Hampshire Hospitals NHS
NHS Foundation Trust

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).

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

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 venesection (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 aymptomatic. 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

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

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

1) Br J Haematol. 2005 Jul;130(2):174-95. Guidelines for the diagnosis, investigation and management of polycythaemia/erythrocytosis. McMullin MF et al.