Thalassaemia and Abnormal Haemoglobins in Pregnancy

1. Introduction

Haemoglobinopathies (comprising the thalassaemias and abnormal haemoglobins) are hereditary disorders which affect the balance of globin chain synthesis and/or the structure of haemoglobin.

Thalassaemia is an inherited condition that affects the production of haemoglobin, which carries oxygen in our blood. It appears in the following forms:

<table>
<thead>
<tr>
<th>Type</th>
<th>Description</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>thalassaemia minor</td>
<td>carrier form - one member of the gene pair is not working properly</td>
<td>no effects on health</td>
</tr>
<tr>
<td>beta thalassaemia major</td>
<td>both members of the beta gene pair are not working</td>
<td>severe anaemia</td>
</tr>
<tr>
<td>alpha thalassaemia major</td>
<td>both members of the alpha gene pairs are not working</td>
<td>Barts Hydrops</td>
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</tbody>
</table>

These disorders are detected in approximately 4% of patients of reproductive age attending the Women's. In almost half of these cases, the abnormality is not evident following simple full blood examination and is only detected by haemoglobin electrophoresis.

Thalassaemias and abnormal haemoglobins have been described in every ethnic group.

They are most frequent in people originating from:
- the Mediterranean basin
- the Middle East
- Africa
- Asia
- Polynesia
- the Subcontinent.

As the inheritance of thalassaemia syndromes is autosomal recessive, the heterozygous carrier states are essentially asymptomatic. Most people are unaware of their carrier state. Depending on the mutation, homozygous or compound heterozygous thalassaemia syndromes may result in adverse maternal outcomes, stillbirth, transfusion dependency, or sickling syndromes.

The aim of screening couples for thalassaemia is to detect those at risk of having children with severe disease, with a view to offering prenatal diagnosis and the option of termination of pregnancy in the event of a positive diagnosis, or to facilitate the early diagnosis and treatment of affected children.

Screening protocol: RWH link -- copy

Further information: www.thalassaemia.org.au/