LZTR1

The LZTR1 gene provides instructions for making a protein whose exact function is unknown. The LZTR1 protein is made in cells throughout the body. Within cells, it is found in the Golgi apparatus, which is a structure in which newly produced proteins are modified. Studies suggest that the LZTR1 protein may help stabilize this structure. Researchers suspect that this protein may also be associated with the CUL3 ubiquitin ligase complex, which is part of the cell machinery that breaks down (degrades) unneeded proteins.

Based on its role in several tumor types, the LZTR1 protein is thought to act as a tumor suppressor.

Schwannomatosis and neurofibromatosis type 2 (NF2) are both characterized by the development of multiple schwannomas but represent different genetic entities. Whereas NF2 is caused by mutations of the NF2 gene, schwannomatosis is associated with germline mutations of SMARCB1 or LZTR1.

Kehrer-Sawatzki et al., studied 15 sporadic patients with multiple non-intradermal schwannomas, but lacking vestibular schwannomas and ophthalmological abnormalities, who fulfilled the clinical diagnostic criteria for schwannomatosis. None of them harboured germline NF2 or SMARCB1 mutations as determined by the analysis of blood samples but seven had germline LZTR1 variants predicted to be pathogenic. At least two independent schwannomas from each patient were subjected to NF2 mutation testing. In five of the 15 patients, identical somatic NF2 mutations were identified (33%). If only those patients without germline LZTR1 variants are considered (n = 8), three of them (37.5%) had mosaic NF2 as concluded from identical NF2 mutations identified in independent schwannomas from the same patient. These findings imply that a sizeable proportion of patients who fulfil the diagnostic criteria for schwannomatosis, are actually examples of mosaic NF2. Hence, the molecular characterization of tumours in patients with a clinical diagnosis of schwannomatosis is very important. Remarkably, two of the patients with germline LZTR1 variants also had identical NF2 mutations in independent schwannomas from each patient which renders differential diagnosis of LZTR1-associated schwannomatosis versus mosaic NF2 in these patients very difficult [1].

1)
Kehrer-Sawatzki H, Kluwe L, Friedrich RE, Summerer A, Schäfer E, Wahlländer U, Matthies C, Gugel I, Farschtschi S, Hagel C, Cooper DN, Mautner VF. Phenotypic and genotypic overlap between mosaic NF2 and schwannomatosis in patients with multiple non-intradermal schwannomas. Hum Genet. 2018 Jul 13. doi: 10.1007/s00439-018-1909-9. [Epub ahead of print] PubMed PMID: 30006736.

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