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USHER III INITIATIVE

Newsletter

Featuring excerpts from Dr. Ed Stone's remarks on Capitol Hill

A Letter from Cindy Elden

Co-founder, Usher III Initiative

Dear Friends,

Greetings! 2015 has been an exciting year at the Usher III Initiative and I look forward to sharing some of the activities that have been keeping us busy. Before I do so, I want to thank you for your continued interest in our mission to find treatments or cures for the blindness caused by Usher syndrome, type 3. We couldn't do what we do without the support of our friends!

In the last newsletter I reported that our researchers and board met in late 2014 and decided on a course forward. The positive results in Usher III Initiative funded labs at Case Western Reserve University, University of Florida and University of California, San Francisco have been most encouraging. The rescue of hearing in Usher3 mice treated with our small molecule and encouraging data from gene therapy testing were signals that we should take the next step to move both of these treatments forward to get them into the clinic and to all Usher III patients!

I am happy to report that our efforts to engage a company to develop the gene therapy treatment have been successful in the early stages and we are encouraged that this will move forward. A firm which specializes in gene therapy is currently in talks with the University of Florida researchers who developed this technology with Usher III Initiative funding. We are very excited about this development as this company has a highly experienced team with an exemplary track record in human gene therapy trials. It is our hope they will not only be best suited to continue refining the gene therapy treatment but will also be best equipped to move this into the clinic and to Usher III patients in the most safe and efficient manner

We are also currently endeavoring to identify the most appropriate, similarly qualified bio-pharmaceutical partner for our small molecule. Earlier this year we engaged the services of Allison Formal who specializes in forging alliances and partnerships to advance treatments and cures for rare disease. She is actively engaging firms in the rare disease, ophthalmic and small molecule space to discuss our unique novel compound and how it potentially can be used to treat Usher III as well as other disease indications. We hope to report exciting news on that front in the very near future.

Cindy Elden

News from the laboratories this year has been both productive and positive. We have two significant publications coming out soon in top industry journals! While we cannot provide details at this time, be on the lookout on our website, Facebook page and Twitter for updates and links to these very exciting journal publications. It is our hope that these highly important articles will raise further awareness about work being done on Usher III and encourage a potential suitor to move forward to partner with us for access to our small molecule compound.

Finally, I reported last year our plans to investigate new areas of therapy for Usher III. In 2015 we have spent a good deal of time researching the restorative therapy and regenerative medicine landscapes for opportunities to develop a therapy for Usher III. It is a search we have taken seriously as we consider various facets of each approach to treatment including risks, opportunities, timing, advantages of the technology, the existing funding landscape, etc. Look for more updates in 2016 as we refine our list and begin to explore steps forward.

If you are wondering how you can help, I would love to remind you about the Usher registry! The Food and Drug Administration (FDA) and potential industry partners will require access to a certain number of patients for clinical trials. An identifiable patient population is important in attracting interest from the pharmaceutical industry and obtaining FDA approval to move into clinical trials.

Our mission is to put this research forward to the industry, and we need your help to move this ahead!
If you have not already, please go to the link below and enroll on the patient registry! www.usher-registry.org

As if getting us through trials quickly wasn't already enough of a reason to register and spread the word about the registry to others, the registry can also help Usher patients and researchers in other ways. Mark Dunning, the Chairman of the Usher Syndrome Coalition, provides a compelling example of how the registry has become an extremely valuable tool to researchers in a short period of time in his recent blog. Thanks to the data in the registry, researchers were able to find sufficient evidence of a connection between Usher syndrome and other illnesses to warrant further investigation.

This is an area where YOU can help! As the year comes to a close, please consider which of your contacts can HELP us in our mission to spread the word. Please forward this newsletter, share our website (usheriii.org), "Like" us on Facebook (www.facebook.com/Usher3Initiative), or Tweet about us on Twitter (www.twitter.com/usheriii). It has been a productive year, and we hope you will continue to support our efforts at the Usher III Initiative.

We wish you and your family a wonderful holiday season and a happy healthy new year.

Thank you for your continued interest and support!

Warm regards,

Cindy Elden

Please visit www.usheriii.org/donate or mail your tax deductible gift payable to the Usher III Initiative to the following:

Usher III Initiative
191 N. Wacker Dr., Suite 2090
Chicago, IL 60606

Ways to get involved!

The Usher III Initiative's top priority is to develop a treatment for patients with Usher III. Because Usher III is an orphan disease that does not affect a large population, it is important that each and every patient join our efforts to prepare for clinical trials. We hope that gathering patient information will help all patients gain access to treatments more quickly.

WE URGE ALL USHER III PATIENTS to take the following steps to be the most proactive patient you can be:

1. **JOIN THE USHER SYNDROME REGISTRY,** which was launched online by the Usher Syndrome Coalition. The patient registry is critically important to clinical trials. You can access the registry at www.usher-registry.org. If you have already registered and would like to help advance the prospects of full participation by all Usher patients in the registry, please share information about it with your contacts. This includes all patients, relatives of patients, caregivers, advocates and support organizations that interact with patients such as schools, clinics and rehabilitation centers. The more data in the database, the more powerful it is in terms of providing valuable information to researchers, funders and government agencies! The database is compatible with JAWS and other readers. It is currently available in English, Spanish, Hebrew, and Dutch. German, Portuguese, French, Swedish versions are in development.

2. **GET A BLOODTEST (GENOTYPING)**. We encourage all Usher Syndrome patients to find out specifically which gene is causing their Usher disease. Many people with Usher have only received clinical diagnosis (this means diagnosis based on your symptoms and not based on a definitive blood test). As a result, some Usher patients have been misdiagnosed as Usher II when a blood test and genotyping may reveal they are actually Usher III. The only way to know for sure is to be screened for one of the many genes that can cause Usher disease. Screening can be done through a medical geneticist locally or through one of the organizations listed on the Genetic Testing Resources page of our website.

We want to thank those of you who have supported our efforts in the past. We are determined to achieve our goal of developing a treatment in the fastest, most efficient way possible and we hope that this newsletter is helpful in outlining how we will be successful!

Edwin M. Stone, MD, PhD

Spotlight on…

Dr. Ed Stone, University of Iowa, Carver College of Medicine

Dr. Stone is a vitreoretinal surgeon with a special interest in hereditary diseases of the retina. He is the Director of the Wynn Institute for Vision at the University of Iowa. In March of this year Dr. Stone was part of a contingent of Usher patients, family and experts who visited legislators on Capitol Hill in Washington DC to make the case for increased federal funding for Usher research. Our partners at the Usher Syndrome Coalition, who recently succeeded in getting Usher Syndrome added as a new category in the NIH categorical spending list for research conditions and disease categories, were integral in setting up this hearing. The following are excerpts from Dr. Stone's remarks to the committee:

On the economics of Usher syndrome research: If a corporate entity can have a business model that will make a safe and effective therapy for Usher Syndrome and if third party payers will pay for it, that is fabulous. Let's go do that. But if there is one of the other types that doesn't fall into the commercial viability spectrum then we have to do that some other way and I think that other is going to be philanthropically assisted academic laboratories at universities.

The importance of genetic testing: A central thing in all these treatments [for Usher syndrome] is genetic testing. The reason for that is that there are a bunch of different subtypes of Usher syndrome and frankly there are a bunch of other different inherited eye diseases that look sort of like Usher syndrome. Many of the treatments that we want to offer our patients depend upon knowing exactly what the person has because the treatment is very mechanistically driven. I just want to update you on where the state of play is for genetic testing of Usher syndrome nowadays. If you got on the web and looked around and called people you would find genetic tests for Usher syndrome ranging from about $450 to more than $7000. If you do everything that you know how to do in 2015, you will find a disease-causing mutation in about 79% of people who have the clinical features of Usher syndrome. Part of that is because there are still some more rare genes to be found and part of that is deafness is common so you can just have deafness and retinitis pigmentosa together accidentally every once in awhile...when I started in this business almost 30 years ago that number was zero. We could not molecularly diagnose a single person.

The work in the Stone lab: Every patient that comes in gets a blood sample and a skin biopsy. The blood sample we use to genotype the person to try to find their gene. We then establish patient derived cell lines from their skin. So what do we use these cell lines for? One thing we use them for is we can actually make retinal tissue, actual living retina... It used to be that if we found a very unusual set of mutations in a patient's sample, something we had never seen before, we would wonder does this thing really harm the retina or not? Sometimes you just couldn't be sure. Now we can actually study the living retina of a patient with an unusual mutation and convince ourselves whether it causes disease or not... now we actually have the cells from the affected individuals growing in the laboratory and we can put the proposed gene therapy into the actual human cells and show that we restore the existence of the protein and sometimes the function.

For people who have lost their photoreceptor cells, we can actually differentiate these cells into retinal precursor cells just like are present in the developing human retina and transplant these into the retina to restore function. So you have people who have lost their photoreceptors and need them back, that's the stem cell transplantation [treatment].

On the other hand, we need to make gene therapies for every one of these subtypes of disease as rapidly as possible so that we can arrest the disease with gene therapy. There will be some people in between who may need both things. They may need some gene therapy to stop the disease they have now but have already lost enough that when this gets really far enough along we want to give them some cells back.

Where do we go from here? Now that we can make photoreceptor cells, we have shown we can put them in via needle in a so-called bolus injection, is that all we need to do? The next steps in our mind are to put some of these cells into people. Of course the people we are going to put them in are completely blind. Why do we want to put cells into completely blind people? Two reasons. Do no harm. We don't want to endanger any vision that someone has now with an unproven technology. Secondly it will be easier to tell whether you've got any sort of response if you don't have any light perception at all and you get restoration of light perception.

In summary: There are a bunch of people out there that were told to go home and go blind. They are out there still. They don't even know they have these diseases. Nobody is talking to them about any of this stuff. We need to go find all of them and genotype them. Then we need to get going with viral mediated gene therapy for the early disease and cell mediated therapy for the late disease.

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Looking Ahead…

- **Email updates**…If you are not currently on our mailing list would like receive updates and exciting newsflashes via e-mail, please visit our website (www.usheriii.org), provide your e-mail address and click on the "Get Notified" button.
- **Facebook**...Another way to stay in touch with us and other Usher III patients and families is via our Facebook page. Please "like" it at this link and you will receive notification of our posts via your newsfeed.
- **Twitter**…we can also be followed on Twitter! Our handle is @UsherIII (https://twitter.com/UsherIII)
- **Support us through your Amazon purchases…** If you shop on Amazon, you can designate the Usher III Initiative as your charity of choice and Amazon will donate a portion of all your eligible purchases to the Usher III Initiative! Click through to smile.amazon.com and follow the steps to designate us as your preferred charity and DON'T FORGET to order through AmazonSmile, in the future!

Contact Us…

We would love to hear from you! Please feel free to contact us with any questions or comments:

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