

GENETICS

# 1000 GENOMES QUICK BROWSER

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We did not anticipate that the first 1,000 human genomes could be such quickly browsed ;-) but this is what the [project website](#) says. By today 4 individuals are on display and I wonder how it will look like with 996 more lines??

The screenshot shows the 'Region in detail' view of the 1000 Genomes Quick Browser. At the top, there's a navigation bar with 'Region in detail' and 'Reseq'. Below this, a location bar shows 'Location: 8 : 62107544 - 62112543' with a 'Go>' button. The main content area displays 'THIS STYLE: Location of SNPs' and 'THIS STYLE: Resequencing coverage'. It notes that there is no resequencing coverage at this position. Below this, it shows the 'Homo\_sapiens' entry for 'chromosome:NCBI36:8:62107544:62112543:1'. The sequence data is presented in two blocks. The first block shows the reference sequence (REF: 36) and the first four individuals (LKG\_NA12878\_pilot2, LKG\_NA12891\_pilot2, LKG\_NA12892\_pilot2, LKG\_NA19240\_pilot2) with their respective coverage (1). The second block shows the reference sequence (REF: 36) and the same four individuals with their respective coverage (121).

Individual	Coverage
REF: 36	1
LKG_NA12878_pilot2	1
LKG_NA12891_pilot2	1
LKG_NA12892_pilot2	1
LKG_NA19240_pilot2	1


  

Individual	Coverage
REF: 36	121
LKG_NA12878_pilot2	121
LKG_NA12891_pilot2	121
LKG_NA12892_pilot2	121
LKG_NA19240_pilot2	121

Nature has asked some people [about their forecast of personalized medicine 2020](#) – no idea why they asked this [guy](#)

... common genetic variation seems to have only a limited role in determining people's predisposition to many common diseases. Second, gene variants that are very rare in the general population can have outsized effects on predisposition.[...] If so, here's one confident but uncomfortable prediction of what personalized genomics could look like in 2020. The identification of major risk factors for disease is bound to substantially increase interest in embryonic and other screening programmes.

I am pleased to read that the community finally accepted the view that there is no value of current GWAs. We will see if rare variants will explain more although I have no idea how we can provide any statistical proof with N=1. At least the consequences are horrible if this will lead to embryonic screening programs.

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