HUMAN MELANOMA CELL LINES HARBORING MUTATIONS IN GRM3, GRIN2A, TRRAP, PLCB4, OR MITF

Published date: Feb. 13, 2012

Technology description

Melanoma cell lines with defined mutations for development of cancer therapeutics.
Melanoma is an aggressive malignancy of the pigment-producing cells, melanocytes, and accounts for the majority of deaths resulting from skin cancer. In order to develop personalized treatments for advanced disease, it is important to identify genetic alterations leading to melanoma. Through the use of exon capture, massively parallel sequencing, as well as whole-exome sequencing, NHGRI's investigators have identified specific genes mutations associated with melanoma, namely glutamate receptor metabotropic 3 (GRM3, a member of the G-protein coupled receptor family), glutamate receptor ionotropic N-methyl D-aspartate (NMDA) 2A (GRIN2A, a subunit of a glutamate-gated ion channel implicated in neurological disorders), transformation/transcription domain-associated protein (TRRAP, adapter protein that is part of multiprotein chromatin complexes), phospholipase C beta 4 (PLCB4, which catalyzes formation of second messenger molecules), and microphthalmia-associated transcription factor (MITF, important,inter alia, in development and differentiation of melanocytes). Dr. Samuels also generated human melanoma cell lines harboring mutations in GRM3, GRIN2A, TRRAP, PLCB4, or MITF (some lines with mutations in a single gene and others with mutations in multiple genes, including protein tyrosine kinase ERBB4).

Application area

The discovery of these mutations, generation of the cell lines, and the investigation of the related molecular pathways could be used in the development of potential therapeutics to limit tumor growth and improve individualized patient care. These cell lines can also be utilized to further understand the involvement of the identified genes in melanoma progression.

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