Beneficial ‘natural’ gene mutation could be effective in treating sickle cell

Sickle cell disease (SCD) patients have a genetic defect that leads them to produce hemoglobin S, or sickle hemoglobin, instead of normal hemoglobin. This impairs the ability of red blood cells to bind to oxygen molecules in the lungs, and consequently to deliver oxygen to tissues throughout the body.

…

A benign genetic mutation in the fetal globin gene is called the British-198 mutation, because it was first discovered in a British family in 1974. Fetal hemoglobin has high affinity to oxygen, and commonly shuts down after birth. However, those carrying the “naturally occurring” British-198 gene variant had high levels of fetal hemoglobin in adulthood, with levels reaching up to 20% expression compared to the 1% found in most children and adults.

…

Seeing therapeutic potential, researchers […] tested if they could introduce the British-198 mutation into human cells to induce fetal hemoglobin production.

…

“Our laboratory has shown that introducing the beneficial mutation British-198 into blood cells using this technology substantially boosts their production of fetal hemoglobin,” [said Merlin Crossley, the study’s senior author.] “Because this mutation already exists in nature and is benign, this ‘organic gene therapy’ approach should be effective and safe to use to treat, and possibly cure, serious blood disorders.”

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: Natural’ Gene Mutation May Offer Way of Treating Sickle Cell Disease, Study Says