We Must Accept Finite Disappointment, But Never Lose Infinite Hope

Hadeel Ahmad Al-mogrbi. 3rd year medical student

Libyan International Medical University

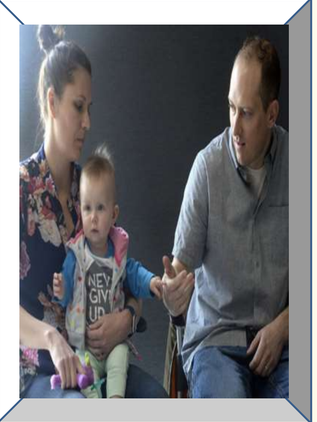
Introduction

- Spinal muscular atrophy is a genetic disorder of motor neurons, the cause of most forms of SMA is the deficiency of SMN protein.

- full-length,functional SMN protein using genetic instructions carried in the SMN1 gene & identical neighboring gene, called SMN2, typically direct cells to produce a short, partially functional version of the SMN protein.

- The great deal of SMA research is aimed at increasing SMN levels, improving muscle strength and function through development of various strategies. [1]

Blakeley was born in 2011 & diagnosed with SMA. Unfortunately there was no treatment to save her. But today there is a hope for patients with SMA. [2]



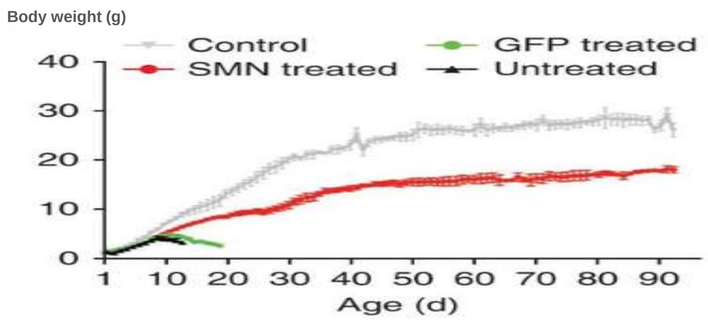
Studies

- **Raising SMN levels through gene therapy.**
- **Prenatal transplantation of human amniotic fluid stem cells .**
- **Raising SMN levels using antisense oligonucleotides.**

Data Analysis

➢ Gene therapy

Scientists have successfully used gene therapy to treat very young mice with SMA-like disease.[3]

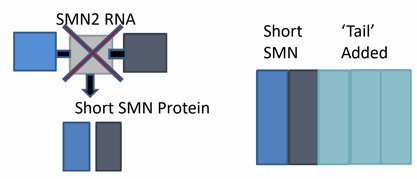


➢ Stem cells transplantation

Engrafting enriched neural cells help produce neurotrophic factors that are responsible for the growth of mature neuron cells.[4]

➢ Making Sense of Antisense

Antisense oligonucleotides are small molecules that can bind to RNA to change how it is spliced. A potential therapy for SMA is to design ASOs that will change the splicing of SMN2 to make more functional SMN protein.[5]



Results

- ❑ A non-replicating adeno-associated virus capsid is used to deliver a copy of human SMN gene (AVXS-101 provided by Avexis) to the patient's own cells.[6]
- ❑ Prenatal stem-cell therapy preserves the time window to treat disease in utero with much less cell number. It could be applied as the additional therapy combined with antisense oligonucleotide in the future.[4]
- ❑ ASOs Showed dramatic improvement of motor function and a significant increase in survival.

FDA on Dec. 23, 2016, approved the antisense therapy Nusinersen (Spinraza) for the treatment of SMA.[5]

Recommendations

- Using CK-107 in Patients With SMA.[7]
- Efficacy of RG7916 in type1 SMA Infants.[8]
- Using Branaplam in Type 1 SMA.[9]

Conclusion

- Researches have focused on strategies to increase the body's production of SMN protein, lacking in the chromosome 5-related forms of the disease.
- The approaches include methods to help motor neurons survive in adverse circumstances and to maximize the child's independence and quality of life.

References

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