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Genetics and Genomics of Erythrocytosis

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Deadline for manuscript submissions:
closed (30 October 2021)

Message from the Guest Editors

Erythrocytosis is a blood disorder characterized by an increased mass of red blood cells. More rarely, erythrocytosis has a known genetic background. Polycythaemia Vera (PV) is caused by somatic mutations, mainly in JAK2, while congenital or familial erythrocytosis (ECYT) is a rare disorder caused by germline mutations in several genes. ECYT1 is associated with EPOR gene variants, ECYT2-5 with defects in the oxygen sensing pathways (variants in VHL, EGLN1, EPAS1, EPO) and ECYT6-8 with increased affinity of haemoglobin for oxygen (variants in HBB, HBA1, HBA2, BPGM). Many patients with idiopathic erythrocytosis remain undiagnosed, indicating that the genes and molecular pathways involved in disease development are not yet fully understood.

This Special Issue will include a selection of original papers and reviews focusing on the genetic background and molecular mechanisms involved in the development of erythrocytosis, including polycythaemia vera and congenital erythrocytosis. Recent advances in genetic and clinical diagnostics will be reviewed. Current and new regulatory networks, epigenetic origins, and pathophysiological mechanisms will be discussed.

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Message from the Editor-in-Chief

Genes are central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fastmoving field. There is a need for good quality, open access journals in this area, and the *Genes* team aims to provide expert manuscript handling, serious peer review, and rapid publication across the whole discipline of genetics. Starting in 2010, the journal is now well established and recognised.

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