South West Strategic Clinical Network
Somerset, Wiltshire, Avon and Gloucestershire (SWAG) Cancer Services

Erythrocytosis

Definition

Raised Hct (>0.52 in males, >0.48 in females) persisting for more than 2 months.

Types

1. Apparent erythrocytosis

   Increased Hb/Hct but normal red cell mass. Can be due to diuretics, excessive ethanol, smoking, obesity. Relative erythrocytosis refers to a true state of dehydration.

2. Absolute polycythaemia

   Due to real increase in red cell mass (>25% of mean predicted value). Any Hct >0.6 in men or >0.56 in women is considered absolute erythrocytosis. It can be divided into three categories:

   - Primary: polycythaemia rubra vera (PRV)
   - Secondary: causes of apparent erythrocytosis and erythropoietin (EPO) driven (see table below)
   - Idiopathic: when primary and secondary causes have been excluded

Central hypoxia	COPD, OSA, OHS, CO poisoning.
Renal hypoxia	ESRF, RAS, renal cysts, hydronephrosis
Tumour producing EPO	RCC, HCC, uterine, parathyroid, cerebellar, meningioma, phaeochromocytoma
Exogenous	Anabolic steroids
Post renal transplant erythrocytosis	

OSA obstructive sleep apnoea, OHS obesity hypoventilation syndrome, CO carbon monoxide, ESRF end stage renal failure, RAS renal artery stenosis, RCC renal cell carcinoma, HCC hepatocellular carcinoma

Table 1. Common causes of secondary erythrocytosis

History

Most patients with erythrocytosis are diagnosed incidentally and are asymptomatic. It is important to take a history and examine for causes of secondary and apparent erythrocytosis.

Symptoms & Signs

Erythrocytosis can cause excessive sweating, hyperviscosity, pruritus thromboses (particular at unusual sites), facial plethora and hepato/splenomegaly.

Investigations
Investigations in primary care should include:

1. FBC & blood film
2. Ferritin (low in PRV)
3. U&Es & LFTs
4. Oxygen saturations

Management
Ensure modifiable risk factors are addressed such as ethanol intake, hypertension, medications (thiazide diuretics) and smoking. If suspicious of secondary causes consider appropriate investigations and referral as necessary (ie respiratory team if chest disease, cancer referral pathways). Treatment of underlying cause should be sought.

Referral to Haematology
Please refer to clinical haematology if persistent, unexplained erythrocytosis. Symptoms of hyperviscosity may need prompt treatment, therefore please discuss with clinical haematology.

Summary of Polycythaemia rubra vera (PRV)

- Myeloproliferative neoplasm (MPN) with clonal disorder of erythroid progenitors.
- Median age of presentation ~ 60 years.
- >95% of cases are associated with a positive mutation in the JAK-2 gene in exon 14, V617F.
- Ferritin and erythropoietin levels usually low.
- Increase in risk of thromboses (both arterial and venous), haemorrhage, with risk of progression to secondary myelofibrosis or AML.
- Standard treatment includes venesection (to keep Hct <0.45) and low dose aspirin.
- Pharmacological cytoreduction occasionally also used.

References
Guidelines for the diagnosis, investigation and management of polycythaemia/ertythrocytosis. McMullin et al. British Journal of Haematology. 2007. 138;6. 821-822