

# Beta-Thalassemia and HB lepore heterozygotes: phenotype-genotype correlation

Citation for published version (APA):

de Sousa Ribeiro, M. L. (1997). *Beta-Thalassemia and HB lepore heterozygotes: phenotype-genotype correlation*. [Doctoral Thesis, Maastricht University]. Universiteit Maastricht. <https://doi.org/10.26481/dis.19970613ms>

## Document status and date:

Published: 01/01/1997

## DOI:

[10.26481/dis.19970613ms](https://doi.org/10.26481/dis.19970613ms)

## Document Version:

Publisher's PDF, also known as Version of record

## Please check the document version of this publication:

- A submitted manuscript is the version of the article upon submission and before peer-review. There can be important differences between the submitted version and the official published version of record. People interested in the research are advised to contact the author for the final version of the publication, or visit the DOI to the publisher's website.
- The final author version and the galley proof are versions of the publication after peer review.
- The final published version features the final layout of the paper including the volume, issue and page numbers.

[Link to publication](#)

## General rights

Copyright and moral rights for the publications made accessible in the public portal are retained by the authors and/or other copyright owners and it is a condition of accessing publications that users recognise and abide by the legal requirements associated with these rights.

- Users may download and print one copy of any publication from the public portal for the purpose of private study or research.
- You may not further distribute the material or use it for any profit-making activity or commercial gain
- You may freely distribute the URL identifying the publication in the public portal.

If the publication is distributed under the terms of Article 25fa of the Dutch Copyright Act, indicated by the "Taverne" license above, please follow below link for the End User Agreement:

[www.umlib.nl/taverne-license](http://www.umlib.nl/taverne-license)

## Take down policy

If you believe that this document breaches copyright please contact us at:

[repository@maastrichtuniversity.nl](mailto:repository@maastrichtuniversity.nl)

providing details and we will investigate your claim.

# STELLINGEN

behorende bij het proefschrift

## **$\beta$ -THALASSEMIA AND Hb LEPORE HETEROZYGOTES: PHENOTYPE-GENOTYPE CORRELATION**

van

Maria Leticia de Sousa Ribeiro

1. Hb H disease when associated with the heterozygous state for Hb New York, has a phenotype which is more severe while the number of erythrocytes with Hb H inclusion bodies is lower.
2. The coinheritance of a  $\delta$ - and  $\beta$ -thalassemia allele should be considered in the presence of a hypochromic microcytic anemia with normal Hb A<sub>2</sub> levels in Mediterranean populations.
3. The phenotypic differences associated with point mutations in the proximal and distal CACCC boxes of the promoter region of the  $\beta$ -globin gene depend on the different binding specificities and affinities of these two motifs for erythroid factors.
4. The single strand conformation polymorphism (SSCP) procedure is a useful technique for the screening of mutations in hemoglobinopathies.
5. Prenatal determination of the fetal RhD type by DNA amplification is a reliable and rapid method for the management of Rh alloimmunization.
6. The homozygous state for the dominant form of Hereditary Spherocytosis, Band 3 Coimbra mutation (488 Val→Met), is associated with hydrops fetalis, and a severe, transfusion dependent, phenotype with metabolic acidosis.
7. The association of a homozygosity for a Pyruvate Kinase deficiency and a heterozygosity for Hb Lepore Baltimore ( $\delta^{68Leu}\text{-}\beta^{84Thr}$ ) creates a phenotype of mild chronic hemolytic anemia with hypochromia, microcytosis and splenomegaly.
8. The implementation of molecular technology in the developing countries is essential for the prevention and control of emerging and endemic infectious diseases.
9. The use of erythropoietin to treat anemia in premature infants reduces their transfusion needs.
10. In infants with neuroblastoma, the hyperdiploid tumor DNA is associated with a favorable prognosis, while N-myc amplification is associated with a poor prognosis.
11. It is easier to climb a mountain than to come down.