

New information about the causes of 'floppy baby' syndrome discovered

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(Medical Xpress) -- New information on the potential cause of Spinal Muscular Atrophy (SMA), known as "floppy baby syndrome", has been discovered by cell biology experts at the University of St Andrews.

A team lead by Dr Judith Sleeman, lecturer in Cell and Development Biology, has uncovered some new information involving the way SMA stops genes from working properly.

In laboratory models of SMA, researchers at the University of St Andrews have uncovered differences in the movement of key parts of a molecular 'machine' called the spliceosome, vital for the way genes work.

This "machine" helps to decode the DNA molecules that carry genetic instructions and removes sections which are not needed. This process goes wrong in conditions such as SMA.

The discovery of these differences in molecular movements may help to explain what goes wrong in [cells](#) to cause SMA.

[Spinal Muscular Atrophy](#) (SMA), 'floppy baby syndrome', is the leading genetic cause of death in children.

It is a type of motor neuron disease, affecting 1 in 6,000 births. The most severely affected children die before the age of two.

The gene responsible for SMA is known, but it is still not clear how problems with this gene damage the cells of the body and why motor neurons, responsible for sending messages to the muscles, are particularly sensitive. The more researchers can find out about exactly how cells are damaged in SMA, the better the chance of finding treatments.

The information stored in the genes of every cell

needs to be interpreted before it can be used by the cell. This is carried out by complex molecular 'machines'. These are extremely dynamic and need to work quickly and accurately.

The research will be published in the *Journal of Cell Science*.

Dr Sleeman said: "The genetic defect that causes SMA has been known for nearly 20 years, but how this defect leads to the symptoms is still not understood.

"Problems with the splicing of messenger RNA, an essential step in decoding genes, have been seen in SMA.

"Our work explains how these problems might be caused. We hope that this will provide an important clue to help unravel how cells are damaged in SMA and, in time, contribute to the development of treatments for this devastating condition."

Provided by University of St Andrews

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