3p deletion syndrome

Morisawa K, Sato T, Shimoyamada M, Mizuno R, Ohashi H, Watanabe-Hisazumi H, Takeshima K, Kojima A, Sato S, Hasegawa T, Takahashi T. Adapted whole-body surveillance for von Hippel-Lindau-associated tumors in 3p deletion syndrome with VHL deletion: A case report. Pediatr Blood Cancer. 2022 Apr 19:e29732. doi: 10.1002/pbc.29732. Epub ahead of print. PMID: 35441425.

3p deletion syndrome is a condition that results from a chromosomal change in which a small piece of chromosome 3 is deleted in each cell. The deletion occurs at the end of the short (p) arm of the chromosome. This chromosomal change often leads to intellectual disability, developmental delay, and abnormal physical features.

Individuals with 3p deletion syndrome typically have severe to profound intellectual disabilities. Most have delayed development of language skills as well as motor skills such as crawling and walking. While affected individuals learn to walk in childhood, their language ability usually remains limited. Some individuals with 3p deletion syndrome have obsessive-compulsive disorder (OCD) or features of autism spectrum disorders, which are conditions characterized by impaired communication and social interaction.

The VHL gene and tumor suppressor genes for renal cell carcinoma (RCC) have been mapped to the short arm of chromosome 3, although the genes have not yet been identified. An RCC cell line, KC12, was established from a VHL patient. Molecular genetic analyses in conjunction with cytogenetic studies revealed that the short arm of chromosome 3 distal to the D3S4 locus at 3p11 was lost in the RCC cell line as a result of an unbalanced translocation between chromosomes 3p and 5q. Structural and numerical aberrations, including those on chromosome 3p, were not detected in T-lymphocytes from the patient, suggesting that the inherited mutation of the VHL gene at 3p25-26 in this patient was too subtle to be detected by either Southern blot or karyotype analysis. Since no permanent RCC cell line has been established from a VHL patient, this cell line will be a useful source for analyzing the VHL gene at 3p25-26 and tumor suppressor gene(s) at 3p13-21 [1)].

Case reports

a 24-year-old woman with 3p deletion syndrome associated with cerebellar hemangioblastoma at the age of 16 years old. She presented dysmorphic facial features, growth retardation and severe psychomotor retardation associated with 3p deletion syndrome. We identified de novo 3p deletion encompassing p25 by using array-based comparative genomic hybridization, where causative gene of von Hippel-Lindau (VHL) disease located. Surgical therapy for cerebellar hemangioblastoma was performed, and histological examination was consistent in cerebellar hemangioblastoma. She showed no other tumors associated VHL disease till 24 years old. This is the first case report of a patient with 3p deletion syndrome whose cerebellar hemangioblastoma may be associated with VHL disease. Repeat imaging studies were recommended for the patients with 3p deletion syndrome [2)].

1)

Kohno T, Sekine T, Tobisu K, Oshimura M, Yokota J. Chromosome 3p deletion in a renal cell carcinoma

cell line established from a patient with von Hippel-Lindau disease. Jpn J Clin Oncol. 1993 Aug;23(4):226-31. PMID: 8105119.

2)

Suzuki-Muromoto S, Hino-Fukuyo N, Haginoya K, Kikuchi A, Sato H, Sato Y, Nakayama T, Kubota Y, Kakisaka Y, Uematsu M, Kumabe T, Kure S. A case of 3p deletion syndrome associated with cerebellar hemangioblastoma. Brain Dev. 2016 Feb;38(2):257-60. doi: 10.1016/j.braindev.2015.07.005. Epub 2015 Sep 11. PMID: 26365017.