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

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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 (-α3.7, -α4.2, -αMED, -αSEA, -(α)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™ washing station (Fujirebio, Gent, Belgium). The assay detects 21 different α-globin mutations: 7 deletions, the αααanti-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, αααanti-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 -α3.7/-α4.2, whilst the remaining 2 were wildtype.

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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.

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.