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

Endolymphatic Sac Tumors (ELST) in VHL Patients – Evaluation of Screening Methods in a National Study

Louise Mølgaard Poulsen, Steen Gimsing, Michael Kosteljanetz, Carsten Alfred Brandt, Hans Ulrik Møller, Carsten Thomsen, Marie Luise Bisgaard
Department of Cellular and Molecular Medicine, University of Copenhagen, Denmark

Introduction: ELSTs are found in 11-16% of vHL patients, but it is uncertain whether recommended audiologic screening is adequate for tumour detection. The study aims at evaluating ELST screening strategies and patterns in audiometric parameters in VHL patients.

Material and Methods: We included 40 out of 42 VHL mutation carriers over 15 years of age known in Denmark. Subjects were interviewed about ELST-related symptoms and referred to audiologic examination and inner ear MRI. Audiological work-up comprised otoscopy, pure tone and speech audiometry, tympanometry, determination of stapedial reflex thresholds, and in some cases auditory brainstem response and vestibular testing. MRIs were carried out by a 3T MR scanner with high resolution imaging. 3D T2 weighted TSE images were acquired of the 8. cranial nerve, inner ear and endolymphatic sac with 0.6 mm isotropic resolution. Results were blinded and evaluated independently by two Ear-Nose- Throat- and Radiology specialists. Median age at examination was 39 years (range: 15-65).

Results: So far, prevalence of MRI-visible ELST among VHL mutation carriers is 5% (2/39). One ELST patient had subjective and objective audio-vestibular symptoms prior to MRI. The second ELST patient, however, had no symptoms and unremarkable audiometric findings. Assuming MRI findings as a measure of true ELST occurrence, sensitivity of audiometry in ELST diagnosis was 50% and specificity 19%. Conclusion: The study is ongoing and results including all subjects will be presented. Preliminary results indicate that MRI is superior to other methods of ELST detection. hemangioblastoma.