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ABSTRACT

VHL in Brazil: Genetics, Biobanking and VHL Family Care

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The von Hippel-Lindau disease is a genetic disorder caused by germline mutation in the VHL gene. Although the clinical manifestations are limited to a particular number of organs and systems, including hemangioblastomas in the CNS and retina, pheochromocytomas and paragagliomas, clear cell renal cell carcinomas (ccRCC), pancreatic NETs, ELSTs, among others, the variety of manifestations vary not only between families, but among members in a family. The first study of VHL in Brazil was published by our group in 2003 presenting the germline VHL mutations of the first 20 families studied. In order to characterize the spectrum of manifestations of VHL patients of Brazilian families, give them appropriated support, and free access to genetic testing, this study was extended at the Brazilian National Cancer Institute (INCA). In 2009 the Brazilian Alliance on VHL (ABVHL) was founded. So far, around 50 VHL families have been identified, and 44 of them have their germline VHL mutation characterized. Interesting, the germline mutation Phe76del was identified in three distinct VHL families in Brazil. A network of reference medical specialists and medical centers has been developed, and genetic services around the country have started seeing VHL patients and families. Orientation and second opinion are remotely accessible to the public, doctors, and researchers. In 2007, a hereditary tumor bank was created with the collaboration of VHL patients and their doctors providing high quality tissue to the ongoing genetics studies of our and other labs.