TRRAP AND GRIN2A MUTATIONS FOR THE DIAGNOSIS AND TREATMENT OF MELANOMA

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Technology description

Melanoma is the most deadly form of skin cancer. Despite years of research, metastatic melanoma has poor prognosis and is often fatal. Because of few therapeutic options, there is a great need for new clinically relevant targets. Using whole-exome sequencing of matched normal and metastatic tumor DNA, researchers at NHGRI have identified several novel, somatic and tumor-specific alterations. In particular, the investigators found a recurrent "hotspot" mutation in the transformation/transcription domain-associated protein (TRRAP, an adaptor protein found in multiprotein chromatin complexes) gene and found the glutamate receptor ionotropic N-methyl D-aspartate 2A (GRIN2A, a subunit of glutamate-gated ion channel) gene to be highly mutated.
These results enhance the understanding of the biology of melanoma and hopefully can lead to improved patient care. Because glutamate pathway antagonists have been previously shown to limit tumor growth, further investigation into this pathway, as well as development of such inhibitors is warranted.

Application area

These mutations can be used in diagnosing patients as having melanoma or being susceptible to developing the cancer, as well as predicting the prognosis of a diagnosed person. The findings can also aid in selecting personalized therapy for a subject diagnosed with melanoma.

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