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Bio 201

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Human chromosome project

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 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, and 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. This is something that is still being researched and exactly how it is inherited but it has been linked directly to the disease. There is also research being done about the linkage of this gene to a nephrotic syndrome but little research has been done so far and it has not been confirmed yet.

There are thousands of genes in the human chromosome and this information only includes two of those many genes!

Citations:

Dictionary.com. (n.d.). Retrieved November 18, 2019, from https://www.dictionary.com/.

human chromosome. (n.d.). Retrieved November 18, 2019, from https://www.omim.org/search/?index=geneMap&start=1&sort=chromosome_number+asc,+chromosome_sort+asc&search=&limit=10&chromosome=9.