What is Ogden Syndrome?

Back to Basics: Genetics 101

It's been a long time since that high school Science class. Here's a quick refresher!

SCIENCE CORNER

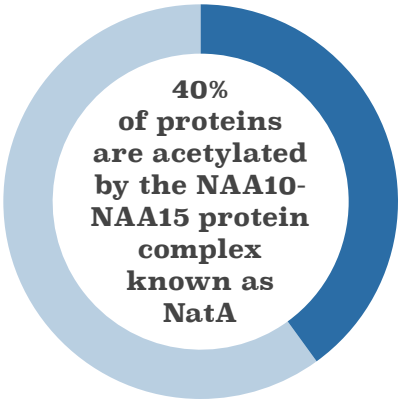
Our bodies are made up of cells. Within each cell is a nucleus. Within each nucleus there are 23 pairs of chromosomes (you get one from mom and one from dad!). Each chromosome contains anywhere from hundreds to thousands of genes and these genes contain instructions for making proteins. The directions for making these proteins are spelled out by a sequence of 4 bases which are represented by the letters A,T,G,C. A's pair with T's and G's pair with C's. The entire Human Genome contains about 3 Billion of these base pairs. We all have multiple spelling errors on our genome and most of the time these errors are inconsequential but if the error occurs on a vital gene in a crucial spot the instructions to make proteins are messed up with devastating consequences.

The 4 Nucleotide Bases

- Adenine
- Thymine
- Guanine
- Cytosine

What does the NAA10 gene do?

Ogden Syndrome is caused by a single spelling error on the crucial NAA10 gene.



NAA10 is a protein coding gene essential for normal cell function. NAA10 encodes the enzyme known as N-Terminal-Acetyltransferase 10 which along with NAA15 makes up the NatA complex. The NatA complex catalyzes a very crucial process known as Acetylation within our cells. Acetylation is a chemical event that involves the attachment of an acetyl-CoA group to a protein. It is through this attachment mechanism that the cell regulates protein stability, activity and localization. Mutations to the NAA10 gene changes the structure of the protein which makes it less effective at N-Terminal acetylation than a non-mutated protein. This reduced activity that sometimes effects NAA10 stability is what causes the symptoms of Ogden Syndrome to manifest.

3 FAST FACTS

1

IN OUR CELLS...

The NAA10 protein is made and generally remains in the cytosol.

2

ON OUR CHROMOSOME...

The NAA10 gene is located on the X chromosome at position Xq28.

3

IN OUR DNA...

The entire NAA10 gene is almost 6000 nucleotide bases long!

What are the Odds?

Ogden Syndrome is Ultra Rare! So rare in fact the odds of diagnosis are 1/11 MILLION

X-Linked Disorders?

Since Ogden Syndrome is caused by a variant on the X Chromosome females who have 2 X chromosomes are generally less severely affected than males who only have 1

Symptoms of Ogden Syndrome

- Intellectual Disability
- Developmental Delay
- Dysmorphic Features/Aged Appearance
- Failure to Thrive/Slow Growth
- Feeding/Swallowing Difficulties
- Cardiac Anomalies
- Seizures
- Autism Spectrum Disorder

HELPFUL RESOURCES

Ogden C.A.R.E.S. - www.ogdencares.org

Ogden C.A.R.E.S. Facebook- https://www.facebook.com/profile.php?id=100067718586138

NAA10 Families Together Website- www.naa10gene.com

NAA10 Families Together Facebook- https://www.facebook.com/NAA10FamiliesTogether

NAA10 Families Together Private Support Group- https://www.facebook.com/groups/1119372051470633

Ogden Syndrome-NAA10 Family Support Group- https://www.facebook.com/groups/1607746136179261160774613617926