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**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.

1. 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:

1. Primary: where there is an intrinsic problem with the bone marrow (see below)
2. Secondary: where there is external influence driving erythropoiesis in the bone marrow
3. 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).

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| 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, phaeochromocytoma, 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 asymptomatic. It is important to take a history and examine for secondary causes. Erythrocytosis can cause excessive sweating, hyperviscosity, pruritus, thrombosis (including at unusual sites), facial plethora and hepato/splenomegaly.

**Investigations**

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| 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.