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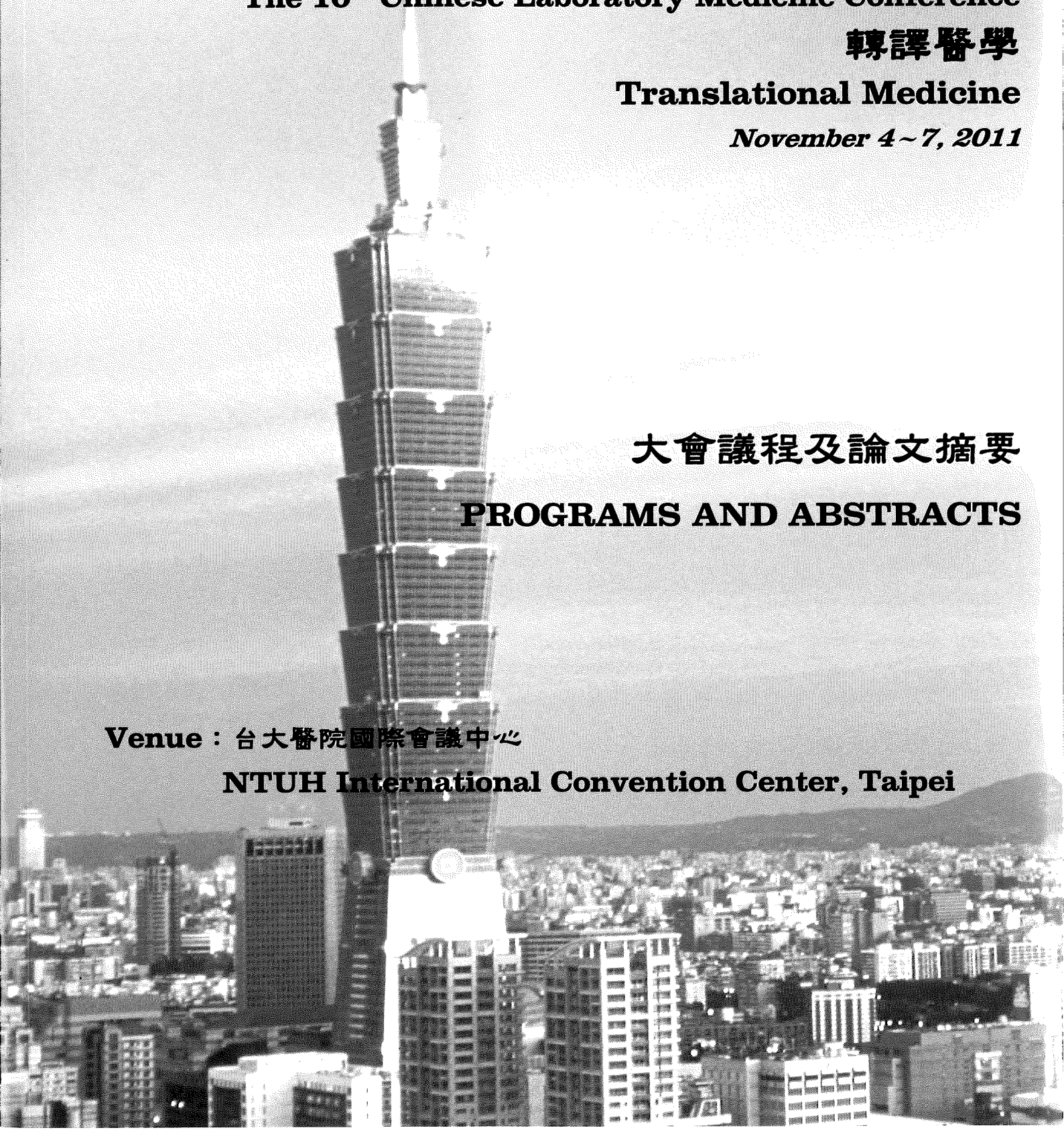

大會議程及論文摘要

PROGRAMS AND ABSTRACTS

Venue：台大醫院國際會議中心

NTUH International Convention Center, Taipei

Organizer： ***台灣醫事檢驗學會***

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SL21

醫學檢驗室在轉譯細胞治療法中的作用（Roles of Medical Laboratory in Providing Translational Cell Therapy）

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Cells have long been used as adjuvant therapy in addition to primary treatment. Recently stem cell therapy was regarded as a treatment modality for patients with otherwise incurable disease by conventional regimens. Bone marrow transplantation becomes a promising regimen for a variety of life-threatening diseases. With the advent of recombinant technology and the availability of cell separators, haematopoietic stem cells (HSC) are collected from the peripheral blood of patients/donors having undergone cytokine mobilization. Umbilical cord blood are also quality HSC. The manoeuvres of HSC therapy depends much on the innovative laboratory technologies. ABO-incompatibility occurs frequently in HLA-matched transplant. Avoidance of intravascular haemolysis at the time of infusion of major ABO-mismatched HSC products can be achieved by red cell depletion or plasmapheresis of recipients to remove circulating ABO-antibodies to a safe titre. T-cell purging and positive selection of HSC are conducted to alleviate graft-versus-host disease in HLA-mismatched transplants. HSC autografts harvested from tumour-bearing patients at clinical remission may have occult tumour cell dissemination. *Ex-vivo* tumour cell purging is performed to eradicate any residual tumour cells that may attribute to graft-mediated disease relapse upon re-infusion. Positive selection is also an effective means to enrich HSCs and negatively deplete tumour cells in autografts. The discovery of stem cell plasticity raises the hope that non-haematological disorders might become amenable to stem cell therapy to replenish the cell loss and degeneration. Preliminary results of randomized controlled phase-I/II trials of bone marrow-derived mesenchymal stem cells and UCB for stroke and spinal cord injury, respectively, are reported.

SL22

Tetrahydrobiopterin Synthesis Deficient Hyperphenylalaninemia in Chinese

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Phenylketonuria (PKU) and hyperphenylalaninemia (HPA) are caused by the deficiency of phenylalanine-4-hydroxylase or its essential cofactor tetrahydrobiopterin (BH_4), which is also required for the hydroxylation of other aromatic amino acids. The most common form of BH_4 deficiency is caused by 6-pyruvoyl-tetrahydropterin synthase (PTPS, gene symbol: *PTS*) deficiency (MIM 261640) and requires different treatment from the classical PKU (MIM 261600). Early diagnosis and proper treatment starting at neonatal period will prevent mental retardation and result in normal intellectual development. Neonatal screening for PKU started in January 1984 in Taiwan. The overall incidence rate of PKU was about 1/31,400 in Taiwan with 19% of them were BH_4 deficient. More than 90% of these BH_4 deficient PKU were caused by the mutations of *PTS* gene. Long-term supplementations with BH_4, 5-hydroxytryptophan and L-dopa have been shown beneficial to all PTPS deficient PKU patients.

The PKU incidence in northern Chinese (1/11,000) is about three times higher than that in southern Chinese. But BH_4 deficiency is more frequently observed in southern Chinese (20 ~ 30% of PKU) than that in northern Chinese (7% of PKU). Twenty-eight missense, 3 nonsense, 3 splicing and 2 deletion mutations in the *PTS* gene were identified in 156 PTPS deficient Chinese families. The c.166G>A and c.84-291A>G mutations were found to associate with mild clinical form of PTPS deficiency. The c.155A>G, c.259C>T, c.272A>G, c.286G>A and c.84-291A>G mutations account for about 75% of the *PTS* mutant alleles. The c.259C>T and c.84-291A>G mutations were widely observed in both southern and northern Chinese. The c.155A>G mutation was a common mutation detected in southern Chinese while the c.286G>A and c.272A>G mutations were predominant found in northern Chinese. Founder effects were suggested for these common Chinese mutations by studying closely linked short tandem repeat (STR) marker.

Notably, these common mutations in Chinese were also found across in other East Asian populations and share the same STR haplotypes. The STR linkage study indicates that these frequently observed mutations in the *PTS* gene of East Asian individuals all originated from Chinese populations. On the other hand, the c.200C>T and c.317C>T mutations found in Chinese as well as other populations might occur in several independent events at the mutation hot spots in the *PTS* gene.