GENETICS AND GENODERMATOSES

COMPARISON OF GENETIC MUTATIONS BETWEEN SQUAMOUS CELL CARCINOMA AND ACTINIC KERATOSIS ON THE RIGHT CHEEK IN A SAME PATIENT USING WHOLE GENOME SEQUENCING

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Background: Actinic keratosis (AK) is considered as a strong independent risk factor for squamous cell carcinoma (SCC) development. However, the difference of somatic mutation and copy number alteration between AK and SCC has not been fully understood

Objectives: To find out the differences of genomic aberrations between AK and SCC and possible genetic mutations for progression from AK to SCC in a same cancerization field.

Methods: Frozen AK and SCC samples on the right cheek and paired blood sample were obtained from a 90- year-old female Korean patient by total excision. The genetic differences were evaluated by whole genome sequencing (WGS) analysis.

Results: The somatic mutation burden was lower in AK than in SCC. In AK, eight mutations in five genes (CACNA1D, LRP1B, NOTCH1, FAT4, and PTPRT) overlapped with the top 20 mutations of SCC defined by the COSMIC database, while, in SCC, ten mutations in nine genes (AFF3, PREX2, ROS1, TP53, GRIN2A, KMT2D, NOTCH1, TERT, PDE44DIP) did with those mutations. Copy number alterations of oncogenes and tumor suppressor genes are only observed in SCC, not in AK.

Conclusion: WGS study showed no common genetic mutation of oncogenes or tumor suppressor genes between AK and SCC in the same patient. Although in a same cancerization field, AK and SCC showed different genetic mutation profiles. Further studies to find out the pathway of progression from AK to SCC are needed.