



## How Smoking Changes Your DNA

A team of researchers has sought to identify mutation signatures caused by smoking. The researchers looked at somatic mutations and DNA methylation in 5,243 cancer genome sequences. These genome sequences belonged to cancer classes that were more likely to occur in smokers. The researchers were able to compare 2,490 samples from smokers with 1,063 samples from individuals who had never smoked. The team of scientists identified mutational signatures and estimated the impact of each signature on cancer development. Mutational signatures 2, 4, 5, 13, and 16 (as defined by the Catalogue of Somatic Mutations in Cancer, COSMIC) were elevated in smokers when compared to those who had never smoked. Signature 4 has a lot in common with another signature which is due to exposure to benzopyrene (a chemical present in tobacco smoke). The researchers also found that smoking a pack of cigarettes a day for a year would lead to 150 mutations accumulating in the lungs, 97 in the larynx, 39 in the pharynx, 23 in the mouth, 18 in the bladder, and 6 in the liver. [This study](#) sheds some light on exactly how smoking mediates an assault on your DNA.

## Cystic Fibrosis in African Populations

Cystic fibrosis (CF) is an autosomal recessive disease. CF patients are usually underweight for their age and have frequent lung infections. Initially it was assumed that CF could only affect Caucasian children. This is not true. CF affects patients of all ethnicities; however, this misconception lingers and as a result CF patients who are not of European descent are more likely to be misdiagnosed. This may explain why European CF patients live to be about 40 years, Canadian CF patients live to 50 years but South African CF patients have a life expectancy of just 20 years.

CF is unique in that causative mutations are population specific, often varying by country or ethnicity. This means that each country would have to invest in identifying which mutations are relevant to its people. Only [12 of the 54 African nations](#) have any published effort toward identifying the mutations relevant to their people. Similarly in the African Diaspora, only [6 of 42 countries](#) in the Americas have done any such investigation. Investing in this kind of research is important, as current commercial genetic tests for CF are all Eurocentric and thus less effective in diagnosing African patients.

## **Genotyping 500,000 People**

Data from 500,000 UK Biobank participants have recently been made available to researchers. Affymetrix genotyped these samples between 2013 and 2014. The SNP chip used examined over 800,000 genomic regions. The UK Biobank project is funded by the MRC, the Wellcome Trust, the Department of Health, the Welsh and Scottish governments, the British Heart Foundation, Cancer Research UK, and Diabetes UK. In addition to genetic data, the study collected phenotypic data. This includes information about the participants' health, their diet, occupational history, mental health, activity levels, and key body measurements. Together, the genotyping data and the participants' phenotypes can help researchers answer many disease-related questions. The data set may also help to answer questions about how genetics, lifestyle, environment, and diet interact to affect our health.

Interested in analysing the data from half a million people? Send an email to [access@ukbiobank.ac.uk](mailto:access@ukbiobank.ac.uk).

## **May the Force Be With You**

Like many of us, Neuroskeptic is interested in publishing. This blogger chose to submit an article to the *International Journal of Molecular Biology: Open Access*, *Austin Journal of Pharmacology and Therapeutics*, and the *American Research Journal of Biosciences*. All four published Neuroskeptic's work.

The problem? He based it on what we know of mitochondria and filled it with Star Wars references.

The paper refers extensively to midi-chlorians. Midi-chlorians are tiny intelligent life forms that exist in symbiosis with all living things according to the Star Wars mythology. The Force speaks through midi-chlorians. [The paper](#) includes fun text such as:

- "Beyond supplying cellular energy, midichloria perform functions such as Force sensitivity..."
- "Involved in ATP production is the citric acid cycle, also referred to as the Kyloren cycle after its discoverer"
- "Midi-chlorians are microscopic life-forms that reside in all living cells—without the midi-chlorians, life couldn't exist, and we'd have no knowledge of the Force. Midichlorial disorders often erupt as brain diseases, such as autism."
- "midichloria DNA (mtDNRey)" and "ReyTP".

The journals have since retracted the article. The fact that it was accepted at all shows what can happen when peer review is missing. It also helps shine a light on the problem with predatory journals.

**Compiled by: Dr. Cheryl Stewart**



**Dr. Cheryl Stewart**  
**Postdoctoral Fellow**  
**University of Pretoria**

**1. What is your current area(s) of research?**

I am using sequencing and bioinformatics to identify mutations in South African cystic fibrosis patients.

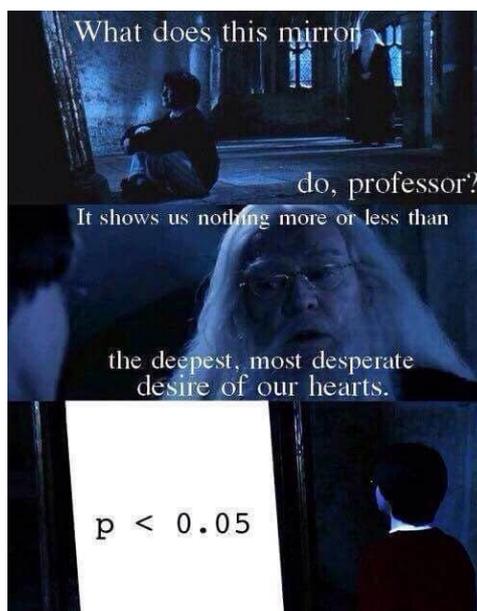
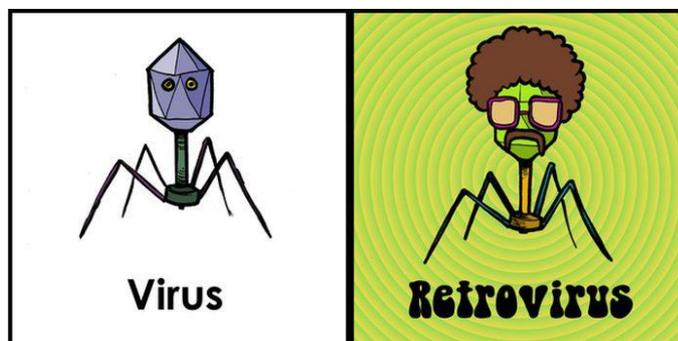
**2. What is your background and how did you become interested in your field of research?**

I have a PhD in Biotechnology. I have been fascinated with DNA since my days as an undergrad when I learned that DNA calls the shots.

**3. What piece of advice would you give to somebody beginning their career in human genetics?**

There are always more questions than answers because genetics is incredibly complex – that is what makes it so fascinating. At the same time, to unravel that complexity you will need a healthy dose of grit!

4. Do you know any science jokes you can share?



5. What does your typical working day look like?

My current project involves a lot of bioinformatics and stats so I spend most days in front of a computer.

6. How do you like to relax?

By reading a compelling book series.