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### 應用 HRM 分析方法快速偵測糖尿病患 Hb Iraq-Halabja 基因突變

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### Application of the rapid HRM analysis to detected Hb Iraq-Halabja in Diabetes patient

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BACKGROUND:
Hemoglobin A1c (HbA1c) is used routinely to monitor long-term blood sugar control in people with diabetes, as HbA1c is related directly to risks for diabetic complications. Hb gene disorders are common hereditary diseases: e.g., alpha- or beta-thalassemia and Hb variant- related disease. More than 1000 abnormal Hb variants have been identified. This study was application of the rapid HRM analysis to detected Hb Iraq-Halabja in diabetes.
METHODS and MATERIALS:
In the present case study was a 50-year-old Taiwanese man and following a long time history of diabetes. We modify Shih et al.(2009) establish rapid identification of HBB gene mutations by HRM analysis.
RESULTS:
In this study, CBC data showed RBC 4.73x106 /μL, Hb 16.4 g/dL, mean corpuscular value (MCV) 100.5 fL, mean corpuscular hemoglobin (MCH) 34.6 pg, mean corpuscular hemoglobin concentration (MCHC) 34.4 g/dL, Ferritin 290.1 ng/ml and hemoglobin electrophoretic pattern (HbA 97.5%, HbA2 2.5% , HbA1C 2.5%). Herein, we application rapid HRM method to detected and find new Hb variant. In addional, we confirmed Hb β10 (A7) Ala -> Val (GCC -> GTC) (Hb Iraq-Halabja) with directly sequencing and PCR-RFLP
CONCLUSIONS:
This study demonstrates that HbA1C values determined by ion-exchange HPLC did not reflect the glycemic state in our patient. The ion-exchange HPLC to measurement HbA1C necessity combine with repeated using additional method e.g. application HRM method or hemoglobin gene whole gene sequence.

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### 利用成人健檢報告資訊評估腎絲球過濾速率之變異因子

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### Utilization of adult health screening information to evaluate the variation factors of estimated glomerular filtration rate

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血清肌酸酐(Serum creatinine, Scr)是最常被用來評估腎臟功能的指標，但因肌酸酐的產生會受個體肌肉質量、飲食、年齡、性別..等多種因素影響，因此並非評估腎臟功能的最好指標。根據美國腎臟基金會建議，檢測腎絲球過濾速率(estimated glomerular filtration rate, eGFR)才能更精準的來判定慢性腎臟病的嚴重度，並定義只要 eGFR 持續低於 60 ml/min/1.73 m2 達 3 個月以上，就可以認定爲慢性腎臟病。
本研究利用 2012 年 1~6 月 1190 位接受成人健檢者之報告資訊，初步依據美國腎臟基金會標準將 eGFR 結果分成腎功能正常、輕度、中度、重度慢性腎衰竭及末期腎臟疾病等 5 種類別(>= 90, 60-90, 30-59, 15-29, <15)，分別與 Scr (< 1.3 mg/dl)之臨床意義比較，除第五類無個案外，其餘 1~4 類別 eGFR 結果爲 102.4 ± 11.0, 76.8 ± 7.6, 51.8 ± 6.6, 22.5 ± 5.5； Scr 結果爲 0.7 ± 0.1, 0.9 ± 1.5, 1.3± 0.2, 2.6 ± 0.9。我們發現在 eGFR 分類上已能呈現輕微功能異常之報告(第 2, 3 類)，但 Scr 數據可能無法提早呈現相同之結果。進一步實際評估影響 eGFR 之可能變異因子(包含性別,年齡, Scr, Glucose, GOT, GPT, TG, Cholesterol (Chol), HDL-C, LDL-C, Urine protein (UP) 十一項)，利用逐步多元迴歸分析方法，保留有統計意義之影響項目，發現性別, 年齡, Scr, UP, Chol 等 5 個項目與 eGFR 之多元相關性達 0.886，影響 eGFR 之變異量達 78.5%。而影響變異量依序爲 Scr (63%), 性別 (10.5%), 年齡 (4.7%), UP (2%), Chol (1%)，但有 21.5% 尚無法由上述項目解釋其變異來源。