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ABSTRACT

Neoplastic Diagnosis Timing Profile in von Hippel-Lindau´s Patients in a Personal Series

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Introduction: von Hipple-Lindau patients present nervous system and retinal hemangioblastomas (HGB) associated with paragangliomas/pheocromocytomas (PGL), endolymphatic sac tumors (ELST), renal cell carcinoma (RCC) and pancreatic neuroendocrine tumors (PNT). The object of this study is to evaluate the presenting timing profile of these tumors.

Material and Method: Age and sequence of imaging confirmed diagnosis of every new tumor were evaluated in a series of 62 patients with a total of 291 diagnosed tumors from 30 families, studied and followed in a neuro-oncological syndrome center.

Results: Among HGBs, Retinal were precocious (initial diagnosis at age 8, median 29); Cerebellar follows (first diagnosed at 13, median 35); Then, Brainstem follows (begins at 8, median 41), and Spinal have a later diagnosis (beginning at age of 9, median 42). PGLs begin at age 11, with median 34. ELSTs begin at 17 years, with median 34. PNTs diagnosis starts at 14, median 42, and RCCs were diagnosed the latest, starting at 20 years with median at 45. No relation has been observed among age of presentation and other clinical or molecular characteristics.

Conclusions: In von Hippel-Lindau´s disease, the neoplastic occurrence begins at early age, 10% of cases before age of 19. A precocious first diagnosis doesn´t predict an aggressive clinical course for subsequent tumors. The temporal profile is not predictable by molecular diagnosis. Molecular diagnosis and clinical and imaging studies should be performed since pediatric age, in order to obtain an early diagnosis and adequate management of these neoplasms.