A006 - POLYCYTHAEMIA (ERYTHROCYTOSIS) IN ADULT PATIENTS

What is polycythaemia?

Polycythaemia means too many red cells in the blood. It may be divided into 'true' (or absolute) erythrocytosis due to an increase in red cells, or an 'apparent' erythrocytosis, when red cell numbers are normal but are instead more concentrated due to less plasma.

An absolute erythrocytosis is then further categorised into primary or secondary according to whether the abnormality arises from the haemopoietic stem cell in the marrow (primary, i.e. polycythaemia rubra vera) or is due to overstimulation of a normal marrow stem cell to produce red cells (secondary, i.e. excessive erythropoietin (EPO) production).

Secondary erythrocytosis is much more common than primary.

Table 1. Causes of erythrocytosis

ABSOLUTE ERYTHROCYTOSIS (true increase in red cells)	
Primary (caused by a clonal abnormality within the haemopoietic stem cell in the marrow)	
• Polycythaemia vera – the *JAK2V617F* mutation is present in approximately 95% of these patients • Other myeloproliferative diseases	
Secondary (caused by an abnormality outside of the marrow)	
• Congenital (rare)	Abnormal haemoglobin with increased oxygen affinity Inherited high erythropoietin levels
• Acquired (increase in EPO)	**Hypoxia**: chronic lung disease, some types of congenital heart disease, sleep apnoea **Renal disease**: tumours, cysts (usually benign), hydronephrosis, post renal transplant **Liver disease**: hepatoma, cirrhosis, hepatitis **Tumours**: bronchial carcinoma, uterine fibroids, cerebellar haemangioma **Endocrine**: Cushing's syndrome, phaeochromocytoma **Drugs** – e.g. testosterone, other anabolic steroids, exogenous erythropoietin
APPARENT ERYTHROCYTOSIS (normal red cell volume, reduced plasma volume)	
• Obesity, fluid loss, diuretics, smoking, hypertension, alcohol, renal disease, psychological stress. • Drugs e.g. diuretics	

What is persistent erythrocytosis?

Raised haematocrit >0.52 males, >0.48 females for >2 months

What are the next steps for patients with persistent erythrocytosis?

Exclude likely secondary causes with full history/examination including:

Hypoxia driven

- chronic lung disease
- smoker's erythrocytosis
- hypoventilation syndromes including sleep apnoea
- right to left cardiopulmonary vascular shunts

Medications e.g. diuretics, testosterone, other anabolic steroids

If no secondary cause identified, check for family history of raised Hb, further investigations to include:

- FBC, differential & blood film
- U&E, LFT
- Ferritin/iron studies
- JAK2 mutation (requested through ICE)

When should I seek further advice or refer to haematology?

- patients who are JAK2 positive
- patients who are JAK2 negative with a very high haematocrit (males >0.6, females >0.56)
- a persistently raised haematocrit and a history of arterial or venous thrombosis
- a persistently raised haematocrit and no secondary cause identified
- a persistently raised haematocrit associated with other FBC abnormalities (thrombocytosis, leucocytosis)

What further investigations are recommended for patients not referred to haematology?

- oxygen saturation
- abdominal ultrasound to look for renal pathology

What follow up is required for patients for whom no cause has been found and who have not been referred to Haematology?

- if no cause for erythrocytosis has been identified, it may be prudent to recheck the patient's FBC every 4 to 6 months to ensure they are not developing a myeloproliferative disorder

Note iron deficiency may mask a rising Hb (but do NOT offer iron replacement)

What other advice should I give?

- patients should be asked to return for FBC if they develop thrombotic (e.g. TIAs) or haemorrhagic symptoms, or significant weight loss or left upper quadrant discomfort.
- lifestyle should be reviewed and additional risk factors for arterial/venous thrombosis should be addressed.

References

BCSH Amendment 2007
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