NEWBORN SCREENING FOR HEMOGLOBINOPATHIES IN MEXICO: EXPERIENCE FROM A PIONEERING PROGRAM ON 174,531 NEWBORNS.

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Hemoglobinopathies

- One of the most common genetic diseases in humans;
- Their high frequency represents a great concern to public health.

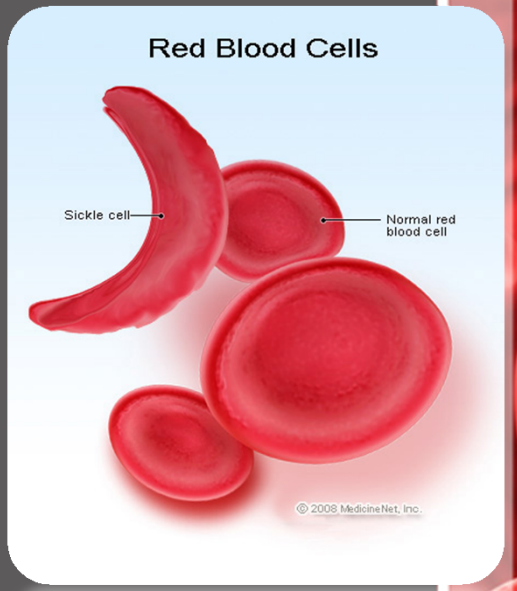
Angastiniotis M, Modell B. Global epidemiology of hemoglobin disorders. Ann N Y Acad Sci. 1998;850:251-69.

- The WHO suggests that at least 5% of the world population are carriers of some genetic disorders of hemoglobin.

Angastiniotis M, Modell B. Global epidemiology of hemoglobin disorders. Ann N Y Acad Sci. 1998;850:251-69.

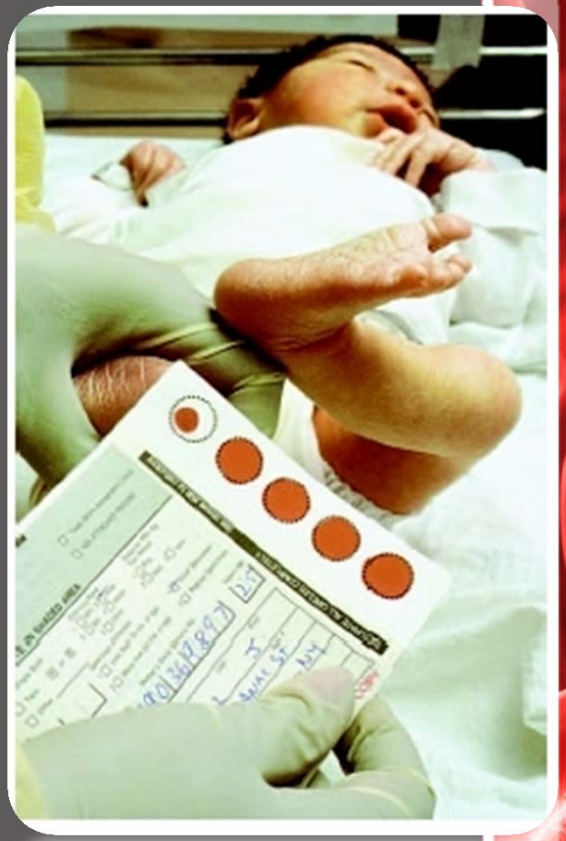
Clinical consequences

- Important cause of morbidity and mortality in childhood.
- Their clinical consequences are:
  - Anemia,
  - Failure to thrive,
  - Repeated infections in infancy,
  - Vascular occlusive disorders,
  - Severe pain,
  - Stroke,
  - Organ failure,
  - Death.



Benefit of HBs NB screening

- Early identification and careful follow-up, coupled with relatively simple interventions **substantially reduces morbidity and mortality**.



Weatherall DJ, Clegg JB, Higgs DR, Wood WG. The Hemoglobinopathies. In Scriver xxx4571-636

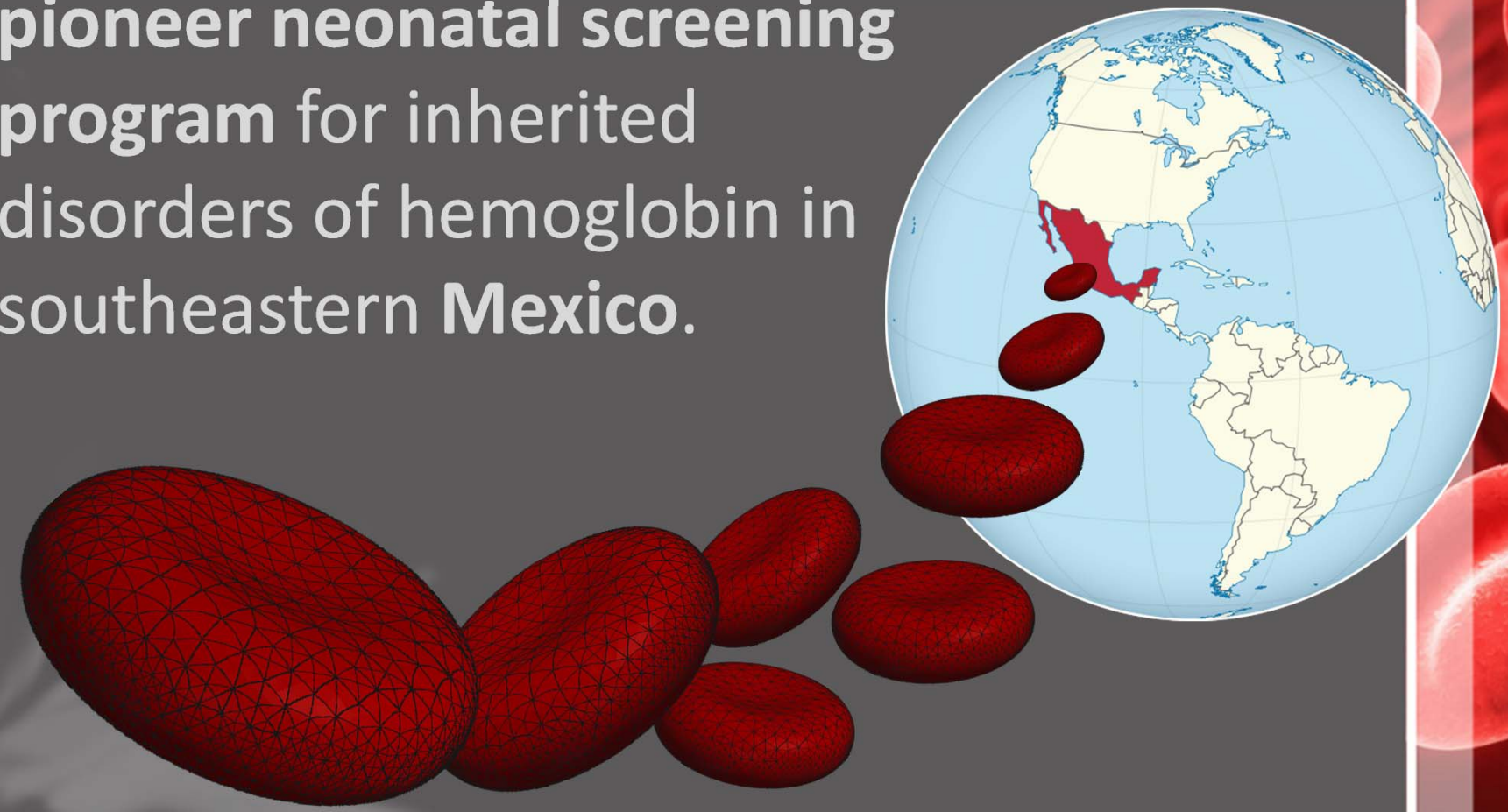
Hemoglobinopathies situation in Latin America

- There are **few** newborn screening programs that include its detection as mandatory.
  - Brazil
- Epidemiology is not well known.

Borrajo GJ. Newborn screening in Latin America at the beginning of the 21st century. J Inherit Metab Dis. 2007:466-81

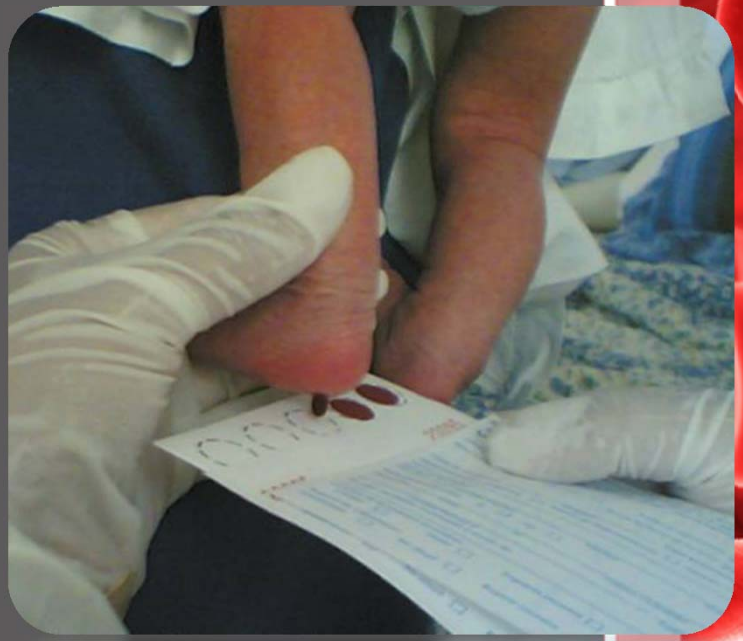
Objective

- To present the results of a **pioneer neonatal screening program** for inherited disorders of hemoglobin in southeastern **Mexico**.



Methodology

- From September 2007 to September 2012;
- Prospective and descriptive study was performed in the south east of Mexico;



Methodology

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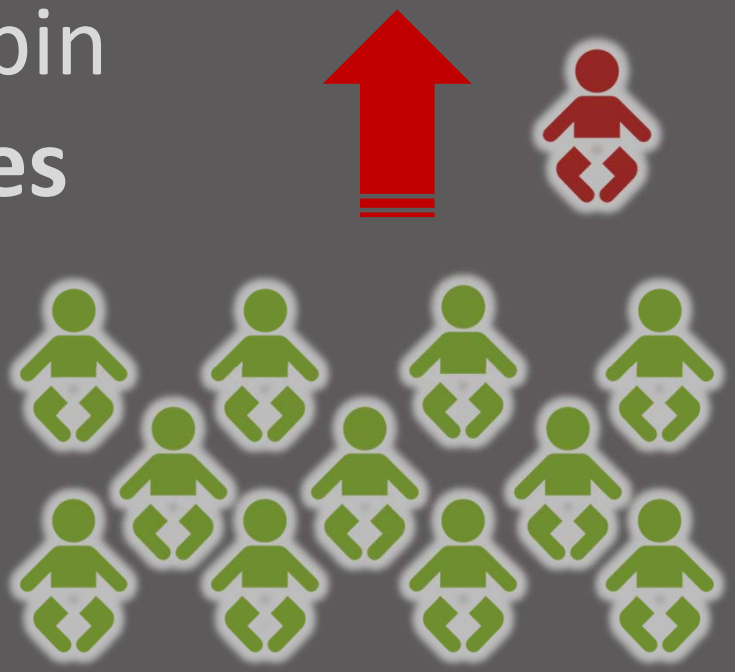
Methodology

- Heel prick dried blood spots from Guthrie cards;
- Analyzed by:
  - **Isoelectric focusing** on agarose gels;
  - Since 2012 the specimens were analyzed also by **HPLC (**Variant nbs by BIO-RAD).

Results

2,853 Hemoglobin variants (**cases and traits**)

1.63%



174,531 Screened NB

Results

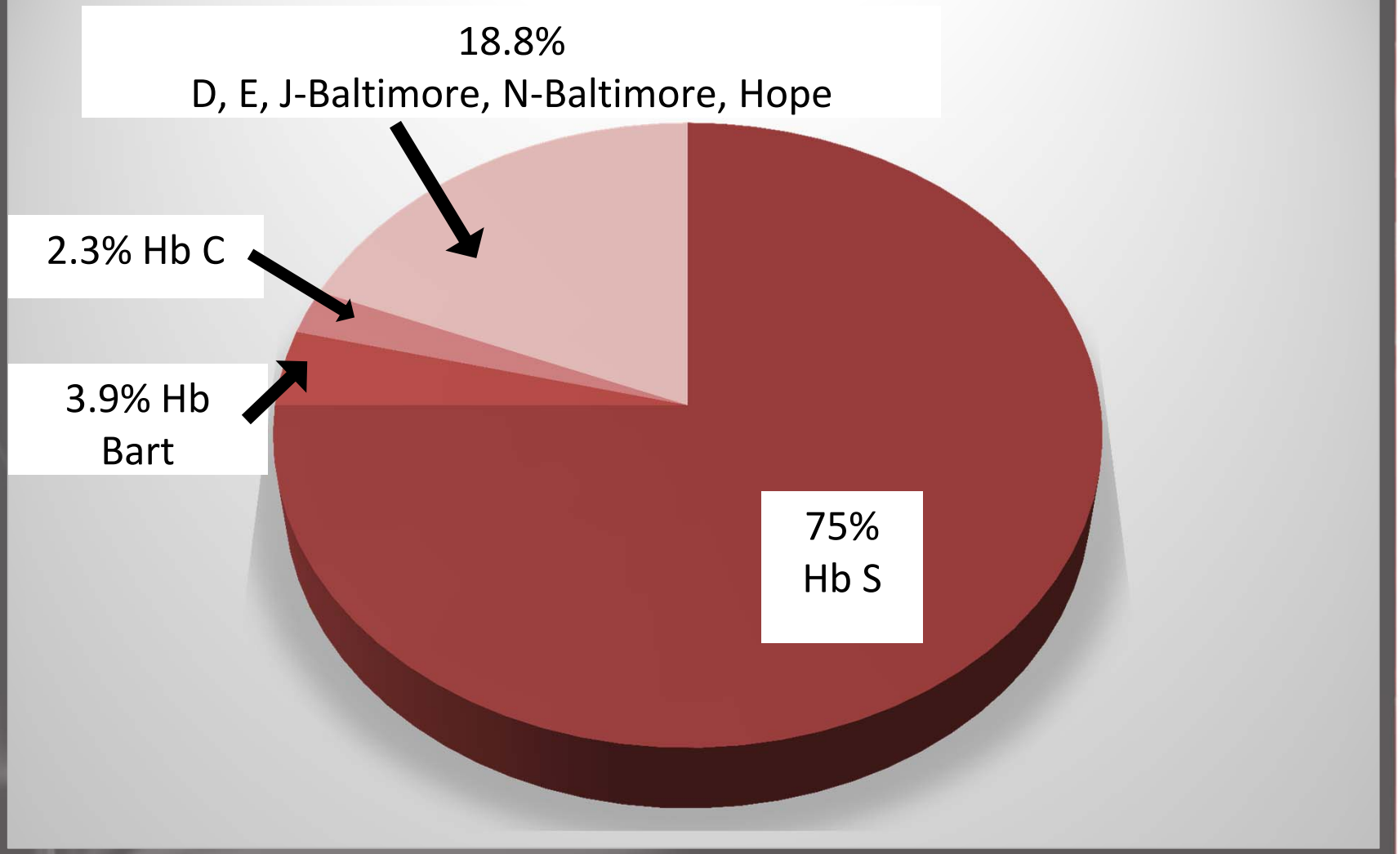
- Twenty two patients (homozygous or double heterozygous,) were diagnosed of having hemoglobinopathies (**1.26 per 10,000 NB**);
  - 17 with sickle cell disease (0.97 per 10,000 NB);
  - 4 beta-thalassemia (0.23 per 10,000 NB)
  - 1 with alfa-thalassemia minor (0.06 per 10,000 NB).

Results

- Birth prevalence of traits and diseases was

16.3 per 10,000 newborns.

Results: Hemoglobine variants found in Southeastern Mexico



Hemoglobinopathies birth prevalence in Hispanic population (per 10,000 nb)

Reference	N	SS/SB/SC	Carriers	Total
Lobo et al. *Pan Am J Public Health* 2003;13:154-9. (Rio de Janeiro)	99,260	0.83	37	ND
Feuchtbaum et al. *Genet Med* 2012:14:937–945 (California) (Hispanics only and hispanic white)	1,183,044	<1.06 (Only SS)	ND	ND
Present study (Southeast Mexico)	**174,531**	**1.26**	**16.3**	**16.6**

Results

- All the affected families received genetic counseling.
- As part of the genetic study of the affected cases, we found two families with other affected siblings with sickle cell disease, without previous specific diagnosis.

Conclusion

- Newborn screening for hemoglobinopathies is **feasible** in Mexico.

CONCLUSION

The hemoglobinopathies birth prevalence found in our study:

1.2 x 10,000 NBS

The **well known public health impact** of early medical intervention

INCLUSION IN THE MANDATORY PANEL OF NB SCREENING IN MEXICO

Thank you,
and
Happy Birthday to Newborn Screening!!!
1963-2013