What Is exon 7

Some of the information below was copied verbatim from www.brainwork.md

Spinal muscular atrophy (SMA) is a relatively common neurodegenerative disease caused by the loss of the survival motor neuron 1 (SMN1) gene. Humans possess a linked, nearly identical gene, SMN2, which produces a functional SMN protein, but at levels insufficient to compensate for loss of SMN1. Exon 7 is all that differentiates the two genes, but this is sufficient to prevent efficient exon 7 splicing in SMN2.

Surprisingly, there’s not a lot of information readily available on the Internet regarding exon 7, and even though more information needs to be published regarding what exon 7 is and what it does, researchers have known about this for quite some time.

Professor Kevin Talbot spoke about this topic, and gave some great information as to why exon 7 is becoming a popular topic with researchers and pharmaceutical companies.

Biogen's drug Spinraza (Nusinersen), addresses this particular problem.

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Professor Kevin Talbot from the University of Oxford, stated the following to an audience in the UK: “In order to try to improve the life of people with SMA, you don’t actually need to increase the protein by very much. To achieve this, various drugs have now been tested in their ability to upregulate SMN levels, as have therapies aimed at altering the splicing of the SMN2 gene in order to make it behave like an SMN1 gene. That is because SMN2, unlike SMN1, does not bind exon 7, which effectively leaves a link missing in the genetic transcript chain. However, by using an antisense oligonucleotide that prevents binding of splicing repressors, SMN2 is “tricked” into readily including exon 7, and SMN levels increase as a result. This seemingly complex process has now been achieved with a single drug, nusinersen, backed up by excellent results in a phase 2, open-label trial in 20 infants with SMA type 1.”

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Professor Talbot also suggested that “we may only be at the very beginning in terms of effective therapeutic options, including next-generation agents that focus on peptide-mediated oligonucleotide therapy, loading SMA systemically. Alternatively, gene therapy with adeno-associated virus (AAV)-9 is hotly anticipated, following mouse models that have indicated they can rescue SMN function. The beauty of gene therapy lies in its one-shot approach and systemic delivery. However, questions remain regarding toxicity (including the effect on antibodies), dosing and manufacturing, and there may be a defined therapeutic window after which gene therapy does not work.”

Offering his conclusions for the audience, Professor Talbot reasoned that “we are now embarking on a revolutionary era in the treatment of SMA. With improvements to therapies, and effective, early screening, could we be poised to eradicate new cases of SMA within the next couple of decades? Such questions will have to be answered in due course, but for now, the nusinersen story has been an exquisite showcase of potential that should pave the way for more innovative therapies.”

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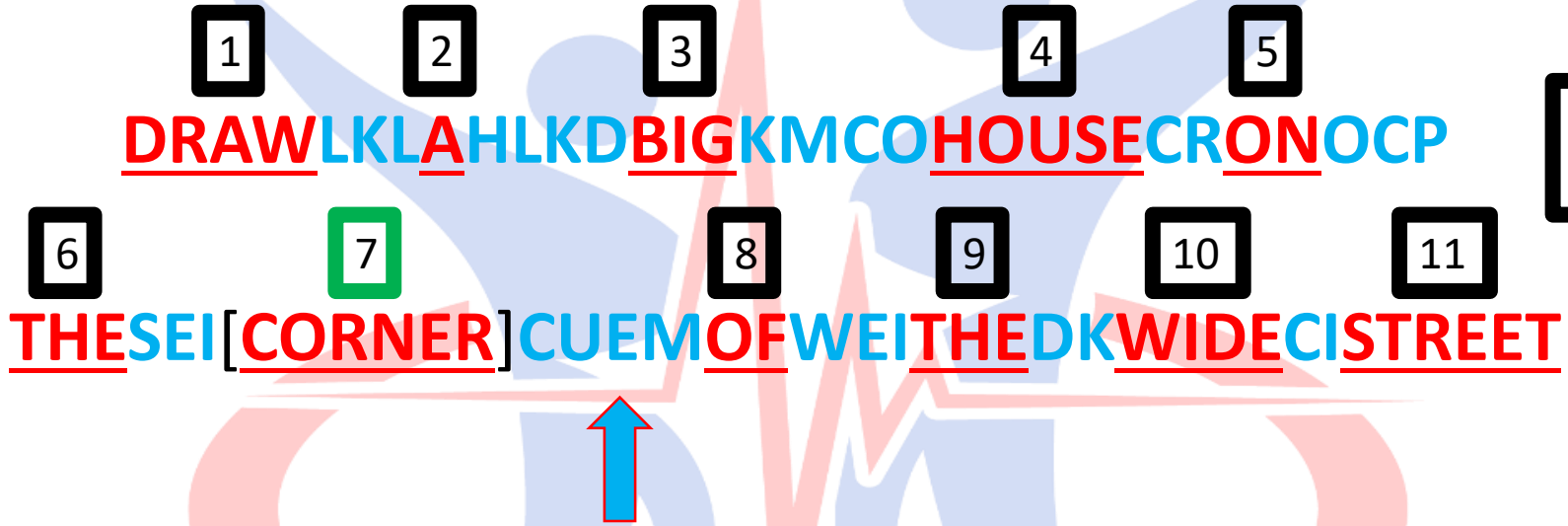
There's a fascinating video that explains in great detail how exon 7 affects those of us with SMA. Unfortunately, due to copyright laws, I'm unable to insert this video in my presentation. After you finish watching my video, please go to my website and click on **More**, then choose **Helpful Links**. I'll provide a link to this video that will explain everything in greater detail.

Explaining exon 7

Some of the information below was copied verbatim from Cold Spring Harbor Laboratory



Introns are in **BLUE**
Exons are in **RED**

Nusinersen is a short sequence of RNA that binds just after exon 7 in the unedited RNA message.

Normal Splicing: **DRAWABIGHOUSEONTHECORNEROFTHEWIDESTREET**

1 2 3 4 5 6 7 8 9 10 11

Faulty Message: **DRAWABIGHOUSEONTHE OFTHEWIDESTREET**

1 2 3 4 5 6 8 9 10 11