Tips & Tricks for the Chromosome Project:

1. start by google imaging your chromosome and getting a good/accurate depiction of the Chromosome and its genes that have been mapped (the gene locations and/or the names of the diseases/disorders that result from those genes being mutated (see below)

2. research info on your chromosome as a whole; number of genes, number of base pairs (nucleotides), any other interesting facts about your chromosome – i.e. does non-disjunctions happen with this chromosome at all? Can you survive it? Crossing over? Etc?

3. choose 5-6 of disorders/diseases on your chromosome (like above) and their associated genes & highlight and research briefly – include that information on your poster

4. from that pool of diseases/disorders – pick 1 to focus on for the remainder of your project – I recommend selecting a gene that is directly associated with a disease/disorder and has a clear mutation that is easily researched, not a gene that is associated with a disease when other genes are also mutated along with it…that makes your project harder…you don't want that ☺

**Note: if you want to send me your chosen gene via email or remind 101 message for me to approve, you can do that anytime this weekend. I'll get back to you before Monday.

5. Now it's time to focus on the gene of interest and use one of the websites below. I like to work backwards and put the syndrome/disease/disorder in google or in pubmed search first to find the gene associated with that syndrome. So if I chose Allgrove syndrome I would determine the gene associated with it in humans is the gene *AAAS. (all names of genes are italicized!)*

http://www.ncbi.nlm.nih.gov/ - if you are at school when using this site; you have to google search pubmed.gov, click on it from google and then click on ncbi (going in the back door) – for some reason there is a filter on this websitehttp://www.ensembl.org/index.html

http://www.omim.org

6. now I use ncbi genbank or fastblast search or the above websites to search for the gene AAAS in *Homo sapiens* and can click on the gene to see # introns/exons, the nucleotide sequence, the location of the gene in my chromosome (called the loci or address…this gene is at 12q13…chromosome 12, q arm, location #13). I can also use this information to determine the mutations that occur in this gene to cause allgrove syndrome

Here was the result of my own research on this gene…

a. I changed the dropdown menu on ncbi to “gene” then typed in AAAS

b. I scrolled down till I found the gene in *Homo sapiens* (click) http://www.ncbi.nlm.nih.gov/gene/8086

c. I scrolled down to the gene map and you can use fastablast or genbank to get the nucleotide sequence of your gene of interest. If you scroll to the bottom of this link you will see the nucleotide sequence. http://www.ncbi.nlm.nih.gov/nuccore/NC_000012.12?report=genbank&from=53307456&to=53321628&strand=true

7. From my gene, I can work forward and determine the name of the protein that is made by AAAS, I like either google or change drop-down on ncbi to “protein”-or- I also like the website uniprotein (uniprot.org = mine is Aladin ☺

Website: http://www.uniprot.org/blast/

http://www.uniprot.org/uniprot/Q9NRG9

8. then from the protein info I can determine what the protein looks like, what it can do, type of protein it is…etc which leads to why if it is non-functioning, why the disease can form…

9. I can also change the drop-down menu to “pubmed health” or just “pubmed” and click on “free full text” to get articles about my disease, protein or gene, depending on what you choose to type in the search engine.

10. For the mutation in the sequence, I like to start with the disease itself. Sometimes they will flat out tell you that there is a point mutation on 1 base pair and tell you how it affects the amino acid sequence. Sometimes you need to dig a little to find this…I can help you with this during in-class research time. Here is what I found out about my disorder and its mutation…

(copied from ncbi search using pub med with the name of the syndrome and “mutation” in the search box) – link below

Exp Ther Med. 2015 Oct;10(4):1277-1282. Epub 2015 Aug 10.

Identification of AAAS gene mutation in Allgrove syndrome: A report of three cases.

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Abstract

Allgrove syndrome (AS) is an autosomal recessive congenital disease, caused by mutations in the AAAS gene, and is characterized by the triad of Addison's disease, achalasia and alacrima. The present study describes three newly diagnosed cases of AS, in which genetic analysis of the AAAS gene was used to identify AAAS gene mutations, to enhance the understanding of the pathogenesis and clinical manifestations of AS in the Chinese population. Two of the cases exhibited homozygous mutations of c.771delG (p.Arg258GlyfsX33) in exon 8 and one case exhibited a homozygous mutation of c.1366C>T (p.Q456X) in exon 15. A review of the current literature suggests that the AAAS c.771delG mutation has only been reported in the Chinese population. Genetic analysis of the AAAS gene in Chinese AS patients at a young age may facilitate an earlier diagnosis and the timely initiation of the appropriate treatment, ultimately improving the patient outcome.

KEYWORDS:

AAAS gene; Allgrove syndrome; Chinese; genetic analysis

http://www.ncbi.nlm.nih.gov/pubmed/26622478

HTH! - JW ☺