# Chromosome 3

- A novel pathogenic germline chromosome 3 inversion in von Hippel-Lindau disease
- Radiation-Induced DNA Damage in Uveal Melanoma Is Influenced by Dose Delivery and Chromosome 3 Status
- SNP array genomic analysis of matched pairs of brain and liver metastases in primary colorectal cancer
- VHL syndrome without clear family history: A rare case report and literature review of Chinese patients
- Common Variants Near ZIC1 and ZIC4 in Autopsy-Confirmed Multiple System Atrophy
- Infundibular hemangioblastoma resection: Video case report
- Adapted whole-body surveillance for von Hippel-Lindau-associated tumors in 3p deletion syndrome with VHL deletion: A case report
- Epigenome-wide data collection in a case of gliofibroma

Chromosome 3 likely contains 1,000 to 1,100 genes that provide instructions for making proteins. These proteins perform a variety of different roles in the body.

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Although inactivation of the von Hippel-Lindau gene (VHL) on chromosome 3p25 is considered to be the major cause of hereditary endolymphatic sac tumors (ELSTs), the genetic background of sporadic ELST is largely unknown. The aim of a study was to determine the prevalence of VHL mutations in sporadic ELSTs and compare their characteristics to VHL-disease-related tumors.

Genetic and epigenetic alterations were compared between 11 sporadic and 11 VHL-disease-related ELSTs by targeted sequencing and DNA methylation analysis.

VHL mutations and small deletions detected by targeted deep sequencing were identified in 9/11 sporadic ELSTs (82%). No other cancer-related genetic pathway was altered except for TERT promoter mutations in two sporadic and one VHL-disease-related ELSTs (15%). Loss of heterozygosity of chromosome 3 was found in 6/10 (60%) VHL-disease-related and 10/11 (91%) sporadic ELSTs resulting in biallelic VHL inactivation in 8/10 (73%) sporadic ELSTs. DNA methylation profiling did not reveal differences between sporadic and VHL-disease-related ELSTs, but reliably distinguished ELST from morphological mimics of the cerebellopontine angle. VHL patients were significantly younger at disease onset compared to sporadic ELSTs (29 vs. 52 years, $p < 0.0001$, Fisher's exact test). VHL-disease status was not associated with an increased risk of recurrence, but the presence of clear cells was found to be associated with shorter progression-free survival ($p = 0.0002$, log-rank test).

Biallelic inactivation of VHL is the main mechanism underlying ELSTs, but unknown mechanisms beyond VHL may rarely be involved in the pathogenesis of sporadic ELSTs [1].

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Schweizer L, Thierfelder F, Thomas C, Soschinski P, Kim HY, Jödicke R, Woltering N, Förster A, Teichmann D, Siewert C, Klein K, Schmid S, Nunninger M, Thomale UW, Onken J, Mühleisen H, Schittenhelm J, Tatagiba M, von Deimling A, Reuss DE, Solomon DA, Heppner FL, Koch A, Hartmann C, Staszewski O, Capper D. Molecular characterisation of sporadic endolymphatic sac tumours and comparison to von Hippel-Lindau disease-related tumours. Neuropathol Appl Neurobiol. 2021 Jun 5. doi: 10.1111/nan.12741. Epub ahead of print. PMID: 34091929.