

PA13-242

SB0465

House	10074-10095	22
Public Health	819-826, 872-889, 919, 973, 1230-1253	52
Senate	2965-2973, 3061-3062	11
		85

H – 1178

**CONNECTICUT
GENERAL ASSEMBLY
HOUSE**

**PROCEEDINGS
2013**

**VOL.56
PART 29
9742 – 10110**

hac/gbr
HOUSE OF REPRESENTATIVES

577
June 4, 2013

Total Number Voting	143
Necessary for Passage	72
Those voting Yea	143
Those voting Nay	0
Absent and not voting	7

DEPUTY SPEAKER RITTER:

The bill is passed in concurrence with the Senate.

Will the Clerk please call Calendar 646?

THE CLERK:

On page 31, Calendar 646, favorable report of the SB 465
Joint Standing Committee on Finance, Revenue and
Bonding, AN ACT REQUIRING NEWBORN SCREENING FOR
ADRENOLEUKODYSTROPHY.

That one year in medical school came in handy.

DEPUTY SPEAKER RITTER:

Representative Johnson.

REP. JOHNSON (49th):

Good evening, Madam Speaker.

I move the Joint Committee's favorable report and
passage of the bill in conformance with the Senate.

DEPUTY SPEAKER RITTER:

The question is acceptance of the Joint Committee's favorable report and passage of the bill in concurrence with Senate.

Representative Johnson, you have the floor.

REP. JOHNSON (49th):

Thank you, Madam Speaker.

This bill would require that newborns be screened for adrenoleukodystrophy; this is a disease. It's a genetic disease that causes severe demyelination of the nervous system and renders its victims completely incapacitated at a very, very young age. A simple blood test will allow the parents and the -- and the medical providers to determine whether or not the infant may be subject to adrenoleukodystrophy and will be able to find a way to treat them.

There's already -- if -- if it's determined that the -- the child has adrenoleukodystrophy before the demyelination of the nervous system then in that circumstance they can remedy the situation and the child will have a normal life.

So I move adoption and with that, I would like to call LCO Number 7708 and be given leave of the Chair to summarize.

DEPUTY SPEAKER RITTER:

Will the Clerk please call LCO 7708, which will
be --

THE CLERK:

Yes --

DEPUTY SPEAKER RITTER:

-- which has been previously designated as Senate
Amendment "A"?

THE CLERK:

Yes, Senate Amendment "A", LCO 7708 introduced by
Representative Johnson, Senator Gerratana, et al.

DEPUTY SPEAKER RITTER:

The Representative seeks leave of the Chamber to
summarize the amendment. Is there objection to
summarization?

Seeing no objection, Representative Johnson, you
may proceed with summarization.

REP. JOHNSON (49th):

Thank you, Madam Chair.

Originally the bill produced a fiscal note. With
the amendment the fiscal is gone away and the -- the -
- the test will be occurring once the Food and Drug
Administration comes up with an approval for test for
adrenoleukodystrophy using dried blood spots by the --
and -- and -- and/or the availability of any other

tests that may be available through the provider institution.

I move adoption.

DEPUTY SPEAKER RITTER:

The question before the Chamber is adoption of the amendment.

Will you remark on the amendment?

Representative Arce, you have the floor, sir.

No. No. You have the floor? No.

Representative Reed. Do you care to remark on the amendment?

REP. REED (102ND):

Yes. Thank you, Madam Chair.

I just want to say this is really very, very important legislation. The family that brought this condition to our attention has an extraordinary who is really incredibly disabled at this point and this loving family has used their tragedy to really tell the world that this could have been avoided had this child been tested at birth.

And Gene and -- I'm saying hi to them because they have been watching and they have been -- they have really learned the legislative process.

Gene and Jack, this is a legacy for Brian. We're really proud of you. We're really honored to be able to have this legislation.

And I want to say to the Public Health Committee, to the Chairs, to the members, thank you so much for making this a committee bill and for your kindnesses to, not only the Kelly family, but to all the fine families who testified.

Thank you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Srinivasan, you have the floor, sir.

REP. SRINIVASAN (31st):

Good evening, Madam Speaker.

I rise in strong support of this bill as -- of the amendment, which obviously will become the underlying bill. We heard in the testimony and we heard in the public hearings how critical -- how essential it is to make the early diagnosis and the big difference -- huge difference between early diagnosis and not picking this up as early as you can.

So ask -- for us to look at this test when it becomes available and make sure that we offer it to our constituents -- to our state is critical in the --

a step in the right direction -- very positive direction in the early diagnosis of this genetic disorder.

Through you, Madam Speaker.

Just one or two questions to the -- to the -- to the -- to the -- to the proponent of the amendment, right. Yep to the proponent.

DEPUTY SPEAKER RITTER:

Please proceed.

REP. SRINIVASAN (31st):

Yep.

Thank you, Madam Speaker.

We heard in public testimony that there was some lab in Maryland -- if I remember correctly -- memory serves me right, that do -- they do have this test and the test was something that they could market and bring to our state. Is this the same test that we are talking about, which is the dried blood spot test?

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Johnson.

REP. JOHNSON (49th):

Through you, Madam Speaker.

This is -- this is the test and the -- I believe you're correct it was in Maryland and -- and they do have a test that's been developed, so this is something that perhaps could be done.

But we added some language with respect to that, including the Food and Drug Administration because I believe Maryland actually did pass a law -- one of the first states in the country to have this test in conformance with the Food and Drug Administration's recommendations, and that was how we were able to limit our fiscal note.

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Srinivasan.

REP. SRINIVASAN (31st):

So through you, Madam Speaker.

If I hear this clearly that this test is already available A, in Maryland and "B", has been approved by the FDA?

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Johnson.

REP. JOHNSON (49th):

Through you, Madam Speaker.

This is in conformance with FDA approval.

DEPUTY SPEAKER RITTER:

Representative Srinivasan.

REP. SRINIVASAN (31st):

Through you, Madam Speaker.

I -- the background noise I couldn't hear the
answer.

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Johnson, would you please repeat
your answer.

REP. JOHNSON (49th):

The -- the amendment puts the requirement for the
test in conformance with the Food and Drug
Administration's requirements.

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Srinivasan.

REP. SRINIVASAN (31st):

Through you, Madam Speaker.

I do understand that it is pending the approval,
but the test that is available in Maryland and that
has already been in use there is that test -- has that
test been a FDA approved test?

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Johnson.

REP. JOHNSON (49th):

Through you, Madam Speaker.

This would be -- there's nothing in this legislation at this point in time that says that the FDA has approved the test. It just says that when they do provide the language that says that they approve this then we will be providing in conformance with their recommendations.

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Srinivasan.

REP. JOHNSON (49th):

Thank you, Madam Speaker.

Knowing the importance of early diagnosis, as we heard from the other Representative as well, and we heard that in the public hearing. What my concern is if this test is already there in Maryland and obviously they would be using something that's only FDA approved, and what is it that is holding us from offering this to our constituents here to -- in our state?

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Johnson

REP. JOHNSON (49th):

Through you, Madam Speaker.

It is -- it is not -- it's -- at this point in time, not a requirement, hence there is no fiscal note. So that is -- that was the object of the change.

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Srinivasan.

REP. SRINIVASAN (31st):

So through you, Madam Speaker.

My -- for us to make it available as a mandate to newborn screening, what would be the next step since it is already available and it is FDA approved? So that is where I'm confused. If there is already there then what is it that we are -- that is holding us back in offering us -- offering this test, which is so critical?

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Johnson.

One moment please, Representative Johnson.

If members of the Chamber could please either take their conversations outside or keep them to a low enough volume that the other members can hear the questions and the answers, it would be appreciated.

Representative Johnson, you may proceed.

REP. JOHNSON (49th):

Thank you, Madam Chair.

The language allows us to move forward just as soon as all the federal and state law is -- is put together in a way -- this is relatively a recent -- a recent test that's been developed and it's just come to the state's attention.

This was not something that was -- that the -- that the Commissioner of Public Health was really aware of and so now we have something with the FDA making the recognition, but in order to make it a requirement we need to make sure that all these things are falling into place first, so we have some procedural things -- some things that need to be ironed out, so we want to make sure this takes effect, and as soon as all these things are ready to go, that these tests will be available.

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Srinivasan.

REP. SRINIVASAN (31st):

Through you, Madam Speaker.

My final question is when this test to make it happen obviously we do have to look at it from a fiscal note point of view, and I know there was some fiscal analysis in the -- in the -- in the bill that has now been, you know, it's -- it's been stricken out.

There's no longer a bill in -- in the amendment, so do we have an idea as to when the fiscal note will be available, or any idea as to what they are charging in the other state, so that we can start preparing and planning, when the test becomes available, meeting all those guidelines, that we can offer it as soon as possible?

Through you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Johnson.

REP. JOHNSON (49th):

Through you, Madam Speaker.

The fiscal note contemplated the fact that the state would have had to do all of the work that the

FDA is now doing. So that is -- that is why the -- the fiscal note was there. A lot of those things have been -- will be addressed through the federal government and we'll be able to proceed.

Through you, Madam Speaker.

Representative Srinivasan.

REP. SRINIVASAN (31st):

Thank you, Madam Speaker.

I do want to thank the Chair for her answers. And I sincerely hope, Madam Speaker, that this test, which is available in our -- in -- in -- in Maryland -- a test that is critical in early diagnosis so that the lives of these children with this genetic disorder are improved because of -- and -- because of early diagnosis that we will take it upon ourselves to make it available as soon as we can.

Thank you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Widlitz, you have the floor.

REP. WIDLITZ (98th):

Thank you, Madam Speaker.

Good evening to you.

DEPUTY SPEAKER RITTER:

Good evening.

REP. WIDLITZ (98th):

Madam Speaker, I rise in strong support of this amendment, which becomes the bill. The public hearing we had before the Public Health Committee was -- was very emotional. Families shared what they have been through this -- the -- the trials and tribulations that they have gone through in trying to take care of their children.

Had they known about this disease. Had they known that there would be a test that would become available they could have avoided some of the -- the suffering of their children.

Unfortunately, by the time the symptoms appear it is, in many cases, too late to treat this disease with any success, so it's very important that as newborns the newborns get the screening so that they can take advantage -- the families can take advantage of all of the treatment that is available, and make a dramatic difference in their children's lives.

It's a disease that affects little boys, although women carry the gene. And the -- up to now really the only way that people were aware that this could happen to their children is if they had another member of their family have the disease and then they had the

genetic testing. So it's -- when this test becomes approved and widely available, this will make an enormous difference for families in being able to care for their children.

And I would just like to give a shout out to the Kelly family from Branford and Gene has been watching CTN and waiting for this bill to come out, so Gene, tonight is your night and we all wish for a quick signature on this bill so we can get moving with helping children.

Thank you very much, Madam Speaker.

DEPUTY SPEAKER RITTER:

Will you remark further on this amendment?

Representative Klarides, you have the floor, ma'am.

REP. KLARIDES (114TH):

Thank you, Madam Speaker.

I, in addition to my colleagues, also rise in strong support of this amendment. You know, it's -- it's a rare occasion that we in this legislature, or as -- as a state in general, are able to do something costs so little, yet makes such a big difference in somebody's life. And educate people to know that all

they need is to go get a simple test and they are -- their lives change dramatically.

And so that's what this bill does. And that's why I'm very proud to have been part of it and support it. And I urge my colleagues to do so, but I have to add one thing to Representative Widlitz comment.

It's not just Gene that's home watching, it's the whole Kelly family, so hello to all of you and thank you for your strength and your family that has allowed us to find out about things like this and be able to help other families.

Thank you.

DEPUTY SPEAKER RITTER:

Thank you.

Representative Sayers, you have the floor.

REP. SAYERS (60TH):

Thank you, Madam Speaker.

I also rise in strong support of this amendment. And I want to give a shout out to the Florian family, who lost their son Joshua to this disease. And I think that what we are doing tonight is going to save so many lives. The treatment is so simple and so easy, only if we know about the disease before the symptoms begin.

The Florian children are friends of my grandchildren, which is why I got to know them. So thank you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Candelora, you have the floor, sir.

REP. CANDELORA (86TH):

Thank you, Madam Speaker.

I also rise in support of this bill. Bills like this truly make us humble. And I think it's a privilege to serve in this General Assembly when we address issues in this -- of this nature. Many of us had the pleasure to meet a very extraordinary family that undertook some extraordinary life events as a result of an illness that they were faced with.

And they brought their information to us to Hartford and become wonderful advocates, very understanding, learned about the process, worked with the legislature and in these difficult economic times came up with a solution that will produce a better quality of life for many of our residents in Connecticut.

And this natal screening is going to go a long way in making sure that this illness never becomes

symptomatic for any family in the state of Connecticut. And so I very proudly stand to support this bill with the rest of my colleagues.

Thank you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Simanski, you have the floor, sir.

REP. SIMANSKI (62ND):

Thank you, Madam Speaker.

I also rise in very strong support of this bill and this is -- if you want to read a story that tugs at your heart, like Representative Sayers, I would ask you to read the testimony of my neighbors Lee and Elizabeth Florian.

Read how their 2-year old son came down with a fever. He got up in the morning sitting in his father's arms drinking some water and he stopped breathing. Shortly thereafter he was taken to the hospital. His three siblings kissed him good-bye. His mom and dad unhooked him from the respiratory and he died in their arms.

And then they went on a four-year quest to try to figure out what was the cause of Joshua's death. And they finally found out it was ALD and that for the fact that he could have been diagnosed if there was a

test available at the time he would still be with us today. They write in their testimony, now our only hope is that you can feel his story leading you to do all you can to pass Bill 465 and protect the children of Connecticut.

So I would urge everyone in this -- in this House her to please vote yes for this bill, so that the Florian's can say Joshua they heard your story and because of you there are other children who will now be alive.

Thank you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Representative Kokoruda.

REP. KOKORUDA (101ST):

Thank you, Madam Speaker.

I also stand in support of this bill. All of us that live in the Shoreline certainly have met the Kelly family. And we've heard tonight of other families also. The day they came to Hartford there was another mother with her son there that was fortunate enough to have been screened because, as Representative Widlitz said, it had been in their family and they had their son screened -- had the

screening and to think that he's going to have a full and wonderful life.

So the Kelly's really stepped up and tonight we get the opportunity to do the same.

Thank you, Madam Speaker.

DEPUTY SPEAKER RITTER:

Will you remark further on the amendment? Will you remark further on the amendment?

If not, I will try your minds. All favor, please signify by saying, aye.

REPRESENTATIVES:

Aye.

DEPUTY SPEAKER RITTER:

Opposed, nay.

The ayes have it and the amendment is adopted.

Will you remark further on the bill as amended? Will you remark further?

REP. JOHNSON (49th):

Yes.

DEPUTY SPEAKER RITTER:

Representative Johnson.

REP. JOHNSON (49th):

Thank you, Madam Speaker.

I just wanted to thank my colleagues here in the House for their good remarks and also Representative Widlitz for the idea of the amendment. It really helped move this bill forward and so I appreciate everybody's support and I look forward to saving the children of the state of Connecticut.

Thanks.

DEPUTY SPEAKER RITTER:

Will you remark further on the bill as amended?
Will you remark further?

If not, will staff and guests please come to the Well of the House? Will members please take their seats; the machine will be opened.

THE CLERK:

The House of Representatives is voting by roll.
The House of Representatives is voting by roll. Will members please return to the Chamber immediately?

DEPUTY SPEAKER RITTER:

Have all the members voted? Have all the members voted?

Will the members please check the board to determine if their vote is properly cast?

If all the members have voted, the machine will be locked and the Clerk will take a tally.

The Clerk will please announce the tally.

THE CLERK:

In concurrence with the Senate, Senate Bill 465
as amended by Senate "A"

Total Number Voting	142
Necessary for Passage	72
Those voting Yea	142
Those voting Nay	0
Absent and not voting	8

DEPUTY SPEAKER RITTER:

The bill is passed in concurrence with the
Senate.

Will the Clerk please call Calendar 287?

THE CLERK:

On page nine, Calendar 287, favorable report of
the Joint Standing Committee on Environment,
Substitute House Bill 5027, AN ACT PROHIBITING THE
SALE OF DOGS AND CATS OBTAINED FROM SUBSTANDARD
DOMESTIC ANIMAL MILLS AND REQUIRING A STANDARD OF CARE
APPLICABLE TO ANIMAL IMPORTERS.

Another rough one.

DEPUTY SPEAKER RITTER:

Representative Gentile.

REP. GENTILE (104th):

S - 661

**CONNECTICUT
GENERAL ASSEMBLY
SENATE**

**PROCEEDINGS
2013**

**VOL. 56
PART 10
2837 - 3149**

cah/med/gbr
SENATE

25
May 23, 2013

to be followed by Calendar Page 45, Calendar 488, Senate Bill 1153 and then Calendar Page 15, Calendar 489, Senate Bill 871 and then, Madam President, have two additional goes to mark, Calendar Page 2, Calendar 49, Senate Bill 523, Calendar Page 43, Calendar 400, Senate Bill 1137.

Thank you, Madam President.

THE CHAIR:

Thank you, sir.

Mr. Clerk.

THE CLERK:

On Page 36, Calendar 152, Senate Bill Number 465, AN ACT REQUIRING NEWBORN SCREENING FOR ADRENOLEUKODYSTROPHY, Favorable Report of the Committee on Public Health.

THE CHAIR:

I'm impressed.

Sen -- Senator Gerratana.

SENATOR GERRATANA:

Thank you, Madam President. I gave him a good thumbs up.

THE CHAIR:

There you go, absolutely.

SENATOR GERRATANA:

Madam President, I move acceptance of the Joint Committee's Favorable Report and passage of the bill.

THE CHAIR:

Motion is on pass -- accept -- passage and acceptance.

Will you remark, Ma'am?

cah/med/gbr
SENATE

26
May 23, 2013

SENATOR GERRATANA:

Yes, thank you, Madam.

This bill adds Adrenoleukodystrophy to the list of generic and metabolic diseases for which hospitals and other institutions must test newborns under the Department of Public Health's Newborn Screening Program.

Madam President, because of much discussion around the legislation, many of us Legislators did come together and we came up with a new version of the underlying bill so if the Clerk would please call LCO 7708 and I be allowed to summarize.

THE CHAIR:

Mr. Clerk.

THE CLERK:

LCO Number 7708, Senate "A", offered by Senator Gerratana, et al.

THE CHAIR:

Senator Gerratana.

SENATOR GERRATANA:

Thank you, Madam President.

I move adoption.

THE CHAIR:

Motion is on adoption.

Will you remark, Ma'am?

SENATOR GERRATANA:

Yes, Madam President, thank you.

cah/med/gbr
SENATE

27
May 23, 2013

This is a strike-all amendment and there has been much discussion in our Legislature about this horrible disease, a genetic disease that can strike particularly young men usually around six, or seven or eight years old from -- young boys I should say in Connecticut and around our country.

It is essential to diagnose ALD early, as early as possible, as soon -- as close to the birth of a child as you possibly can. All the evidence indicates that the two currently available treatments and you may have heard of Lorenzo's Oil, there was a famous movie on -- that was made regarding a young boy and his parents and how they found a way to treat it and also stem cell implantation but the only way to know whether Adrenoleukodystrophy is present is through newborn screening.

Now the amendment before us we decided to come up with a way because there were some challenges to requiring this in our state. Right now there is no feasible way to do the testing and the testing still has to go through somewhat of a process including the inclusion on the -- I'm getting the correct designation, the recommended uniform screening panel approved by the federal government.

So because we felt compelled after meeting Mr. -- Dr. and Mrs. Kelley I should say and their extraordinary son Brian, we thought that it would be very, very appropriate to work out a way and we did so with the amendment. So there are some conditions starting on line 39, on and after the occurrence of the following, and then once they are met, then this law can be enacted and we can do the newborn screening.

You know I talk a little bit about the technicalities of this amendment and what it will do and the work that went into it but I must say that in Public Health Committee this year we have had many people who have come before us, we had many long exhausting hearings, but hearing from the families that are affected and seeing their children who came up with mom and dad in most cases to testify we were extremely moved and worked very hard to get this enacted.

Thank you, Madam President.

cah/med/gbr
SENATE

28
May 23, 2013

THE CHAIR:

Thank you.

Will you remark?

Senator Fasano.

SENATOR FASANO:

Thank you, Madam President.

I'd like to thank Senator Gerratana for working on this bill to achieve the goal that we have in front of us as well as Senator Meyer and other folks in the -- in the House, particularly Representative Sayers, Representative Klarides, Representative Widlitz and Representative Reed because it took a lot of us to get together to figure out how to do this the best way.

Jack and Jean Kelley I have known because I went to college with Jack and played football with Jack and when Jack was dating his wife Jean at a nursing school in Vermont some weekends we spent on a road trip visiting them up at the nursing school and so I got to know them very well. They went to my wedding and I went to theirs and our kids are roughly the same age and they're excellent people and Ryan is a very special individual.

And Ryan was diagnosed as a result of a sledding accident in which he hit his head and they did an MRI and on the MRI that image appeared. But for that sledding accident Ryan may not be alive today. And there was another person who came to testify who was a doctor and when her son was born she had a cousin who had this disease so she knew to test her child.

Her child was tested early on, received all the appropriate treatment. Now her child is the captain of a soccer team, is on his way to a prestigious university and graduated with high honors. That cousin died at the age of 13.

What this tells you is the disease is curable, not in the sense of being curable that a person lives an

cah/med/gbr
SENATE

29
May 23, 2013

adequate life, I'm talking about 100 percent curable and a test done at birth gives the opportunity to cure it and it doesn't cost millions and millions of dollars to cure it once you find it.

You take some medicine that I'm not precise in what it is and you have a series of MRI scans and then when you see that the brain is starting to change, you go back on the medicine and it clears up. It's amazing. It's amazing because these people can be cured. About two children every year in Connecticut are born with this disease and if not detected, they will die before they reach 20.

In particular a lot of times it's misdiagnosed as other diseases, as other behavioral issues, and that turns out to be a problem. We had a parent who came up and said their child was diagnosed with a behavioral issue and literally she was driving home and her son's head dropped. She saw that in the backseat, brought her child to a neurologist who diagnosed this very disease. The child is alive but his prognosis is not good for the next few years.

So we can cure it. So this is one of the rare bills that come in front of this Chamber that we can make an absolute definitive difference in someone's life.

I'd also like to con -- to thank the Commissioner of Public Health. She was kind enough to entertain us at a tour of her facility, meet with the scientists, talk the issue out, come up with a bill that put triggers in it such that when this is approved by medical scientists in terms of an absolute test, the State of Connecticut be one of the first to jump on it and say we're saving lives.

I want to thank this Chamber. I want to thank Senator Gerratana. I want to thank Senator Meyer here in the Chamber for their hard work. I want to thank Senator Welch for allowing me to bring the bill in front of the Public Health Committee. I saw Senator Welch early on, talked to him about the issue, he brought it to the Committee, they got a public hearing as a result of -- of his argument in front of the Committee of a disease that no one really knows about so I want

cah/med/gbr
SENATE

30
May 23, 2013

to thank him for his belief in me and belief in this -
- in this bill.

Thank you, Madam President.

THE CHAIR:

Thank you.

Will you remark? Will you remark?

Sen -- Senator Kissel.

SENATOR KISSEL:

Thank you very much, Madam President, great to see you
this afternoon.

THE CHAIR:

Same here, sir.

SENATOR KISSEL:

I would like to commend Senator Fasano and Senator
Gerratana, Senator Welch and all the others, Senator
Meyer for participating and allowing this to go
forward.

Back in February 27th of this year two of my
constituents from Granby, Elizabeth and Lee Florian,
came and testified, submitted testimony about their
beautiful, funny, charming and dashing and handsome
blond little boy named Joshua Seth Florian and as it
turned out unfortunately there was no screening to
test Joshua for Adrenoleukodystrophy.

And so what they did is they said had they had the
screening available, as this bill will allow going
forward, they wouldn't have lost their little boy at
two years two months and two days. It takes a lot for
a mom and dad to submit testimony and -- and come to
this building and -- and testify. It takes an awful
lot to come and testify about the loss of your child.

And there's not a single blessed thing that anybody in
this Circle can do to bring that little boy back but

cah/med/gbr
SENATE

31
May 23, 2013

there is one thing we can do this afternoon. We can remember then in the heart of winter on a February cold day these folks came and said can we make it better for the people going forward so that when they have their own little Joshua Seth or their own little girl that a fairly simple process can be employed to screen for this disease and as was stated on the record that the magical marvels of medicine can be brought to bear to try to save lives.

And when you get right down to it, we pass a lot of bills, we add to the statute books but if some of those things can directly result in a mom or a dad's heart not being broken because they've lost a two year old, then we've had a good day.

And so I'm proud to stand shoulder to shoulder with the Florians. I commend them for coming to Hartford and testifying in favor of this bill. Senator Gerratana just spoke to me and she recalled that once upon a time when we were in the majority, I was the Vice-Chair of the Public Health Committee and she wondered if I missed the Committee and I said there are many times I do, there are many times I do.

I think that Committee and I think this Chamber can stand proudly this afternoon and for those reasons, for Elizabeth and Lee Florian and to the memory of their two year old son Joshua Seth Florian, I am proud to stand in support of this legislation.

Thank you, Madam President.

THE CHAIR:

Thank you.

Will you remark further?

Senator Welch.

SENATOR WELCH:

Thank you, Madam President.

I -- I don't think there's much to add just to the compelling testimony and heartfelt stories that were

cah/med/gbr
SENATE

32
May 23, 2013

shared before me by Senator Gerratana, Fasano and Kissel. Just to say that it's -- you often find yourself in a difficult place when the establishment of medicine is slow to embrace what are some very obvious answers and solutions and so I'm grateful that we can all stand here today and -- and encourage that and make a difference and, you know, when you're a -- a State Senator or a State Legislator and you come to Hartford and you're asked, you know, what difference have you made? What have you done to make Connecticut a better place?

Bills like these are the ones that are at the top of the list and so I'm thankful and humbled and honored to have been a part of this process and I want thank Senator Fasano just for bringing this to everybody's attention and of course Senator Gerratana for leading the charge.

Thank you, Madam President.

THE CHAIR:

Thank you.

Will you remark? Will you remark?

If not, I'll try your minds. All in favor of Senate "A", please say aye.

SENATORS:

Aye.

THE CHAIR:

Opposed?

Senate "A" is adopted.

Senator Gerratana.

SENATOR GERRATANA:

Thank you, Madam President.

cah/med/gbr
SENATE

33
May 23, 2013

If there's no objection, I'd like to move this item to the Consent Calendar.

THE CHAIR:

Seeing no objection, so ordered.

Mr. Clerk.

THE CLERK:

On Page 45, Calendar 488, Substitute for Senate Bill Number 1153, AN ACT CONCERNING CONTRACT COMPLIANCE REQUIREMENTS FOR THE METROPOLITAN DISTRICT OF HARTFORD COUNTY, Favorable Report of the Committee on Judiciary.

THE CHAIR:

At this point I will call on Senator Coleman.

Good afternoon, sir.

SENATOR COLEMAN:

Good afternoon, Madam President, thank you for your patience and your recognition.

I'd like to move acceptance of the Joint Committee's Favorable Report and passage of the bill.

THE CHAIR:

The motion is on acceptance and passage.

Will you remark, sir?

SENATOR COLEMAN:

Madam President, Metropolitan District is an entity that exists in the City of Hartford and it's currently involved in a \$2 billion clean water project. Many of the streets of the City of Hartford are being renovated. The whole project consists of separation of the stormwater sewer from the sanitary sewer.

cah/med/gbr
SENATE

121
May 23, 2013

Mr. Clerk.

THE CLERK:

On Page 2, Calendar 49, Senate Bill 523; Page 15,
Calendar 489, Senate Bill Number 871.

On Page 35, Calendar 44, Senate Bill Number 809; on
Page 36, Calendar 152, Senate Bill 465.

On Page 37, Calendar 177, Senate Bill 972 and on Page
40, Calendar 293, Senate Bill 814.

Page 41, Calendar 359, Senate Bill 1099 and Calendar
377, Senate Bill 889.

On Page 43, Calendar 400, Senate Bill 1137 and on Page
45, Calendar 488, Senate Bill 1153.

THE CHAIR:

Thank you.

Please announce that the machine is open on the first
Consent Calendar.

THE CLERK:

Immediate roll call has been ordered in the Senate.
Senators please return to the Chamber. Immediate roll
call on today's Consent Calendar ordered in the
Senate.

THE CHAIR:

Have all members voted? If all members have voted,
please check the board to make sure your vote is
accurately recorded. If all members have voted, the
machine will be closed and the Clerk will announce the
tally.

THE CLERK:

Today's Consent Calendar.

Total Voting

36

cah/med/gbr
SENATE

122
May 23, 2013

Voting Yea	36
Voting Nay	0
Absent, not voting	0

THE CHAIR:

Consent Calendar 1 passes.

Senator Looney.

SENATOR LOONEY:

Thank you, Mr. President.

Mr. President, before moving to the item which will be marked for the order of the evening, I believe the Clerk is in possession of Senate Agenda Number 2 for today's session.

THE CHAIR:

Mr. Clerk.

THE CLERK:

The Clerk is in possession of Senate Agenda Number 2. It's dated Thursday, May 23, 2013. Copies have been made. They are on Senators' desks.

THE CHAIR:

Senator Looney.

SENATOR LOONEY:

Thank you, Mr. President.

Mr. President, I move all items on Senate Agenda Number 2 dated Thursday, May 23, 2013 to be acted upon as indicated and that the Agenda be incorporated by reference into the Senate Journal and the Senate Transcript.

THE CHAIR:

So ordered.

**JOINT
STANDING
COMMITTEE
HEARINGS**

**PUBLIC
HEALTH
PART 3
681-1010**

2013

from Michelle Obama to the NFL Play60 program is looking for opportunities to get our kids outside.

SENATOR GERRATANA: Thank you, Madam. Thank you for your diligence. I know last year we did pass legislation that requires, I think it's 20 minutes of physical activity in our school system which is absolutely appropriate. But I thank you very much.

Are there any other questions? If not, thank you for coming today.

And, again, just for some consideration, I'd like to call Senator Fasano up. He has with him a family with children. I think it's Jean and Jack Kelley. Senator Fasano.

SENATOR FASANO: Senator Gerratana, Representative Johnson, Representative Miller, Representative Sri (sic) -- I can't say it right. Doc, I'll call you Doc.

SENATOR GERRATANA: Srinivasan.

SENATOR FASANO: Srinivasan. I just want to thank you for giving us the consideration allow us to go out of order. I appreciate it. This is regarding Committee Bill number 465, a bill regarding Adrenoleukodystrophy otherwise known as ALD.

I have with me Dr. Jack Kelley and his wife Jean Kelley, as well as their son. Brian. I've known Jack all my life, all my adult life. We met each other in college and played ball together.

Their son, Brian, had -- has ALD. And the thing about ALD is that it was diagnosed until after the age of 5. Prior to that

time, Brian was a healthy boy who skied, did activities, and you would never know he had the disease.

The reason what or what this bill does, it asks to have this Legislature have a bill passed which requires testing of newborns of this disease. If it is caught early, there are other things you can do to offset some of the impacts from the disease. Rather than me sort of reiterating what the family has told me, I would ask the indulgence of the Committee and of the Chairs and ranks to allow Jean and Jack Kelley to tell you why it is important.

They started an organization called Brian's Hope to bring awareness of this situation. The awareness of the new testing. I will tell you in New Jersey is looking at the issue in their legislative session this year. Other states have done it. And if it would indulge the Chair, I will turn the microphone over to Jean Kelley.

SENATOR GERRATANA: Thank you. Yes. We do have for members of the public, a three minute time limit. And, so, we will be doing that. But, please, proceed.

JEAN KELLEY: Okay. Thank you very much, Senator Gerratana, Representative Johnson, and Public Health Committee members. Thank you for providing us with the opportunity to appear here today. We are honored to speak in support of this vital bill.

We are especially excited at the possibility of making a change in the outcome of the lives of boys who have Adrenoleukodystrophy in their families.

Early detection of Adrenoleukodystrophy will help avoid undue suffering and the enormous emotional and financial costs associated with the treatment and care of the full onset of the disease. That is possible now. We have a test that detects ALD from newborn blood spots.

Brian was diagnosed with the cerebral demyelization form of ALD at the age of 6 after suffering a sled riding accident with a head injury. While sledding one day, he hit his head on a wood pile. At the Yale ER, a CT Scan and follow up MRI led to Brian's diagnosis. Within six months of Brian's diagnosis, he lost his mobility, speech, ability to eat, and most of his vision.

Since there was no newborn screening at the time of Brian's birth and, although, this is an X-linked hereditary disease; there were no red flags in our family. Had his condition been detected at birth, early intervention would have, likely, altered the outcome. It is truly a parents' worst nightmare. We know this. Our experience with our warrior, Brian, has taught us much.

Despite his challenges, there is much Brian is able to do. In his very quiet way and with his tenacity to persevere, he has inspired us to move on and impact others in a positive way such as this.

Adoption of ALD newborn screening with be Brian's legacy so his many difficulties in life were meant to help others and were not in vain. Early detection is crucial. The misdiagnosis of newborns with ALD will most importantly result in the needless suffering and loss of lives. At best, ALD results in

neurological impairment, developmental delays, long-term feeding issues, and costly therapies. There's my bell.

It is completely within our power to keep babies born. And I wish you -- urge you, your leadership in expertise and public health to support this important bill.

SENATOR GERRATANA: Mrs. Kelley, thank you so much. We did raise it as a Committee bill. We feel that after hearing from you and meeting with you, that this is -- and Senator Fasano, of course, that this is an issue that our Committee should take up. So, I think it's well on its way, if you will. And I had the pleasure of meeting with you and also with Brian.

Do we have any questions?

Yes. Senator Bartolomeo.

SENATOR BARTOLOMEO: Thank you. And thank you for being here today.

Can you explain to us with early detection what then changes? What's the intervention?

JOHN KELLEY: John Kelley, Brian's father from Branford. If you pick it up at birth and you follow the boys and you treat them with a special diet including Lorenzo's Oil which some of you may have seen in movies in the past and a special diet, over 70 percent can remain asymptomatic. And that was done in a study at Johns Hopkins. And one of our testimony -- some of the testimony later is going to be from Ann Moser who runs the lab there. And her husband who has passed away did a lot of the clinical stuff in the country and in the world.

The newborns would get MRIs every six months. And if in the percent it becomes symptomatic meaning you find something on the MRI, but they still see normal, they can get a bone marrow transplant and the vast majority of them will live good lives without the problems.

So, it's not only easy to do because they get the blood spots already. Obviously, there's a cost involved which you people know more about than I do. But the other end of it is just Brian, alone, and this has been in the testimony submitted to you folks. Brian, alone, from I think sixth grade through high school between the Federal Government, the State of Connecticut, the Town of Branford spent almost \$1 million for his education. And he's one boy. And you take that to the number of people that have it that don't even get diagnosed, the health costs, that doesn't include Medicaid, Husky-type costs. We have a woman testifying later with a son with ALD that was not caught early enough. And she can tell you the number of hours and nursing she has authored also through statement.

SENATOR BARTOLOMEO: Thank you. And I commend you for your courage to be here today. Thank you.

SENATOR GERRATANA: We all thank you and thank you for bringing this issue up.

Senator Kane.

SENATOR KANE: Thank you, Madam Chair.

In your testimony you said boys. It only

affects boys?

JEAN KELLEY: Yes. It's genetically passed. Boys have a 50/50 chance. We have an older son who is healthy. He's 29. And our daughter that we pass on our trait to our daughter, our daughter is a carrier as well. So, it manifests in boys, but it is passed through woman.

JOHN KELLEY: I think the easy explanation, so, you have to have it somewhere in your family either in the past or a mutation. And once the mother has it, the enzyme that Brian lacks is only on the X chromosome. So, boys only get one copy of that. So, if they get the copy of it that mom doesn't have it, that's how they get the disease.

SENATOR KANE: And how many people does this affect? I don't know if you said that?

JOHN KELLEY: One in 17,000 births.

SENATOR KANE: Thank you. Thank you very much. Thank you, Madam Chair.

JOHN KELLEY: Senator, there's 37,000 births a year -- roughly, 40,000 births a year in Connecticut.

JEAN KELLEY: And some who do have Adrenoleukodystrophy it wasn't found. We lost a boy in our state a few years ago. And it wasn't found until several years later. And you're going to hear testimony from his mom. So, it's more frequent than that.

SENATOR GERRATANA: Thank you. Representative Widlitz.

REP. WIDLITZ: Thank you, Madam Chair. And thank you so much for being here today.

I think the whole town of Branford is solidly behind Brian's Hope. I've heard from so many people. And it's such a credit to you and your family to have this outpouring of response to this bill.

I really had never heard of this disease before. And I assume that most of the Committee members really were not familiar with it. So, it's rather startling to know that there is a test that could actually avoid a lot of the suffering that your family has gone through. So, we hope we'll take this one all the way. And give you great credit and encouragement for continuing because you've really done a wonderful thing for children who will be born into the future and who knows what you've actually accomplished by bringing this to light today.

JEAN KELLEY: Thank you.

REP. WIDLITZ: So, thank you so much.

JEAN KELLEY: Brian is sleeping. But he knows what we're doing and he knows we're trying to save lives of other boys and keep other boys from suffering such as he. So, thank you very much for the opportunity.

SENATOR FASANO: Madam Chair, if I may. We have other people signed up. You'll hear them later. But what we tried to do is there's a few parents here, but there are also some experts in the area of the testing, what it means, the cost et cetera, so that the idea that we were trying to do is not to repeat story after story, but to give the Committee

valuable information so they could get a better handle on how the tests are done, the costs of the tests, and what other states have done.

SENATOR GERRATANA: Good. And I'm sure we'll hear from those individuals which will also be very helpful.

Dr. and Mrs. Kelley, thank you so much for coming today. There's one person who would like to ask you some questions, also, and that's Rep -- oh, no, I'm sorry. He's all set. I think our Committee is too. Thank you for coming.

JEAN KELLEY: Thank you very much.

JOHN KELLEY: Thank you.

SENATOR FASANO: And thank you, again, for the courtesy.

SENATOR GERRATANA: Absolutely.

Next is Representative Brenda Kupchick.

REP. KUPCHICK: Thank you very much for the opportunity to the Chairs, Vice Chairs, and ranking members.

I, actually, am here to just talk about a bill that I submitted, an internship program for autistic adults. It's 5360. I submitted testimony from a constituent in my district who brought this issue to my attention of the lack of opportunities for autistic adults to have internship programs in different areas so that they can learn other things.

But I also, with your indulgence, wanted to

HB5240

much all the research that you are doing and taking proactive stands here on the issue. We certainly thank you for that.

If there are no further questions.

REP. SANCHEZ: And I just wanted to state the doctors have -- could not make it today, but they will be submitting written testimony.

SENATOR GERRATANA: Oh, good. Thank you, again.

REP. SANCHEZ: Thank you.

SENATOR GERRATANA: Next is Eliza Florian. Are you coming up with Lee, also?

LEE FLORIAN: Yes.

ELIZABETH FLORIAN: Yes.

SENATOR GERRATANA: Okay.

ELIZABETH FLORIAN: If that's okay.

SENATOR GERRATANA: Just -- oh, okay. And just make sure that you state your name in the record. Thank you.

My name is Elizabeth Florian and I'm from Granby, Connecticut.

LEE FLORIAN: My name is Lee Florian. I'm from Granby. We are here speaking in support of Senate Bill 465. This is our son, Joshua Seth Florian. And he's the youngest of our four children.

ELIZABETH FLORIAN: Josh was funny and he was charming. For someone so tiny, he had a deep laugh that sounded like a pirate. We always said he'd be a politician someday

because he was very friendly. And he loved to smile and waive at everyone he passed.

On June 26th, 2008, Joshua came down with a fever. Like any normal toddler, he woke up in the morning and asked for a drink. Just like other mornings, my husband, Lee, put him on his hip and handed him a cup. Joshua handed the cup back and then suddenly collapsed in his father's arms and was not breathing.

That morning our children witnessed the extreme heroic effort to save their brother's life. But Joshua could not be saved that day because none of the doctors knew what was wrong with him.

Our children who sit behind us, Emma, Ben, and Adam kiss their brother goodbye and we took Joshua off life support. Joshua died in my arms. He was just two years, two months, and two days old.

To add to our trauma, when we left the hospital, we were told because there was no cause of death, this was now a police investigation. But that investigation ended when the medical examiner diagnosed our son with adrenal failure of an unknown origin.

Our grief took us on a marathon search for the origin of this adrenal failure. We are trying to find reassurance that our other three children would not die suddenly one morning.

Three years into our search, we were led by good friends to Dr. Raymond who kindly agreed to test the last remaining drop of our son's blood. Three years after Joshua died, the mystery was solved and we were

diagnosed with adrenal failure caused by Adrenoleukodystrophy or ALD.

According to Dr. Gerald Raymond who diagnosed us, the world's expert on ALD, newborn would have given the early diagnosis needed to save Joshua's life. Had this testing been implemented when Joshua was born six years ago, Joshua would be here in this meeting himself. With Joshua's natural confidence, he would look you in the eye and ask himself if you would please support this bill for newborn testing for ALD.

Joshua, as young as he was a leader. And, now, it is our only hope that you can feel his story leading you to support this bill, 465, and protect children.

SENATOR GERRATANA: Thank you, both, for your testimony, Mr. and Mrs. Florian. I'm so sorry for the loss --

LEE FLORIAN: Thank you.

SENATOR GERRATANA: -- to you and your family. But we appreciate you very much coming up today and testifying. Oh, what a little cutie. And saying hi to your children back there too. And as you heard, this is a Committee bill. So, it certainly has gotten our attention.

Are there any questions or if not, thank you all.

ELIZABETH FLORIAN: And we just would like to comment that this is another face of ALD that is not known, but there are many more kids that might be dying of adrenal failure and wouldn't be able to go through the tenacious process we went through to get

62
lk/gbr PUBLIC HEALTH COMMITTEE

February 27, 2013
10:30 A.M.

this diagnosis. And, so, this could be
credibly important.

SENATOR GERRATANA: Right, of course. Thank you.

LEE FLORIAN: Thank you very much for hearing us.

SENATOR GERRATANA: Thank you, sir.

LEE FLORIAN: My youngest son, Adam, did ask that
I read something he wanted me to read last
night --

SENATOR GERRATANA: Sure.

LEE FLORIAN: -- if that's okay with the
Committee.

SENATOR GERRATANA: Please proceed.

LEE FLORIAN: My son Adam in his words says "That
if my baby brother could be here for us, for
even one more hour, he would want most to
touch his hand one more time." He told us
that. "And I would not want anyone else to
feel the pain and suffering that I have felt
over these few years. And I want this
testing for ALD to happen because I don't
want any more families to lose their
siblings and suffer as I have. Please help
other families out and vote for the bill on
ALD. No other family in our state should
feel the way I have felt over these long
years of just memories of my lost brother.
Signed, Adam Florian." Thank you very much.

SENATOR GERRATANA: Thank you, too. Take care.

Next to testify is Representative Lonnie
Reed.

REP. REED: Good afternoon to Committee Chairs,

SB465

Senator Gerratana, and Representative Johnson and the Vice Chairs, Senator Slossberg, Representative Miller and the ranking members, Senator Welch, and Representative Dr. Srinivasan and to all members of the Public Health Committee.

I am -- my name is Lonnie Reed. I represent Branford. And I know you've already met the Kelley family as well as these other wonderful families. I am so grateful to you for raising this bill and signing for this bill. And I know you're going to see it home to a really tremendous conclusion.

The Kelleys wanted me to talk to you a little bit just so we have more data about the actual financial cost in an education system for a child who the miracle of their warrior, Brian, surviving this long has been quite something. And it's been, obviously, the societal costs have been huge and the medical costs.

But they asked the Branford school system to also tally up the additional price paid to educate Brian given his disabilities. And some of the line items include special transportation costs of \$46,000 a year, special computer, hardware, software, and assist of technology evaluation that came to \$35,000 for Brian's 12 years in school. And the grand total of additional costs came to about a million dollars.

So, clearly, human costs, societal costs, and ability to prevent this from happening to other families -- Brian is experienced in the Branford school system. This is probably one of the most beloved and awesome families in Branford. And Brian has inspired his classmates and his educators in

ways that you never learn in a classroom. And he's teaching us all something now. And he's telling us to do what this Committee is doing and what I'm sure the entire Legislature will do which is to pass this bill, not only for Brian as a legacy, but for all the children yet to come. Thank you so much.

SENATOR GERRATANA: Thank you, Representative. Thank you for your testimony. And I will ask are there any questions or follow up? If not, thank you.

REP. REED: Thank you.

SENATOR GERRATANA: Thank you for coming today.

Next is Dr. Salzman followed by Commissioner Macy.

AMBER SALZMAN: Senators, Representatives, and members of the Public Health Committee, my name is Dr. Amber Salzman. I'm here in support for Senate Bill 465.

Adrenoleukodystrophy, ALD, is a genetic disorder with an estimated incidents of one in 17,000. It's known to affect all ethnic groups. The disorder has two components. The adrenal gland is primary adrenal insufficiency or Addison's Disease. This occurs in 90 percent of affected males. Individuals with adrenal insufficiency are at risk for severe decompensation at the time of inspection or other physiological stresses. The Addisonian Crisis that you just heard about may result in substantial morbidity or even death.

Because of the nature of adrenal insufficiency, there's often a substantial

delay in diagnosis and many individuals will have repeated events before being appropriately diagnosed and treated.

In terms of the neurological presentation, 35 percent of boys will develop an inflammatory demyelination condition referred to as childhood cerebral disease between the ages of four and ten years of age. Boys with initial -- with initially normal development will begin to manifest neuropsychiatric features such as changes in attention, learning, and behavior.

Often misdiagnosed as ADHD, learning disorders or autisms, boys eventually go on to have progressive neurological findings. They're usually diagnosed by evidence of widespread abnormalities on the MRI.

The disease is strikingly aggressive and affected individuals die within two to three years of presentation. It needs to be highlighted that while the ADL clinical disease is very rapid in progression, the MRI abnormalities perceived these onset by months. The only available treatment for cerebral disease is allergenic bone marrow transplants. This therapy can arrest the progressive demyelization, but is of absolutely no benefit when the disease is well advanced.

To be effective, a bone marrow transplant must be performed at a very -- when there are very early changes on the MRI prior to seeing symptoms. The optimal candidates are those who have undergone monitoring with MRI from an early age.

My son, Spencer, was born with ALD. But he's now a very health 13-year old boy who

is a loving young man, top student, and start of his school swim team. After many years of misdiagnosis, Spencer's cousin, Oliver, was diagnosed with ALD. Spencer was one-year old when this occurred. This led to Spencer being tested and learning that he, too, was born with ALD.

When he was two-years old, he had a cord blood transplant that stopped the disease. If newborn screening were available at the time his cousin Oliver was born, Oliver would be alive today. It took years of Oliver going to doctors to find out what was going on. And by then it was too late to help Oliver since transplants do not work at that stage of the disease. Oliver died at the age of 12, a few years after he was diagnosed. Spencer is alive and healthy because Oliver was the ALD screen for our family.

Given an accurate and affordable newborn screen now exists, an early diagnose will follow life-saving treatment, families should never again have to suffer the painful losses that our family did. All babies born with ALD should be identified at birth so they, too, can be saved as Spencer was.

SENATOR GERRATANA: Thank you so much, Dr. Salzman. Are there any other questions? Thank you.

I have been reading your testimony. I am going to -- Commissioner Macy, I apologize. I wanted to finish the testimony on Senate Bill 465. We have Ann Moser followed by Lydia Giordano. If Ann would come up followed by Lydia and then Commissioner Macy.

ANN MOSER: Good morning. Thank you for the opportunity to speak in support of the Senate Bill 465. My name is Ann Moser. I'm a biochemist and research associate at the Kennedy Krieger Institute and the department of neurology at Johns Hopkins Medical School in Baltimore, Maryland.

My late husband, Hugo Moser, was a neurologist. And he was an expert in inherited diseases of the nervous system of which ALD is one. And in the early 80's, my husband and I worked as a team and we developed the first clinical test to diagnose ALD in boys. However, they were already symptomatic at that time when the clinical test was done. It was Hugo's dream to develop a newborn screening test for ALD, so, that treatment could be initiated at a very early age.

So, in 2006, we and our team developed a newborn screening test that used the blood spot which is obtained at birth on all newborns. And we piloted this test in Maryland. We screened with consent, we screened 5,000 newborns and we did not find one positive. But you have to remember, ALD is one in 17,000 births. But by not finding any positives in the 5,000, we believe that the false positive rate is very low.

So, we are going to the state of Connecticut to, please, consider this bill and make ALD newborn screening available to the -- all babies born in Connecticut.

If you have any questions, I'm happy to answer.

SENATOR GERRATANA: Thank you, Ms. Moser. Are

you a medical professional or researcher?

ANN MOSER: I'm a biochemist, a researcher.

SENATOR GERRATANA: A biochemist. Thank you for your research and, also, your husband, particularly, in this field. It sounds like a simple enough test. I was following your testimony as you were reading and presenting. And we do appreciate that because very often members have questions as to, you know, how this came about and how it was discovered as well as the procedure. So, we do appreciate that.

Are there any -- yes, Representative Srinivasan.

REP. SRINIVASAN: Thank you, Madam Chair. Thank you very much for your testimony. Could you elaborate for us a little bit more on the screening test that will be -- that you have been doing and that you are suggesting that we do here in Connecticut?

ANN MOSER: Well, the screening test is done on the dry blood spot that is obtained from every newborn. And it's a test which we use an instrument which is called a Tandem mass spectrometer. The instrument which we are proposing to use in the State of Maryland costs about half a million dollars. But that includes high-throughput capacity because we have 70,000 births in Maryland. So, we need an instrument with very high throughput capacity to be able to use only one instrument and one technician to run the test in the State of Maryland.

REP. SRINIVASAN: Thank you, Madam Chair. So, elaborating on this further, I understand the instrument, I understand the cost and

all of that. But what does the test show when the screening is done? Is it just a positive/negative --

ANN MOSER: Yes.

REP. SRINIVASAN: -- or is it more elaborate? I know you did say there are no false positives which is good -- which is great. Obviously, we don't want to unnecessarily a family of anything they don't have -- the child does not have. But the accuracy rate of the testing and what does the test basically tell you if we were to do the screening tests?

ANN MOSER: Well, if we have a positive test, that is, we're looking at a certain fat in the blood that has a hallmark for this disease. And if we get a positive test, then we can notify the geneticists or physician who would then bring the family in and we would do follow-up testing on that baby. And the follow-up testing would be the gold standard test for ALG which are the plasma fatty acid tests.

And, in addition, we have the gene test, so, we could detect the mutation in the ALD gene in that baby and then do further testing on that baby's family to find out if they're additional cases within that family. Since it's an X-linked disorder, you can identify any other family members at risk.

REP. SRINIVASAN: Thank you. And just one follow-up question. So, if a diagnosis is made through this elaborate process, then early intervention would be with the bone marrow would be the treatment of choice in that particular situation as it is right now?

ANN MOSER: Well, initially, that baby boy -- if it's a baby boy, he would be screened for Addison's Disease periodically. And if he has adrenal insufficiency which is Addison's Disease, he would be treated with steroid therapy. And at times of strep, like, high fever, he would be given additional hormones in order to meet the stress of an illness and, thereby, save his life.

And then at starting at the age of one, this baby boy would have periodic evaluations by a pediatric neurologist. And most pediatric neurologist recommend an MRI every year after the age of two. I think the age of two is the earliest known time when a boy with ALD would develop brain disease.

REP. SRINIVASAN: Thank you very much. Thank you, Madam Chair.

SENATOR GERRATANA: Thank you, Representative.

Yes, Senator Bartolomeo.

SENATOR BARTOLOMEO: Thank you. How common is it that there would be a child would pass away prior to having symptoms be known?

ANN MOSER: How common? It's too common. The incidents of Addisonian Crisis that you heard about to. It's very common. And it can be prevented by adequate following by an endocrinologist.

SENATOR BARTOLOMEO: So, do you have anyway of estimating for us how often a child would pass away without presenting symptoms and it would later be found out versus a child who by the time is diagnosed is experiencing symptoms, but it's too late for the preferred

71
lk/gbr PUBLIC HEALTH COMMITTEE

February 27, 2013
10:30 A.M.

treatment? Do you know what I'm saying?
We've seen two different scenarios and I'm
trying to --

ANN MOSER: Well, we diagnose -- in our
laboratory, we diagnose about 50 boys with
ALD every year. But we're getting samples
from all over the country. And all those
individuals or most of those individuals are
symptomatic and it's too late. Some of them
are at the border zone and they are referred
for bone marrow transplantation. However,
if they're already having neurological
symptoms, the outcome isn't as good as those
who are identified before they have overt
neurological symptoms.

SENATOR GERRATANA: Thank you. I do have a
question, however. I am reading the
Department of Public Health's testimony on
this. And I think it's a good question.
They say that currently there are no public
health laboratories that screen newborns for
ALD.

ANN MOSER: That's correct.

SENATOR GERRATANA: So, how would the State of
Connecticut do this?

ANN MOSER: How would they do it?

SENATOR GERRATANA: Yes, in other words, the
protocol would be set up by, usually, the
way it is cited, it would be a newborn and
the test would be done. But who would do
the testing, in other words, the analysis of
the blood test?

ANN MOSER: The newborn state -- for the state
lab who does newborn screening in the State
of Connecticut would do the test.

72
lk/gbr PUBLIC HEALTH COMMITTEE

February 27, 2013
10:30 A.M.

SENATOR GERRATANA: They, also, say that they would have to develop the method and I guess there's --

ANN MOSER: Correct. But we're --

SENATOR GERRATANA: -- there's a minimum of 5,000 dry blood spots would have to be tested before the method could be validated?

ANN MOSER: Yes. We can help with the validation. Our laboratory is a CAP certified laboratory and can help up with the setup of the test.

SENATOR GERRATANA: I see. Okay. Thank you very much for that. There's no other questions. Thank you for coming today and giving your testimony.

ANN MOSER: Well, it's my pleasure. Thank you very much.

SENATOR GERRATANA: And just to complete that Lydia Giordano is the last person to testify on this bill.

Yes, Commissioner Macy is to follow. Thank you.

LYDIA GIORDANO: Shall I start?

REP. JOHNSON: Yes.

SENATOR GERRATANA: Oh, yes, please. Just put -- state your name for the record. Thank you.

LYDIA GIORDANO: Hi, my name is Lydia Giordano. I'm from Stamford, Connecticut. Where do I begin? Steve had issues in learning when he was a toddler, so, I had him in special

SB465

educations to help him. I knew something was wrong. I thought maybe ADHD, but doctors told me he was too young to be diagnosed. Steve was very sickly as a little boy, asthma at six months, allergic to diary until five years old. I was struggling wit Steve's energy all the time.

My doctor says usually around six-years old, the boys tend to mellow down. At six, I took him to see someone and they told me he had ADHD. They put him on Concerta. He was only on it for about four months. I felt that I could control him during summer months.

He was not a disrespectful boy, he was just very hyper and all over the place. Summer is over now and he's going to first grade. Since he was in school and the principal used to call me almost every other week for something Steve was doing in class. He didn't do that well that year, so, he was going to repeat first grade once again but in a new school for learning disabilities.

September 28th, 2006, was the first day Steve attended and the last day. We were in the van and his head had dropped. I said to Steve, I said, "Are you okay?" He picked up his head and said, "Yeah, Mommy, I'm okay." I didn't feel comfortable. I called my pediatrician. That same night she had me come in and did the neurological test follow my hand follow this. She said, "I want you to go to the emergency room." She said, "I'm going to have a CT Scan ordered." She didn't tell me what she thought it might be. She just thought something wasn't right.

So, I'm sitting there with his father, Steve playing as always waiting for the CT Scan

results. The doctor comes back down and he looks at me and he has the kiss of death of on his face. And I said, "Was my son's CT Scan bad?" He said, "Mom, it's not good." He said, "We have to admit your son."

They took him upstairs, asked me a lot of questions about his milestones, how he was as a baby and everything. I didn't know at that time. They already knew Stevie had Adrenoleukodystrophy. Half the brain had already demyelinated. So, there's nothing, really, basic we can do.

So, I -- after he was in the hospital -- two weeks after diagnoses, Stevie could not walk. For Halloween, he was in a wheelchair. Two weeks after that, Stevie did not talk. We was on our Make-A-Wish trip which he was already in Disney World four months earlier when no symptoms, being a happy -- playing happily little kid. He cried the whole time in Disney World. He hated it because he couldn't communicate to us. He was literally, like, (makes sound) like that. He couldn't barely eat.

Two weeks after that, I called Duke to find out if he can be a bone marrow transplant, so, I brought him in. All the doctors came around. I thought Stevie's coordination was off because he wasn't walking. He was on the floor trying to reach. And it turned out Steve was already blind by then.

I asked the doctors, I said, "Is anything you can do for Steve?" And they said, "Mom, take him home and enjoy the time you have left with him." I said, "How long does he have to live?" They said, "Three to six months."

So, I brought him home, had a feeding tube put in. Steve almost died in December of 2007. They gave him an emergency trach. After the trach, I learned to take care of him with the trach. He was in rehab for four months. I learned how to take care of trach, change it, whatever. And Steve's father and me separated. We was in a happy, healthy relationship for 18 years. But he had lost everything. He lost his son. We lost our home. He just couldn't handle it. One day he just didn't come home. He stayed at his mother's house.

Steve's father become depressed and gave up hope. September 28th, 2009, three years exact to the date my son got sick, his father dies of a heart attack at 43 years old. My life has never been the same. Steve has now lived six years and has -- life has been hard. This is not how I thought I would spend my golden years.

I ask you to please pass this bill so no other family would go through the mental and physical challenges that I endure everyday.

SENATOR GERRATANA: Mrs. Giordano, thank you for your bravery in coming here today and testifying. Certainly, your story is a long and arduous one. But I think you're a very, very brave and courageous women. And we, you know, listening, I'm laughing rather than crying here. But in listening, it certainly hestered me and I'm sure other members of the Committee.

Are there any other comments or questions?

LYDIA GIORDANO: I think the other lady asked how long do usually the boys live once they've been diagnosed once they can't get the bone

marrow transplant. From a lot of parents that I've met on Facebook, I'm trying to be there for them. Usually, if their boys have the symptoms, usually don't live more than a year or two. So, I feel really guilty sometimes. I'm on Facebook and I still have Steve. But I try to be there for the mothers that are going through whatever they're going through. And Facebook has been amazing because I didn't know anybody the day Stevie got sick.

SENATOR GERRATANA: It is a good support for you. I know, social media is just --

LYDIA GIORDANO: And Brian's Hope.

SENATOR GERRATANA: -- amazing.

LYDIA GIORDANO: Brian's Hope. I moved to Connecticut and I just met --

SENATOR GERRATANA: Mr. and Mrs. Kelley --

LYDIA GIORDANO: Kelley.

SENATOR GERRATANA: Dr. and Mrs. Kelley.

LYDIA GIORDANO: And they're the reason why I'm here today and now I don't feel so alone up in Connecticut.

SENATOR GERRATANA: Well, thank you and thank Steve for coming out today and testifying before our Committee. We, certainly, do appreciate all of your efforts. We'll keep you in mind.

LYDIA GIORDANO: Okay. Thank you.

SENATOR GERRATANA: Thank you, ma'am.

SENATOR GERRATANA: Well -- thank -- thank you for that. And we'll go down the list and we'll come back to you. Thank you. Thank you, Jim.

Any other questions? I guess not.

Okay. Next is Representative Lemar.

REP. LEMAR: Thank you, Senator Gerratana, Representative Srinivasan. I appreciate your consideration of our testimony today. I'm joined by two individuals, Heather Aaron who is the Executive Director of Leeway and former Representative William Diffen.

There are four items on your agenda. I want to briefly reference three and then fully testify on one referencing House Bill 5538 which was introduced by the entire New Haven delegation which would establish a pilot program for school-based health clinics in New Haven. There are three people who are going to testify in great detail about those. So, I'll reserve my testimony for written form. You'll hear later from the New Haven Board of Education from Community Providers Association of Connecticut and the Director of Public Health in New Haven to the strong merits of that program.

Secondly, Senate Bill 465 requiring newborn screening for ADP. You heard a great deal from the Kelley family and Senator Pisano earlier today.

And 5746, AN ACT CREATING A TASK FORCE ON CHILDHOOD OBESITY. In a separate committee, I've introduced a bill this year that would establish a penny per ounce tax on sugar, sweet, and beverages and redirecting funds

HB6003

testimony.

MARIO GARCIA: Thank you.

REP. JOHNSON: Representative Claire Janowski.
Welcome.

REP. JANOWSKI: Thank you. Good afternoon,
Representative Johnson, Chairwoman, and
members of the Public Health Committee. I
am State Representative Claire Janowski from
the 56th District of Vernon and Rockville.
And, although, I am here to speak on behalf
of Bill number 6004, AN ACT CONCERNING
HEALTHCARE SERVICES FOR THE UNINSURED AT
COMMUNITY HOSPITALS, having been here for a
while and listening to some of the
testimony, I have signed onto a couple other
bills and I would like to express my support
for those as well. And that would be Bill
number 360, AN ACT CONCERNING PEDIATRIC
AUTOIMMUNE OR PANDAS SYNDROME. I believe
it's important to catch and treat this very
early because it can lead to other problems
in adult life.

And, also, would like to learn my support to
Bill number 465, AN ACT REQUIRING NEWBORN
SCREENING FOR ALD. I believe that's, also,
a very important bill.

With regard to H.B. 6004, concerning health
care services for the uninsured at community
hospitals, I am here to testify in support
of funding two hospitals to enable them to
continue providing healthcare services for
the uninsured.

As you are aware, this is something that is
being slated to be reduced in the Governor's
budget proposal. I would believe the cuts
would be of particular harm to small

**JOINT
STANDING
COMMITTEE
HEARINGS**

**PUBLIC
HEALTH
PART 4
1011-1347**

2013

Statement of Elizabeth Florian and Lee Florian RN

Senate Bill 465, An Act Requiring Newborn Screening For Adrenoleukodystrophy

February 27, 2013

Senator Gerratana, Representative Johnson, and Public Health Committee Members

The youngest of our four children, Joshua Seth Florian, was a funny, charming and dashing handsome blonde boy with big blue eyes. My husband Lee and I always said he would be a politician someday because he loved to smile and wave at everyone he passed like he was riding on a float in a small town parade. He had a distinctive laugh, for someone so tiny; hearty and deep like a pirate. On June 26, 2008 Joshua came down with a fever, like any normal toddler. He woke up at dawn and asked for a drink, as he did on many mornings. My husband sat him on his hip, and handed him a cup. Joshua handed the cup back and then suddenly fell back in his father's arms and stopped breathing. The trauma of that day is hard to talk about, but that morning we and our three other children, Emma, Ben, and Adam, witnessed the heroic efforts to save their brother's life. Just hours after Joshua took his last drink of water, our children kissed their brother goodbye and Joshua was taken off life support, he died in our arms. He was 2 years 2 months and 2 days old.

Joshua's life could not be saved that day because none of the doctors knew what was wrong with him. Had Joshua been diagnosed, he could have been given a simple, inexpensive medication to treat his symptoms. When we left the hospital that day, we were told that because there was no cause of death, this was now a police investigation and could take up to 10 months. But that investigation never happened because the next morning, the medical examiner told us it was Addison's Adrenal insufficiency of an unknown origin.

Our grief took us on a marathon search for the origin of this disease. We wanted reassurance that our other children would not suddenly die one morning. After 4 years of intense, personal scientific research, countless dollars in medical tests, and meeting with many empathetic doctors, we found Doctor Raymond who kindly agreed to test the last remaining drops of our son's blood as one last reassurance to us. Last Christmas, we found out our son Joshua had a very rare presentation of Addison's adrenal insufficiency caused by Adrenoleukodystrophy, or ALD. According to Dr. Raymond the world expert on ALD, Newborn Screening would have given the early diagnosis needed to save Joshua's life. Had this testing been implemented when our son was born 6 years ago, Joshua could have been here in this meeting himself. With Joshua's natural confidence, he would have looked you in the eye and asked you directly if you would please allow the State of CT to do Newborn screening for ALD. But Joshua will never get that chance and his short life will have to serve as his testimony to the importance of testing for this disease. And we, his family, now must live with the painful fact that our son could have been protected by the knowledge that comes from a simple newborn screening test that costs \$1.50. Young as he was, Joshua had the personality of a leader. Now our only hope is that you can feel his story leading you to do all you can to pass Bill 465 and protect Connecticut's children. Our family is urging you to act so that what happened to our son will not happen to another child in the state of CT.

Support for Senate Bill 465: An Act Requiring Newborn Screening For X-linked Adrenoleukodystrophy (ALD)

Senator Gerratana, Representative Johnson, and Public Health Committee Members

Thank you for the opportunity to speak in support of the Senate Bill 465. My name is Ann Moser. I am currently a research associate at the Kennedy Krieger Institute and the Dept. of Neurology at Johns Hopkins Medical School in Baltimore, Maryland. My late husband, Dr. Hugo Moser, and I developed an interest in studying ALD in 1978 when the group at Albert Einstein in NYC reported that patients with ALD had increased very long chain fatty acids (VLCFA) mainly of 26 carbons chain length (C26:0) in brain and adrenal cholesterol esters. In the early 1980's Hugo's research team at the Kennedy Krieger Institute developed gas chromatographic assays of the very long chain fatty acids, first in cultured cells and later in plasma, to diagnose patients with ALD. After the plasma C26:0 assay became available, many families with ALD were identified and therapy trials began. One of the most important, and available life-saving therapies for ALD is **hormone replacement** for those ALD patients with Addison's disease.

Since the early 1990's, **bone marrow transplantation** was shown to be effective in halting the central nervous system demyelination if done at the **first signs of progressive brain dysfunction**. By 2010 several hundred ALD boys have benefited from bone marrow and umbilical cord cell transplantation as well treatment for their Addison's disease.

It was Hugo Moser's dream to identify boys with ALD early, at a time before Addison's disease and brain dysfunction occurred. In 2005 Hugo suggested to the national newborn screening committee that ALD be added to the list of disorders that would possibly benefit from newborn screening, however, at that time there was no test for ALD utilizing the sample collected on all newborns, the heel stick blood spot on filter paper.

In order to develop a newborn test for ALD, Hugo and I contacted Walter Hubbard, Ph.D. at the Dept. of Clinical Pharmacology at Johns Hopkins. Walter is an expert in liquid chromatography tandem mass spectroscopy (LC/MSMS) of lipids and he was interested in helping us devise a test for ALD utilizing the newborn dried whole blood spot (DBS). In January of 2006, we determined that the C26 content of the lyso phosphatidylcholines (lyso PC) was 5 to 10 fold higher in whole venous blood spots from ALD patients when compared with controls. The ALD newborn DBS had a 5 to 15 fold increased C26:0 lyso PC with no overlap when compared with 500 anonymous newborn DBS. These findings were published in 2009. Since that time we have developed a high throughput LC/MSMS screening procedure and have published a combined extraction of the C26:0 lyso PC with that of the acyl carnitines. Recently together with the MD State Newborn Screening Lab, we have completed the screening of 5000 consented newborns born in 3 local Baltimore hospitals and did not find one positive, thus we believe that using our procedure the false positive rate will be low.

We are here today on the behalf of all ALD researchers, the ALD support groups who have donated funds to ALD newborn screening, and many ALD families worldwide to request that ALD be added to the uniform panel of screening tests performed on all newborns.

Thank you for your time and consideration of this important, life-saving request.

Ann B. Moser, Kennedy Krieger Institute, 707 North Broadway, Baltimore, MD 21205, tel: 443-923-2761, mosera@kennedykrieger.org

PEDIATRIC BLOOD & MARROW TRANSPLANT PROGRAM
 STEM CELL LABORATORY
 ROBERTSON CLINICAL & TRANSLATIONAL CELL THERAPY PROGRAM
 CAROLINAS CORD BLOOD BANK

Joanne Kurtzberg, MD
 Telephone (919) 668-1119
 Laboratory (919) 668-1177
 FAX (919) 668-1183
www.cancer.duke.edu/pbmt
kurtz001@mc.duke.edu
 Page 1 of 2

February 20, 2013

RE: Newborn Screening for Adrenoleukodystrophy

To Whom It May Concern:

SB 465

I am writing to express my strongest support for the implementation of newborn screening for adrenoleukodystrophy (ALD) a genetic disorder of fatty acid metabolism, which causes adrenal insufficiency, neurological deterioration and death. The majority of pediatric patients who die from the disease have the cerebral form presenting in mid to late childhood.

Hematopoietic stem cell transplantation with related and unrelated donors can prevent progression of the cerebral form ALD *IF performed before the patient has manifested clinical symptoms of the disease*. Conversely, if transplantation is performed in symptomatic children, the disease usually progresses and the child either dies or stabilizes in a severely debilitated state.

Most families are unaware of the fact that they are at risk for having a child with ALD until a child in the family becomes symptomatic. Usually it's too late to help that index case. While subsequent pregnancies or births can be screened, this approach fails to rescue children who are the first case in their family. Newborn screening for ALD would identify these children before they manifested symptoms enabling them to have access to curative therapy with hematopoietic stem cell transplantation.

Another important reason to identify these children with newborn screening testing is because some will present with life-threatening problems due to adrenal crises which are secondary to adrenal insufficiency which also develops in babies and children with ALD. In many children adrenal insufficiency occurs before the onset of neurological symptoms. We have seen a child present in adrenal crisis as early as 6 months of age due to ALD. As adrenal crisis is also life-threatening, and preventable, it is a second compelling reason to screen newborns for ALD.

Over the course of my 20+ year career, I have seen hundreds of children with ALD who were diagnosed to late for treatment and who subsequently died months to years after devastating neurological deterioration. Newborn screening is an intervention that could have saved these children's lives.



PEDIATRIC BLOOD & MARROW TRANSPLANT PROGRAM
STEM CELL LABORATORY
ROBERTSON CLINICAL & TRANSLATIONAL CELL THERAPY PROGRAM
CAROLINAS CORD BLOOD BANK

Joanne Kurtzberg, MD
Telephone (919) 668-1119
Laboratory (919) 668-1177
FAX (919) 668-1183
www.cancer.duke.edu/pbmt
kurtz001@mc.duke.edu
Page 2 of 2

Pilot studies using Mass Spectroscopy for newborn screening for ALD have been shown to be effective and ready for additional testing. The next step would be implementation in a state in the U.S. with a large number of births. The state of Connecticut meets these criteria and could move forward to implement newborn screening for ALD.

Babies who screen positive would have to undergo mutation testing. If mutation testing was also positive, twice yearly screening of adrenal function (through blood tests) from birth and neurologic function (with brain MRI and developmental assessments, +/- BAER) from 3 years of age would allow for boys with ALD to be identified in the pre-symptomatic state.

In summary, I endorse, with the highest level of enthusiasm, the idea that the state of Connecticut under take, the task of adding newborn screening for ALD to their current panel. Please do not hesitate to contact me if I can provide additional information in support of this request.

Sincerely,



Joanne Kurtzberg, M.D.
Jerome Harris Distinguished Professor of Pediatrics
Professor of Pathology
Chief Scientific Officer and Medical Director, Robertson Clinical & Translational Cell Therapy Program
Director, Pediatric Blood & Marrow Transplant Program
Director, Carolinas Cord Blood Bank at Duke



Lydia Giordano

Where do I begin... Steve had issues with his learning When he was a toddler so I had Special education services to help him. I knew something was wrong I thought maybe ADHD but doctors told me he was too young to be Diagnose. Steve was very sickly.. asthma at 6 months.. allergic to dairy until 5 years old..i was always stuggling with steve s energy.,My doctor says usually around 6 years old the boys tend to mellow down. At six I took him to see someone and they told me he had ADHD and we put him on conserta.. He was only on it for about four months because I felt I can control him during the summer months.. He was not a disrespectful boy he was just very hyper all over the place. Summer is over and now he's going to first grade and Stevie was called into the principal office almost every other week for something he was doing in class . He didn't do that well that year he was going to repeat first-grade once again but in the new school For learning disabilities.. September 28, 2006 was his first day and his last We were in the Van and his head had dropped ..I asked him are you okay Steve picked his head up and said I ok mommy.i called my pediatrician.. That same night she had sent me to the emergency room. She did not know what was wrong with Steve but she felt something was different. She had order a CAT scan at the emergency room Sitting there with his father Steve playing as always.. Waiting for results from the CAT scan .The doctor came towards us.The look was like the kiss of death on his face.. I said was Steve CAT scan.. bad. He said mom is not good. it was confirmed two days later he had X-linked Adrenoleukodystrophy. Within two weeks Steve stop walking. Steve was in a wheelchair one month later for Halloween ..two weeks after that Steve could barely talk by the second month. And by the third month we went to see if Steve could recieve bone marrow transplant at duke in north carolina and I did not know Steve was blind by then.. duke said Steve Was not a candidate for bone marrow transplant I asked the doctors how long does he have to live? I was told 3 to 6 months .. So I took him home got a feeding tube and the trache eventually when he almost died in December 2007 since then Steve has a trache g tube and no mobility.. I was in a happy and healthy relationship for 18 years this disease devastated his father Steve Conley Sr.one day he didn't come home he just stayed at his mothers .I was taking care Steve jr alone with home health aides and then I got nurses We lost our house his father own in new jersey..financially he could not afford the Mortgage .because of loss income from me.. Steve father became depressed & gave up hope.. September 28, 2009 three years exact to the day my son got sick his father dies of a heart attack 43 years old and my life has never been the same since.. Steve has live now 6 years.. And life has been hard.. This was not how I thought I would spend my golden years.. I asked of you please pass this bill so no other families would have to go to the pain that I go through every day..

SB465



State of Connecticut

HOUSE OF REPRESENTATIVES
 STATE CAPITOL
 HARTFORD, CONNECTICUT 06106-1591

REPRESENTATIVE VINCENT J. CANDELORA
 EIGHTY-SIXTH ASSEMBLY DISTRICT

LEGISLATIVE OFFICE BUILDING, ROOM 4200
 300 CAPITOL AVENUE
 HARTFORD, CT 06106-1591

TOLL FREE. (800) 842-1423
 CAPITOL (860) 240-8700
 Vin.Candelora@housegop.ct.gov

DEPUTY REPUBLICAN LEADER

MEMBER

FINANCE, REVENUE AND BONDING COMMITTEE
 REGULATIONS REVIEW COMMITTEE
 PLANNING AND DEVELOPMENT COMMITTEE
 LEGISLATIVE MANAGEMENT COMMITTEE

Public Health Committee
Public Hearing
Wednesday, February 27, 2013
Testimony In Support of,

SB 465, An Act Requiring Newborn Screening For Adrenoleukodystrophy (ALD)

Good Morning Co-Chairs Senator Gerratana, Representative Johnson and Ranking Members Senator Welch and Representative Srinivasan and members of the Public Health Committee. My name is Representative Vincent Candelora. I am testifying in support SB 465.

Thank you for bringing the proposed bill forward for a public hearing.

SB 465 calls for the pre-screening of ALD in newborns. Pre-screening for this devastating disease not only is most effective in saving the life of a child, but it is cost effective as well. Newborn screening allows babies with ALD to be identified and treated before they become sick, resulting in cost savings by preventing serious health problems.

Currently screening of newborns has saved or improved the lives of more than 12,000 children.

Thank you for your consideration on this important matter.

Vincent Candelora
 State Representative
 86th District

Amber Salzman, PhD
The Stop ALD Foundation
amber@stopald.org
1.610.659.1098

Support for Senate Bill 465: An Act Requiring Newborn Screening For Adrenoleukodystrophy

X-linked adrenoleukodystrophy (ALD) is a genetic disorder with an estimated incidence of 1:17,000 and it is known to affect all ethnic groups. The disorder has an adrenal gland component and a neurological presentation. The disorder in the adrenal gland is primary adrenal insufficiency or Addison's disease. This occurs in 90% of affected males. Individuals with adrenal insufficiency are at risk for severe decompensation at the time of infections or other physiologic stresses. The Addisonian crisis may result in substantial morbidity or even death. Because of the protean nature of adrenal insufficiency there is often a substantial delay in diagnosis and many individuals will have repeated events before being appropriately diagnosed and treated.

In terms of ALD neurologic presentation, 35% of boys will develop an inflammatory demyelinating condition referred to as childhood cerebral disease (CCALD) between the ages of 4 and 10 years of age. Boys with initially normal development will begin to manifest neuropsychiatric features such as changes in attention, learning, and behavior. Often misdiagnosed as ADHD, learning disorders, or autism, boys eventually go on to have progressive neurologic findings. They are usually diagnosed by evidence of widespread abnormalities on MRI. The disease is strikingly aggressive and affected individuals die within 2-3 years of initial presentation.

It needs to be highlighted that while the ALD clinical disease is very rapid in progression, the MRI abnormalities precede disease onset by months. The only available treatment for cerebral disease is allogeneic bone marrow transplant. This therapy can arrest the progressive demyelination, but is of no benefit when the disease is well-advanced. To be effective, a bone marrow transplant must be performed when there are early changes on MRI, but prior to neurologic findings. The optimal candidates are those who have undergone serial monitoring with MRI from an early age.

Because of the ability to intervene both for the endocrine (Addison's) and neurologic issues, it is critical to identify pre-symptomatic individuals. Newborn screening of dried blood spots is a near universal method of testing individuals and allows the diagnosis of a wide variety of genetic disorders. A newborn screen for ALD has been developed and piloted, and over 50,000 newborn dried blood spot samples have been screened to validate the test.

My son, Spencer, was born with ALD, but is now a healthy 13-year-old boy who is a loving young man, top student, and star of his school's swim team. After many years of misdiagnosis, Spencer's cousin Oliver was diagnosed with ALD. Spencer was one year old when this occurred. This led to Spencer being tested and learning that he too was born with ALD. When he was

two years old he had a cord blood transplant that stopped the disease. If newborn screening were available at the time his cousin Oliver was born, Oliver would be alive today. It took years of Oliver going to doctors to find out what was going on and by then it was too late to help Oliver, since transplants do not work at that stage of the disease. Oliver died at the age of 12, a few years after he was diagnosed. Spencer is alive and healthy because Oliver was the ALD screen for our family. Given an accurate and affordable newborn screen now exists and early diagnoses will allow life saving treatment, families should never again have to suffer the painful losses that our family did. All babies born with ALD should be identified at birth, so they too can be saved as Spencer was.

In summary, there exists over 30 years of experience in the diagnosis and evaluation of ALD. It is well established that the outcomes are vastly improved by the early identification and clinical monitoring of affected individuals. Therapies are available that can improve the quality of life for affected individuals. In fact, the most common manifestation of ALD, adrenal insufficiency, can be treated with steroid replacement, a simple and inexpensive medication. Finally, there is a methodology that allows us to accurately identify individuals off of the newborn blood spot.

Based on the information that we already have, we can estimate screening newborns for ALD will have a significant impact on the acute and sometimes catastrophic adrenal presentation and will alter the population of boys who are judged ineligible for bone marrow transplant because they are too advanced when diagnosed with ALD. It is for these reasons that we ask you to require that all newborns are screened for x-ALD.

Thank you for your time and consideration of this important, life-saving request.



Stopping the
progression of ALD

Jean Kelley DIRECTOR

Statement of Jean Kelley and John Kelley, M.D.

SENATE BILL 465, AN ACT REQUIRING NEWBORN SCREENING FOR ADRENOLEUKODYSTROPHY

February 27, 2013

Senator Gerratana, Representative Johnson, and Public Health Committee Members

Thank you for providing us with this opportunity to appear here today. We are honored to speak in support of this vital bill. We are especially excited at the possibility of making a change in the outcome of the lives of boys who have Adrenoleukodystrophy and their families. Early detection of Adrenoleukodystrophy will help avoid undue suffering and the enormous emotional and financial costs associated with the treatment and care of the full onset of the disease. That is possible now. We have a test that detects ALD from newborn blood spots.

Adrenoleukodystrophy, or ALD, is a life threatening disorder that often causes adrenal gland failure, as well as neurologic dysfunction, affecting the brain and/or spinal cord. ALD has 3 characteristics of phenotypes: adrenal insufficiency that all get at a very young age, cerebral demyelination in about 40% of the children which leads to death, and AMN as an adult if they live to that age. It is truly a parent's worst nightmare. We know. This was our experience with our warrior, Brian.

Brian was diagnosed with the cerebral demyelination form of ALD at the age of 6 after suffering a head injury. While sledding one day, Brian hit his head on a woodpile. At the Yale ER, a CAT Scan and follow up MRI led to Brian's diagnosis. Within 6 months of Brian's diagnosis he lost his mobility, speech, ability to eat and most of his vision. Since there was no newborn screening at the time of Brian's birth and although this is a X-linked hereditary disease, there were no red flags in our family. Had his condition been detected at birth, early intervention would likely have altered the outcome.

Despite his disabilities there is much Brian is able to do. In his very quiet way and with his tenacity to persevere, he has inspired us to move on and impact others in a positive way, such as this. Adoption of ALD newborn screening will be Brian's legacy so his many challenges in life were meant to help others and were not in vain.

Early detection is crucial. The missed diagnosis of newborns with ALD, will most importantly, result in the needless suffering and loss of lives. At best, ALD results in neurological impairment, developmental delays, long-term feeding issues, and costly therapies, all at huge costs to the health care system and tremendous emotional pain for the families of these precious children. Physicians, hospital administrators, and advocates clearly understand that early detection of newborn diseases and disorders saves lives and dramatically reduces health care costs and burdens associated with deferred diagnosis. Add to this, the very expensive cost of educating children with special needs as a result of the disease and the financial cost of ALD can easily run into the millions of dollars per child.

Newborn screening for ALD is a simple and cost-effective solution to these problems; a simple newborn screening, costs \$1.50 per child and would be part of the existing newborn screening panel. The savings—both in dollars and in needless suffering—are dramatic.

It is completely within our power to keep babies born with the potential for this devastating disease well, setting them up for a normal and healthy adulthood. Treatment is most effective if begun before symptoms of ALD appear. For this reason, it is vital that parents have the information so that they can make the best possible medical choices for their children. This one intervention could save lives.

We believe this testing should become a standard of care for all newborns, prior to discharge from the hospital. We urge you to leverage your leadership and expertise in public health to support this important bill. SB#465 will save lives and money. It is that simple. The time is right...there is an accepted method and treatments. Let's make this happen.

Screening Newborns for Adrenoleukodystrophy

Adrenoleukodystrophy

Adrenoleukodystrophy (ALD) is a life threatening disorder that often causes adrenal gland failure, as well as neurologic dysfunction affecting the spinal cord and/or brain.

The disease is passed down from parents to their children as an X-linked genetic trait.¹ It therefore affects mostly males, although some women who are carriers can have milder forms of the disease. It affects approximately 1 in 17,000 people from all races. The condition results in the buildup of very-long-chain fatty acids in the nervous system, adrenal gland, and testes, which disrupts normal activity. There are three major categories of disease:

- Childhood cerebral form – appears in mid-childhood (at ages 4 - 8)
- Adrenomyelopathy -- occurs in men in their 20s or later in life
- Impaired adrenal gland function (called Addison disease or Addison-like phenotype) – adrenal gland does not produce enough steroid hormones

Newborn Screening

Connecticut law requires newborns to be screened in the hospital for a number of diseases including cystic fibrosis, severe combined immunodeficiency, critical congenital heart disease, HIV, sickle cell and other tests that determine if the newborn has inborn metabolic or other disorders (Section 19a-55).

The Federal government is currently monitoring a study underway at the Mayo Clinic's Biochemical Genetics Laboratory to develop the best possible screening test using dried blood spots from anonymous newborns to test for ALD. We expect the results of this work could be completed and forwarded to the federal panel that will consider newborn screening protocols in May 2013. A larger study would then occur before a final recommendation is made to the Uniform Screening Panel in 2014.

ALD met all the criteria to be included in the uniform newborn screening panel and the initial preliminary results from the Mayo clinic were 12 positives out of 42,000 samples. Due to the extensive time this will take for each state to implement, too many children will be put at significant risk, many of them dying.

¹ www.nih.gov

The legislation we are seeking will add screening for Adrenoleukodystrophy to the existing newborn screening panel.

Life-saving Test

- *It is an inexpensive and sensitive screening test.
- *It is an opportunity to detect complications from the disease before there are symptoms.
- *There are treatments, which if given in the early phase, dramatically improve the outcome.
- *If treatment is delayed until the condition is apparent clinically, the outcome is much worse.
- *Up to this point, early, often life-saving treatment has been available only to those diagnosed because a relative suffered the disease.

Brian Kelley

Brian Kelley is a 24-year old, young man from Branford who was diagnosed with ALD at the age of 6. Unfortunately, Brian was already symptomatic. There were no red flags in his family. Had the newborn screening for ALD been in place at the time of Brian's birth, he would not be facing the great challenges he does today. With great courage and tenacity, Brian has taught much to many in his very quiet way for the past 18 years. We hope we can honor Brian by making a difference in the lives of others diagnosed with ALD.

Connecticut Facts

Connecticut Birth Rate: 37,708 per year

Price of test: \$1.50-2.00 per test

Cost per year to add ALD newborn screening: \$56,500-74,000

Mission Statement of the Connecticut Department of Public Health

To protect and improve the health and safety of the people of Connecticut by:

- *Assuring the conditions in which people can be healthy
- *Preventing disease, injury, and disability, and
- *Promoting the equal enjoyment of the highest attainable standard of health, which are a human right and a priority of the state

**The time is right.... there is an accepted method and treatments.
Let's make this happen.**

HAMLET M HERNANDEZ,
Superintendent

MARY PERRARO, Ed.D.
Assistant Superintendent

TASHIE S. ROSEN
Chief Financial Officer



BRANFORD PUBLIC SCHOOLS

1111 Main Street, Branford, CT 06405-3717

203.488.7276 • Fax 203.315.3505

www.branford.k12.ct.us

Cost Estimate for 12 years of Brian's Education

Paraprofessional Salary-Gross Pay- \$21,254.00/ Extended School Year-\$2,360.00 per year

Transportation-School Year-\$45,974.00/ Extended School Year-\$5,080.00 per year

OT/PT Therapy-based on 3 hours a week-\$3,195.3/Extended School Year-\$1,065.12 per year

Assistive Technology Evaluation-\$20,000 (based on 12 years of education)

Computer Software/Hardware-\$15,000 (based on 12 years of education)

Wheelchair- (2) at \$30,000.00 (this is often a covered cost by insurance)

\$927,266 is the estimated total without the cost of the chairs.

Progress Made toward Universal Newborn Screening for X-linked adrenoleukodystrophy (ALD)

History and recent developments:

It was Hugo Moser's dream to identify boys with ALD early, at a time before Addison's disease and brain dysfunction occurred. In 2005 Hugo suggested to the national newborn screening committee that ALD be added to the list of disorders that would possibly benefit from newborn screening, however, at that time there was no test for ALD utilizing the sample collected on all newborns, the heel stick blood spot on filter paper.

In order to develop a newborn test for ALD Hugo and I contacted Walter Hubbard, Ph.D. at the Dept. of Clinical Pharmacology at Johns Hopkins. Walter is an expert in liquid chromatography tandem mass spectroscopy (LC/MSMS) of lipids and he was interested in helping us devise a test for ALD utilizing the newborn dried whole blood spot (DBS). We first used LC/MSMS to measure the total lipid C26 fatty acid content of the DBS and also the C26 content of other lipids such as ceramides and sphingomyelins, but found that the naturally high red blood cell C26 content interfered and gave many false positives. Finally in January of 2006, we determined that the C26 content of the lyso phosphatidylcholines (lyso PC) was 5 to 10 fold higher in whole venous blood spots from ALD patients when compared with controls. This finding was published in the *Molecular Genetics and Metabolism* in 2006. There was still much more work to be done to validate the assay. We contacted Walter Shaw at Avanti Lipids and paid for the custom synthesis of an authentic C26:0 lyso PC standard and a 4 deuterium labeled C26:0 lyso PC as an internal standard. With IRB permission, we obtained the newborn blood spots from known ALD patients born in the states of CA and MI. At the same time we also tested anonymous leftover newborn DBS from the States of MD, CA, the CDC and Costa Rica and found no positives. The ALD newborn DBS had a 5 to 15 fold increased C26:0 lyso PC with no overlap when compared with the anonymous newborn DBS. These findings were published in *Molecular Genetics and Metabolism* 2009. Since that time we have developed a high throughput LC/MSMS screening procedure and have published a combined extraction of the C26:0 lyso PC with that of the acyl carnitines. Recently together with the MD State Newborn Screening Lab, we have completed the screening of 5000 consented newborns born in 3 local Baltimore hospitals and did not find one positive, thus we believe that using our procedure the false positive rate will be low.

From the beginning of our interest in developing ALD newborn screening, our colleagues Drs. Piero Rinaldo, Silvia Tortorelli and Dieter Matern at the Mayo Clinic were very supportive of our initial efforts and made significant contributions to the design of our study and to our method of analysis. In addition at the newborn screening lab at the Mayo Clinic, they developed their own rapid high throughput method of LC/MSMS analysis of the newborn DBS for ALD and obtained funds to do 100,000 anonymous newborn DBS from the state of California. To date they have analyzed 60,000 and have found 20 positive samples. The plan is to confirm these positives by DNA sequencing of the ALD gene in these positive newborn DBS samples. Steven Steinberg, PhD and colleagues at the DNA Diagnostic Lab at Johns Hopkins will provide the DNA analyses. We expect to find that most of the positives, both males and females, will have a mutation in the ALD gene, but there may be a few positives from newborns with Zellweger spectrum disorders and these samples will be analyzed and confirmed by the Peroxisomal Lab at the Kennedy Krieger Institute.

In order to have ALD newborn screening confirmed for inclusion in the panel of disorders recommended for newborn screening nationally, Amber Salzman, PhD and Charlie Peters, MD prepared the nomination form for the inclusion of ALD. The nomination was reviewed and on September 13, 2012 Gerald Raymond, MD, Amber Salzman's 12 year old son, Spencer Barsh, 14 year old Taylor Kane, daughter of Jack Kane who died with ALD, and I testified before the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (HRSA) in Washington on behalf of many advocate organizations including The Stop ALD Foundation, ALD/AMN Global Alliance, Be A Hero Become A Donor, Fight ALD, The Myelin Project, Run4ALD, ELA and the ULF. Unfortunately the HRSA Review Committee did not recommend inclusion of ALD in the Recommended Uniform Screening Panel at this time. The following quotes are from a letter from Joseph A. Bocchini Jr, MD, Chairperson of HRSA dated 10/1/12.

"The Committee recognizes ALD as a medically important disorder that deserves serious consideration, possessing a well-established case definition as well as screening, diagnostic, and treatment protocols. However, at this time the Committee has decided to not send the nomination forward to the external review group

The Committee's decision is based primarily on the determination that sufficient prospective data is not yet available from the large pilot study presently underway at the Mayo Biochemical Genetics Laboratory (MBGL).

After the additional data from the MBGL study is made available to the Committee for evaluation, we encourage you to contact us to facilitate an expedited review. The Committee will then determine whether the new data provides sufficient support for the Committee to request a formal review of the scientific evidence by the external condition review group."

Based on the letter from HRSA we are proceeding with our plan to confirm the positive newborn DBS from the pilot study at Mayo. It is our hope that sufficient data will be available for the May 16th 2013 meeting of HRSA and that the Review Committee will accept the nomination that ALD newborn screening be forwarded to the external review group where it will take up to a year before the final recommendation of ALD to the Uniform Screening Panel.

Yesterday I forwarded 120 anonymous venous DBS from ALD, ALD heterozygotes, and Zellweger spectrum disorders from our collection of consented research samples to Silvia Tortorelli, MD at the Mayo Clinic so that the positive sample data collected by the Mayo Clinic can be augmented. We thank all the patients and their families for their willingness to provide samples for the ALD DBS screening data.

Ann Moser, January 9, 2013

NEWBORN SCREENING UNIFORM PANEL			
NOMINATION FORM FOR PROPOSED CONDITION			
Name of Proponent Charlie Peters, MD	Advocate Organizations: The Stop ALD Foundation, ALD/AMN Global Alliance, Be A Hero Become a Donor, Fight ALD, The Myelin Project, Run4ALD, ELA, ULF	Date May 4, 2012	
Condition	X-linked Adrenoleukodystrophy (ALD)		
Type of Disorder	Adrenal insufficiency and neurodegeneration		
Screening Method	High throughput screening assays and tandem mass spectrometry of dried blood spots		
Treatment strategy	Hormone replacement therapy for adrenal insufficiency, hematopoietic cell transplant (HCT) for cerebral demyelination		

CONDITION	Comment	Gene:	ABCD1	Locus:	Xq28	OMIM	300100
-----------	---------	-------	-------	--------	------	------	--------

*Note: Please reference each statement, listing references below (p.2)

Incidence	Determined by clinical identification. In the US, the estimated combined male and female frequency of ALD is 1:17,000 [1].
Timing of clinical onset	Distinct phenotypes exist with virtually all males developing adrenal insufficiency, some as early as 1 yr [2]. All phenotypes can occur in a kindred [1] with 31-35% of affected males having the demyelinating childhood cerebral form (CCER) [1] with typical onset between 4 and 8 yrs [1]. Boys develop normally until the onset of dementia and progressive neurologic deficits which lead to a vegetative state and death often within 3 yrs [1]. Forty to 46% of males with ALD present in mid-adulthood with slowly progressive paraparesis, sensory, and sphincter disturbances involving spinal cord long tracts (adrenomyeloneuropathy, AMN) [1]. At least 30% of men with AMN develop cerebral involvement that is similar to CCER [1]. Fifty per cent of heterozygous females develop overt neurologic disturbances resembling AMN, with a mean age of onset of 37 yrs [1].
Severity of disease	Untreated adrenal insufficiency can be fatal and untreated CCER is fatal. Earlier onset of CCER correlates with more severe, rapidly progressive clinical manifestations [3]. Boys with parieto-occipital lobe disease demonstrate visual and/or auditory processing abnormalities, impaired communication skills and gait disturbances, prior to death. The neuropsychological consequences have been described [1]. Boys with frontal lobe involvement have signs/symptoms similar to ADHD and are often misdiagnosed, prior to death. The extent of demyelination can be quantitated using the MRI severity score of Loes [4]. The presence of gadolinium contrast enhancement in areas of cerebral demyelination on brain MR imaging is highly positively predictive of clinically significant disease progression [3,5].
TEST	Comment
Screening test(s) to be used	Pilot 1: Analysis by tandem mass spectrometry with or without chromatographic separation of lysophosphatidyl choline (LPCs) species (C20 to C26) [6][13]. Pilot 2: Analysis of these compounds can be multiplexed with other analytes (e.g., acylcarnitines, lysosomal enzymes).
Modality of screening	Dried blood spots, the same specimen and collection modalities that are currently used for newborn screening tests
Clinical validation	<i>Pilot study 1:</i> 5,000 newborns completed by Kennedy Krieger Institute (KKI). <i>Pilot study 2:</i> 100,000 anonymous newborn specimens to be analyzed at Mayo Clinic for analytical validation (25,000 as of April 2012). <i>Clinical validation of the test (KKI):</i> 16/16 ALD newborn blood spots; 2/2 PBD (peroxisomal biogenesis disorders) newborn blood spots; 0/0 ALD carrier newborn blood spots; 105/105 ALD not newborn blood spots, 66/66 PBD not newborn blood spots, 95/118 ALD carrier not newborn blood spots [6]. <i>Clinical validation of the test (Mayo Clinic):</i> 20/20 ALD newborn blood spots; 2/2 PBD newborn blood spots; 3/5 ALD carrier newborn blood spots, 12/12 ALD non-newborn blood spots; 6/6 PBD non-newborn blood spots; 9/12 ALD carrier non-newborn blood spots [7].
Laboratory performance metrics	<i>Pilot study 1:</i> true positive cases: 0; false positive cases: 0.[14] <i>Pilot study 2:</i> true positive cases: to be determined (TBD) false positive cases: TBD (expected: << 0.1%)
Confirmatory testing	Analysis of very long-chain fatty acids in plasma by GC/MS. Elevated in 99.9% of affected males and 85% of heterozygous females. Mutation analysis of the ABCD1 gene. In addition, the KKI Peroxisomal Diseases Laboratory has set up analyses of plasmalogens and peroxisomal bile acid intermediates on dried blood spots [7].
Potential harms of screening and testing	Patients affected with peroxisomal biogenesis disorders and 70-85% of ALD heterozygous females will be detected by this assay. Post analytical tools based on the R4S model are available to discriminate these cases from females affected with other peroxisomal disorders [7].

NOMINATION OF CONDITION (page 2)	
TREATMENT	Comment
Modality	Maintenance and stress-dosing adrenal hormone replacement therapy is the standard of care for primary adrenal insufficiency including that associated with ALD [2]. HCT is the only effective long-term treatment for CCER; however, to achieve optimal survival and clinical outcomes, HCT must occur prior to manifestations of symptoms [7-10]. Gene therapy experimental treatment has been shown to be safe and efficacious [11].
Urgency	It is imperative to implement by 3 months the following: (a) adrenocortical function testing to detect adrenal insufficiency and by 3 yrs (b) serial neuroimaging to detect the earliest evidence of demyelination [9,12]; therefore timely diagnosis is critical. The recommended evaluation for boys 3 to 15 yrs is comprehensive neurologic, neuropsychological, neuroradiologic, and adrenal function evaluations at diagnosis with serial monitoring at least every 6 months during the 1 st decade of life and annual monitoring in the 2 nd decade [9,12]. The goals are: (a) to detect cerebral disease early, prior to the development of neuropsychological and/or neurologic signs/symptoms, and (b) to identify adrenal insufficiency, a potentially life-threatening condition, and to treat it. Monitoring is essential and critical since phenotype cannot be predicted and there is no genotype-phenotype correlation. Early knowledge of an ALD diagnosis is critical for the treatment of a patient during the narrow therapeutic window [9].
Efficacy (Benefits)	Reports have described the initial success of HCT for a patient with CCER [7], long-term beneficial effects of HCT [8], and large international HCT experience [9]. By using this monitoring strategy (see above), timely and effective HCT can be achieved i.e., 95% 5-year survival, with excellent clinical outcomes compared to 54% survival for a similar group not treated by HCT [10]. Of note, boys in the untreated group progressed to a vegetative state and death. Survival for transplanted patients is 92% for boys with <u>early stage</u> brain disease compared with 45% at 5 years for patients with <u>late stage</u> disease [9]. Identification of ALD can lead to timely diagnosis of adrenal insufficiency and initiation of hormone replacement therapy [2]. A metabolic crisis due to unrecognized and consequently untreated adrenal insufficiency can be fatal or result in significant morbidity with long-term sequelae including profound, rapid neurologic deterioration in boys with CCER [2,9].
Availability	Adrenal hormone therapy is available to treat adrenal insufficiency. Due to volunteer bone marrow donor registries, umbilical cord blood banks, HLA-matched related donor(s) and autologous hematopoietic cells (HCs), a suitable source of therapeutic HCs is available for all patients [11,12]. This applies to patients with CCER as well [9,11].
Potential harms of treatment	Adrenal hormone therapy for adrenal insufficiency has no adverse effects but, rather, can be life-saving [2]. While HCT carries risk of morbidity (e.g., acute and chronic GVHD, cardiac, pulmonary, GI, skeletal and endocrine complications) and mortality, HCT safety is markedly enhanced when it is performed prior to clinical manifestations and at an early stage of disease in CCER patients [9,12].

KEY REFERENCES (Specific citations – limit to 15)

1	Moser HW, Mahmood A, Raymond GV. X-linked Adrenoleukodystrophy. <i>Nat Clin Pract Neurol</i> 2007; 3:140-51.
2	Dubey P, Raymond GV, Moser AB, et al. Adrenal insufficiency in asymptomatic adrenoleukodystrophy patients identified by very long-chain fatty acid screening. <i>J Pediatr</i> 2005;146:528-32.
3	Moser HW, Loes DJ, Melhem ER, et al. X-linked adrenoleukodystrophy: overview and prognosis as a function of age and brain magnetic resonance imaging abnormality. A study involving 372 patients. <i>Neuropediatrics</i> 2000;31:227-39.
4	Loes DJ, Hite S, Moser H, et al. Adrenoleukodystrophy: a scoring method for brain MR observations. <i>AJNR Am J Neuroradiol</i> 1994;159:1761-6.
5	Melhem ER, Loes DJ, Georgiades CS, Raymond GV, Moser HW. X-linked adrenoleukodystrophy: the role of contrast-enhanced MR imaging in predicting disease progression. <i>AJNR Am J Neuroradiol</i> 2000;21:839-44.
6	Hubbard WC, Moser AB, Liu AC, et al. Newborn screening for X-linked adrenoleukodystrophy (X-ALD): validation of a combined liquid chromatography-tandem mass spectrometric (LC-MS/MS) method. <i>Mol Genet Metab</i> 2009 97:212-20.
7	Aubourg P, Blanche S, Jamabaque I, et al. Reversal of early neurologic and neuroradiologic manifestations of X-linked adrenoleukodystrophy by bone marrow transplantation. <i>N Engl J Med</i> 1990;322:1860-6.
8	Shapiro E, Krivit W, Lockman L, et al. Long-term effect of bone-marrow transplantation for childhood-onset cerebral X-linked adrenoleukodystrophy. <i>Lancet</i> 2000;356:713-6.
9	Peters C, Charnas LR, Tan Y, et al. Cerebral X-linked adrenoleukodystrophy: the international hematopoietic cell transplantation experience from 1982 to 1999. <i>Blood</i> 2004;104:881-8.
10	Mahmood A, Raymond GV, Dubey P, Peters C, Moser HW. Survival analysis of hematopoietic cell transplantation for childhood cerebral X-linked adrenoleukodystrophy: a comparison study. <i>Lancet Neurol</i> 2007;6:687-92.
11	Carter N, Hacein-Bay-Abina S, Bartholomae CC, et al. Hematopoietic stem cell gene therapy with a lentiviral vector in X-linked adrenoleukodystrophy. <i>Science</i> 2009;326:818-23.
12	Appelbaum FR, Forman SJ, Negrin RS, Blume KG, eds. <i>Thomas' Hematopoietic Cell Transplantation</i> . 4 th ed. Oxford: Wiley-Blackwell, 2004, chapters: 11-13, 15-17, 19, 21-23, 25, 27, 29, 30-31, 33, 34, 36-39, 46-48, 77, 80-108.

13	Sandlers Y, Moser AB, Hubbard WC, Kratz LE, Jones RO, Raymond GV. Combined extraction of acyl-carnitines and 26:0-lysophosphatidylcholine from dried blood spots; prospective newborn screening for X-linked adrenoleukodystrophy <i>Molec Genetics Metabol</i> , 2012;105:416-420.
14	Statement from Kennedy Krieger

Submission Check list		Submit Nominations to:
	Cover letter by proponent	Sara Copeland, MD Acting Chief, Genetics Services Branch Division of Services for Children with Special Health Needs Maternal and Child Health Bureau 5600 Fishes Lane, Room 18-A-19 Rockville, MD 20857 301-480-1312 - fax 301-443-1080 - phone
	Nomination form	
	Copy of references listed on this form	
	Formal conflict of interest statement by proponent	
Contact information (proponent)		
Charlie Peters, MD <u>cjpeters1982@earthlink.net</u> (612) 760-6192		



DEPARTMENT OF HEALTH AND HUMAN SERVICES

Secretary's Advisory Committee on Heritable
Disorders in Newborns and Children
5600 Fishers Lane, Room 18A19
Rockville, Maryland 20857
(301) 443-1080 – Phone
www.hrsa.gov/advisorycommittees

October 1, 2012

Charles Peters, M.D.
48055 252nd Street
Garretson, SD 57030

Amber Salzman, Ph.D.
The Stop ALD Foundation
500 Jefferson Street, Suite 2000
Houston, Texas 77002-7371

Dear Drs. Peters and Salzman:

JB465

The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (Committee) appreciates your nomination of X-linked Adrenoleukodystrophy (ALD) for inclusion in the Recommended Uniform Screening Panel (RUSP) for state newborn screening programs. As part of the formal review process, the Nomination and Prioritization Committee conducted a preliminary review of the nomination package, and results were presented for discussion and a decision during the September 2012 Committee meeting. A copy of the presentation is enclosed.

The Committee recognizes ALD as a medically important disorder that deserves serious consideration, possessing a well-established case definition as well as screening, diagnostic, and treatment protocols. However, at this time, the Committee has decided to not send the nomination forward to the external condition review group.

The Committee's decision is based primarily on the determination that sufficient prospective data is not yet available from the large pilot study presently underway at the Mayo Biochemical Genetics Laboratory (MBGL).

After additional data from the MBGL study is made available to the Committee for evaluation, we encourage you to contact us to facilitate an expedited review. The Committee will then determine whether the new data provides sufficient support for the Committee to request a formal review of the scientific evidence by the external condition review group.

Please contact Lisa Vasquez (lvasquez@hrsa.gov) when you are prepared to submit any additional data or if you have any questions or concerns.

Thank you for your nomination of ALD for inclusion in the RUSP for state newborn screening programs.

Sincerely yours,

A handwritten signature in black ink, appearing to read "Joseph A. Bocchini Jr.", written in a cursive style.

Joseph A. Bocchini Jr., M.D.
Chairperson

Enclosure:
SACHDNC Nomination and Prioritization Workgroup Presentation: ALD



State of Connecticut
 HOUSE OF REPRESENTATIVES
 STATE CAPITOL
 HARTFORD, CONNECTICUT 06106-1591

Representative Lonnie Reed
 Chair - Energy and Technology Committee
 102nd Assembly District
 Branford

Legislative Office Building
 Hartford, Connecticut 06106
 860-240-8585 or 800-842-8267
 Lonnie.Reed@cga.ct.gov

Testimony of Representative Lonnie Reed
In support of SB 465 Act Requiring Newborn Screening for Adrenoleukodystrophy
February 27, 2013

Good morning to Committee Chairs, Sen. Gerratana and Representative Johnson; the Vice Chairs, Sen. Slossberg and Rep. Miller and the Ranking Members, Sen. Welch and Rep. Srinivasan, and all members of the Public Health Committee.

My name is Lonnie Reed, State Representative for Branford. I am here to support SB 465 and to talk to you about one of our town's most beloved and awe inspiring families.

Jean and John Kelly and their warrior son Brian are asking you – asking us all – to help other families avoid the devastating, life changing health challenge that has befallen them.

Brian Kelly appeared to be a totally healthy, rambunctious little boy until a sledding accident at the age of six revealed that he had Adrenoleukodystrophy (ADL), a gene-linked, inherited, deteriorating condition that can lead to total incapacitation and early death. It is a condition that can now be detected and often controlled with interventional therapies by adding ADL to the diseases that are easily tested for in newborns.

You will undoubtedly hear more about the human costs of ADL.

I want to talk just a bit about the financial costs that we all pay.

The Kellys asked the Branford School system to tally up the additional price paid to educate Brian, given his disabilities.

Some of the line items include special transportation costs of \$46,000 per year; Special Computer Hardware/Software and Assistive Technology Evaluation that came to \$35,000 for Brian's twelve years in school. The grand total of extra costs came to almost \$1-Million Dollars.

The human costs and the societal costs could have perhaps all been prevented if there had been the automatic testing of newborns for ADL.

I have to say that throughout his years in our Branford schools, Brian's strength, determination and refusal to give up inspired his classmates and his educators, teaching them valuable lessons they would not have learned otherwise.

Brian is still teaching us and he is asking us today to please vote for SB 465, Act Requiring Newborn Screening for Adrenoleukodystrophy, ADL. What a legacy that would be for Brian and his family and all for Connecticut families who could be spared the heartbreaking, life altering destruction caused by ADL.

I am grateful to you for hearing this bill. I ask you to please keep supporting it and to help it become law. Thank You.



STATE OF CONNECTICUT

DEPARTMENT OF PUBLIC HEALTH

**TESTIMONY PRESENTED BEFORE THE JOINT COMMITTEE ON PUBLIC HEALTH
February 27, 2013**

Jewel Mullen, MD, MPH, MPA, Commissioner (860) 509-7101

**Senate Bill 465 – AN ACT REQUIRING NEWBORN SCREENING FOR
ADRENOLEUKODYSTROPHY**

The Department of Public Health is opposed to Senate Bill 465

Adrenoleukodystrophy (ALD) is an X-linked inherited disorder of very long chain fatty acid metabolism which results in a build-up of these fatty acids in the nervous system, adrenal glands and testes. The incidence of ALD is approximately 1/17,000 births, predominately affecting boys. The disease manifests itself in three forms: a cerebral form affecting children between the ages of four to eight; adrenomyelopathy, affecting men in their twenties; and adrenal gland failure. Other than treatment of adrenal gland failure with steroids, treatment for the other forms of ALD is still experimental and involves diet very low in very long chain fatty acids, supplemented with other oils (Lorenzo's oil). Bone marrow transplant is also being considered.

In October 2012, the Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children responded to a letter from The Stop ALD Foundation indicating that they would not nominate the disorder for external review because there was not sufficient data to support inclusion on the recommended Newborn Screening (NBS) panel. However, they also noted that the Mayo Clinic is undergoing a large pilot study testing for ALD, to be completed at the end of 2013, the results of which will be used by the Committee to decide whether to move ALD into the nomination and prioritization process.

Currently, there are no public health laboratories that screen newborns for ALD. As such, there are no standard methods available for the test. Consequently, it would take an enormous effort for the Connecticut Newborn Screening program to develop and validate an in-house method to test for ALD. Time will be required for method development and validation to enable routine screening to be implemented by the NBS laboratory. A minimum of 5,000 dried blood spots would have to be tested before the method could be validated. The Laboratory tests all newborns in Connecticut for over 40 disorders on its NBS panel. The addition of another disorder for routine screening, including method development and validation, would require additional staff, equipment and supplies. These resources are not included in the Governor's proposed budget.

Analysis for this disorder also presents additional problems with the identification of many individuals with one normal gene and one gene for the disorder, as well as secondary target disorders that will be inevitably identified through screening. This carrier and secondary disorder identification may prove problematic for treatment and follow-up at the level of the overseeing physician.

Thank you for your consideration of the Department's views on this bill

*Phone: (860) 509-7269, Fax: (860) 509-7100
Telephone Device for the Deaf (860) 509-7191
410 Capitol Avenue - MS # 13GRE
P.O. Box 340308 Hartford, CT 06134
An Equal Opportunity Employer*



State of Connecticut
HOUSE OF REPRESENTATIVES
 STATE CAPITOL
 HARTFORD, CONNECTICUT 06106-1591

REPRESENTATIVE KIM FAWCETT
 ASSISTANT MAJORITY LEADER
 ONE HUNDRED THIRTY-THIRD ASSEMBLY DISTRICT

LEGISLATIVE OFFICE BUILDING
 ROOM 4033
 HARTFORD, CT 06106-1591

CAPITOL 860-240-8585
 TOLL FREE 1-800-842-8267
 FAX 860-240-0206
 E-MAIL: Kim.Fawcett@cga.ct.gov

VICE CHAIRMAN
 SELECT COMMITTEE ON CHILDREN

MEMBER
 FINANCE, REVENUE & BONDING COMMITTEE
 ENERGY AND TECHNOLOGY COMMITTEE

Testimony of State Representative Kim Fawcett, 133rd District
 Public Health Committee, Public Health Hearing
 February 20, 2013

Proposed S.B. No. 466 An Act Concerning Continuing Education Requirements For Medical & Sister Bill H.B. No. 6238

Senator Gerratana, Representative Johnson and distinguished members of the Public Health Committee, thank you for the opportunity to comment on the SB 466, An Act Concerning Continuing Education Requirements For Medical Professionals. This bill mimics in content a similar bill I sent to this committee, HB 6238.

Doctors throughout our state are required each year to take continuing education requirement credits to maintain their professional licensing and ability to practice in our state. For many years this continuing education system has been onerous and unreasonable for our medical professionals. While no one disagrees that some form of continuing education is necessary and helpful, mandating the actual course selection has proven less beneficial, even antiquated.

Doctors are required to take the same five courses every two years, and over time are wasting time and resources repeating the courses over and over. The repeated courses include:

A) Infectious diseases, including, but not limited to, acquired immune deficiency syndrome and human immunodeficiency virus, B) risk management, C) sexual assault, D) domestic violence and e) cultural competency

Our suggestion is to allow doctors to complete continuing education requirements in areas of study that are more current and targeted to the medical fields in which they work.

I look forward to working with the members of this committee to develop a more appropriate continuing education system for our state.