People missing Alzheimer’s gene provide clue to disease

The 40-year-old man showed up in Dr. Mary Malloy’s clinic with sadly disfiguring symptoms.

His hands, elbows, ears and feet were blemished with protruding pustules and tuber-like welts, some so painful it was hard for him to walk. He suffered from a rare genetic condition called dysbetalipoproteinemia, which caused his cholesterol levels to soar so high that pools of fatty tissue seemed to bubble up under his skin.

But there was something else about this patient. He was missing a gene that, when present in one form, greatly increases the risk of developing Alzheimer’s disease.

Malloy, who co-directs the Adult Lipid Clinic at the University of California, San Francisco, and her colleagues saw an opportunity to answer an important neurological riddle: Does the absence of the gene — named apolipoprotein E, or APOE, after the protein it encodes — hurt the brain?

If a person with this rare condition were found to be functioning normally, that would suggest support for a new direction in Alzheimer’s treatment.

It would mean that efforts — already being explored by dementia experts — to prevent Alzheimer’s by reducing, eliminating or neutralizing the effects of the most dangerous version of APOE might succeed without causing other problems in the brain.

Read the full, original story: Tactic in Alzheimer’s fight may be safe, study finds