# 1 in 5 healthy adults may carry rare genetic disease mutations

A new pair of studies is the first ever randomized clinical trials evaluating whole genome sequencing in healthy people. They suggest that sequencing the genomes of otherwise healthy adults can, for about one in five people, turn up risk markers for rare diseases or genetic mutations associated with cancers.

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The first genome screening study looked at 100 healthy adults who initially reported their family history to their own primary care physician. Then half were randomly assigned to undergo an additional full genomic workup, which cost about $5,000 each and examined some 5 million subtle DNA sequence changes, known as single-nucleotide variants, across 4600 genes.

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Of the 50 participants whose genomes were sequenced, 11 had alterations in at least one letter of DNA suspected to cause—usually rare—diseases, researchers [reported] in The Annals of Internal Medicine. But only two exhibited clear symptoms.

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Another paper published…on the preprint server bioRxiv, which has not yet undergone peer review, yields similar results…Michael Snyder, director of the Stanford Center for Genomics and Personalized Medicine in Palo Alto, California, and colleagues found that 12 out of 70 healthy adults, or 17%, unknowingly had one or more DNA mutations that increased the risk for genetic diseases.

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Both studies suggest that physicians should look at genes beyond the ACMG's 59 top priorities, Snyder says. He argues that whole-genome sequencing should be "automatically" incorporated into primary care. "You may have some super-worriers, but I would argue that the information is still useful for a physician to have."

**The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: One in five 'healthy' adults may carry disease-related genetic mutations**