CHEK2

Neurofibromatosis, including type 1 and type 2, is inherited dominant disease that causes serious consequences. The genetic mechanism of these diseases has been described, but germline mutation of checkpoint 2 kinase gene, together with other DNA repair related genes, has not been fully elucidated in the context of neurofibromatosis.

Li et al., reported identical germline mutation of CHEK2 gene (p.R180C) in a 7-year-old Tibetan boy with NF1, and in a 12-year-old Chinese girl with NF2.

Both patients underwent operation to obtain tumor tissue, and peripheral blood of their family was tested.

Identical germline mutation of CHEK2 gene (p.R180C) was detected in both patients, and germline mutations of POLE, MUTYH and ATR were also detected.

This is the first article to describe CHEK2 mutation in both NF1 and NF2. This article highlights a possible role of CHEK2, in association with other germline genetic mutations, in tumorigenesis of NF1 and NF2 [1)].

1)
Li Q, Zhao F, Ju Y. Germline mutation of CHEK2 in neurofibromatosis 1 and 2: Two case reports. Medicine (Baltimore). 2018 Jun;97(23):e10894. doi: 10.1097/MD.0000000000010894. PubMed PMID: 29879026.