CHROMATOGRAM INTERPRETATION AIDS

- Software to assist in the interpretation of chromatograms
- Tosoh Bioscience offers an online clinical interpretation platform under the supervision of highly knowledgeable experts in the field

Example chromatograms: Normal, ß-Thalassaemia trait, HbS trait, HbE trait

References:
2. Haemoglobinopathies on the move: Is Europe ready? Report by group of experts from the European Network for Rare and Congenital Anaemias (ENERCA) and the Thalassemia International Federation (TIF) in collaboration with the International Organization for Migration (IOM), Migration Health Division, Regional Office Brussels
Haemoglobinopathies are among the most common hereditary diseases of the world’s population. About 4.5% of all human beings carry a gene for a thalassaemia or haemoglobin anomaly. The areas in which such abnormalities were originally most common extend from Africa over the Mediterranean basin and the Near and Middle East to Southeast Asia and the Indian subcontinent. Global migration in the modern period has led to a continual spread of these anomalies to all regions of the world, with the result that they are rapidly becoming more common in the industrialised regions of Northern and Central Europe as well.

Variants of thalassaemias and sickle abnormal haemoglobins interact to produce a wide range of clinical disorders. The diagnosis of thalassaemia major can be reliably made if at least one of the parents is a carrier of the disease. Variants of the α-globin chain, the β-globin chain, and the δ- and γ-globin chain are the major genetic defects in thalassaemia.

LABORATORY DIAGNOSIS

Diagnosis of Beta-Thalassaemia and other types of haemoglobinopathies should be done based on clinical symptoms if available and a number of laboratory tests, such as MCV, MCH, total red cell count, HbA, HbA2 and iron markers.

As a guideline, the below scheme can be used (adapted from Mosca et al. J.Clin.Pathol. 2009 – with permission)

- Quantitative determination of HbA and HbA2 in 5 minutes
- Chromatographic separation between HbA2 and HbF
- High resolution chromatogram thanks to Tosoh’s over 40 years’ experience in HPLC
- Full reagent traceability
- Easy to use and intuitive instrument
- Highly reliable system
- Instrument connectable to open laboratory automation lines

TOSOH G11 BETA-THALASSAEMIA SOLUTION

<table>
<thead>
<tr>
<th>HbA2</th>
<th>MCV</th>
<th>MCH</th>
<th>RBC</th>
<th>HbF</th>
<th>Iron Markers</th>
</tr>
</thead>
<tbody>
<tr>
<td>reduced</td>
<td>&lt; 2.3%</td>
<td>normal</td>
<td>normal</td>
<td>&lt;1%</td>
<td>normal</td>
</tr>
<tr>
<td>normal</td>
<td>normal</td>
<td>normal</td>
<td>normal</td>
<td>&lt;1%</td>
<td>normal</td>
</tr>
<tr>
<td>increased</td>
<td>&gt; 4.1%</td>
<td>normal</td>
<td>normal</td>
<td>&lt;1%</td>
<td>normal</td>
</tr>
</tbody>
</table>

As a guideline, the below scheme can be used (adapted from Mosca et al. J.Clin.Pathol. 2009 – with permission)

- Quantitative determination of HbA and HbA2 in 5 minutes
- Chromatographic separation between HbA2 and HbF
- High resolution chromatogram thanks to Tosoh’s over 40 years’ experience in HPLC
- Full reagent traceability
- Easy to use and intuitive instrument
- Highly reliable system
- Instrument connectable to open laboratory automation lines

Molecular confirmation:
- α-Gap-PCR
- β-Gap-PCR
- δ-gap DNA
- γ-gap DNA
- α-gap DNA
- β-gap DNA
- γ-gap DNA
- δ-gap DNA
- Gap-PCR
- Δ-gene
- Δ-MLPA
- Δ-MLPA
- MLPA
- α-MLPA
- β-MLPA
- Gap-PCR
- Δ-gene
- Δ-MLPA
- Δ-MLPA

Variant of the α-globin gene
- Normal
- Reduced
- Increased
- α-thalassaemia
- δ-thalassaemia
- β-thalassaemia
- γ-thalassaemia

Variant of the β-globin gene
- Normal
- Reduced
- Increased
- β-thalassaemia
- δ-thalassaemia
- β-thalassaemia
- γ-thalassaemia

Variant of the δ-globin gene
- Normal
- Reduced
- Increased
- δ-thalassaemia
- β-thalassaemia
- β-thalassaemia
- γ-thalassaemia

Variant of the γ-globin gene
- Normal
- Reduced
- Increased
- γ-thalassaemia
- δ-thalassaemia
- β-thalassaemia
- β-thalassaemia

Variant of the δ- and γ-globin gene
- Normal
- Reduced
- Increased
- δ-thalassaemia
- β-thalassaemia
- β-thalassaemia
- γ-thalassaemia

Variant of the α- and β-globin gene
- Normal
- Reduced
- Increased
- α-thalassaemia
- β-thalassaemia
- γ-thalassaemia
- δ-thalassaemia

Variant of the α- and δ-globin gene
- Normal
- Reduced
- Increased
- α-thalassaemia
- δ-thalassaemia
- β-thalassaemia
- γ-thalassaemia

Variant of the α- and γ-globin gene
- Normal
- Reduced
- Increased
- α-thalassaemia
- γ-thalassaemia
- β-thalassaemia
- δ-thalassaemia

Variant of the α- and β-globin gene
- Normal
- Reduced
- Increased
- α-thalassaemia
- β-thalassaemia
- δ-thalassaemia
- γ-thalassaemia

Variant of the α- and δ- and γ-globin gene
- Normal
- Reduced
- Increased
- α-thalassaemia
- δ-thalassaemia
- γ-thalassaemia
- β-thalassaemia

Variant of the δ- and β-globin gene
- Normal
- Reduced
- Increased
- δ-thalassaemia
- β-thalassaemia
- γ-thalassaemia
- α-thalassaemia

Variant of the γ- and β-globin gene
- Normal
- Reduced
- Increased
- γ-thalassaemia
- β-thalassaemia
- δ-thalassaemia
- α-thalassaemia

Variant of the δ- and γ-globin gene
- Normal
- Reduced
- Increased
- δ-thalassaemia
- γ-thalassaemia
- β-thalassaemia
- α-thalassaemia

Variant of the α-, β-, δ-, and γ-globin gene
- Normal
- Reduced
- Increased
- α-thalassaemia
- β-thalassaemia
- δ-thalassaemia
- γ-thalassaemia

Variant of the α-, β-, δ-, γ-, and ε-globin gene
- Normal
- Reduced
- Increased
- α-thalassaemia
- β-thalassaemia
- δ-thalassaemia
- γ-thalassaemia
- ε-thalassaemia

Variant of the α-, β-, δ-, γ-, ε-, and ω-globin gene
- Normal
- Reduced
- Increased
- α-thalassaemia
- β-thalassaemia
- δ-thalassaemia
- γ-thalassaemia
- ε-thalassaemia
- ω-thalassaemia

Variant of the α-, β-, δ-, γ-, ε-, ω-, and η-globin gene
- Normal
- Reduced
- Increased
- α-thalassaemia
- β-thalassaemia
- δ-thalassaemia
- γ-thalassaemia
- ε-thalassaemia
- ω-thalassaemia
- η-thalassaemia