# Delta Beta (δβ) thalassemia: Learning from External Quality Assessment

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## Introduction

**External Quality Assessment Programs (EQA)** evaluate retrospectively the laboratory results, assessing their performance and competence.
They play a key role in the continuous training process of the professionals, contributing not only for precise and accurate results, but also to a correct interpretation.

**δβ thalassemia** results from a deletion in genes delta and beta of chromosome 11. Although its definitive identification demands genetic analysis, the hematologic evaluation allows the presumptive identification. The hematologic phenotype of heterozygotes for δβ-thalassemia is identic to β-thalassemia carrier, with microcytosis and hypochromia where the percentage of $HbA_2$ is not increased and Hb F is usually high, varying from 5 to 20%.

In 2018, the **National External Quality Assessment Program (PNAEQ)**, sent a sample **that simulated a carrier of delta beta (δβ) thalassemia**, in order to evaluate the performance of the participants registered in the Hemoglobinopathies Program.

## Objective

Evaluate the performance of PNAEQ's participants in the quantification of $HbA_2$ and HbF, and interpretation of results of a sample that simulated an δβ-thalassemia carrier.

## Methods

PNAEQ organizes, in collaboration with an expert work group, three rounds/year, with control and real patient samples, and case-studies for the evaluation of haemoglobinopathies.
One sample prepared by PNAEQ from whole blood and umbilical chord blood in order to simulate normal HbA2 and increased HbF, was sent to 17 participants in the 1st round of 2018.
The **quantitative results**, for HbA2 and HbF, were statistically evaluated for all the participants, according to the hierarchy (all, method, equipment)
The **qualitative results** for Hb fraction identification and interpretation, were evaluated according to the expert laboratories results.

## Results

The sample simulated **a 13 year old girl**, sent to a consult to study of **microcytic and hypochromic anaemia**. The blood count is shown in table 1.
15 of the17 labs answered and a summary from the results sent, is presented bellow:

| | |
|---|---|
| RBC (x1012/L) | 5,31 |
| Hb (g/dL) | 10,5 |
| Ht (L/L) | 0,335 |
| MCV (fL) | 63,0 |
| MCH (pg) | 19,8 |
| MCHC (g/dL) | 31,4 |
| RDW (%) | 18,0 |

**Table 1** – Hemogram values sent with the sample

### 1-Hb fraction identification

- 2 /15 labs did not identify HbF
- 3 /15 labs did not identify HbA
- 1/15 labs did not identify $HbA_2$
- 11/15 identified Hb A and Hb F

The identification of HbA is important for the characterization of the heterozygosity status as well as the increase of HbF for the phenotype hematologic characterization.

### 2-Quantitative essays

**$HbA_2$ - Normal** – Participant mean results = **2,2%** (minimum= 2,0% ;maximum= 2,7%)
**HbF- Increased-** Participant mean results =**16,2%**(minimum= 11,5% ;maximum=21,1%)

### 3-Interpretation:

Only 5 from 15 participants chose the option of δβ-thalassemia carrier and 11 advised a consult with an haematologist.

In the simulated case it was essential to make the presumptive identification of δβ-thalassemia carrier to clarify the microcytic and hipocromic anemia, as well as recomend the study of the partner in adulthood and identification of risk couple to forward to genetic counseling.

## Conclusion

The laboratory performance is evaluated not only by the evaluation of result precision and accuracy, as well as by the knowledge e competence demonstrated in the result interpretation. The results show that it is necessary to continue with the performance evaluation process and continuous training for External Quality Assessment in Hemoglobinopathies, aiming for improvement of performance of clinical results as well as result interpretation. The improvement of the performance will contribute for the correct genetic advise of patients and their family as well as clinical follow-up.

**References:**
Delta beta thalassemia:a rare hemoglobin variantBlood Res 2016;51:204-14; Barbara J.Bain. Haemoglobinopathy Diagnosis . Blackwell Science Ltd 2001