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Analysis of the oxygen sensing pathway genes in familial chronic myeloproliferative neoplasms and identification of a novel EGLN1 germ-line mutation

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Ph-negative chronic myeloproliferative neoplasms (MPNs) include polycythaemia vera (PV), essential thrombocythaemia (ET) and primary myelofibrosis (PMF) (Swerdlow et al, 2008). Despite the discovery of increasing numbers of genetic aberrations in MPNs (Oh et al, 2010; Tefferi, 2010), the oncogenic causes of this group of clonal hematologic neoplasms are complex and largely unknown. In addition to JAK2V617F, found in most PV patients and in about half of the patients with ET or PMF (Baxter et al, 2005), JAK2-exon 12 mutations have also been found in patients with isolated erythrocytosis (IE) or in JAK2V617F-negative PV cases (Bernardi et al, 2009). Familial clustering of MPNs supports the evidence that pathological phenotype is driven by yet undefined susceptibility genes (Bellanné-Chantelot et al, 2006; Landgren et al, 2008) and that multiple genetic lesions are involved in the pleiotropy of these neoplasms, including the existence of a pre-JAK2 mutated clone (Jäger & Kralovics, 2010).