SKIN CANCER (OTHER THAN MELANOMA)

HYDROA VACCINFORME-LIKE LYMPHOPROLIFERATIVE DISORDER: A STUDY OF CLINICOPATHOLOGY AND THE WHOLE EXOME SEQUENCING

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Introduction: Hydroa vaccinforme-like lymphoproliferative disorder (HVLPD) has recently been recognized as a chronic EBV+ lymphoproliferative disorders. At present, the etiology and pathogenesis of HVLPD are still unclear.

Objective: In order to further understand HVLPD in the clinicopathology and to understand the mutations of the exogenous gene in the whole genome and the enrichment of gene mutations on pathways.

Materials and Methods: we performed this retrospective study of 19 cases of HVLPD in West China Hospital (WCH), Sichuan University. Immunohistochemical staining, in situ hybridization for EBER1/2, T-cell receptor gene rearrangement and WES technology were performed. All the cases were followed-up and the prognosis related factors were analyzed.

Results: After verification, five driver genes STAT3, IKBKB, ELF3, CHD7, KMT2D and three mutation genes ELK1, RARB and HPGDS were screened.

Conclusions: 1 HVLPD is a group of cutaneous lymphoid proliferative diseases with unique clinicopathological features. The facial swelling, blisters, vacciniform scars, fever and waxing and waning clinical process have important implications for the diagnosis of the disease. The prognosis of this disease is different case by case. The age of onset is more than 15 years old, the first site is facial, and the increase of HBDH and LDH in peripheral blood may be related to poor prognosis. 2 Due to the HVLPD infiltrating lymphoid cells are often without atypia and mitosis happens occasionally, it is easily misdiagnosed as an inflammatory disease. They can be identified by clinical manifestations, tumor cell immersion, peripheral blood EBV detection, immunohistochemical phenotype, EBER1/2 in situ hybridization and TCR gene rearrangement tests. 3 The known driver genes STAT3, IKBKB, ELF3, CHD7, KMT2D, the mutation genes ELK1, RARB and HPGDS, and their enriched pathways may be related to the development of HVLPD, and even may be the driver genes of the disease.