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CME article

Patients with high haemoglobin and negative for JAK2 mutation

KHBP Fernandopulle[1]

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Diagnosis of JAK2 positive polycythaemia vera (PV) is fairly straightforward as it requires only the dual criteria of a high haematocrit (>0.52 in men, >0.48 in women) or raised red cell mass (>25% above predicted) and a mutation in JAK2 (JAK2 V617F mutation is seen in over 95% and an exon 12 mutation in most remaining).

The difficulties in diagnosis are encountered when patients with high haemoglobin and haematocrits are JAK2 negative.

This CME is designed to refresh your knowledge on investigating patients who are JAK2 negative.

1. Regarding JAK2 negative polycythaemia vera
   a. Requires a haematocrit of >0.52 in men and >0.48 in women.
   b. Splenomegaly is a diagnostic criterion.
   c. Elevated WBC is included as a diagnostic criterion.
   d. Some patients display BCR-ABL1 gene mutation.
   e. Bone marrow shows trilineage expansion of haemopoiesis.
2. Regarding investigating for secondary causes of polycythaemia
   a. Serum erythropoietin levels are high in Chuvash erythrocytosis.
   b. Arterial oxygen saturation of <92% is associated with absolute erythrocytosis.
   c. Normal arterial oxygen saturation results are seen in patients with high affinity haemoglobin.
   d. TET2 gene mutations are seen in patients with congenital erythrocytosis.
   e. Serum calcium levels are increased in parathyroid adenomas.
3. Secondary causes for polycythaemia include,
   a. sleep apnoea
   b. hydronephrosis
   c. papillary carcinomas of the thyroid gland
   d. diuretics
   e. post kidney transplant

Answer Grid

Q1	a	b	c	d	e	Q2	a	b	c	d	e	Q3	a	b	c	d	e
	T	T	T	T	T		T	T	T	T	T		T	T	T	T	T
	F	F	F	F	F		F	F	F	F	F		F	F	F	F	F

The answers and the discussion are given in page 28

[1]*Senior Lecturer, Consultant Haematologist, Department of Pathology, Faculty of Medical Sciences, University of Sri Jayewardenepura, Sri Lanka.*

Correspondence: E-mail: fernandopulle@sjp.ac.lk