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.

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

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;
The study included newborns from the states of Tabasco,
Methodology

- The study included newborns from the states of Tabasco, Yucatan
Methodology

- The study included newborns from the states of Tabasco, Yucatan and Chiapas.
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)

174,531 Screened NB

1.63%
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: Hemoglobin variants found in Southeastern Mexico

- 75% Hb S
- 18.8% D, E, J-Baltimore, N-Baltimore, Hope
- 2.3% Hb C
- 3.9% Hb Bart
<table>
<thead>
<tr>
<th>Reference</th>
<th>N</th>
<th>SS/SB/SC</th>
<th>Carriers</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Feuchtbaum et al.</strong> <em>Genet Med</em> 2012:14:937–945 (California) (Hispanics only and hispanic white)</td>
<td>1,183,044</td>
<td>&lt;1.06 (Only SS)</td>
<td>ND</td>
<td>ND</td>
</tr>
<tr>
<td><strong>Present study</strong> (Southeast Mexico)</td>
<td>174,531</td>
<td>1.26</td>
<td>16.3</td>
<td>16.6</td>
</tr>
</tbody>
</table>
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