Abstract

Background: Familial cancer syndrome is a group of hereditary disorders in which a person is exposed to certain types of cancer much more than the general population.

Aim: This study was conducted with the aim of investigating the genetic cause of breast, thyroid, stomach and leukemia cancers in a super family living in Ardabil.

Materials and Methods: The statistical population of the current study included 28 members of a superfamily living in Ardabil, who had a history of cancer in the family, and all members were included in the study by census method. After checkiing the clinical records of the family members, genomic examination of the members was done by WES method. Based on this, NTHL1 gene mutations were investigated by Snger based PCR-sequencing method and finally segregation analysis was performed in healthy and sick family members.

Results: Of the 28 people studied, 11 (39.3%) were men and 17 (60.7%) were women. The minimum age was 13 years and the maximum age was 76 years. Clinical history was reported in 9 (32.1%) of the studied subjects. NTHL1 mutation was observed in (50%) of 14 subjects. Mutation was reported in 7 (50%) people with clinical history and in 7 (50%) people without clinical history. There was a statistically significant difference in mutation in women with clinical history and without clinical history ($p = 0.015$) and NTHL1 mutation was more in women with clinical history.

Conclusion: In general, our study shows that NTHL1 mutation is more common in women who have a clinical history.

Keywords: NTHL1 - cancer – pedigree