What is Angelman Syndrome?

Angelman syndrome (AS) is a rare neurogenetic disorder that affects approximately one in 15,000 people or approximately 500,000 individuals worldwide. Children and adults with AS typically do not speak verbally. They have balance issues, gross and fine motor impairments and often debiltating seizures. Some individuals never walk. Disrupted sleep cycles also can be a serious challenge to the individual and caretaker(s).

Uniqueness of Angelman Syndrome

While Angelman syndrome is not only a rare disorder, it is also unique because it involves an imprinted gene. We receive two copies of each gene, one from our mother and one from our father. Genes make proteins that perform various functions in our bodies. A small handful of our genes are imprinted, meaning only one parent's copy is making a protein. One of our imprinted genes is the *UBE3A* gene which is on chromsome 15. The copy of *UBE3A* that we receive from our mother is active in the neurons of our brain, yet the copy we receive from our father is silenced in these neurons by something called the *UBE3A-antisense*. In the rest of our body the father's copy is not silenced and is active.

If an individual is born missing the copy of the *UBE3A* gene from their mother, or the copy they get from their mother isn't functioning properly, they have Angelman syndrome. The individual still has a copy of the *UBE3A* gene from their father, but like in all of us, it is turned off, or silenced, in the neurons of the brain by this imprinting phenomenon.