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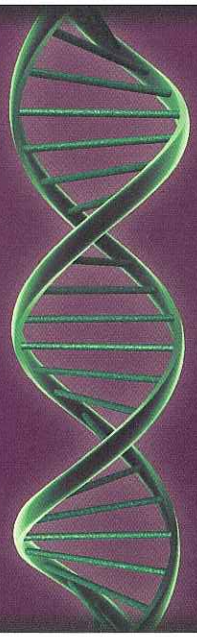

PROGRAM & ABSTRACTS

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in the lab of the Nobel Laureate Arthur Kornberg at Department of Biochemistry, Stanford University. He is currently a chair-professor of Biomedical Sciences and Head of the Department of Health Technology and Informatics of PolyU. The identification of NPM's role in transcriptional regulation charts new courses for understanding of gene regulation and tumorigenesis. He has published over 80 scientific papers.

Position held and Education:

2008-2009 Head (2009-), Chairprofessor, Health Technology & Informatics, PolyU

1993-2008 Associate professor (1989-1993), Professor (1993-), Pharmacology, College of Medicine, Chang Gung University

1986-1989 Postdoct Fellow of Professor Arthur Kornberg (Nobel Laureate of Physiology and Medicine), Biochemistry, Stanford University, California

1982-1986 Ph.D. Pharmacology. Baylor College of Medicine, Texas

1980-1982 MA Organic Chemistry, Rice University, Texas

1977-1980 BA Chemistry, Southern Illinois University at Carbondale, Illinois

Awards and Honors:

2003 Research Excellence Award, Chang Gung University

1999 Outstanding Cancer Research Award (Chinese Oncology Society)

1999 Outstanding Research Award (Pharmacological Society)

1993, 1994 National Science Council Outstanding Research Award

1993 Ministry of Education Outstanding Teacher Award

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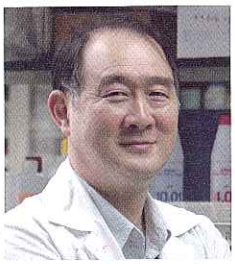

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Title: TETRAHYDROBIOPTERIN SYNTHESIS DEFICIENT HYPERPHENYLALANINEMIA IN EAST ASIAN POPULATIONS

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Phenylketonuria (PKU) and hyperphenylalaninemia (HPA) may be caused by deficiency of phenylalanine-4-hydroxylase or tetrahydrobiopterin (BH_4), the essential cofactor required in the hydroxylation of aromatic amino acids. The most common forms of BH_4-deficiency is caused by 6-pyruvoyl-tetrahydropterin synthase (PTS) deficiency (MIM 261640) and require 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-deficiency and more than 90% of BH_4-deficiency was caused by PTS deficiency. Long-term supplementations with BH_4, 5-hydroxytryptophan and L-dopa have shown beneficially to all PTS-deficient PKU patients.

Twenty-eight missense, 3 nonsense, 3 splicing and 2 deletion mutations on the PTS gene were identified in 148 PTS-deficient Chinese families. Among these mutations, the c.155A>G, c.259C>T, c.272A>G, c.286G>A and IVS1-291A>G mutations account for about 77% of the mutant alleles. The c.166G>A and IVS1-291A>G mutations were found to associate with mild clinical form of PTS-deficiency. The c.155A>G mutation was found to be the common mutation in southern while the c.286G>A and c.272A>G were common in northern Chinese. The c.259C>T and IVS1-291A>G mutations were common in both southern and northern Chinese. Besides IVS1-291A>G, founder effect was suggested for these common Chinese mutations by studying closely linked short tandem repeat (STR) markers.

In other East Asian populations, we had identified 3 mutations (243G>A, 259C>T, 286G>A) in 6 Japanese PTS-deficient patients, 7 mutations (68G>C, 155A>G, 259C>T, 272A>G, 317C>T, 347A>G, IVS1-291A>G) in 7 Korean patients, 3 mutations (155A>G, 259C>T, 116-119del) in 4 Malaysian patients, 2 mutations (58T>C, 382T>A) in 3 Filipino patients, and 3 mutations (200C>T, 147T>G, 259C>T) in 3 Thai patients. Notably, the c.259C>T mutation was found across Chinese, Japanese, Korean, Malaysian and Thai and shared a common STR marker suggesting a founder effect of c.259C>T mutation in these populations. The 243G>A and 58T>C, which were only detected in the Japanese and Filipino, respectively, were also found to have founder effects. On the other hand, the 200C>T might occur several times independently at the mutation hot spot in the PTS deficient patients.