# GAS CHROMATOGRAPHIC ANALYSIS OF ABNORMAL URINARY ORGANIC ACIDS IN PHENYLKETONURIA

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# 苯酮尿症尿中不正常有機酸之氣體層析分析

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Reprinted from the Journal of the Formosan Medical Association
Vol 84, No 11, pp 1240～1250, November, 1985

臺灣醫學會雜誌第84卷第11號第1240～1250頁抽印本
中華民國74年11月發行

J Formosan Med Assoc 84; 1240-1250, 1985

# GAS CHROMATOGRAPHIC ANALYSIS OF ABNORMAL URINARY ORGANIC ACIDS IN PHENYLKETONURIA

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**Phenylketonuria (PKU) is an inherited metabolic disease due to a defect in the phenylalanine hydroxylating system. Analysis of the abnormal urinary metabolites is in the interest of clinical investigation of the disease.**

**A gas chromatographic system for analysis of abnormal urinary organic acids was studied. Urinary organic acids were oximed and extracted by ethyl acetate and diethyl ether before they were trimethylsilylated. The organic acid derivatives were chromatographed on both 10% OV-1 and 10% OV-17 columns. The methylene unit, which was calculated by calibrating with hydrocarbon standards, was found to be a better index than the retention time for identification of the derivatives. The major abnormal organic acids, such as phenylpyruvate, phenyllactate, phenylacetate, and 2-hydroxyphenylacetate, were confirmed in the urines of phenyleketonurics. With the standard extraction procedure, the detection limits for the abnormal metabolites were 25 μg/mg creatinine in urine, which was about 10 times more sensitive than the conventional chemical methods. The changes of urinary organic acid profile after dietary management were clearly demonstrated. The results indicated that this gas chromatographic system was a sensitive and specific method for analysis of abnormal urinary organic acids and could be applied to aid the diagnosis and monitoring the dietary managment of phenylketonuria.**

Key words: *phenylketonuria, PKU, phenylacetic acid, phenyllactic acid, phenylpyruvic acid, 2-hydroxyphenylacetic acid, gas chromatography.*

(*J Formosan Med Assoc* 84; 1240-1250, 1985)

Phenylketonuria (PKU) is caused by a genetic defect in the enzyme system that catalyzes the conversion of phenylalanine to tyrosine.[1,12,19] The disease is due to an autosomal recessive trait and is characterized clinically in the untreated patient by severe neurological findings that include agitated behavior, hyperactivity, hypertonicity of muscle, fits and severe mental retardation.[1,2,12,19] It has been reported that early diagnosis with proper dietary therapy could prevent the mental retardation caused by PKU. Control of phenylalanine homeostasis with maintenance of normal nutrition is the goal of PKU treatment. A low phenylalanine diet is the basis for prevention of mental retardation in classical PKU homozygotes.[1,7,12,13,23]

It has been shown by neonatal screening that the frequencies of PKU were from 1/5,000 to 1/300,000 in different

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Received August 5, 1985.
Accepted Sepember 10, 1985.