. S. Wong, L. Xu, J. B. Jonas, O. Parssinen, J. Wedenoja, S. P. Yip, D. W. Ho, C. P. Pang, L. J. Chen, K. P. Burdon, J. E. Craig, B. E. Klein, R. Klein, T. Haller, A. Metspalu, C. C. Khor, E. S. Tai, T. Aung, E. Vithana, W. T. Tay, V. A. Barathi, P. Chen, R. Li, J. Liao, Y. Zheng, R. T. Ong, A. Doring, D. M. Evans, N. J. Timpson, A. J. Verkerk, T. Meitinger, O. Raitakari, F. Hawthorne, T. D. Spector, L. C. Karssen, M. Pirastu, F. Murgia, W. Ang, A. Mishra, G. W. Montgomery, C. E. Pennell, P. M. Cumberland, I. Cotlarciuc, P. Mitchell, J. J. Wang, M. Schache, S. Janmahasathian, R. P. Igo, Jr., J. H. Lass, E. Chew, S. K. Iyengar, T. G. Gorgels, I. Rudan, C. Hayward, A. F. Wright, O. Polasek, Z. Vatavuk, J. F. Wilson, B. Fleck, T. Zeller, A. Mirshahi, C. Muller, A. G. Uitterlinden, F. Rivadeneira, J. R. Vingerling, A. Hofman, B. A. Oostra, N. Amin, A. A. Bergen, Y. Y. Teo, J. S. Rahi, V. Vitart, C. Williams, P. N. Baird, T. Y. Wong, K. Oexle, N. Pfeiffer, D. A. Mackey, T. L. Young, C. M. van Duijn, S. M. Saw, J. E. Bailey-Wilson, D. Stambolian, C. C. Klaver and C. J. Hammond (2013).

"Genome-wide meta-analyses of multiancestry cohorts identify multiple new susceptibility loci for refractive error and myopia." Nat Genet **45**(3): 314-318.

12. Wang, C. C., E. Billett, A. Borchert, H. Kuhn and C. Ufer (2013). **"Monoamine oxidases in development."** Cell Mol Life Sci **70**(4): 599-630.

13. Wong, E. H., L. Cui, C. L. Ng, C. S. Tang, X. L. Liu, M. T. So, B. H. Yip, G. Cheng, R. Zhang, W. K. Tang, W. Yang, Y. L. Lau, L. Baum, P. Kwan, L. D. Sun, X. B. Zuo, Y. Q. Ren, X. Y. Yin, X. P. Miao, J. Liu, V. C. Lui, E. S. Ngan, Z. W. Yuan, S. W. Zhang, J. Xia, H. Wang, X. B. Sun, R. Wang, T. Chang, I. H. Chan, P. H. Chung, X. J. Zhang, K. K. Wong, S. S. Cherny, P. C. Sham, P. K. Tam and M. M. Garcia-Barcelo (2013). **"Genome-wide copy number variation study in anorectal malformations."** Hum Mol Genet **22**(3): 621-631.

14. Xuan, C., L. M. Lun, J. X. Zhao, H. W. Wang, B. Z. Zhu, S. Yu, Z. Liu and G. W. He (2013). **"PTPN22 Gene Polymorphism (C1858T) Is Associated with Susceptibility to Type 1 Diabetes: A Meta-Analysis of 19,495 Cases and 25,341 Controls."** Ann Hum Genet.

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16. Zheng, S. Y., M. Wu, J. S. Huang, M. J. Mai, T. B. Chen and G. W. He (2012). **"Use of antispastic nicardipine and nitroglycerin (NG) cocktail solution increases graft flow during off-pump coronary artery bypass grafting."** J Cardiovasc Surg (Torino) **53**(6): 783-788.

17. Xuan C, Jia KG, Wang BB, Bai XY, Gao G, Yang Q, Wang ZL, Liu XC, Ma X, He GW. **"Identification of Two Novel Mutations of HOMEZ gene in Chinese Patients with Isolated Ventricular Septal Defect."** Genetic Testing and Molecular Biomarkers. 2013 (in press)