



SMS RESEARCH FOUNDATION

October 2017 News & Events

Greetings!

Our 2017 Challenge Grant ran from January 15 - July 15 and was a tremendous success, raising \$165,000 for Smith-Magenis Syndrome (SMS) research! Our anonymous donors contributed \$70,000, which was matched by an additional \$70,000 from a variety of sources. An extra \$25,000 was also raised during this time, for a grand total of \$165,000. The entire amount will be allocated to the new SMS gene therapy project at Baylor College of Medicine. Thank you to everyone who contributed to the success of this fundraising campaign!

Donate
Here

This newsletter provides an update on phase 1 of the gene therapy research project, as well as information on upcoming fundraisers and recent events. If you are interested in hosting an event, please [contact us](#), and we will help you every step of the way.

Thank you for your continued support as we strive to support research that will improve the quality of life of those with Smith-Magenis Syndrome.

Sincerely,
SMSRF Board of Directors

Research Update

The SMSRF has embarked on a gene therapy project to replace the dysfunctional copy of the RAI-1 gene. This is a proof-of-concept phase performed with an SMS mouse model to determine if the neurocognitive and behavioral issues identified in these mice (which mimic those of humans with SMS) can be improved or reversed with the addition of a normal copy of the RAI-1 gene that will lead to an increase in the level of RAI-1 protein critical in brain function and development.

The first challenge identified is that the size of the RAI-1 gene is too large for a typical viral vector. We have developed a series of shorter versions that can fit by removing noncritical portions of the gene, thus leading to a more easily packaged RAI-1 gene that can still produce a functional RAI-1 protein. We are currently testing these shorter versions to prove that they fit in the vector and also produce the correct amount of protein.

Once this crucial step has been accomplished, the gene vector will be injected into the brain of SMS mice and then additional studies will be developed to prove that the protein was delivered to the brain at the correct levels and study the subsequent effects on the mice.

This work is being performed primarily at the Baylor College of Medicine under the leadership of Sarah Elsea PhD and in collaboration with Sanofi Pharmaceutical, a leader in gene therapy.



Baylor
College of
Medicine



SMS Awareness Day

SMS Awareness Day is coming up on Friday, November 17. This date signifies the 11th part of the 17th chromosome where the SMS deletion is located.

This year's hashtag campaign is #17for17. Stay tuned for more information about how you can get involved!



International Partnerships

We are thrilled that Pas à Pas avec Alexia, an SMS foundation based in France, has donated \$25K (USD) to the SMSRF toward the gene therapy project. A small french foundation, called Des Reves Pour Quentin, contributed \$5K to that total amount. We are incredibly thankful for their support and willingness to support this important project.



Upcoming Events

Marine Corps Marathon Run Arlington, VA - October 22

On October 22, **Missy Longman** will be running the Marine Corps Marathon in Arlington, Virginia, to raise awareness and funding for the SMS Research Foundation.

SMS parents donated \$25 to have their child's name printed on the back of Missy's race shirt (see image at right). She will "carry" our kids through the streets of our nation's capitol as a reminder that we are on this journey together.

If you'd like to support Missy's race to raise awareness and funding for SMS, please visit her [event page](#).



6th Annual 5K Walk for SMS Santa Monica, CA - November 12

Susan and Paul Diamond will be hosting their 6th Annual 5K Walk on November 12, in honor of their son, Jeremy.

This is a fun event for the whole family with a drum circle, face painting, art projects, music, lunch, a silent auction, fire trucks, and more!

For more information, or to register and donate, go to the [5K Walk for SMS event page](#).



Recycle for Research Wilton, CT - through June 2018

Ben and Cole Iannuzzi, in support of their sister Sydney, have started a recycling campaign in their town of Wilton, Connecticut, to raise money for the SMSRF. If you live in the area, please consider supporting their cause by dropping off your empty water bottles so they can recycle them for SMS research.



For pick up or drop off information, contact Ben and Cole at jmiannuzzi@msn.com. You can follow them on Facebook at [Recycle for Research](#) or on the SMSRF [website](#).

Recent Events

Golf Fundraiser

Thank you to **Monica and Bill Mason**, who hosted their 6th Annual Emily Mason Golf Tournament on September 15, 2017 in Atglen, Pennsylvania. Their event raised over \$14,000 for SMS research this year! There were 70 golfers, plus family and volunteers in attendance. The golfers played 18 holes, followed by dinner, raffles and prizes.

Bill Mason is pictured to the right with his daughter, Emily, who is 14 years old with SMS.



Beerfest Fundraiser

The Connecticut Hops For Hope Brewfest took place on September 2, and raised over \$35,000 for SMS research!

Thank you to SMS parents, **Stephen and Hoang Wood**, for all of your hard work organizing such a tremendously successful event! The Brewfest featured more than 36 different craft beers and drew 560 attendees.

Stephen Wood is pictured at the far right with his son, Damian, who is 10 years old with SMS.



Movie Fundraiser

Thank you to the **Iannuzzi Family** for raising over \$25,000 at their Lights, Camera, Research movie fundraiser June 17! The event featured the *Cars 3* movie, and was well attended by friends and local SMS families.

Jennifer Iannuzzi is pictured with her daughter, Sydney, who is 12 years old with SMS.



Comedy Fundraiser

Thank you to the **Frist and Ayres Families** who co-hosted their second annual comedy night at the Pittsburgh Improv on April 8, and raised \$12,600 for SMS research!

Thank you to everyone who sponsored, donated, and attended a night of laughter for SMS!



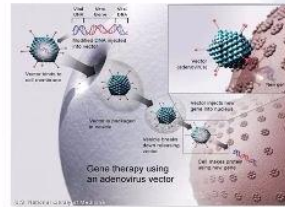
Webinar

Did you miss the webinar explaining our new gene therapy project? Or do you want to watch it again to gain a better understanding?

Our Scientific Advisory Panel members, Chris Iannuzzi and Gianluca Pirozzi, give a detailed presentation of the project.

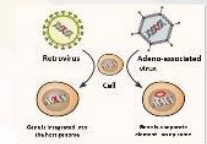
[Watch the webinar here.](#)

Viral Vectors for Gene Delivery



- Viral vectors can be used as vehicles to deliver exogenous genetic material into target cells.
- The genome of the virus is modified to express the gene(s) of interest in the recipient cells.

- Depending on the virus, the gene can either be stably integrated into the host genome or maintained episomally.



**DONATE to SMS
REARCH**

The mission of the SMS Research Foundation is to support research to improve the knowledge and understanding of SMS so that viable therapeutic options can be developed in order to improve the quality of life of those living with Smith-Magenis Syndrome.

The SMS Research Foundation is a 501(c)(3) nonprofit organization founded in June 2010.
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