

Gerard Vockley, MD, PhD

Doctor

Chief, Division of Medical Genetics
Director, Center for Rare Disease Therapy
Professor of Pediatrics, University of Pittsburgh School of Medicine
Professor of Human Genetics, University of Pittsburgh Graduate School of Public Health
Cleveland Family Professor of Pediatric Research, University of Pittsburgh

Services

[Center for Rare Disease Therapy](#)
[Medical Genetics](#)
[Metabolic Disease](#)

Locations and Directions

Pediatric Medical Genetics

Specialty Care Center Hermitage

80 E. Silver St.
Sharon, PA 16146 United
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(724) 347-4596

[Map](#)

Pediatric Medical Genetics

Children's Hospital of Pittsburgh of UPMC

4401 Penn Ave.
Pediatric Medical Genetics -
3rd Floor
Pittsburgh, PA 15224 United
States
(412) 692-5070

[Map](#)

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Biography

Research Interests

- Metabolism of Short Chain Acyl-CoAs and Its Deficiency, Principal Investigator
- Molecular Characterization of Acyl-CoA Dehydrogenases, Principal Investigator

Biography Summary

Gerard Vockley, MD, PhD, is an internationally recognized leader in medical genetics and the field of inborn errors of metabolism. He joined Children's Hospital of Pittsburgh as chief of the Division of Medical Genetics in 2004 and was named professor of pediatrics at the University of Pittsburgh School of Medicine and professor of human genetics at the university's Graduate School of Public Health.

Dr. Vockley, a native of Homestead, Pa., earned a bachelor's degree in biology from Carnegie Mellon University in 1978 and completed his medical degree and a doctorate in genetics at the University of Pennsylvania School of Medicine, Philadelphia, in 1984. Following his residency in pediatrics at the Denver Children's Hospital in 1987, he completed a fellowship in pediatrics and human genetics at Yale University School of Medicine.

In 1991, Dr. Vockley joined the faculty of the Mayo Clinic School of Medicine, where he was engaged in teaching, clinical service and research until he moved to Children's Hospital of

yong.xie@outlook.com

From: Vockley, Gerard
<vockleyg@upmc.edu>
Sent: Thursday, April 24, 2014 3:53 AM
To: Yong Xie
Subject: Re: help on possible FAOD

Thank you for the information. It is certainly abnormal, but is not consistent with ACAD9 deficient in which newborn screening has been normal in all cases known to me. The DNA sequencing data shared with me by Dr. Merritt really does rule out ACAD9 deficiency. I suggest you work with Dr. Merritt to find the correct one.

Dr. Vockley

Jerry Vockley, M.D., Ph.D.

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Genetics, Graduate School of Public Health Chief of Medical Genetics
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From: Yong Xie
<yong.xie@outlook.com<mailto:yong.xie@outlook.com>>
Date: Thursday, April 24, 2014 at 2:32 AM
To: Jerry Vockley <vockleyg@upmc.edu<mailto:vockleyg@upmc.edu>>
Subject: RE: help on possible FAOD

Dear Dr. Vockley,

I think we didn't send you Jianhua's newborn screen last time, which may be valuable for research. His C-18, C-18:1, and C-16 are extremely high (6X, 3X, and 10X compared to SCH's maximum values respectively while other acylcarnitine values are quite normal (only C-3 at 1X).

We still think it's almost certain that Jianhua has ACAD9 even after Dr. Merritt told us that his liver problem was very likely from his immune system during our Apr-11 visit. The only reason we cannot make a decision to visit you immediately is Jianhua's health, he is still too weak. In the meantime, we start worrying about his brother. Jianhua's older brother, Zhenghua, may also have the same problem because we observed low platelet level on him a couple times. (Blood spots beneath his skin, unexplained reason. Tested once with low platelet) I am not sure if I can get Zhenghua's NBS as Jianhua's. (Pure luck, a nice person sent it to us last year.)

If you think you can use Jianhua's medical record for research, we would like to release all medical information to you any time.

Best Regards

Yong

-----Original Message-----

From: Evans (Grechen), Annette [mailto:annette.evans@chp.edu]

Sent: Wednesday, April 09, 2014 4:34 AM

To: Yong Xie

Subject: Re: help on possible FAOD

Yong,

You are welcome. All the best to you and your family.

Regards,

Annette

Annette M. Evans

Sr. Administrative Assistant

Medical Genetics/PKU Depts.

Children's Hospital of Pittsburgh

One Children's Hospital Drive

4401 Penn Avenue

Pittsburgh, PA 15224

annette.evans@chp.edu<mailto:annette.evans@chp.edu>

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On 4/8/14 10:13 PM, "Yong Xie"
<yong.xie@outlook.com<mailto:yong.xie@outlook.com>> wrote:

>Annette,

>

>It's good to know that Dr. Vockley had talked to our physician, Dr.

>Merritt. We hope that we can get a diagnosis soon. It's just been a

>year since we rushed our baby to SCH on April 9, 2013. It took us a

>long time to figure out what is wrong and who are the experts in this
field.

>

>If we still cannot get a final diagnosis, we will certainly need Dr.

>Vockley's help. Please convey our appreciation to Dr. Vockley.

>

>Thanks a lot.

>

>Yong

>

>

>

>

>-----Original Message-----

>From: Evans (Grechen), Annette [mailto:annette.evans@chp.edu]

>Sent: Tuesday, April 08, 2014 4:51 AM

>To: 永谢

>Subject: Re: help on possible FAOD

>

>Good Morning Yong,

>

>I spoke with Dr. Vockley and he will be unable to see your baby during

>his brief trip to Seattle.

>

>He has, however, spoken to Dr. Merritt. Dr. Vockley made a few

>suggestions to Dr. Merritt and he is very comfortable with the care
>plan

>by Dr. Merritt. He believes there is no need for your visit to

>Pittsburgh but you can schedule a visit if you feel it is absolutely

>necessary.

>

>Sincerely,

>Annette

>

>

>Annette M. Evans

>

>Sr. Administrative Assistant

>
>Medical Genetics/PKU Depts.
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>Children's Hospital of Pittsburgh
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>

>

>From: 永谢

><yong.xie@outlook.com<<mailto:yong.xie@outlook.com><<mailto:yong.xie@outlook.com>>>>

>ok.com%3c<mailto:yong.xie@outlook.com>>>>

>Date: Monday, April 7, 2014 4:02 PM

>To: Annette Evans

><annette.evans@chp.edu<<mailto:annette.evans@chp.edu><<mailto:annette.evans@chp.edu>>>>

>@chp.edu%3c<mailto:annette.evans@chp.edu>>>>

>Subject: FW: help on possible FAOD

>

>Annette,

>

>This is Yong Xie. My wife, Liying and I are wondering if it is possible

>that our baby gets treated by Dr. Vockley while he is in Seattle. We

>may be able to fly over a couple of times in summer to meet Dr.

>Vockley; however, it's very hard for us to stay there long, especially

>in winter because his health condition and financial reason. (Insurance

>coverage and other related cost.)

>

>Liyong sent Dr. Vockley an email about 10 days ago; and we think Dr.

>Vockley's reply is very informative. (Below, the attached emails.) We

>also think Dr. Vockley is willing to be his doctor, and we have no

>reservation on using his medical information for research. Actually, we

>hope that we can know our baby better through Dr. Vockley's help.

>

>

>If we can do any kind of arrangement, please let us know.

>

>Thanks

>

>Yong

>

>

>_____

>From:

>liyong.lc.chen@hotmail.com<mailto:liyong.lc.chen@hotmail.com<mailto:liyong.lc.chen@hotmail.com>>

>liyong.lc.chen@hotmail.com%3cmailto:liyong.lc.chen@hotmail.com>>

>To:

>yong.xie@outlook.com<mailto:yong.xie@outlook.com<mailto:yong.xie@outlook.com>>

>yong.xie@outlook.com%3cmailto:yong.xie@outlook.com>>

>Subject: FW: help on possible FAOD

>Date: Mon, 7 Apr 2014 06:51:42 -0700

>

>

>

> _____

>From:

>liying.lc.chen@hotmail.com<mailto:liying.lc.chen@hotmail.com<mailto:liy

>ing.lc.chen@hotmail.com%3cmailto:liying.lc.chen@hotmail.com>>

>To:

>gerard.vockley@chp.edu<mailto:gerard.vockley@chp.edu<mailto:gerard.vock

>ley@chp.edu%3cmailto:gerard.vockley@chp.edu>>

>Subject: RE: help on possible FAOD

>Date: Tue, 1 Apr 2014 22:17:23 -0700

>

>Dear Dr. Vockley,

>

>Thanks a lot for your kind reply. Jianhua's treating physician is Dr.

>Lawrence Merritt at Seattle Childrens' Hospital. We have forwarded your

>message to him. He may contact you soon. We will bring Jianhua to see Dr.

>Merritt in 10 days. Hope we will learn your recommendations from him.

>Jianhua's health is still delicate. He has had 3 significant RSV

>infections since born. His most recent one was 2 weeks ago. He had high

>fever and difficulty to breathe. He sweated much but ate or drank

>nearly nothing. After several sleepless nights, we had to rush him to

>the ER due to his dehydration. He just recovered for a day or two. At

>the moment we dare not take him far from home even though we would very

>much like to take him to see you. We will bring him to Pittsburg to be

>your patient after he grows stronger.

>

>Thanks again for your email. We will keep you posted on Jianhua's

>progress. We really appreciate your help.

>

>Sincerely yours

>

>Liyong

>

>

>

>

>

>

>> From:

>>

vockleyg@upmc.edu<mailto:vockleyg@upmc.edu<mailto:vockleyg@upmc.edu%3

>> cmailto:vockleyg@upmc.edu>>

>> To:

>>

liying.lc.chen@hotmail.com<mailto:liying.lc.chen@hotmail.com<mailto:|

>> iying.lc.chen@hotmail.com%3cmailto:liying.lc.chen@hotmail.com>>

>> Subject: Re: help on possible FAOD

>> Date: Sun, 30 Mar 2014 13:52:37 +0000

>>

>> Ms. Chen,

>>

>> it is not possible for me to comment directly on your child's

>>condition as I have not had the opportunity to evaluate him

>>personally. I can say that the type of liver disease you describe has
>>not been described in fatty acid oxidation defects and that the
>>acylcarnitine profile and exome sequencing results are not consistent
>>with such a diagnosis, including ACAD9 deficiency. If your physicians
>>would be interested in my opinion I would be happy to speak with
them directly.

>>

>> Regards,

>>

>> Dr. Vockley

>> _____

>> Jerry Vockley, M.D., Ph.D.

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>> Professor of Pediatrics, School of Medicine Professor of Human

>> Genetics, Graduate School of Public Health Chief of Medical Genetics

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>> ailto:gerard.v>

>> o

>> ckley@chp.edu<mailto:ckley@chp.edu>>

>>

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Thank you.

>>

>>

>> On Mar 30, 2014, at 2:08 AM, Liying Chen

>><liying.lc.chen@hotmail.com<mailto:liying.lc.chen@hotmail.com<mailto:liying.lc.chen@hotmail.com%3cmailto:liying.lc.chen@hotmail.com>>>
<mailto:

>>liy

>>ing.lc.chen@hotmail.com<mailto:ing.lc.chen@hotmail.com>>>
wrote:

>>

>> Dear Dr. Vockley,

>>

>> This is Liying Chen, mother of a very sick 18-month old baby. I am

>>writing to you to ask for your help in finding out if my baby has

>>fatty acid oxidation disorders (FAOD). We may save him from future

>>liver transplantation if we can manage and treat him properly with your help.

>>

>> Our baby, Jianhua Xie, was diagnosed with micro nodular liver

>>cirrhosis in April 2013 and was put on liver transplant waiting list.

>>In June 2013, he was changed to on hold in the list because he improved so much.

>>The only change in the two months was that his doctor switched him to

>>a MCT formula Pregestimil.

>>

>> His doctors did extensive tests on him last year trying to find out

>>what causes his liver damage, but we have not received a definite

>>answer. A possible cause proposed by several doctors was a metabolic

>>disease. Actually, they tested his blood acylcarnitine, urine organic

>>acid, and plasma amino acid profiles within his first several days in

>>PICU. None of them were normal. However, his doctors considered those

>>results not evident enough to determine a clear diagnosis. For example

>>in his blood acylcarnitine profile C16:0 and C16:1 levels were just

>>slightly higher than normal values while others were within range.

>>

>> Since his whole exome sequencing (WES) showed a deleterious mutation

>>in his ACAD9 gene in December 2013, we have begun to work with his

>>biochemical genetics doctor to do more test on our baby. The latest

>>MitoMetPlus aCGH test shows negative result. His doctor thought he

>>should take some other tests instead of genetic tests. He also said

>>you are the expert in the field and your suggestions will be very helpful.

>>Therefore, I am writing to you for your help.

>>

>> These files are attached for your review:

>>

>>

>> 1. Acylcarnitine Profile Blood, 04-12-2013 2. Urine Organic Acids,

>> 04-11-2013 (Comments: no MCT supplements used then) 3. Amino Acids

>> Plasma, 04-09-2013 4. Genetic, Whole Exome Sequencing, focused

>> report, 12-2013.

>> 5. Genetic, MitoMetPlus aCGH, 03-2014.

>> 6. Surgical pathology Final Report, 04-18-2013 7. Pathologic

>> Diagnosis from MGH, 07-2013 8. Op Note, 04-18-2013

>>

>>

>> Jianhua went to ER on 04-09-2013, and then he was transferred to PICU

>>on 04-10-2013. Both genetic tests were done at Baylor College of

>>Medicine.

>>

>> We'd like to ask some questions:

>>

>>

>> 1. Based on the results of his blood acylcarnitine, urine amino acid,

>>and liver surgical pathology, can we say that our baby has FAOD?

>> 2. Is

>>f<<http://www.mayomedicallaboratories.com/test-catalog/Overview/81927>>a

>>tty

>> acid oxidation probe

>><<http://www.mayomedicallaboratories.com/test-catalog/Overview/81927>>

>>assay<<http://www.mayomedicallaboratories.com/test-catalog/Overview/819>

>>27> a good test for our baby? Where is a good place to do the test?

>>We only found one such test from Mayo Medical Laboratories. Will they

>>be able to do the test reliably since ACAD9 gene is so new?

>> 3. Are there any other tests which our baby may take? Could you

>>please recommend to us both the test names and the lab names?

>> 4. In the MitoMetPlus aCGH report, we found the technique is MitoMet

>>V3.1 oligonucleotide microarray. A friend once suggested that our baby

>>take CMA test V9.1. Are these two tests using the techniques of the

>>same version? Is it possible that MitoMet V3.1 is not up-to-date? If

>>so, what shall we do?

>> 5. Some tissue samples from his surgical liver biopsy may still be

>>available. Are those liver tissue samples useful to do the FAO probe

>>assay test mentioned in question 2?

>> 6. He developed RSV type symptom (coughing, wheezing, high

>>temperature,

>>etc.) in December 2013 after we fed him full eggs for two weeks. But

>>his pediatrician couldn't find any infection. He recovered after we

>>stopped giving him eggs. Is it related to FAOD? Do you have any comments?

>> 7. We gave him formulas rich in high oleic sunflower oil since he was

>>born. He developed many problems after a month and eventually went to

>>ER at 6 month old. He also responded badly to Nutramigen formula,

>>which has more vegetable oil (26%). We are wondering if vegetable oil

>>is more difficult for him to metabolize than animal fat. We want to

>>know if he will be able to eat a little red meat even if he has FAOD.

>> 8. Do you know any young patient who has grown out of micro-nodular

>>liver cirrhosis? In other words, have you ever heard of a miracle like

>>this?

>>

>>

>> We have a lot more of his medical records for you to review if you

>>are interested. We will also be glad to sign any forms to release the

>>information to you if you are willing to use his medical record and

>>genetic data for research. We will really appreciate your kind help.

>>We hope you will take Jianhua as your patient.

>>

>> Sincerely yours,

>>

>> Liying Chen

>> <Acylcarnitine Profile Blood, 2013-04-12.pdf><Amino Acids Plasma,

>>2013-04-09.pdf><Generic, Whole Exome Sequencing, Focused,

>>2013-12.pdf><Genetic, MitoMetPlus aCGH.pdf><Op Note,

>>2013-04-18.pdf><Pathologic Diagnosis from MGH, 2013-07.pdf><Surgical

>>Pathology Final Report, 2013-04-18.pdf><Urine Organic Acids (Urine

>>Succinylacetone), 2013-04-11.pdf>

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