

ADNI Full Genetic Sequences Now Available for Download

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DNA data miners, fire up your engines. The Alzheimer's Disease Neuroimaging Initiative (ADNI) released whole genome sequences for 809 study participants on September 18. Data from the multimillion-dollar effort, which includes people with Alzheimer's, people with mild cognitive impairment (MCI), and healthy controls, are available to investigators using the ADNI database.

"We are very excited," said Robert Green of Brigham and Women's Hospital in Boston, who heads ADNI's sequencing working group. "The advantage of the sequence is that you have an opportunity to explore unique and rare variants that you would not see in single nucleotide polymorphism [SNP] studies."

Between summer 2012 and spring 2013, ADNI completely sequenced DNA from 818 people: 128 with AD, 415 with MCI, 267 controls, and eight people whose variable symptoms over time led investigators to label them "uncertain diagnosis." Any given DNA sequence in the database was read an average of 40 times, Green said.

Before releasing the data, the scientists put it through what Green called "rudimentary" quality control. They checked the integrity of the data and compared the sequences with previously identified single nucleotide polymorphisms from the subjects. The researchers have temporarily removed some sequences until they can double-check their quality, and omitted others because of concerns about patient consent, bringing the number of sequences released to date to 809.

Researchers registered with ADNI can obtain the data in two forms. They can acquire the near-200 terabytes (that is, 200 million megabytes) of raw sequence information by mail, since it is too much to download over the Internet, or, as many researchers will likely prefer, they can work with smaller, downloadable files that identify where each person's genome differentiates from the norm, Green said. Users will be able to cross-reference the sequences with ADNI's other data, such as brain images and biomarkers.

Thus far, the National Institute on Aging's national genetics data repository has requested the data. Researchers can find out how to access ADNI's sequences here. Green said ADNI itself has not yet commenced data analysis, and that plans were too early stage to discuss at present.—

Amber Dance.

REFERENCES

External Citations

Alzheimer's Disease Neuroimaging Initiative

national genetics data repository

here

FURTHER READING

Papers

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