9º Simpósio Médico Internacional de VHL
9th International Medical Symposium on VHL

III Encontro de Famílias com a Síndrome de VHL
3rd VHL Family Meeting

Rio de Janeiro • October 2010

ABSTRACT

Role of Pregnancy on Hemangioblastomas in von Hippel-Lindau Disease: A Retrospective French Study

Caroline Abadie, Isabelle Coupier, Sophie Bringuier-Branchereau, Grégoire Mercier, Sophie Deveaux, Stéphane Richard

French National Cancer Institute Network, Centre de Référence Cancers Rares INC, Service d'Urologie, Hôpital de Bicêtre, AP-HP, Le Kremlin-Bicêtre, France

Von Hippel-Lindau disease (VHL) is a dominantly inherited disorder predisposing to highly vascularized tumors including hemangioblastomas of the central nervous system (CNS) and the retina. The disease results from germ-line mutations in the VHL tumour-suppressor gene that plays a key role in the cellular response to tissue hypoxia and angiogenesis. CNS and retinal hemangioblastomas occur in about 75% and 60% patients, respectively. Only a few case reports and one study on a small population of VHL patients (Grimbert et al., Am. J. Gyn. Obst. 1999,180:110-1) were previously interested in a potential deleterious role of pregnancy on hemangioblastomas which express progesterone receptors.

We present a retrospective and comparative French study in 269 women from 172 families coming from the national VHL clinical database. The aim was to analyse the onset of new hemangioblastomas and potential tumoral complications in patients according to their gestational status. Available data of imaging follow-up of CNS and retina were collected in 176 women with at least one pregnancy (group 1) and 93 women with none (group 2). More complications of hemangioblastomas in group 1 (p=0.031) were noted. This appeared not to be correlated with cerebrospinal or retinal topography. To our knowledge, this work represents the first study analysing the effect of pregnancy in a very large series of women with VHL. This study underlies the necessity of follow-up patients closely during pregnancy. Thus, a magnetic resonance imaging without injection is systematically required during the fourth month of pregnancy in each patient with previously known CNS lesions.