Aniah Matthews
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Sulkosky
ENGW 105

**Introduction:**

For this assignment the original paper was written for a genetics class, which is an upper level biology course. Being that this was written as a project to turn in and possibly share with the class who had a clear understanding of what the human genome is and the general idea of what we were talking about this paper may not be suited for someone who is not in the field as they would most likely not understand it much. As well as doing this project for a genetics class everyone had to do a different gene, which in research was able to give them an even deeper understanding of the human genome.

The purpose of the original content was to do research on a specific chromosome, and be able to speak about the many genes that are dependent on that chromosome. Along with this we were to target a couple of genes on that chromosome and talk about their functions in the body.

**Original draft:**

The body is made of 23 pairs of chromosomes with the twenty third being what determines what gender you are, male or female. On each of the chromosomes there are hundreds of genes which code for many different things, some of them are linked to diseases, some are not. On the ninth chromosome there are over 600 genes but not all of them have diseases linked to them such as the gene located on locus number 613632. This gene has been linked to two functions. The first function of this is for the VCA module, an activator to be stimulated in the cell to be able to regulate the protein actin by using a polymerization to put these proteins together. The gene is then able to regulate this protein with something called the ARP ⅔ complex that keeps the levels in the cytoskeleton where they should be. This complex is

made of seven subunits to make up this protein complex structure. The second function of this gene is to shuttle molecules away from the axon termini, the nerve endings of the branches that send signals to the next branch, this is called retrograde transport. The molecules that are being transported is something called mannose-6 phosphate receptor which is a molecule that moves proteins that target enzymes to lysosomes (the trashcan or cleaner of the cell). This gene gets help from another component called FAM21 which aids in the endosomal localization in cells. This is a rather large gene which contains 11 exons and has a span of 15kb, compared to the average 6.4 exons and an average length of 3kb. In the animal model when wash was washed out of the genome was lethal to the unborn eggs. When it was tried with mice who had tamoxifen-dependent loss of wash, which is estrogen the mice showed weight loss and severe anemia, or low iron count. It was then concluded that the mice needed WASH to be able to function normally, even though it has not been linked to any diseases in humans.

Another gene on this chromosome is at locus 607704, this gene is called KANK1 which is also called the kidney ankyrin repeat-containing protein. This protein is found to be suppressed in most renal tumors and in kidney tumor cells as well. This gene has a heterozygous (a dominant and recessive) component where the heterozygosity was taken out and the active gene was the one deleted. In 9 out of 10 tumors this occurrence was shown and the inactive gene was maintained leading researchers to believe that this was what was causing the tumors to grow. When the researchers screened for this in drosophila they found a gene similar to this one found in humans and named it dKANK this resulted in abnormalities which included structural problems and an abnormal diaphragm along with decreased number of vacuoles and lysosomes in the cells. This determined that the flies needed the gene to be able to function correctly. In the adult rat kidney research found the gene in podocytes, which are cells that wrap around

capillaries and help with the filtering of blood in the kidney, this was also found to work with synaptopodin, an actin- associated protein. This gene is even larger than the first one, having 18 exons and spanning 275kb. This gene has alternative splicing and can make different versions of the gene as well. This protien has been linked to the disease known as Cerebral palsy and spastic quadriplegic when looked at a 4- generation family. It was found that the deletion of the KANK1 gene was something that had an effect on whether the offspring would inherit the gene or not. However, this deletion was also found in healthy fathers without the disease but in the affected offspring, as well as in healthy female relatives yet the individuals who did inherit it inherited the deletion from their fathers.

**Revision analysis:**

For the revision of my essay I would like to include a slide in which introduces the human genome for the new audience to be able to understand what the rest of the slide show consists of. To change the paper to a more hostile audience I am putting emphasis on the benefits of getting your own genome completed while using the examples on the paper to show the important information the genome can tell someone. The removal of deeper explanations I think would keep the essay more to the point and be able to follow a bit better rather than all of the technical terms that are very confusing.

**Revised draft:**

Original word count: 654

Revised word count:465

30% decrease

Slide 1:

Text: ”why you should get your full genome done” “the body is made up of 23 pairs of chromosomes”

Images: chromosomes

Spoken:The body is made of 23 pairs of chromosomes with the twenty third being what determines what gender you are, male or female. On each of the chromosomes there are hundreds of genes which code for many different things, many of which can show you what may be in store for you or your children and may even prepare you or affect your decision to have kids. ~~some of them are linked to diseases, some are not.~~

Slide 2:

text: On the ninth chromosome there are over 600 genes but not all of them have diseases linked to them such as the gene located on locus number 613632.

Spoken: This gene has been linked to two functions. The first function of this is for the VCA module, an activator to be stimulated in the cell to be able to regulate the protein actin by using a polymerization to put these proteins together. The gene is then able to be regulated ~~this protein with something called the ARP ⅔ complex~~ that keeps the levels in the cytoskeleton where they should be. ~~This complex is made of seven subunits to make up this protein complex structure.~~ The second function of this gene is to shuttle molecules away from the axon termini, the nerve endings of the branches that send signals to the next branch, this is called retrograde transport.

~~The molecules that are being transported is something called mannose-6 phosphate receptor which is a molecule that moves proteins that target enzymes to lysosomes (the trashcan or cleaner of the cell).~~ ~~This gene gets help from another component called FAM21 which aids in the endosomal localization in cells. This is a rather large gene~~ ~~which contains 11 exons and has a span of 15kb, compared to the average 6.4 exons and an average length of 3kb~~

-Slide 3:

Text: the WASH gene is important to your health overall

In the animal model when WASH was washed out of the genome was lethal to the unborn eggs. When it was tried with mice who had tamoxifen- dependent loss of wash, which is estrogen the mice showed weight loss and severe anemia, or low iron count.

It was then concluded that the mice needed WASH to be able to function normally, even though it has not been linked to any diseases in humans, just yet.

~~Another gene on this chromosome is at locus 607704, this~~

Slide 4:

Text: the ~~gene is called~~ KANK1 gene which is also called the kidney ankyrin repeat-containing protein.

Spoken: This protein is found to be suppressed in most renal tumors and in kidney tumor cells as well. This gene has a heterozygous (a dominant and recessive) component where the heterozygosity was taken out and the active gene was the one deleted. In 9 out of 10 tumors this occurrence was shown and the inactive gene was maintained leading researchers to believe that this was what was causing the tumors to grow.

~~When the researchers screened for this in drosophila they found a gene similar to this one found in humans and named it dKANK this resulted in abnormalities which included structural problems and an abnormal diaphragm along with decreased number of vacuoles and lysosomes in the cells. This determined that the flies needed the gene to be able to function correctly. In the adult rat kidney researeh found the gene in podocytes, which are cells that wrap~~

~~around capillaries and help with the filtering of blood in the kidney, this was also found to work with synaptopodin, an actin- associated protein.~~

Slide 5:

Text: The kank1 gene can save lives

Spoken: ~~This gene is even larger than the first one, having 18 exons and spanning 275kb. This gene has alternative splicing and can make different versions of the gene as well.~~ This protien has been linked to the disease known as Cerebral palsy and spastic quadriplegic when looked at a 4- generation family. It was found that the deletion of the KANK1 gene was something that had an effect on whether the offspring would inherit the gene or not.

Slide 6:

Text: it is better to know then be left in the dark, blind

spoken: ~~However,~~ this deletion was also found in healthy fathers without the disease but in the affected offspring, as well as in healthy female relatives yet the individuals who did inherit it inherited the deletion from their fathers.