Spinal Muscular Atrophy (SMN1 Linked)

Spinal muscular atrophy (SMA) can be caused by mutations in a number of genes. One of these is the SMN1 gene, which encodes for the spinal motor neuron protein. This protein is required for the maintenance of motor neurons in the spinal cord and brainstem. SMA is divided into different types: types 1-4 of SMA are all caused by mutations of the SMN1 gene, and all involve muscle weakness. Type 1 SMA first occurs before 6 months. Affected babies are unable to hold their heads up or sit up; they typically have difficulty swallowing and breathing, so they tend not to survive beyond the age of two. Type 2 SMA first occurs between 6 months and a year. Babies can sit up, but do not go on to stand or walk unaided in the usual manner. Type 3 SMA first occurs in older children. They can normally walk unaided, but may find climbing stairs or other similar tasks difficult. They may need to use a wheelchair by mid-life. Type 4 SMA first occurs in adulthood. Sufferers have some muscle weakness, tremors, and mild breathing problems.

The incidence of all types of spinal muscular atrophy is around 1 in 6,000 to 1 in 10,000 births. It is estimated that around 1 in 40 to 1 in 50 people is a carrier. Although the incidence varies somewhat from country to country, it does not seem to be highly present in any ethnic group. The faulty gene is transmitted in an autosomal recessive manner, typically requiring both parents to be asymptomatic carriers.

Sources

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