Good performance of the α-Globin StripAssay® for diagnostics of alpha-thalassemia

Ilse A.M. Luijten- de Vrije[1], Henk Martens[2], Jacqueline Leuvenink[2], Cornelis (Ronald) J.J. Huijsmans[1] Adriaan J.C. van den Brule[1]

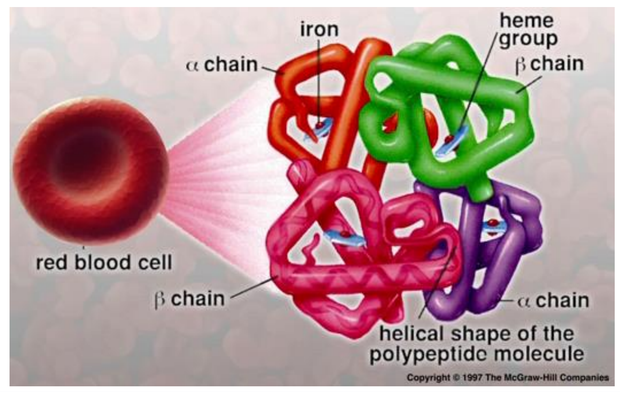
1 Laboratory for Molecular Diagnostics

2 Laboratory of Clinical Chemistry and Hematology

Jeroen Bosch Hospital, 's-Hertogenbosch, The Netherlands.

Introduction

Alpha-thalassemia is a blood disorder characterized by a decrease in α-globin chain production due to deletion or mutation of one or more of the four α-globin genes. This results in reduced oxygen carrying capability of the red blood cells. In this study we evaluated a commercial reverse-hybridisation strip assay, the α-Globin StripAssay® (ViennaLab-Goffin Molecular Technologies, Beesd, Netherlands), in comparison to our in-house conventional PCR.



Methods

A cohort of 40 archival DNA samples, extracted from EDTA-blood using either the QIAamp DNA mini kit (QIAGEN, Hilde, Germany) or MagNA Pure 96 (Roche, Basel, Switzerland) , was selected. All samples were previously tested for the presence of 5 α-Globin deletions (-$\alpha^{3.7}$, -$\alpha^{4.2}$, --^{MED}, --^{SEA}, -$(\alpha)^{20.5}$) using a set of conventional in-house PCR's.

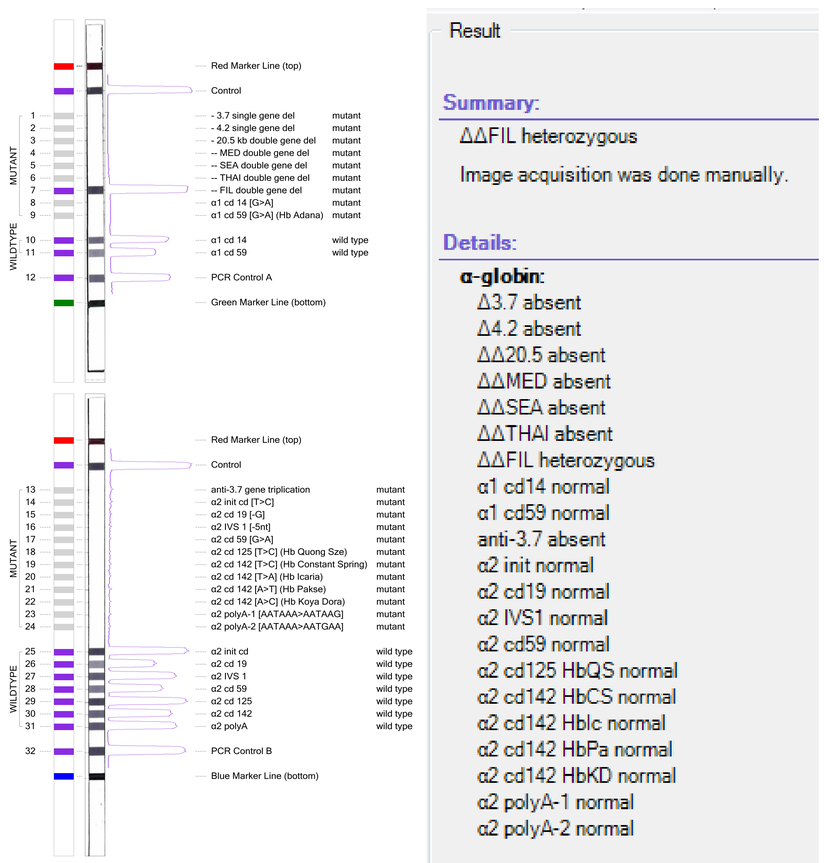
The cohort was subsequently re-evaluated using the α-Globin StripAssay® in combination with the automated Tendigo^{tm} washing station (Fujirebio, Gent, Belgium)

The asssay detects 21 different α-globin mutations: 7 deletions, the ααα$^{anti-3.7}$ gene triplication, two α1 and eleven α2 point mutations that can occur in different combinations.

Data interpretation was performed both manually and by using the StripAssay® evaluator software v2 (ViennaLab, Wien, Austria).

Results

Of the 40 samples, 16 were α-Globin wildtype in both assays. In another 16 samples, the 2 assays identified the same deletions. Five samples showed additional mutations using the StripAssay®, that were not covered in our in-house assay (--^{FIL}, ααα$^{anti-3.7}$ gene triplication and α2 IVS1-5nt (n=3)). Finally, 3 samples were found invalid in the StripAssay®, most likely due to no adequate NA input . In one of these samples, results from the in-house assay indicated -$\alpha^{3.7}$/-$\alpha^{4.2}$, whilst the remaining 2 were wildtype.



		α-globin stripAssay			
		invalid	wildtype	mutated	total
in-house	wildtype	2	16	5	23
	mutated	1	0	16	17
	total	3	16	21	40

n=3 α2 IVS1-5nt
n=1 --^{FIL}
n=1 ααα$^{anti-3,7}$
Mutations not covered by in-house assay

Conclusion

The α-globin StripAssay® showed good concordance with our in-house PCR assay and detected several additional α-Globin mutations because of its more comprehensive mutational coverage. The assay is user friendly and less labour intensive than our in-house assay, especially in combination with the evaluator software.

Ilse Luijten - de Vrije, Bachelor of Science, E-mail: i.d.vrije@jbz.nl, Tel.nr. 0031-73.553.3894