SASHG
Southern African Society for Human Genetics

Sugar cravings? It's in your genes!

Many studies have warned about sugar intake and its contribution to diseases such as type 2 diabetes, obesity and cardiomyopathy. It is known that some people are more likely to find it hard to stop sugar intake, while for others, it is just a matter of a decision. Previous work done by von Holstein-Rathlou *et al*. (2015) in rodents has shown that FGF21 (a hormone secreted by the liver) suppresses the intake of sweets. A follow-up study on primates also supported this hypothesis (Talukdda *et al.*. 2016). More recently a study by Soberg *et al*. (2017), investigated whether FGF21 has the same effect in humans. The research was conducted on 6,514 Danish subjects. A statistically significant association was found between *FGF21* rs838133 and increased consumption of candy (sweets), as well as a nominal association with alcohol intake. Also mentioned in the study is that the level of FGF21 increased acutely after consumption of sucrose and was elevated in individuals who disliked sweets. According to the authors, the data presented suggests the liver may secrete hormones that influence eating behaviours – thus providing insight to a potential hormonal basis of "sweet tooth". More work will be carried out to confirm these findings.

Links to the studies:

http://www.cell.com/cell-metabolism/abstract/S1550-4131(15)00618-X

http://www.sciencedirect.com/science/article/pii/S1550413115006233

http://www.cell.com/cell-metabolism/fulltext/S1550-4131(17)30214-0

DNA in dirt?

Scientists from the Max Planck Institute in Germany have recovered ancient hominin DNA from sediment in caves. In an article published in Science, researchers examined seven archaeological sites across Europe and Russia with known hominin occupation but in which no macroscopic skeletal remains were previously found. Using just a teaspoonful of dirt and a targeted enrichment technique, they were able to recover mitochondrial DNA which matched with known Neandertal and Denisovan sequences. From the DNA recovered, only about 0.05% - 10% was identified as mammalian while the rest remains. Previous studies were able to recover DNA from sediment which corresponded to the sequences of animals and plants but were not able to reliably identify hominin DNA. Finding ancient bones is extremely difficult but with the use of this technique, a clearer picture will be constructed with regard to the occupation and movement of ancient species and the building of our evolutionary history. The journal article pertaining to this discovery can be found at the following URL:

http://science.sciencemag.org/content/early/2017/04/26/science.aam9695

There are 6500 differentially expressed genes between him and her!

A recent paper published in BMC Biology by the Weismann Institute of Science has reported that there are 6500 differentially expressed genes between men and women. For this study, the group analysed RNA sequencing data from 544 adults (part of the GTEx-project) and revealed genes that are differentially expressed in the reproductive tracts and tissues common in both sexes. For instance, they found that the genes highly expressed in the skin of men were related to growth of body hair and those responsible for building muscles were also highly expressed in males; while genes responsible for fat storage were more highly expressed in women. In general, the study identified significant association between sex-specific gene transcription, reduced selection efficiency, and accumulation of deleterious mutations, which might affect the prevalence of different traits and diseases. Indeed men and women have different selection pressures. The study suggests the need for a better understanding of both sexes at the genetic level, as this may play a role in disease or response to treatment.

You can read more about the study at:

https://bmcbiol.biomedcentral.com/articles/10.1186/s12915-017-0352-z

Compiled by:

Lindiwe Lamola and Shareefa Dalvie

Lindiwe Lamola

PhD Candidate

University of Cape Town

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

I have an interest in rare genetic syndromes (disorders). My PhD project investigated the genetic factors underlying Lynch syndrome and Constitutional Mismatch Repair Deficiency Syndrome; both are inherited cancer syndromes.

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

I completed my BSc at the University of the Free State and subsequently joined the Division of Human Genetics at the University of Cape Town; where I completed both my honours and Masters and currently PhD. I have always had an interest in cancer genetics ever since my grandmother was diagnosed with cancer many years ago. Having worked on projects involving the understanding of cancers, I have gained much needed insight, but I also have learned that the road is complicated and much still needs to be elucidated.

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

The road is not easy; however what we do (no matter how small) is a step closer to a better understanding of diseases or disorders.

4. **How do you like to relax?**

I read a lot and I like to take walks on the beach.

5. **Do you have any secret talents?**

I am proficient in mirror writing.

Shareefa Dalvie

Lecturer

University of Cape Town

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

Currently, I am doing a trauma genome-wide association (GWAS) and meta-analysis as part of the Psychiatric Genomics Consortium (PGC) working group on Post-traumatic Stress Disorder (PTSD). I also have an interest in population structure and ancestry.

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

I completed a BSc in Genetics & Development and Physiology at UCT. I then went on to do an Honours, Masters and PhD in Human Genetics (also at UCT). Subsequently, I did a post-doc for 2 years at the Department of Psychiatry and Mental Health. For my honours, I worked on a project looking at the genetic basis of bipolar disorder. Since then I've been hooked on psychiatric genetics. I enjoy working on complex disorders (and the challenges that these types of phenotypes bring) and there is still so much left to do in the field.

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

Nothing worth doing is easy. There will be good and bad days but in the end the pay-off is always worth it.

4. What does your typical working day look like?

I work at the Department of Psychiatry and Mental Health and Division of Human Genetics at UCT. My typical day involves doing some analyses (mostly computational), meeting with postgraduate students whose projects I co-supervise, and skypes or telecons with international collaborators.

5. How do you like to relax?

Reading a good book, being in nature (preferably simultaneously).