More information about ASPREE-G

Q: Why is ASPREE studying genetics?

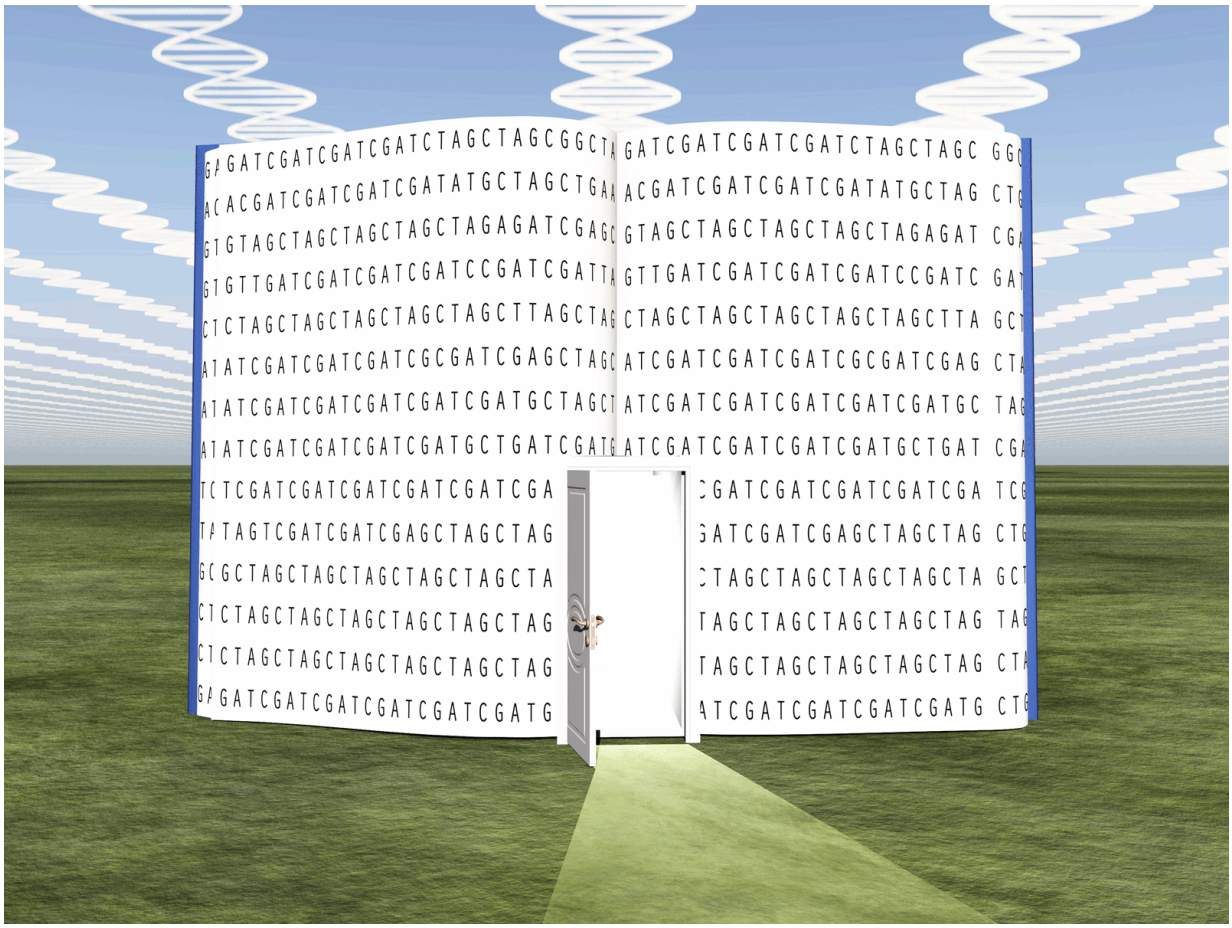
A: Since the ASPREE Healthy Ageing Biobank began in 2010, more than 15,000 Australian and U.S. ASPREE participants generously donated blood or saliva samples for future genetic research. The explosion in technology, reductions in cost and new funding opportunities, has launched genomics projects sooner than originally anticipated.

ASPREE is a unique resource for genetic research because unlike other studies:

- the ASPREE Healthy Ageing Biobank has the largest collection of biospecimens from healthy older adults in the world (most genetic studies look at the genes from people with a specific disease)
- all samples are of very high quality, maximising research opportunities
- each sample is associated with unprecedented health and lifestyle information to help bridge the link between our health and what is happening at a genetic level

It is important to note that genetic studies on ASPREE Biobank samples are for **research purposes only** and are **not** intended to be medical investigations or diagnostic tests.

Q: What is the difference between genetics and genomics?



A: Genetics generally relates to the study of single genes that are inherited and passed down from one generation to the next. Abnormalities or changes to genes can lead to inheritable diseases such as Huntington's disease.

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Genomics is the study of a person's entire set of genes. A process called whole genome sequencing reveals all the DNA on each gene in a given individual. Whole genome sequencing is comparable to reading and remembering the position of every letter in a 20,000 page book.

Imagine you had two large books – each book of 20,000 pages represents a set of genes from your mother and one from your father. Every page in a book represents one single gene. The letters on each page represents the order of DNA on the genes. Genomic sequencing 'reads' every letter of every word on every page in one fell swoop.

Armed with the complete set of genetic information, researchers now have the means to discover how genes behave and interact with other genes, and how environmental factors such as diet, may affect gene function.

This may mean that in some studies, researchers seek to identify a 'variation' (a change or 'mutation') in a genetic pattern equivalent to finding one changed letter in a 20,000 page book!

Genomics also enables researchers to look for patterns in genes that may be associated with good health versus patterns associated with disease. For instance, samples from the ASPREE Healthy Ageing Biobank are contributing to The Medical Genome Reference Bank (detailed below). The group of genetic patterns associated with good health in the Medical Genome Reference Bank will help to highlight in other samples, genetic variations that may be associated with disease.

Genomic sequencing generates very large amounts of data requiring extensive interpretation and analysis. It can take many years before scientists make discoveries that may change medical care.

Q: What are genetic mutations?

A: Genetic mutations are changes, variations or 'glitches' in a person's DNA akin to a spelling mistake. Everyone has a number of 'glitches' in their DNA and the vast majority have no effect on health. These are called 'silent mutations'.

Mutations can cause problems if they interfere with how a gene functions. But not all mutations are bad for you; some genetic variants can help preserve good health. For instance, US researchers discovered a mutation that seems to help protect some people from developing diabetes.

The Resilience Project (further details below) is another study which aims to determine why, in rare cases, some people have genetic mutations that would usually cause serious disease, remain otherwise healthy.

Q: How is my privacy protected?

A: ASPREE takes very seriously the responsibility for protecting the confidentiality and privacy of ASPREE participants whose biobanked samples are involved in medical research. All ASPREE genomic research projects:

- are tightly governed by ASPREE and human research ethics committees and restricted to highly ethical, non-commercial, public-good studies
- must have formal ethics approval from the Alfred Hospital Human Research Ethics Committee (HREC) and associated institutions
- de-identify all samples, meaning the donor's identity is never available to collaborating research organisations or laboratories and no individual can be identified in reports of the findings.

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Q: What type of genetic research is being undertaken?

A: Genetic studies on Biobank samples contribute to a growing body of research enhancing our knowledge about genetics and health.

Because genetic research is a specialised field, ASPREE is collaborating with national and international scientific groups with the leading technologies and expertise to provide high quality analysis and interpretation of genetic information.

All ASPREE samples used in genomic studies (present and in the future) are given a de-identifying code to protect the donor's identity. No individual can be identified in reports of the findings.

Two major projects established so far are:

1. **The Medical Genome Reference Bank Project:**

ASPREE and the Sax Institute's '45 and Up' Study in NSW collaborated with the Garvan Institute of Medical Research in Sydney to develop a Medical Genome Reference Bank. Funded by the NSW government and the Garvan Institute, this reference bank will provide genetic information from the genomes or DNA of healthy people. This reference bank or library of genomes from well older adults can be used as a comparison by researchers to identify, more easily, genes that cause disease. Approximately 4,000 genomes from healthy people aged 75 years and over were sequenced and analysed in this project.

The Garvan Institute in Sydney is one of the most respected research facilities in Australia and is recognised nationally and internationally. With their expertise, the reference bank will form the first genetic profile of this calibre of healthy older people in the world. This initiative, which began in 2016, has made considerable progress and will continue to generate scientific publications for many years to come.

2. **The Resilience Project:**

This project is a collaboration between the ASPREE Biobank and the Icahn Institute at the Mount Sinai School of Medicine in New York. The Icahn Institute has world-leading expertise and laboratory facilities to map and analyse very specific genetic information in a genome. It has been awarded grants from the NIH (National Institutes of Health), which is the same government agency that funded the ASPREE Clinical Trial in Australia and the US.

Called the 'Resilience Project', scientists at The Icahn Institute examined approximately 700 genes from ASPREE Biobank participants in Australia and the US for the presence of genetic variations associated with disease. The aim is to identify and understand why healthy older people with these genetic variations remain free of disease when others do not.

Additionally, the Resilience Project may also help reveal to ASPREE researchers genes that can reliably predict the onset of cardiovascular disease, cancer, dementia and depression in older adults.

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It is expected to take several years to undertake the analysis of all findings from the Resilience Project.

Q: Are donors notified if their sample is included in a genetic study?

A: Samples donated to the Healthy Ageing Biobank form a collective resource (as far as we are aware this is the biggest collection of blood samples from healthy older adults) to promote medical advancements for the benefit of the whole community.

The research focus is on new genetic discoveries that one day may improve medical diagnosis and treatment for future generations. These projects will vary in size, scope and duration and it will not always be feasible to identify each and every participant who has or has not had their sample included in a genetic study.

Q: Are donors notified if they have a genetic variant thought to be linked to disease or associated with an increased risk of disease?

A: In the rare event that a participant is found to have a medically significant genetic finding the ASPREE Executive Committee will work closely with the overseeing HREC (Human Research Ethics Committee) to determine the best path of action. If the HREC decides that participants should have the option to receive the results and a participant chooses to receive the results, a genetics counsellor will support the participant, and possibly the participant's family members, to make an informed decision about whether any steps may be required to further investigate the genetic finding.

Q: Could the results be used by insurance companies?

A: In Australia, a moratorium lasting until 2024 prevents insurance providers from requiring a disclosure of genetic results in applications for life insurance of AU$500,000 or below.

If an individual is not informed of their genetic data, there is no obligation to ever disclose research findings to insurers. Genetic research by ASPREE will not be passed on to insurance companies.

Q: Where can I find out more information about genomics?

A: A great place to start is this NIH website.