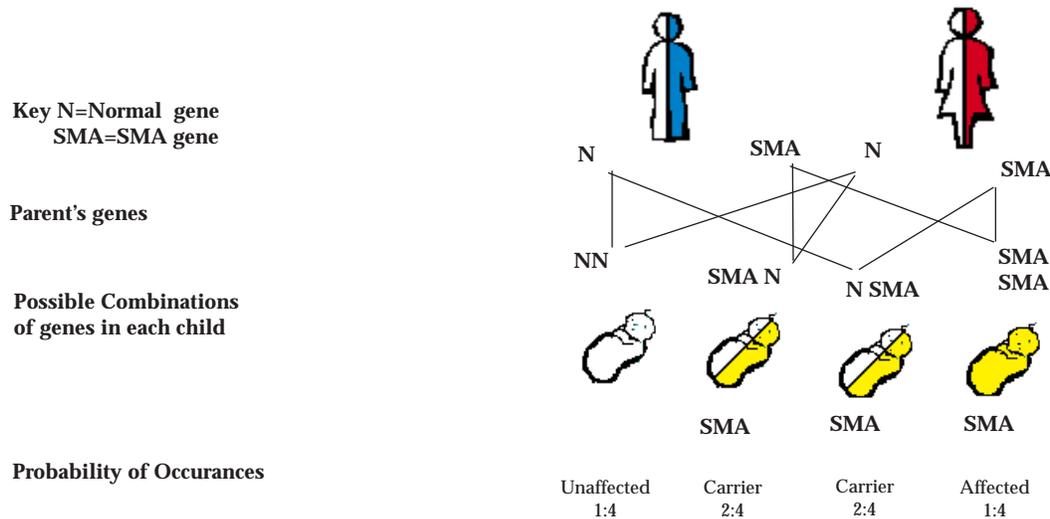




## WHAT CAUSES SMA?

Spinal Muscular Atrophy is an autosomal recessive disease. In order for a child to be affected by SMA, both parents must be carriers of the defective gene and both must pass this gene on to their child. Although both parents are carriers, the likelihood of passing this gene along to a child and having an affected child is 25%, or 1 in 4.



SMA is caused by a mutation in the survival motor neuron gene 1 (SMN1). This SMN1 gene is responsible for the production of a protein that is essential to the proper working of the motor neurons. People who are missing both copies of SMN1 have SMA, while carriers are missing only one copy. There is a similar copy of SMN1 called SMN2 that even people with SMA always have. The SMN2 gene differs by producing only a little SMN protein. This small amount of SMN protein is not sufficient for the normal function of the motor neuron. The lack of this protein causes the motor neurons in the spinal cord to degenerate resulting in SMA.

### How is SMA diagnosed?

90-95% of all cases of SMA can be detected through a blood test to screen for SMN deletion. For carriers there is a prenatal test available that allows for prenatal testing with 98% reliability.