EuroSciCon

8th Annual Conference on
Clinical Research & Biomarkers

July 19-20, 2018
Prague, Czech Republic

Biomark J 2018, Volume 4
DOI: 10.21767/2472-1646-C1-003

COMMON ETIOLOGIC PATHWAYS IN PLATELET-MEDIATED ASPIRIN RESPONSIVE ERYTHROMELALGIA AND ARTERIOLAR PLATELET THROMBOPHILIA IN JAK2, MPL OR CALR MUTATED THROMBOCYTHEMIA AND INCURABLE NAV1.7 NEUROPATHIC ERYTHROTHERMALGIA DUE TO GAIN OF FUNCTION MUTATIONS IN THE SCN9A GENE

Jan Jacques Michiels

Goodheart Institute in Nature Medicine, Bloodcoagulation, Phlebology & Vascular Medicine Science Center Rotterdam, Netherlands

The nosologic classification of burning, painful, red congested extremities in the 1990s includes aspirin responsive erythrothermalgia and microvascular disturbances in thrombocythemia versus aspirin resistant primary erythermalgia, an incurable autosomal dominant Nav1.7 neuropathic erythrothermalgia due to gain of function mutations in the SCN9A gene. Both conditions should not be confounded with aspirin resistant erythermalgia secondary to cutaneous vasculitis, systemic lupus erythematosus, rheumatiod arthritis, associated with hypertension or elicited by vasoactive drugs. Michiels between 1975 and 2017 discovered common etiologic pathways in platelet-mediated aspirin responsive erythromelalgia in JAK2-thombocythemia in patients with essential thrombocythemia (ET) and polycythemia vera (PV) and incurable neuropathic inherited erythrothermalgia in neuropathic Nav1.7 chanellopathy of dorsal root ganglia. The two disorders are defects in the thermoregulatory and pain-sensing nociceptive afferent sensory C-fibers neurons and blood flow regulating system in the arteriole-capillary arterio-veneus (AV) shunt. Aspirin responsive von Willebrand factor-platelet-mediated erythromelalgic arteriolar inflammation and thrombosis followed by microvascular ocular, cerebral and coronary thrombosis in thrombocythemia (EMT) patients are caused by prostaglandin endoperoxides released from spontaneous activation, release reaction and aggregagation of hypersensitive constitutively activated platelets due to a gain of function mutations in the JAK2, TPO, MPL and CALR genes (Sticky Platelet Syndrome). Incurable inherited erythrothermalgia (IE) and Paroxsysmal Erythromelalgic Extreme Pain Disorders (PEPD) are incurable dominant congenital neuropathic pain conditions caused by gain of function mutations in the SCN9A gene as the cause of Nav1.7 sodium channel protein in the dorsal root ganglion (DRG) nociceptive neurons of afferent C-fibers in the subcutaneous arteriole-capillary arterio-veneus (AV) shunt. Targeted curative management of JAK2-thrombocythemia in ET and PV patients with aspirin responsive erythromelalgia and its microvascular and major thrombotic complications caused by a gain of function mutation in JAK2, MPL, CAL or TPO gene is feasible when adequately diagnosed in time. Inherited erythromelalgia (IEM) is a congenital dominant neuropathic Nav1.7 channelopathy caused by a gain of function mutation in the SCN9A gene, and remains incurable and a great suffer for affected patients recently labeled as the Man on Fire Syndrome in medicine anno 2016, that cries for targeted initiatives to develop novel effective analgesics by pharmaceutical companies.

goodheartcenter@outlook.com