**What is spinal muscular Atrophy?**

* A rare neuromuscular condition that usually causes early death. The condition also causes loss of motor neurons in the anterior horn of the spinal cord and brain.
* This leads to wasting and weakness of the muscles that humans use to walk, sit, and control the movement of the head. In the worst cases the breathing and swallowing muscles are affected.

**Causes**

* When someone is affected by Spinal Muscular Atrophy, the SMN1 gene is mutated via deletion at exon 7, an exon is a part of a gene that will be a part of the final mature RNA.
* This causes reduced SMN (survival of motor neurons) proteins because SMN1 can’t code them properly. SMN plays a big role in the survival of motor neurons. All people with SMA can still form 10-20% of the normal SMN amount, this results in slow death of motor neuron cells in the anterior horn of spinal cord and the brain.
* The severity of the condition is based on how well the SMN2 genes can make up for the loss of SMN proteins. Healthy people carry 2 SMN2 gene copies but people with SMA can have 1-4 and maybe even more (the more they have the more the severity lowers).
* As a result of this, muscles that need these neurons now have lower input from the central nervous system. This causes movement to be much harder. There is a high chance affected people will die early in life. If they survive their spines can be quite misaligned. It affects 1 in 6000-10000 people.

**Different types**

* Type I (also named Werdnig-Hoffman disease) is a severe form of SMA. Children with Type I SMA show symptoms at birth or in the first few months of life. All affected children are developmentally delayed. All affected children have breathing and swallowing problems. This can result in gagging or choking. Many cannot support their heads. Death can occur in the first few weeks and usually they don’t live past 2.
* Type II develops at around 6-12 months and is evident by muscle weakness. These children can sit normally but might not be able to get into position on their own. People with Type II SMA cannot walk or stand alone. Life expectancy is shortened but many live well into adulthood.
* Type III symptoms start between early childhood and the teen years. They can stand and walk without help, but walking and stairs will become harder over time and many will need a wheelchair later on. Life expectancy is almost normal.
* Type IV symptoms start after 30 years of age. Symptoms include mild/moderate muscle weakness and tremors but also minor breathing trouble. Most of the time only proximal muscles are affected.

**Inheritance**

* SMA is inherited by autosomal recessive means and both parents have to be carriers for the child to be affected. Carriers only have one SMN1 gene with mutations.
* Even if both parents are carriers doesn’t mean the child will be affected. There is a 2/4 chance they will carry it, a 1/4 chance they won’t carry it and a 1/4 chance they will have it.
* Both copies of the SMN1 gene have mutations

**Research and technology**

* In 2016, Nusinersen was the first approved drug for treating SMA. It costs 125k per injection and is 750k for the first year and 375k a year after
* It is administered by intrathecal injection directly into the nervous system and changes the SMN2 genes and makes them function like the SMN1 gene.
* Drug stopped the progression of SMA and in 60% of the babies that had type 1 SMA showed significant improvement to motor function.
* Side affects include: rashes, severely low salt levels, risk of ear and respiratory infections, and chances of scoliosis. In older subjects the side affects include headaches and back pain. It may also stunt growth.