STAT morning rounds
REPORTING FROM THE FRONTIERS OF HEALTH AND MEDICINE

By Megan Thielking

Lab Chat: Scientists scour the genomes of healthy people

Scientists scoured the DNA of healthy babies and adults in two genome-sequencing projects called MedSeq and BabySeq — and have turned up some surprising results. Here's what Dr. Robert Green of Brigham and Women's Hospital told me about the new findings, which are being presented at a conference this week.

What did the studies look at?

They are randomized clinical trials of genome sequencing in healthy adults and newborns. We concentrated on monogenic disease, which are due to a single gene, both dominant conditions like BRCA and recessive conditions like cystic fibrosis. We looked at close to 5,000 genes and found that a remarkably high percentage of ostensibly healthy people carry risk markers for these rare conditions.

What's the takeaway from that?

Remember that these are healthy people. They have no symptoms and no complaints. The fact that they have a mutation doesn't mean they're going to get a disease. ... We also went back and found that about one-quarter of those with a risk marker had features of the disease that hadn't been previously noticed or had not been previously imagined to be connected to a genetic condition.