National Human Genome Research Institute (NHGRI)

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Human Melanoma Metastasis Cell Line with Isocitrate Dehydrogenase 1 (IDH1) R132 Mutation

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Key Words

Melanoma, Glioma, AML, Isocitrate Dehydrogenase 1, Cell Lines, Small Molecule Inhibitors

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Summary

Isocitrate dehydrogenase 1 (IDH1), which during the citric acid cycle converts isocitrate to α-ketoglutarate while reducing nicotinamide adenine dinucleotide phosphate (NADP+) to NADPH, was previously identified as mutated in a large percentage of progressive gliomas and acute myeloid leukemias (AML). These mutations are found at the R132 residue. Cancers associated with the IDH1 mutation result in the accumulation of 2-hydroxyglutarate, which can serve as a diagnostic and prognostic marker. In order to further understand the biology of the IDH1 and identify specific inhibitors, a cell line harboring the mutation would be extremely beneficial. NHGRI researchers developed a human melanoma metastasis cell line harboring the R132C mutation by using low passage cell lines derived from a panel of pathology-confirmed metastatic melanoma tumor resections, paired with apheresis-collected peripheral blood mononuclear cells.

Potential Commercial Applications

This particular cell line could be used to further decipher the biology of IDH1 (using both *in vitro* and *in vivo* techniques), including its role in cell growth, motility, invasion, and metabolite production. The line could also assist with high throughput drug screening to identify inhibitors of IDH1, and thus potential therapeutics for cancers, such as glioma, AML, and melanoma.

Related Article

Lopez et al., *IDH1^{R132} Mutation Identified in One Human Melanoma Metastasis, but Not Correlated with Metastases of the Brain*, 398 Biochemical and Biophysical Research Communications 585 (2010).
http://www.sciencedirect.com/science/article/pii/S0006291X10012829