

A clinical trial of a promising new drug for melanoma [reported in this week's \*New England Journal of Medicine\*](#)

highlights the emerging importance of using genetic mutation diagnosis to determine what drug treatments might be effective in specific patients. The

*NEJM*

study focused on melanoma patients with a specific type of mutation, called V600E, in the gene BRAF; this mutation is seen in over half of all melanoma patients. Patients with the mutation showed promising responses to the Plexikon-developed drug PLX4032. Response was marked by an increased window of months before the melanoma progressed again. The study is in very early stages, and there remains a risk that patients will as often happens build up resistance to the drug. However this opens the door to the growing trend in cancer of using drug combinations - perhaps combining PLX4032 with another drug that targets a different mutation.

The use of genetic mutations to select drug therapies is a growing field that could have huge value especially in aggressive cancers like melanoma where there may be a limited window in which the right drug can be selected for a patient. Also, this could potentially reduce the side effects of drugs, since the drug is selectively targeting the cells with the gene mutation, rather than healthy cells. Melanoma is a malignant tumor that arises from melanocytes (pigment) cells in the skin. It is an aggressive cancer affecting 60,000 American a year, and accounts for three-quarters of all skin cancer related deaths. Like many other cancers, the cell signaling pathways that are disrupted in melanoma and lead to tumor formation are common to those cell signaling pathways disrupted in neurofibromatosis - the drug targets including BRAF are on the 'Ras pathway'. It is important to monitor progress and approaches in these other conditions to see what we can learn about candidate future treatments for neurofibromatosis. For neurofibromatosis as we advance in identifying candidate drugs, the goal is to develop enough knowledge about genetic mutations to use this information to select the most appropriate drugs.