Name Period Date

Proteus Syndrome



In conjunction with neurofibromatosis type I, this is the disease that likely afflicted Joseph Merrick, the socalled Elephant Man. It’s a condition in which bones, skin, and other tissues are overgrown. Individuals typically have organs and tissues that grow out of proportion with the rest of their body, and because the overgrowth varies and exhibits no apparent order, it can result in strange and imbalanced features. Signs of the disorder don’t usually appear until about 6 to 18 months after birth. The severity of proteus syndrome varies from individual to individual, and it occurs in less than one in one million people. And in fact, only a few hundred documented cases have ever been reported. The disorder results from a mutation in the AKT1 gene (which regulates cell growth), causing mosaicism; as cells grow and divide, some cells exhibit the mutation while others do not. The resulting mixture of normal and abnormal cells is what causes the overgrowth.

Write a 3 sentence summary of this article: