

Gene Mutation Detection Study

In a small study conducted in April 2019, LexaGene's open-access LX technology successfully identified mutations directly from cheek swabs, as announced ([read news](#)).

With this study, LexaGene demonstrates the utility of the LX technology to identify single mutations within the human genome directly from DNA samples such as buccal (cheek) swabs, and the instrument's capability in looking for other types of genetic targets aside from bacterial targets [as previously studied](#).

- To exemplify the power of LexaGene's open-access technology, its scientists purchased DNA samples of nine individuals whose genomes were sequenced as part of the Human Genome Project (HGP).
- Each of the nine purchased DNA samples are known to have a unique combination of genetic variations at locations within the AKT1 and COMT genes.
- Each of these nine DNA samples were processed using LexaGene's technology, which confirmed the identity of the variations in these two genes with 100% concordance with HGP sequences.

Application of genetic testing for pharmacogenomics – prescribing medications, including cannabis:

To further test the LX technology for applicability to pharmacogenomics research use, cheek swabs were collected from 8 individuals and analyzed by the LX analyzer technology, which generated unambiguous results for all 8 samples. The allele frequency for both AKT1 and COMT mutations were close to 50% for both our samples (AKT1 63%:38%, COMT 50%:50%) and the population in general (AKT1 43%:58%, COMT 63%:37%)⁵ and support the need for pharmacogenomic testing prior to prescribing medicines, including cannabis.

The targeted mutations are known to influence the manner in which the brain responds to cannabis. Daily smoking of cannabis has been linked to increasing one's risk of developing a psychotic disorder.¹ This risk varies with one's genetic makeup and the AKT1 and COMT genes have been implicated in these disorders.²

Understanding the influence drugs have on individuals (i.e. personalized genomic-based medicine) is a high priority for the pharmaceutical industry and drug consumers.

- 1 Hall et al., Cannabis use and the risk of developing a psychotic disorder. World Psychiatry. 2008 Jun; 7(2): 68–71.
- 2 Morgan et al., AKT1 genotype moderates the acute psychotomimetic effects of naturalistically smoked cannabis in young cannabis smokers. Translational Psychiatry volume6, pagee738 (2016)
- 3 <https://www.genome.gov/11006943/human-genome-project-completion-frequently-asked-questions/>
- 4 http://www.genomenetwork.org/resources/whats_a_genome/Chp4_1.shtml
- 5 <http://www.internationalgenome.org/>

Appendix:

The human genome is comprised of approximately 3 billion base pairs³—99.9% of which is conserved across the human race. The remaining ~ 3 million base pairs differ from person to person⁴ making each of us unique (barring identical twins, where the number is much less, though generally not zero).

Some of these variations are in genes that influence very noticeable traits, such as height, weight, and hair, eye, and skin color, whereas others are much less noticeable, affecting intellectual aptitude, hearing, smell, sight, food preferences and the like. For medical relevance, some variants make individuals more or less prone to cancer and infectious disease. Likewise, some affect the ability to metabolize drugs, some are known to be associated with drug dependence (i.e. addiction), and others make individuals more prone to developing health problems after prolonged drug use.