pd53 - Poster Discussion
Glycated haemoglobin: Where does it fit?

Abstract: D-0812

Undetectable HbA1c in a type 2 diabetic woman: Case report of a rare haemoglobin variant.

R.P Radermecker[1], V. Thielen[1], J.P. Chapelle[2], N. Paquot[1]

[1] *CHU Sart Tilman University of Liege, Department of Diabetes Nutrition and Metabolic disorders, Liege, Belgium*
[2] *CHU Sart Tilman University of Liege, Department of Clinical Chemistry, Liege, Belgium*

Background: Haemoglobin A1c (HbA_{1c}) is defined by the International Federation of Clinical Chemistry (IFCC) working group as haemoglobin that is irreversibly glycated at one or both N-terminal valines of the beta chains. Various factors may affect the accuracy of HbA_{1c} measurements according to the assay method used and haemoglobin variants represent one of them. Hb C is caused by the substitution of glutamic acid for lysine at position 6 of the ß-globin chain and is commonly found in West Africa and the Caribbean regions. Hb D is caused by the substitution of glutamine for glutamic acid at position 121 of the ß-globin chain. It is usually found in the Sikhs of the Punjab region of the Indian subcontinent.

Objective: To report a case of a diabetic patient with clinically silent haemoglobin variant, causing undetectable HbA_{1c} concentration measured by high performance liquid chromatography (HPLC) method.

Case Summary: Our patient was a Caucasian 64-year-old female with type 2 diabetes mellitus for 5 years treated by diet, sulfonylureas and metformin. She had a history of breast cancer treated by surgery and curietherapy. Her mother had a type 2 diabetes. The patient lived in Germany before coming for the first time in our unit. During the hospitalization for check up, determination of HbA_{1c} level were performed. HbA_{1c} concentrations measured using a HPLC method (Adams ARKRAY, KYOTO, Japan distributed through Menarini, Italy) were undetectable. An electrophoresis of hemoglobin reported: HbA 0% (normal range 94,5-98,5), HbA2 2,4% (1,5-3,5), HbC 43,1% (0%), HbD 53,8% (0%). A research of ß-thalassemia with study of ß-Globin HBB gene was performed using Polymerase Chain Reaction amplification and promoter sequencing of HBB gene methods. The results confirmed an haemoglobin variant C/D with 2 mutations (p.E6K and p.E122Q).

Conclusions: Clinical laboratories should be aware of limitations of the HbA_{1c}assay method used, such as a potential interference with haemoglobin variant as illustrated by our case. Alternative methods for monitoring glycaemic control in these patients should be considered as, for instance, dosage of fructosamine or use of continuous glucose monitoring system.