Angelman Syndrome (AS) is a neurological disorder that affects about 1 in every 12,000-20,000 people [1]. Outcomes of AS include severe mental and motor impairments, such as ataxia. AS ataxia is caused by a disruption of neuronal *UBE3A* expression, and 10% of these cases involve an *SNRPN* mutation [2][3]. *SNRPN* overlaps the imprinting center (IC) of chromosome 15, which is known to control neuronal expression of *UBE3A* and is independent of expressional control through methylation [4]. It is suggested that *SNRPN* is involved in regulating imprinting, affecting *UBE3A* expression, but it is unknown how it may do so [5].

My **primary goal** is to determine the role of *SNRPN* in AS ataxia.

My **hypothesis** is that *SNRPN* is produces a non-coding transcript that interacts with the chromosome 15 imprinting center, regulating neuronal *UBE3A* expression independent of methylation, and deletions in this gene result in a non-functional transcript.

My **long-term goal** is to determine what other genetic expressions may be affected by *SNRPN* deletions.

**[1]** Angelman Syndrome: Genetics Home Reference [<https://ghr.nlm.nih.gov/condition/angelman-syndrome>](https://ghr.nlm.nih.gov/condition/angelman-syndrome)

**[2]** Heck, D. H., Zhao, Y., Roy, S., LeDoux, M. S., & Reiter, L. T. (2008). Analysis of cerebellar function in Ube3a-deficient mice reveals novel genotype-specific behaviors. *Human Molecular Genetics*, *17*(14), 2181-2189. doi:10.1093/hmg/ddn117

<<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2902285/>>

**[3]** Farber, C. (1999). The chromosome 15 imprinting centre (IC) region has undergone multiple duplication events and contains an upstream exon of SNRPN that is deleted in all Angelman syndrome patients with an IC microdeletion. *Human Molecular Genetics*, *8*(2), 337-343. doi:10.1093/hmg/8.2.337 [<https://academic.oup.com/hmg/article/8/2/337/585544/The-Chromosome-15-Imprinting-Centre-IC-Region-Has>](https://academic.oup.com/hmg/article/8/2/337/585544/The-Chromosome-15-Imprinting-Centre-IC-Region-Has)

**[4]** Williams, C. A., Driscoll, D. J., & Dagli, A. I. (2010). Clinical and genetic aspects of Angelman syndrome. *Genetics in Medicine*, *12*(7), 385-395. doi:10.1097/gim.0b013e3181def138

<<https://www.ncbi.nlm.nih.gov/pubmed/20445456>>

**[5]** Dittrich, B., Buiting, K., Korn, B., Rickard, S., Buxton, J., Saitoh, S., … Horsthemke, B. (1996). Imprint switching on human chromosome 15 may involve alternative transcripts of the SNRPN gene. *Nature Genetics*, *14*(2), 163-170. doi:10.1038/ng1096-163

<<https://www.ncbi.nlm.nih.gov/pubmed/8841186> >