Erythrocytosis

Presentation

Definition

Raised haematocrit persisting for more than 2 months (>0.54 in males, >0.48 in females)

Clinical Findings

Types of Erythrocytosis

1. Apparent erythrocytosis
   Increased Hb/Hct but normal red cell mass due to a reduction in plasma volume. This can be due to diuretics, excess alcohol, excess caffeine, smoking and obesity.

2. Absolute erythrocytosis
   Due to a real increase in red cell mass. Any Hct >0.6 in men or >0.56 in women is considered absolute erythrocytosis. This can be divided into three categories:
   a) Primary: where there is an intrinsic problem with the bone marrow (see below)
   b) Secondary: where there is external influence driving erythropoiesis in the bone marrow
   c) Idiopathic: when primary and secondary causes have been excluded

Causes

Secondary causes

- The treatment is dependent on identifying the underlying condition and treating it appropriately, such as:
  - Consider oxygen supplementation in COPD
  - Consider referral for assessment of sleep apnoea and possible CPAP
  - Recommend cessation of smoking
- Venesection only warranted if previous history of vascular or venous insults or deemed at very high risk. Aim for Hct <0.54 with venesection (this is not routinely provided by haematology but can be discussed).

<table>
<thead>
<tr>
<th>Central hypoxia</th>
<th>Chronic lung disease, right-to-left cardiopulmonary vascular shunts, Obstructive sleep apnoea, smoking, carbon monoxide poisoning</th>
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</thead>
<tbody>
<tr>
<td>Renal hypoxia</td>
<td>End stage renal failure, renal artery stenosis, renal cysts, hydronephrosis</td>
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<td>Tumours producing erythropoietin</td>
<td>Hepatocellular cancer, renal cell cancer, cerebellar haemangioblastoma, parathyroid, uterine, pheochromocytoma, meningioma</td>
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<td>Exogenous erythropoietin</td>
<td>Anabolic steroids, androgens, post renal transplant</td>
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<td>Congenital (rare)</td>
<td>High oxygen-affinity haemoglobin, VHL mutation, erythropoietin receptor-mediated</td>
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Primary erythrocytosis (polycythaemia vera)

- Myeloproliferative neoplasm (MPN) with a clonal disorder of erythroid progenitors.
- Median age of presentation around 60 years old
- >95% of patient will have a positive JAK2 mutation.
- Ferritin and erythropoietin levels usually low (or low normal).
- May also have a raised WCC or platelet count and may have an enlarged spleen.
- If the JAK2 mutation is negative with a reduced EPO level or ferritin please consider referral still as a bone marrow biopsy may be indicated
- Increased risk of both arterial and venous thrombosis, haemorrhage and risk of progression to myelofibrosis and acute myeloid leukaemia.
- Standard treatment includes venaecion (to keep haematocrit <0.45) and low dose aspirin. Pharmacological cytoreduction may be required (commonly with hydroxycarbamide).
- Cardiovascular risk factors should be addressed.

Symptoms and Signs

- Most patients with erythrocytosis are asymptomatic. It is important to take a history and examine for secondary causes. Erythrocytosis can cause excessive sweating, hyperviscosity, pruritis, thrombosis (including at unusual sites), facial plethora and hepato/splenomegaly.

Investigations

<table>
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<tr>
<th>Investigations in primary care should include</th>
<th>Investigations to consider in primary care</th>
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<tbody>
<tr>
<td>FBC and film</td>
<td>JAK2 mutation</td>
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<tr>
<td>Ferritin</td>
<td>USS of abdomen</td>
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<tr>
<td>Erythropoietin</td>
<td>Lung function test</td>
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<td>Oxygen saturations</td>
<td>Epworth sleepiness score</td>
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<tr>
<td>U+Es and LFTs</td>
<td>Carboxyhaemoglobin (smokers or possible carbon monoxide exposure).</td>
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Referral

- Persistent, unexplained erythrocytosis: routine referral.
- Symptoms of hyperviscosity may need prompt treatment: urgent discussion.
- Urgent referral if no congenital heart disease with Haematocrit (Hct) of >0.60 in men and >0.56 in females.

References
