Next-generation genome sequencing pushes clinical research

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When people talk about the $1,000 genome, they are not speaking about the whole genome, but the exons, the so-called coding regions of the genome. “Six years ago, I was spending $15,000 per exome sequence,” says Gholson Lyon, M.D., Ph.D., a genomic scientist working for the Cold Spring Harbor Laboratory. “Now that costs about $700.”

Whole genome sequencing is more expensive. “We are still not at the $1,000 genome in my opinion,” Dr. Lyon continues. “Almost everyone I’ve talked to is charging $1,500–2,000, and we pay $3,000 because that gets us 60× coverage of the genome, which we have shown is very important to recover small insertions and deletions in the genome ranging in size from 5 to 50 base pairs.”

Dr. Lyon, who studies rare but heritable medical diseases such as Ogden syndrome and TAF1 syndrome, believes that advances in next-generation sequencing technology—better software algorithms, improved methodologies, and lower costs—accelerate his work and the work of others conducting clinical research.

The standard advocated by Illumina, the industry giant, and other sequencing companies is a 30× genome, which means sequencing the genome enough to generate on average 30 reads aligned at each base pair. But according to Dr. Lyon, the 30× genome does not capture all the insertions and deletions.

Read full, original post: Pure Innovation Drives Clinical-Grade NGS