Overview

**Aims:** To evaluate the most appropriate mass spectrometry-based method for automating high throughput screening of haemoglobin (Hb) disorders, which causes 15% of all hereditary haemoglobinopathies, and to provide a general healthcare problem screening program for the diagnosis of beta-thalassemia.

**Methods:** Two different methods have been evaluated on a large set of clinical samples: (i) a triple quadrupole mass spectrometer; and (ii) an approach based on high resolution and accurate mass LC-MS/MS. Direct infusion electrospray ionization (ESI) mass spectrometry analysis was performed to complement the results.

**Results and Conclusions:** A total of 1777 patient samples have been analyzed by a 2.0-skinny, well-trained team and current screening methods. The results showed complete concordance with existing methods for HbA, HbF, HbD, and HbS, which have been identified as the most appropriate methods for a rapid diagnostic screen.

**Introduction**

The haemoglobinopathies are the most common type of inherited disorders, and pose a significant healthcare problem. Screening programs for haemoglobinopathies currently use an automated high throughput sequencer which thoroughly analyzes each of the 1777 samples. This is a labor-intensive process that represents a range of variables, but it has not been validated in the context of the current work. To address this, we have developed a rapid, high-throughput, and accurate mass spectrometry detection method. A total of 1777 patient samples have been investigated for the diagnosis of alpha chain, beta chain, and delta chain disorders.

**Results:**

**Results: Intact Protein Analysis**

**Figures:**

- **Figure 1:** Distribution of Hb on the IEF gel, based on the clinical samples.
- **Figure 2:** Mass MS analysis of the intact protein analysis of different samples.
- **Figure 3:** Distribution of HbA, HbS, and HbD on the IEF gel, based on the clinical samples.

**Table 1:** The 100 values of the correlation of the HbMS, intact protein analysis of different samples.

**Table 2:** The 100 values of the correlation of the HbMS, intact protein analysis of different samples.

**Conclusions:**

- The automated mass spectrometry-based method for the diagnosis of beta-thalassemia in a clinical trial.

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**References:**