Introduction

- Hemoglobinopathy disorders are genetic defects in hemoglobin.
- High mortality and morbidity rates present in thalassemia disorder (β-thal) and sickle cell disease (SCD).
- Higher prevalence of β-thal and SCD in Saudi Arabia compared to neighboring countries.
- In 2011, 0.05% β-thal and 4.5% SCD prevalence.
- Saudi Arabia initiated a mandatory premarital screening and genetic counseling program (PMSGC) in 2004.
- Trends in prevalence rates of β-thal and SCD have not been examined since 2011.

Objectives

- Assess recent trends in β-thal and SCD and their distribution by demographic characteristics and geographic regions using data from the PMSGC program.

Methods

- Secondary data analysis.
- 1,230,582 individuals.
- Data obtained from Department of Genetics of the Saudi Ministry of Health (MoH).
- PMSGC program.
- Included all couples within 13 administrative regions from February 2011 to December 2015.
- Status of β-thal and SCD categorized as positive, negative, and carrier.
- Prevalence rate and 95% (CI) of β-thal and SCD estimated by study year and geographic region.

Results

Population characteristics: 49.7% men; average age 27.8 years (SD 8.85 years) and 50.34% women; average age 22.6 years (SD 6.4).

Table 1. Overall prevalence rate (per 1000) for β-thalassemia and Sickle Cell disorders in all region, Saudi Arabia, 2011–2015

<table>
<thead>
<tr>
<th>Year</th>
<th>Disease</th>
<th>Trait</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>2011</td>
<td>β-thal</td>
<td>3.8</td>
<td>12.9</td>
</tr>
<tr>
<td>2012</td>
<td>SCD</td>
<td>45.8</td>
<td>49.6</td>
</tr>
</tbody>
</table>

β-thal major: PR (per 1000) ranged from 1 in 2011 to 1.6 in 2015.
β-thal trait: PR (per 1000) ranged from 24.2 in 2011 to 12 in 2015.

SCD: PR (per 1000) ranged from 5.2 in 2011 to 12 in 2015.

β-thal Trait: PR (per 1000) ranged from 3304 in 2011 to 13490 in 2015.

SCD Trait: PR (per 1000) ranged from 113.2 in 2011 to 113.0 in 2015.

Table 2. Prevalence rates (PR per 1000) for β-thalassemia Trait and Sickle Cell Trait in Saudi Arabia form 2011 to 2015

<table>
<thead>
<tr>
<th>Year</th>
<th>1,230,582</th>
<th>Positive test</th>
<th>PR</th>
<th>Confidence Interval</th>
</tr>
</thead>
<tbody>
<tr>
<td>2011</td>
<td>78072</td>
<td>1982</td>
<td>24.2</td>
<td>23.3-25.3</td>
</tr>
<tr>
<td>2012</td>
<td>256601</td>
<td>4057</td>
<td>17.1</td>
<td>16.8-17.3</td>
</tr>
<tr>
<td>2013</td>
<td>285070</td>
<td>2837</td>
<td>10.6</td>
<td>10.2-11.0</td>
</tr>
<tr>
<td>2014</td>
<td>276226</td>
<td>2696</td>
<td>9.7</td>
<td>9.5-10.0</td>
</tr>
<tr>
<td>2015</td>
<td>325871</td>
<td>3057</td>
<td>11.8</td>
<td>11.6-12.0</td>
</tr>
</tbody>
</table>

Table 3. Prevalence rate for β-thal and sickle cell traits and disorders in regions with highest burden

<table>
<thead>
<tr>
<th>Region</th>
<th>Jazan Region</th>
<th>Eastern Region</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prevalence rate (95% Confidence Interval)</td>
<td>32.1 (30.8-33.4)</td>
<td>23.7 (23.1-24.4)</td>
</tr>
<tr>
<td>β-thal major</td>
<td>0.6 (0.5-0.8)</td>
<td>0.4 (0.3-0.5)</td>
</tr>
<tr>
<td>SCT</td>
<td>135.7</td>
<td>114.4</td>
</tr>
<tr>
<td>SCD</td>
<td>6.8 (6.2-7.4)</td>
<td>9.8 (9.4-10.2)</td>
</tr>
</tbody>
</table>

Conclusion

- Over a 5-year period, a decreasing trend in the prevalence of β-thal was observed.
- SCD rates were rather stable over time.
- Compared to 2004–2009 rates:
  - β-thal major rates were similar in 2011–2015.
  - β-thal carrier rates decreased (from 32 to 13 per 1000).
- SCD and SCT rates similar.
- Highest prevalence observed in Eastern region and Jazan region, similar to distribution reported in 2004–2011 study.
- Program is moving towards reaching goals of lowering prevalence of β-thal.

Recommendations

- Rates still especially high among certain regions like Eastern Region and Jazan; these regions should be targeted with intensified awareness programs.
- β-thal and SCD rates in newborns need to be assessed to evaluate program effectiveness.
- More effective genetic counseling programs for at-risk couples.
- Awareness programs among youth about risks and receptivity to genetic counseling.

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