In The Matter Of:

Eric Gwynne, etc. v.
University Hospitals of Cleveland, et al.

Arthur Zinn, M.D.
January 22, 2003

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Original File ***030122AZ.VI*****,** ***59*** *Pages*
Min-U-Script® File ***ID: I689268261***

Word Index included with this Min-U-Script®

IN THE COURT OF COMMON PLEAS
CUYAHOGA COUNTY, OHIO
ERIC GWYNNE, ADMINISTRATOR
of the ESTATE OF EMILY
GWYNNE, deceased,
Plaintiff,
JUDGE GAUL
-vs- CASE NO. 468327
UNIVERSITY HOSPITALS OF
CLEVELAND, et ai.,

Defendants.

Deposition of ARTHUR ZINN, M.D., taken as il upon cross-examination before Pamela S. Greenfield, a Registered Diplomate Reporter, Certified Realtime Reporter and Notary Public within and for the State of Ohio, at the offices of Reminger & Reminger, 1400 Midland Building, Cleveland, Ohio, at 11:00 a.m. on Wednesday, January 22, 2003, pursuant to notice and/or stipulations of counsel, on behalf of the Plaintiff in this cause.

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Drs. Fanaroff, Zinn and Rodriguez.

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WITNESSINDEX

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ARTHUR ZI", M.D., of lawful age, called by the Plaintiff for the purpose of cross-examination, as provided by the Rules of Civil Procedure, being by me first duly sworn, as hereinafter certified, deposed and said as follows:

CROSS-EXAMINATIONOF ARTHUR ZI", M.D.

BY MS. KQLIS:

Q: Doctor, just for identification purposes on the record, would you state your name and your professional address.

A: My name is Arthur Brian Zinn and I work at University Hospitals of Cleveland and Case Western Reserve University.

Q: Doctor, of course you know my name is Donna Kolis and I've been retained to represent the Estate of Emily Gwynne. My purpose today at this deposition is to find out what actual involvement you had in the care and treatment or consultation of this particular patient as well as to talk to you a little bit about LCHAD.

Doctor, I know that you've given a deposition before. I'm correct in that assumption?

A: I've given one deposition.

Q: Was that the deposition that you gave, I believe

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it was August of 2000, perhaps? Do you recall the deposition that you gave?

A: I don't recall the date.

Q: Do you know what case it was involving?

MS. DiSILVIO: Objection. You may answer. You may answer.

A: It was a case of Burdette.

Q: That's the only one you recall giving a deposition in?

A: You need to help me.

Q: Sure.

A: Is a deposition when I'm a defendant or I'm an expert witness?

Q: Sometimesyou could be a defendant, sometimes you could be an expert.

A: Okay.

Q: Sometimes you could be a subsequent treating physician.

A: Thank you. So the only deposition I gave as a defendant was in Burdette.

I've given other depositions, I think two as an expert witness.

Q: All right. We'll get into that in just a second.

Essentially, the ground rules are pretty simple. I ask questions. Hopefully you

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understand the questions, understanding that I'm not a physician. If you answer a question, I'm going to assume you understood it.

Therefore if there's any hesitancy in your own mind as to what information I'm seeking, you get to tell me that. Is that all right with you?

A: Yes.

Q: On occasion, Marilena may interpose an objection. You should wait until she and I resolve our differences before you answer the question. Do you understand that?

A: Yes.

Q: Okay. And so far you're doing wonderful. Each and every question has to be answered orally so that the court reporter can take down your answer. Is that acceptable to you?

A: Yes.

Q: Okay. Doctor, in anticipation of today's deposition, what materials did you review?

A: The medical records.

Q: Specifically what medical records did you review?

A: The ones Ms. DiSilvio prepared and gave me.

Q: Well, I don't know what she prepared and gave you, so you're going to have to tell us **what** medical records those are, facilities, dates,

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whatever you have.
MS. DISILVIO: Here. Let me help you out.
A: I need some help.
Q: If your attorney needs to help you during the deposition that's okay, too, as long as she doesn't answer for you.
A: All I have to do is read what she prepared for me?
Q: That's fine.
A: Thank you. Index to medical records Volumes 1 through 4 University Hospitals of Cleveland.
Q: Have you seen —
A: I'm not done yet.
Q: Oh, I'm sorry. I thought you were done.
A: Index to medical records, Volume 4 of 4 — oh, this is 1 of 4. 4 of 4. Volume 3 of 4.
MS. DISILVIO: He also got Volume 4 but I didn't bring it because I didn't think you'd need it.
MR. MALONE: Or 2 of 4.
MS. DISILVIO: Whatever the other one is.
Q: So you have three volumes of medical records, those include medical records only from

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University Hospitals of Cleveland?
A: Go back to Volume 1.
Q: Okay. That's fine.
A: Volume 1 is, A, University Hospitals of Cleveland — should I keep reading the page to you?
Q: Yes, sir, so I know what you were able to review.
A: Admission, labor and delivery, premature baby girl. Number 2, admission, postpartum respiratory distress syndrome, intraventricular hemorrhage, rule out metabolic disorder.
I don't have Volume 2. I presume it's what came between Volume 3 and 4.
Q: Right.
A: I'm sorry.
Q: That's okay.
A: Volume 3 says **A.** University Hospitals of Cleveland, Number 2, admission, postpartum respiratory distress syndrome, intraventricular hemorrhage, rule out metabolic disorder. B, Dr. Fanaroff/Dr. Rodriguez office records. It goes on to say a little more. You want me to read everything on that or is that satisfactory?
Q: Just as long as I know what records you reviewed, that's okay.

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A: I don't know the detail you need. Number C is Aultman Hospital.
Volume 4, D, E. James Witmer, M.D.
E, Alliance Community Hospital.
F, Children's Hospital Medical Center of Akron.
G, death certificate.
Q: Okay.
MS. DiSILVIO: Volume 2 for your purposes, Donna, is more of the UH chart that I just didn't bring with me today.
Q: That's fine. I have a huge second one that I didn't bring with me today.
All right. Doctor, we're going to mark your vitae which was given to me this morning, although I actually have seen one, Plaintiffs' Exhibit A.

(Thereupon, Plaintiffs' Exhibit A, Zinn CV, was marked for purposes of identification.)

Q: Basically, I don't think that we need to go through and recite what's contained in the vitae since it's self-explanatory but I just want to

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make sure that I am correct about your certifications.
You are board certified in clinical genetics, correct?
A: Which page is that?
Q: Page 2.
A: Yes. Thank you.
Q: And that was in 1982.
A: I'm sorry. Could you repeat yourself?
Q: Sure. You obtained your board in medical genetics?
A: The first board was the American Board of Medical Examiners in 1981.
Q: In terms of genetics, however, you obtained a board certification in 1982 in clinical genetics, right?
A: Correct.
Q: Then in 1996 you obtained an additional board in clinical bio, biochemical genetics; is that right?
A: Right.
Q: How long has that board been available, the one that you obtained in 1996?
A: I think it was first available in **'82.**
Q: Can you just sort of walk me through what made

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you decide to get that board in 1996?
A: My division chief asked me to do it for purposes of being able to accredit our unit as a training center for residents in medical genetics.
Q: Were you seeking the accreditation through the AMA certification program or do you know which program?
A: It, the American Board of Medical Genetics is one of the subboards of the American Board of Medical Specialties, so I don't, I may have the names slightly wrong but —
Q: That's perfectly fine.
A: — it's part of the AMA clinical boards.
Q: And you have such a program in place now; is that correct?
A: Such a program being?
Q: The program that you were seeking this board certification in?
A: Yes.
Q: For residents?
A: Yes.
Q: And when did that program come into existence, if you know, doctor?
A: It preceded 1996 but I don't know the date.
Q: Before we move on to talking some more I guess

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about your background, you indicated that you had served as an expert witness on two occasions; is that correct?
A: Yes.
Q: For whom did you serve as an expert witness?
MS. DISILVIO: Objection. You may answer.
A: I don't know the order. One case was for the United States Department of Justice. I believe that's what it is. It was for an alleged immunization reaction.
Q: So you were a government witness in that matter?
MS. DISILVIO: Objection. You may answer.
Q: If you know.
A: I don't know, actually.
Let me think for a second. A U.S. attorney was the one who obtained my deposition.
Q: And?
A: And the second one was for a case involving a child at University Hospitals of Cleveland.
Q: In that matter, did you offer expert testimony on behalf of the physicians involved in that matter?
A: No. On behalf of the plaintiff.
Q: Do you recall who the plaintiff's attorney was?

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Was it Howard Mishkind?
A: I truly don't remember.
Q: That's all right. And you also indicated that you did give testimony in a case named Burdette. I gather you were a defendant in that matter?
A: As I indicated —
MS. DISILVIO: Objection.
A: Yes.
Q: Do you know what year that case was filed?
A: No.
Q: Do you know if that case was settled on your behalf!
MS. DISILVIO: Objection.
A: Which means?
Q: You can answer.
MS. DISILVIO: You can answer.
A: The case was dismissed with prejudice.
Q: When Emily was a patient in 1999, tell me how your department of genetics was organized at the hospital.
A: I don't know how to answer that question.
Q: Okay. Well, I guess we'll do a preempt. We'll make it a little easier.
A: I'm not trying to be difficult.
Q: I know that you're not.

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A: I just don't know how to answer the question.
Q: Okay. Your department has information which you post on the Internet, a web page. Are you familiar with that web page?
A: I know they have one. I've never looked at it.
Q: Okay. Well, the reason I asked you how your department was organized, the way that they break this out, and I'll mark it Plaintiffs' Exhibit B.

(Thereupon, Plaintiffs' Exhibit B, three-page website printout, was marked for purposes of identification.)

Q: I obtained this information off the Internet and the question I was asking you is it seems that there's a pretty accurate description, I'm hoping, of the services provided by the Center for Human Genetics. I don't know if you've had an opportunity to read it.
A: I have not.
Q: You are listed on the last page as your special interest, and I know that this is a typographical error, it says, "unborn errors of metabolism." I'm assuming it should say inborn errors of metabolism?

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A: It should.
Q: Is that your specialty within the group?
A: That's one of them, yes.
Q: Why don't you tell me, Dr. Zinn, we try to reference at depositions what you were doing at or around the time of the person's hospitalization. I don't really need to know what you do today but back in October/November of 1999, sort of describe for me what your responsibilities were within the group.
A: I'm a clinical geneticist. I primarily see patients. I'm responsible for educating our residents as well as students who, or other residents who rotate through our service. I have some responsibility for academic performance as a member of the department of genetics.
Q: Back in 1999 were you performing any research?
A: I was not — can you define that for me?
Q: Well, I assume research is research. Were you involved in any research projects on an ongoing basis at that time?
A: No.
Q: Are you now —
A: I'm sorry. I may have been. I can't recall.
Q: At this point in time, are you involved in any

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research studies?
MS. DISILVIO: Objection. You may answer. You may answer.
A: No long-term research projects.
Q: Doctor, what is LCHAD?
A: It's an acronym.
Q: And it's an acronym for what, if you want to —
A: No.
Q: Okay.
A: It's an acronym for a long-chain 3-hydroxyacyl-CoA dehydrogenase.
Q: When did LCHAD — and the acronym —
A: Are we done with this?
Q: — is in fact an inborn error of metabolism; is it not?
A: Yes.
Q: When did you become aware of the existence of LCHAD?
A: I can't recall precisely.
Q: Can you tell me *if* it was in the '70s?
A: I think it was probably later than that.
Q: The '80s?
A: Probably.
Q: In the '80s, I presume from looking at your curriculum vitae, you were a clinical geneticist

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at that point in time, correct?
A: Yes.
Q: Prior to Emily Gwynne had you ever seen a child that had LCHAD?
A: No.
Q: How does one confirm a diagnosis of LCHAD?
MS. DISILVIO: Now or then?
Q: In October of 1999. October/November/December.
A: By a combination of clinical studies and laboratory studies.
Q: Tell me about the clinical studies first.
A: You would look for evidence of hepatic dysfunction, cardiac dysfunction or skeletal muscle dysfunction.
Q: And laboratory?
A: You would do a combination of relatively routine laboratory studies.
Q: Such as?
A: A comprehensive chemistry panel.
Q: Consisting of what?
A: It's different things in different hospitals; but basically it's a blood sugar, a series of electrolytes.
Q: What **else?**
A: It depends somewhat on the clinical presentation.

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Q: What about DNA testing?
A: No.
Q: Why?
A: It wasn't clinically available.
Q: Well, I guess we'll just deal with that issue. Is it your testimony, first of all, you were aware of the disorder, I think we've already established that, prior to 1999, correct?
A: Yes.
Q: And can I gather, and maybe I can't but we'll see, that this is something because of your interest in inborn errors of metabolism that you would have continually read the literature and made yourself aware of —
MS. DISILVIO: Objection.
Q: — in terms of advances in diagnoses and treatments?
MS. DiSILVIO: Objection. You may answer.
A: I tend to read in my field of specialty, **yes.**
Q: Is it your testimony under oath, doctor, that on October 28th, 29th, 30th, 31st of 1999 and beginning in the first couple of weeks of November, that there was no place that you could have sent a DNA sample and obtained a result on

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Emily Gwynne?
A I didn't say that.
Q: Okay. Well, that's why I want to be sure about what you are saying.
There were places where you could have sent Emily Gwynne's DNA sample. Is that a fair statement?
MS. DiSILVIO: Objection. You may answer.
A: Certainly there are places I could send her DNA.
Q: For analysis?
A: What kind of analysis?
Q: To determine whether or not she had LCHAD?
A: On a clinical basis? In a clinically useful manner? That's my question. That's why I'm having trouble answering you.
Q: What facilities were you aware of in late 1999 that would take a DNA sample and give you back a result as to whether or not there was an indication that a child had LCHAD?
A: I was aware of no place that would do that on a clinical basis.
Q: So it's your belief that there was no place in 1999 where you could send the DNA on a clinical basis?

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A: That is correct.
Q: What about Washington University?
A: I was unaware that that was possible.
Q: Were you aware, doctor, that there were places that you could send the sample for testing that were on a research basis?
A: Yes.
Q: What facilities were you aware of on a research basis?
A: Washington University.
Q: What about Wake Forest?
A: I did not know that that was available.
Q: Do you know who Dr. Ibdah is?
A: I do not know him personally.
Q: Excuse me?
A: I do not know him personally.
Q: I wasn't asking if you knew him personally but that's inartful question asking on my part.
Prior to Emily's admission to University Hospital, were you aware of Dr. Ibdah's research and writing in the area of LCHAD?
A: He was a junior author on a paper by Dr. Strauss from Washington, yes.
Q: Do you recall the title of that paper?
A: Not in precise words.

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Q: Do you recall what the paper was about?
A: Yes.
Q: What was the paper about?
A: The paper was about the incidence of children with, the mothers of children who have LCHAD deficiency presenting with either acute fatty liver of pregnancy or HELLP syndrome. That's capital H-E-L-L-P.
Q: Were you aware from reading that article that — I take it you read the article?
A: I have.
Q: We establish that first.
I had an opportunity last week to depose the resident who did the consultation with you in this matter, Do you recall that resident?
A: Do you mean Derek?
Q: Yes.
A: Yes. I know Derek.
Q: And you still work with Derek, correct?
A: We still work together.
Q: Derek indicated in his testimony — I'm paraphrasing it since I don't have the transcript with me — that he was in possession of Dr. Ibdah's article at the time of Emily's consult.

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Do you recall looking at the article at the time of Emily's consult?
A: Yes.
Q: Did you, because you looked at the article, did you become aware that Dr. Ibdah had in his facility at Wake Forest the ability to test DNA to look for the particular defect?
A: I was more familiar with Dr. Strauss and he was the senior author and to my knowledge Dr. Strauss was, the work was done in Dr. Strauss' lab that I know about.
Q: Would you agree with me that the presentation of Emily's mother with fatty liver disease in and of itself was an indication that the child should be worked up for LCHAD?
A: Yes.
Q: Are you involved, doctor, at all in helping the State of Ohio make determinations as to which tests should be performed upon children at birth, newborn screening?
A: I'm on the ad hoc newborn advisory committee.
Q: Did the State of Ohio this past June, maybe June 26th, 2002, recommend that LCHAD be tested for?
MS. DiSILVIO: Objection.
Q: As an optional newborn screening test?

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MS. DISILVIO: Objection. You may answer.
A: Yes.
Q: Do you know how that came about?
MS. DISILVIO: Objection. You may answer.
MR. MOSCARINO: Same objection.
Q: Let me ask a better question.
You told me you're on the ad hoc committee. I'm wondering if you had any input or involvement in helping the Department of Health determine that this was a good newborn screening test?
MR. MOSCARINO: Objection to form.
MS. DiSILVIO: Objection. You may answer.
A: I was present at several meetings of the State Newborn Advisory Board.
Q: And at the meetings that you were present, did you participate in any conversations about screening for LCHAD?
MS. DISILVIO: Objection. You may answer.
A: I was not present at the meeting when the recommendation was made. I was present I believe

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at a subsequent meeting where the implications of that were discussed.
Q: Are there recorded minutes from these meetings?
MS. DISILVIO: Objection. You may answer.
MR. MOSCARINO: Can I just have a continuing objection to any kind of committee meetings that went on in 2002 or 2003 with regards to their application to the standard of care in '98?
MS. KOLIS: I would agree that it doesn't apply to the standard of care. I'm simply trying to find out if there is a place where Dr. Zinn may have rendered some testimony.
MR. MOSCARINO: I just don't want to interrupt anymore. I just made an objection to everything in 2002 and 2003.
MS. DiSILVIO: That's obviously my continuing objection, as well, and I'll just join in so I don't have to keep making that objection.
MR. MALONE: And I'll ride on that cart.
A: Having said that, folks?

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MS. DiSILVIO: You can answer.
A: Thank you. There are minutes, I don't believe they are recorded minutes but I'm not in charge of the meeting and I may be inaccurate in that.
Q: That's fine. I mean now that I know what committee you're on, there's mechanisms for me to try to obtain that information.
Who is going to be performing the screening tests for the State for LCHAD?
MS. DiSILVIO: Objection.
Q: If you know.
MS. DISILVIO: You may answer.
A: For the State of Ohio?
Q: Yes.
A: The screening is done through the Department of Health of the State of Ohio.
Q: Do you know in what laboratory? Is there a laboratory that they've selected to —
A: The Department of Health, the State of Ohio.
Q: So they're going to have their own internal laboratory setting to screen for LCHAD?
A: They're not screening for LCHAD per *se*. For their newborn screening.
Q: In October of 1999, October, November and December of 1999, it's my understanding that the

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lab at University Hospital was not able to perform a DNA test that would have indicated whether or not Emily had LCHAD. Is that a correct understanding?
MR. MALONE: Can I have the question read back, please.

(Thereupon, the requested portion of the record was read by the Notary.)

A: It was not performing that test.
If you ask me the able, the key to your question, they were potentially able to but they were not performing that test.
Q: Potentially able meaning that they just hadn't set it up to test for LCHAD, not meaning they were incompetent. I wasn't implying that.
A: Rut you said able.
Q: But they were not doing that test, correct?
A: That is correct.
Q: In the past when you have had situations, not referring to LCHAD but other situations where there were inborn errors of metabolism, did you send those out to other laboratories when UH couldn't perform it?

MS. DISILVIO: Objection. If you can answer that question. An objection as to relevance.

A: Can you repeat the question for me or read it back, I guess.

Q: Yes. She could read it back. That would be fine.

(Thereupon, the requested portion of the record was read by the Notary.)

A: I need to qualify your question, then.

Q: That's fine.

A Whenthere was, do you want metoanswerforwhen there was a suspected case, which is the case for Emily, or when there are confirmed cases?

Q: Suspected.

A: I would send out — then the answer to that question is I would send out samples when there was a laboratory doing that test on a clinical basis.

Q: So you're riot restricted by the hospital from sending out samples when there are tests that can be performed that aren't being performed at UH; is that correct?

MS. DiSILVIO: Clinically available tests?

Q: Or on a research basis.

MS. DISILVIO: Objection. You may answer.

MR. MOSCARINO: Object to the form.

A: That's allowed to my clinical discretion.

Q: That's, I just wanted to make sure that the hospital hadn't formulated some policy that prevented you from doing that if you so elected to do it based on your clinical judgment.

A: The, I'll let it stand.

Q: Sitting here today, do you know whether Emily Gwynne had LCHAD?

A: From everything I've read, she does.

Q: What is it —

A: She did.

Q: I'm sorry. What is it that you read that led you to that conclusion?

A: My understanding is that she had mutational analysis performed. She subsequently had organic gas analysis and subsequently had acylcarnitine analysis, all three of which confirmed the diagnosis.

And in addition she presented with clinical features of that disorder and at the time of autopsy had laboratory findings of that disease consistent with that disease.

Q: I guess that I would like to in-depth probably evaluate the things that you evaluated that make you know that she did have LCHAD.

First of all, were you contacted when Emily was admitted to Akron Children's Hospital?

A: Yes.

Q: Who contacted you?

A: Dr. Gunay.

Q: Is that someone that you knew prior to Emily's admission to Akron Children's?

A: I need to correct. At the time that that was, Meral had married and changed her name. So I

A: So if it's all right, with no disrespect, I'll call her Meral because otherwise I'll get it backwards. And Meral is M-E-R-A-L.

Q: So she contacted you to let you know that Emily had been admitted to the hospital. Is that essentially it? That's how you became aware of

it?

A: Yes.

Q: Did you talk with her after she received DNA analysis back on Emily?

have been after she received her result.

Q: Did she ever send you a copy of the DNA results from Wake Forest?

A: Not to my memory.

MS. KOLIS: What I'm going to do

(Thereupon, Plaintiffs' Exhibit C, five-page 11/7/00 Wake Forest Molecular Genetic Laboratory report, was marked for purposes of identification.)

Q: Doctor, what I'm handing you are laboratory results from Wake Forest University School of Medicine Department of Pediatrics Section of Medical Genetics and they are, there are some

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findings for Emily Gwynne, Eric Gwynne and Kristie Gwynne and then there's some additional things we'll talk about afterwards?

MS. DISILVIO: Why don't you take your time and go through those, doctor.

Q: Yeah, I don't have a problem with that.

A: Yes.

Q: In doing a DNA mutation analysis looking for LCHAD, where is the defect found; do you know?

A: I'm sorry. I was thinking about something. Tell me again.

Q: Sure. Well, I could ask the question probably in a much simpler way. The first sheet of paper in the packet that I've given you are the results for Emily Gwynne?

A: Yes.

Q: Do those results indicate that Emily has LCHAD?

A: It indicates that she's heterozygous for the common mutation consistent with the clinical disease of LCHAD. This testing tests for the genetic basis. She has the genetic basis consistent with that disease.

Q: Have you ever seen, not Emily but prior to me showing you this, have you received reports like this from other laboratories having done testing

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for the mutation?

A: For this mutation?

Q: Yes.

A: I received — could you read the question back to me.

(Thereupon, the requested portion of the record was read by the Notary.)

A: Yes.

Q: The LCHAD mutation, but I might be saying that incorrectly, just to clarify it.

A: You're doing very well. You said it correctly. The answer is yes.

Q: Okay. What other laboratories have you worked with?

MS. DISILVIO: At what point in time?

Q: Okay. Well, let me, that's a good preempt. Prior to October, November and December of 1999, had you ever sent any DNA out for analysis looking for the LCHAD mutation?

A: Yes.

Q: Where did you send them?

A: Strauss.

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Q: At Washington?

A: Uh-huh.

Q: What was the basis of your decision prior to Emily Gwynne in those other instances where you did send DNA to Strauss?

A: It was a mother who presented with HELLP syndrome.

Q: And was that the sole basis for that decision?

A: And, yes. Yes.

Q: When you consulted relative to Emily, and eventually we'll get to the consultation, what information did you have about her mother's pregnancy?

A: That she had acute fatty liver of pregnancy confirmed by biopsy.

Q: And as a result of that, Emily was delivered prematurely, correct?

A: I believe so.

Q: Well, I guess my question I'm asking you, if you can recall it today because I know it's been some time, did you have an opportunity to review Kristie Gwynne's chart at the time that you were consulting with Emily to see what the parameters of her pregnancy were and how it is that this child came to be delivered at 25 weeks?

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A: I reviewed the basic information, yes.

Q: Can you tell me today, doctor, why you elected not to send Emily's DNA for testing for LCHAD?

A: Because the testing wasn't available on a clinical basis.

Q: And you were uncomfortable sending it to a research-based test?

A: First, the previous — yes.

Q: Why?

A: Okay. Three reasons, I think. One is that the federal government has actually tightened up considerably and it's no longer permissible, and our unit is being very careful, you cannot give research based data to patients. You cannot use it for clinical purposes so that in fact there are restrictions on that.

Second, the testing had proved negative in the past, the relative minority of patients with acute fatty liver of pregnancy in fact had LCHAD deficiencies.

Third, not knowing that that was the case, I thought that the primary way of establishing that diagnosis would be to use a broader based approach which is based on metabolite analysis and clinical exam.

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Q: Just so I don't walk away saying I didn't ask that question, at what point did the federal government say that you couldn't use research-based information and share it with the patient?
A: That was evolving over the last few years. I can't tell you exactly when.
Q: That was not in fact the case in October of 1999, would you agree with that?
A: That was not my understanding. But it was only one of four reasons.
Q: Okay. But I just want to be real clear about that.
A: Right. But there was, to skirt — it was not a clinical based test, which did affect my decision.
Q: Why do you believe medically, not scriptly but why do you believe, doctor, that the battery of metabolic analysis that you were looking at proved, quote, unquote, negative during Emily's hospitalization at University Hospital?
A: Because it was negative.
Q: See, I know I was going to ask a lot of inartful questions.
A: Ne. No. It's a perfectly fine one.

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Q: We both agree that this child definitely had the LCHAD mutation?
A: That is correct.
Q: And clinically she manifested signs and symptoms consistent with that at a later point in her life, would you agree with that?
A: Correct.
MS. DiSILVIO: Subsequent to discharge.
Q: Subsequent to discharge from University Hospital?
A: Correct.
Q: Why were those signs and symptoms not present during her hospitalization at University Hospital?
A: I don't know the answer to that.
Q: Well, we'll see if we can get some agreements which is very difficult for doctors to ever want to agree with you, but we could try.
A: Excuse me?
Q: In the interpretation of these tests, the clinical tests, and we're going to go back and sort of talk about what tests were run on Emily. Do you agree that those tests would essentially be negative if Emily was not in LCHAD crisis?
MS. DiSILVIO: What tests?

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Q: Well, that's what I'm going to ask him, what tests would we base it on?
MS. DiSILVIO: Objection,
MR. MALONE: I don't understand the question. I'm totally lost now.
Q: All right. Here's what we'll do: Name each and every test that you recommended be performed on Emily to attempt to make a determination whether she was LCHAD, had LCHAD.
MR. MALONE: I'm going to object. He's already testified that he looked at this patient globally. Clinically there were a number of labs.
MS. KOLIS: Well, we're going to talk about the labs first and we're going to talk about clinical later.
MR. MALONE: I understand. Let me make my objection, Donna, please.
MS. KOLIS: That's fine.
A: Would I, to answer your question, I'm going to go to the consult.
Q: That's fine.
A: On Page 4 of the consult dated 10/30/99, the first test was a plasma carnitine and acylcarnitine profile.

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The second test was urine carnitine and acylcarnitine profile. The third test was urine organic acid analysis. The fourth test was mutational analysis of LCHAD gene; because of the elevated ammonia, ammonia analysis.
An EKG.
Q: Did you write the orders for these tests to be performed?
A: No.
Q: How do these tests, then, become performed if you don't write the orders for them?
A: By providing the house staff with this recommendation and then the house staff or one of the neonatologists, whose care, who were providing the care for Emily.
Q: Is that how it always occurs when you do an in-hospital consultation?
A: If I'm not the primary providing physician, yes.
Q: How do you receive the results of the laboratory tests that you are recommending?
A: I usually receive them from the primary physician.
Q: Do they call you with them? I mean, how does that work?
A: Many times they call me.

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Q: Do you have any notes, personal notes in a folder somewhere or otherwise or in the chart that indicate that you were made aware of any of the results of the tests that were performed on Emily?
A: I have no personal notes to that effect.
Q: What about in the chart?
A: No.
Q: Do you believe you were made aware of the results of these tests?
A: I was made aware of the results of these tests, yes.
Q: How do you believe you were made aware of them?
A: Because I was.
Q: I'm not trying to be difficult; but I don't — subsequent to the date of the consultation, doctor, did you record any further notes in the hospital chart of Emily Gwynne relative to your analysis of the results that came back?
A: No.
Q: So this is the sole note that you wrote — well, let me take that back.
You countersigned a note written by Derek and I believe added something at the end of it; is that right?

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MS. DISILVIO: Are you talking about the consult?
Q: The consult. Let's start with that.
MS. DISILVIO: Are you asking if that's his only note?
Q: Right.
A: Yes. I'm sorry. There's another addendum to Derek's short note which is a prelude to the complete consult.
Q: And that's in the progress notes?
A: Yes, ma'am.
Q: But other than these two places, there are no further notes from you in the chart; is that correct?
A: None that I could find.
Q: Do you know what physician or physicians spoke with you about Emily after the consultation?
A: I know I spoke with several people. I can't tell you everybody I spoke with.
Q: If you can't tell me everyone, can you tell me anyone who you remember speaking to about her?
A: I can do that.
Q: Okay.
A: I spoke with Eilene Stork. I spoke with Rodriguez, Riccardo Rodriguez and I remember

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speaking with a Cynthia Bearer.
Q: Did you ever speak with Dr. Fanaroff at or around the time of Emily's discharge from University Hospital?
A: No.
Q: Going back to your consultation, then, and you can, doctor, take as much time as you want, look anyplace in the chart, the plasma carnitine, and, I can't — the acylcarnitine — I'm getting good at pronouncing these words — profile that was performed. You know the results of that?
A: Yes.
Q: You can look at the results if you want to; but generally speaking, what did they indicate?
A: Can I see them, please? Thank you.
Q: I think that's the one.
A: Yes.
Q: Just for the record, could you indicate what you're looking at?
A: I am looking at a quantitative acylcarnitine profile report on Emily, girl, Kristie. And the test date was, let's see, the date the sample was received was 02-November-99.
Q: This particular test is performed in the Department of Veterans Affairs Medical Center,

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correct?
A: Physically the test is performed at the Veteran's Administration Hospital.
Q: Is there something, based upon the inflection in your voice, is there —
A: You said department. It's not the Department of Veterans Affairs, I thought. Maybe I misheard you. It's performed in a laboratory at the Veteran's Administration Medical Center.
Q: At the clinical pharmacology lab there, correct?
A: That's what it says, yes.
Q: Is that correct?
A: That is the designation for the laboratory.
Q: Would you designate it some other way? Is there something you're trying to tell me?
A: Yes.
Q: Okay.
A: I would designate it as — I'm trying to really answer your question as asked.
Q: That's all right.
A: They do call it department of. I never do. It's the VA, it's the VA branch of the Center for Inherited Disorders of Energy Metabolism.
Q: So you wanted to really call them what they were versus what the stamp says across the top?

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A: I'm sorry. Yes.
Q: So you have those results and those are, I don't know what the name at the bottom says, that Dr. Hoppel, if that's not an L?
A: That's Charles Hoppel.
Q: Is it Charles Hoppel?
A: Yes.
Q: What do those results say to you about this child?
A: The results say that the patient's total carnitine, free carnitine and total acylcarnitines are above the normal range. The acyl, the total acylcarnitine to free carnitine ratio is normal and the individual acylcarnitine profile is normal.
Q: Now, this is one laboratory test that can aid and assist the physician in making a diagnosis of LCHAD, correct?
A: Correct.
Q: Going way back to the question I asked a little bit earlier, why was that test within the normal range given that we now know Emily definitely had LCHAD?
A: The, it's a false negative.
Q: Were you aware in October, November and December

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of 1999 that it was possible to get a false negative using this particular test?
A: The, to answer the question, how do you define possible? No. I'm —
Q: That's okay. Were you aware that there was a false negative/false positive rate of error contained within the analysis of this test?
A: Yes.
Q: What did you believe the percentage was?
A: I thought that percentage was small.
Q: So in your opinion that is a false negative test if it —
A: Speaking when?
Q: As of —
A: Speaking as of that date?
Q: Correct.
A: I thought this was a negative result.
Q: Okay. I'm sorry. Are you —
A: You asked me my time reference.
Q: On that date?
A: When I saw this result, I thought this was a negative result.
Q: And now in retrospect you're saying that was in all likelihood a false negative result?
A: Yes.

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Q: To the best of your knowledge — I want to call you a metabolic geneticist. I probably shouldn't do that. As a geneticist?
A: You may feel free to call me a metabolic geneticist.
Q: To the best of your knowledge, in addirion to potentially being a false negative result on that day, are you, do you believe that all children that have LCHAD will — okay. Let me withdraw the question. I know what I'm trying to ask and I have to ask it the right way.
Going back to the other question that I had, have you reviewed any body of literature in your endeavor to understand LCHAD prior to Emily becoming a patient that indicates that this test may not give you a result that indicates LCHAD if the child has not yet gone into LCHAD crisis?
MR. MALONE: I'm going to object because I think the question is almost incomprehensible; but if he understands it, that's not my client.
A: Could you ask me the question again? I have trouble when there are objections, to make sure I focus, so could you ask me again?
Q: Sure.

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Could that result in fact be a correct result?
A: It is a correct result.
Q: And not —
A: No, excuse me. It is a correct result.
Q: And my question is prior to —
A: To my knowledge, there's nothing to make me think this is not accurate.
Q: Okay. So you think that test result is accurate?
A: I think this test result is accurate.
Q: Why would she have not shown positive signs of LCHAD on that test on that date?
A: Because you can have false negative results.
Q: Okay. We're going backwards. Is there any other reason other than a false negative result in this context that that report wouldn't have shown an indication for LCHAD?
A: That the test is not accurate.
Q: Okay. Hold on, though, to that piece of paper. We're going to go a different way.
What happens to a child that has LCHAD doctor?
MS. DiSILVIO: Clinically?
Q: Yes.
A: The clinical course is variable.

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Q: In general, what is the concern for a child who has LCHAD?

A: The concern is that you will have progressive hepatic and/or cardiac and/or skeletal muscle disease.

Q: And how does that happen?

A: I'm not sure I understand your question.

Q: Sure. How is it that you get progressive hepatic, cardiac and skeletal disease if you have LCHAD?

A: No one knows the precise mechanism.

Q: Do you have an understanding of the mechanism even though you're saying no one knows the precise mechanism?

A: My understanding of the mechanism is it's related to a relative energy deficiency for tissues that require fatty acid oxidation and/or potential toxic metabolites that accumulate secondary to the metabolic block and/or those metabolites impair other pathways.

Q: And indeed it's implied in your answer the condition is progressive, correct?

MS. DISILVIO: Objection. You may answer.

A: I stated that it's a variable course. On average

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it is not thought to be progressive.

Q: Do you know the treatment for a child who has LCHAD once they're diagnosed?

A: I know a treatment.

Q: What is the treatment that you know?

A: The treatment involves a combination of avoiding fasting, of frequent feedings and reduced fat in the diet and some individuals provide supplemental carnitine, but that's still debated.

Q: I don't think I asked this question before and if I did, your attorney will graciously let me know. In anticipation of testifying here today, did you conduct a literature search of any sort regarding LCHAD?

A: Not for myself, no.

Q: Is the answer that you did not conduct a literature search?

A: I, my attorney asked —

MS. DISILVIO: I'm going to object to anything that he may have done for me.

MS. KOLIS: That's fine.

A: My attorney asked me —

MS. DiSILVIO: You don't have to answer that.

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Q: You don't have to tell me that.

A: I didn't know that.

Q: All right. Doctor, do you have an opinion as to generally speaking the life expectancy of a child who has LCHAD if they are diagnosed?

MS. DISILVIO: Objection. You may answer, if you know.

MR. MALONE: I join the objection. I think you need a few more factors in there. Diagnosed and treated?

MS. KOLIS: Right. Diagnosed and treated.

MR. MALONE: What's the clinical situation, what's the age — there's a million variables to that question.

MS. KOLIS: I'm sure there are but I'm going to ask the general question first.

A: I would like a more specific, to answer your question, I would need more specifics.

Q: Have you evaluated the studies that exist as to what the success rate is in treating the children that have been diagnosed with LCHAD strictly along the lines that you discussed before, focusing on the three, avoid fasting, frequent

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feedings and reduction of the amount of fat in a diet?

MS. DISILVIO: Objection. You may answer.

A: I have evaluated various reports on the treatment of LCHAD deficiency.

Q: Based upon your evaluation of those reports, do you have an opinion as to what the anticipated life expectancy can be for a child with an early diagnosis of LCHAD?

MR. MALONE: Objection.

A: How early is early?

Q: Well, let's try someone who is diagnosed within the first four weeks of their life?

MS. DISILVIO: Objection. You may answer.

A: There are no prospective studies to answer that question.

Q: What literature have you reviewed that indicates life expectancies to you in this arena?

A: As of when?

Q: As of — well, let's start first with as of October, 1999?

A: I can't remember all the articles.

Q: Suffice it to say based upon your testimony you

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would have had to have read articles and relied upon those for your understanding because you hadn't conducted some independent study of your own, correct?
A: Correct.
Q: And I think you already answered this but I just want to be perfectly clear. Prior to Emily Gwynne becoming a patient at University Hospitals, had you seen, diagnosed or treated a child with LCHAD?
A: No.
Q: Did you, prior to Emily's discharge from University Hospitals, rule out LCHAD?
A: I'm sorry. Go ahead.
Q: See, your mind is working.
Did you, prior to Emily's discharge from University Hospitals, rule out LCHAD?
A: To a clinically acceptable degree, yes.
Q: Define what you mean when you say to a clinically acceptable degree.
A: I thought the, it was unlikely that she had LCHAD deficiency.
Q: And the basis of your belief was what?
A: The a priori risk that she would have LCHAD deficiency, the results of her clinical

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evaluation by the neonatology service and the results of the laboratory studies performed.
Q: To whom did you communicate that you believed that she did not have LCHAD?
A: I had spoken with the neonatologist along the way.
Q: But you didn't record, once again I'm just, you know, there's no written report in some other file?
A: That's correct.
Q: That you sent a report to anybody in the department of neonatology, Dr. Fanaroff, Dr. Rodriguez, Dr. Stork; is that right?
A: That's correct.
Q: Did you see the second set of quantitative acylcarnitine profile report; do you know?
A: Yes.
Q: You did see it? Do you know when you saw it?
A: I can't tell you exactly.
Q: Let me just see if this refreshes your recollection. I'm not going to even mark this.
Tell me what report I'm handing you.
A: Tell you?
Q: Just for the record so you can identify it, not me.

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A: I'm sorry, It's the same — okay. It's a quantitative acylcarnitine profile report on Gwynne, Emily. Date received, 28-December-99.
Q: Okay. And you believe that you did see that report?
A: Uh-huh.
Q: When do you think you saw that report?
MS. DISILVIO: Objection. Don't guess.
Q: Do you know when you saw the report?
A: I just said I couldn't tell you exactly.
Q: Okay. Was it before or after Emily died?
MS. DISILVIO: Objection. If you know.
A: I would say before.
Q: You would say that it was before? Are those results different than the ones that occurred in November?
MS. DISILVIO: Numerically different?
A: Yes. Well, these are also urine. The other ones you asked me about were plasma.
Q: I'm sorry. That's all right. You can talk about those and I'll give you those, too.
A: Can I talk about the plasma first?

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Q: Sure. You want to switch?
A: Yes, just to compare apples to apples.
Q: Okay.
A: So the answer to your question is — I'd need to change that, then.
This is quantitative acylcarnitine profile. It's on plasma on Emily Gwynne and the date received is 27 December '99.
Q: Okay.
A: And the answer to your last question is yes, they are different.
Q: In what regard are they different?
A: The total carnitine, the free carnitine are both low and whereas the total acylcarnitines and the acylcarnitine to free carnitine ratio is normal.
Q: What do the —
A: In addition, the —
Q: Goahead.
A: The individual acylcarnitine profile, and that's, which only has one species which is acetyl carnitine, is low.
Q: What do the differences in those results mean to you or what did they mean to you at the time when you saw them?
A: It meant that Emily was, had a relatively low

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amount of total and free carnitine and, but the acyl to free ratio was normal suggesting she was not making any increased amounts of atypical or pathopneumonic acylcarnitines. In particular, the acyl, individual acylcarnitine profile at the bottom included only acetyl carnitine, did not include any of the individual acylcarnitines that are pathopneumonic for LCHAD deficiency or any other disorder of fatty acid, long-chain fatty acid oxidation, any other disorder of fatty acid oxidation, any organic acidemia or any other inborn error that might represent itself that might potentially be associated with LCHAD deficiencies.

Q: I'm going to ask you the exact same question I asked you about the November labs.

Why did that test not reveal the possibility of the existence of LCHAD?

A: I don't know.

Q: All right. I'll take it back. Thank you.

I've already asked you about whether you wrote letters to the physicians in this case and the answer was no.

Did you ever, first of all, do you recall talking with Eric Gwynne, Emily's father?

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A: When?

Q: At the time of your consultation in October?

A: I don't have clear recollection of him.

Q: Okay. I guess I asked a different question.

Did you have a conversation with Mr. Gwynne regarding potentially what the concerns were about Emily?

A: My memory is unclear about it.

Q: Do you have a memory of any conversation ever with Eric Gwynne?

A: To answer your question, I don't remember the conversation. I remember the fact that I had a conversation. I don't remember the conversation.

Q: Did you ever contact any of the physicians at Aultman Hospital regarding your conclusions that Emily did not have LCHAD?

A: No.

Q: The plasma carnitine test that we're referring to, the second one?

A: Yes.

Q: That came back after Emily was discharged, correct?

A: Right.

Q: Do you think, not do you think.

Do you recall having a conversation with anyone at University Hospital, any of the attendings after you saw that result in January?

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A: No.

Q: So the answer is no, you didn't have a conversation or you don't recall?

A: You asked me did I recall.

Q: Okay. Do you recall?

A: No.

Q: And the answer is no.

And once again no notes reflecting that you received that test and then discussed this with anyone, correct?

A: Correct.

Q: Doctor, are you critical of anyone involved in the care and treatment of Emily Gwynne starting from the day she was born till the day she passed away?

MR. MALONE: Objection.

MS. DISILVIO: Same objection but you may answer.

A: What do you mean by critical?

Q: Do you feel that the action or inaction of any persons based upon the medical records that you reviewed caused or contributed to Emily's death?

MS. DISILVIO: Objection. You may answer.

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A: No, I'm not critical.

Q: I have to think for a minute.

A: I do, too.

Q: Have you discussed this particular lawsuit with Drs. Rodriguez and Fanaroff?

A: My only discussions were to say I couldn't talk about this with them.

MS. KOLIS: Fair enough. All right, doctor, I don't have any further questions. Anyone else?

MS. DISILVIO: Anyone else?

MR. MALONE: No questions by Drs. Fanaroff and Rodriguez.

MR. MOSCARINO: Off the record.

(Thereupon, a discussion was had off the record.)

MR. MOSCARINO: I have no questions.

MS. DISILVIO: We'll read it. Thank you.

ARTHUR ZI", M.D.

CERTIFICATE

The State of Ohio,) SS:
County of Cuyahoga.)

I, Pamela S. Greenfield, a Notary Public within and for the State of Ohio, authorized to administer oaths and to take and certify depositions, do hereby certify that the above-named witness was by me, before the giving of their deposition, first duly sworn to testify the truth, the whole truth, and nothing but the truth; that the deposition as above-set forth was reduced to writing by me by means of stenotypy, and was later transcribed into typewriting under my direction; that this is a true record of the testimony given by the witness; that said deposition was taken at the aforementioned time, date and place, pursuant to notice or stipulations of counsel; that I am not a relative or employee or attorney of any of the parties, or a relative or employee of such attorney or financially interested in this action; that I am not, nor is the court reporting firm with which I am affiliated, under a contract as defined in Civil Rule 28(D).

IN WITNESS WHEREOF, I have hereunto set my hand and seal of office, at Cleveland, Ohio, this ____ day of _____, A.D. 20____.

Pamela Greenfield, Notary Public, State of Ohio
1750 Midland Building, Cleveland, Ohio **441**15
My commission expires June 30,2003

Lawyer's Notes

D

E

F

G

Lawyer’s Notes

CURRICULUM VITAE
January 1,2003

Name: Arthur B. Zinn, M.D., Ph.D.

SS#: 115-40-8605

Place of Birth: Brooklyn, New York

Date of Birth: February 6, 1949

Work Address: Center for Human Genetics
University Hospitals of Cleveland
11100 Euclid Avenue
Cleveland, Ohio 44106
(216) 844-3936

Home address: 3674 Townley Road
Shaker Heights, Ohio 44122
(216) 751-4972

Education:

1965 - 1969	Brandeis University, B.A. (cum laude, chemistry)
1969 - 1976	Case Western Reserve University, M.D.
1969 - 1977	Case Western Reserve University, Ph.D. (biochemistry)
1977 - 1979	University of Minnesota Hospitals, Pediatrics residency
1979 - 1982	Yale University, Human Genetics fellowship

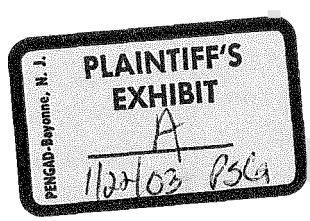
Professional Appointments:

Academic appointments:

1982 - 1992	Assistant Professor of Pediatrics, Case Western Reserve University
1992 -	Associate Professor of Pediatrics, Case Western Reserve University
1993 -	Associate Professor of Genetics, Case Western Reserve University

Hospital appointments:

1982 -	Full-time member of active staff, Rainbow Babies and Childrens Hospital of University Hospitals of Cleveland
1999 -	Tod Children's Hospital, Youngstown, OH



Honors and Awards:

1969 - 1976	Medical Scientist Training Program Award
1983	Frederick C. Robbins Award, March of Dimes, Northeast Ohio chapter
1985	International Travel Award, American Society of Human Genetics
1991	Department of Pediatrics Teaching Excellence Award

Licensure:

1982 -	Ohio

Board certification:

1981	American Board of Medical Examiners
1982	American Board of Medical Genetics (Clinical)
1996	American Board of Medical Genetics (Clinical Biochemical)

Professional Service:

Community activities:

1991 -	Medical Advisory Board, National Organic Acidemia Association
1994 -	Medical Advisory Board, Huntington Disease Society of America, Northeast Ohio Chapter

Journal reviewer:

American Journal of Human Genetics
Clinical Pediatrics
Human Molecular Genetics
Journal of Biological Chemistry
Journal of Clinical Investigation
Journal of Pediatrics
Neurology
New England Journal of Medicine

National collaborative programs:

1991 -	Register of Selected Inherited Metabolic Disorders
1991 -	Value of Bone Marrow Transplantation for Storage Disorders
1994 -	Director, Designated Gaucher Disease Treatment Center

Professional societies:

1982 -	American Society of Human Genetics
1993 -	Society for Inherited Metabolic Disorders

Study sections:

1989	NIH Special Grants Division, ad hoc review committee
1987-1989	American Heart Assn, Northeast Ohio Affiliate (ad hoc)
1989	NIH Special Grants Division, ad hoc review committee
1995	NIH Special Grants Division, ad hoc review committee

Committees:

National:

2001 -	Clinical Genetics Examination Committee, American Board of Medical Genetics

School of Medicine:

1982	School of Medicine Task Force on Genetics
1985 - 1989	Co-coordinator, Area of Concentration in Human Genetics
1989 -	Coordinator, Area of Concentration in Human Genetics
1999 -	Committee on Students

Department:

1983	Search committee for Chief, Division of Pediatric Hematology
1994	Ad hoc committee on graduate education
1995 - 1997	Committee on Appointments and Promotion
1995 -	Coordinator, Clinical Genetics Grand Rounds

Hospital:

1986 - 1992	Co-organizer and member, Disorders of sexual development team
1987 - 1994	Co-director, Center for Inherited Disorders of Energy Metabolism
1994 -	Center for Inherited Disorders of Energy Metabolism
1994 -	Director, Gaucher Disease Treatment Center
1994 -	Director, Huntington Disease Testing Center
1995 - 1997	Liver transplantation committee
1996 -	Director, Biochemical Genetics Training Program
1996 -	Laboratory Liaison Committee
1997 -	Director, Neurogenetics Clinic
1999 -	Co-Director, Metabolism Clinic

Community:

1997 -	Medical Advisory Board, Huntington Disease Society of America, NE Ohio chapter
2000 -	Ad hoc committee on newborn screening, Ohio Department of Health

Teaching Activities:

Courses:

1982 -	Human Genetics Committee (Phase 1 Medical Students), 7-10 hrs per year
1996 -	Biochemistry Committee (Phase 1 Medical Students), 2 hrs/year
1996 -	Genetics and the Law (Medical, Law and Ethics Students), 2 hrs/year
1998 -	Advanced Medical Genetics 527 (Biochemical Genetics), 32 hrs/year
2000 -	Advanced Medical Genetics 527 (Biochemical Genetics), 32 hrs/year

Postgraduate Training:

Sean Phipps, B.S.	Ph.D. Thesis Committee, 7/84 - 6/86
Vickie Zurcher, M.D.	Genetics resident, clinical training, 7/87 - 6/89
Mark Johnson, B.A.	Ph.D. Thesis committee, 7/90 - 6/96
Meral Gunay, M.D.	Genetics resident, clinical training, 7/96 - 6/99
David Everman, M.D.	Genetics resident, clinical training, 7/97 - 6/00
Derek Neilson, M.D.	Genetics resident, clinical training, 7/99 -
Mark Johnson, M.D., Ph.D.	Genetics resident, clinical training, 7/99 -
Linda Jeng, M.D., Ph.D.	Genetics resident, clinical training, 7/99 -
Khusroo Qureshi, M.D.	Genetics resident, clinical training, 7/99 - 6/01
Jeanne Brunger, B.S.	M.S. Thesis Committee member, 6/99 – 6/00
Marni Falk, M.D.	Genetics resident, clinical training, 7/00 -
Rocio Tarvin, M.D.	Genetics resident, clinical training, 7/00 -

Previous grant support:

1. A multinuclear NMR study of animal models of propionic acidemia and methylmalonic acidemia (AB Zinn, PI). Research Initiation Grant, Board of Trustees, Rainbow Babies and Childrens Hospital. Award for $10,000 (total direct costs). July, 1983 - June, 1985.

2. Biochemical studies of defects in the electron transport chain in mitochondrial myopathies (AB Zinn, PI). Robert J. Frackelton Fund, University Hospitals of Cleveland. Award for $10,000 (total direct costs). July, 1985 - June, 1987.

3. Complementation analysis of respiratory chain defects in patients with early-onset dementia (AB Zinn, PI). Alzheimer Center Pilot Grants, University Hospitals of Cleveland. Award of $10,000 (total direct costs). July, 1987 - June, 1988.

4. Complementation analysis of mitochondrial cardiomyopathy (AB Zinn, PI). American Heart Association, Northeast Ohio Affiliate. Award of $17,047 (total direct costs). July, 1987 - June, 1988.

5. Center for Disorders of Energy Metabolism (DS Kerr, Director; AB Zinn, Co-director), Rainbow Babies and Childrens Hospital Center of Care Grant. Award of $72,280 (total direct costs). January, 1989 - December, 1991. Percent effort: 10%.

6. Center for Disorders of Energy Metabolism (DS Kerr, Director; AB Zinn, Co-director), Bureau of Maternal and Child Health. Award of $747,112 (total direct costs). October, 1988 - October, 1993. Percent effort: 10%.

7. Ethics, Genetics, and Alzheimer Disease (Post SG, PI). NIH. Award of 155,377 (total direct costs). April, 1995 - September, 1996. Percent effort: 10%.

Current grant support:

1. Comprehensive Genetic Services in Northeastern Ohio, Ohio Department of Health #525K5 (HF Willard, PI). January 1,2002 - December 31,2002. Award of $194,000 (total direct costs). Percent effort: 50%.

Publications:

Articles (peer reviewed):

1. Kosower NS, Kosower EM, Zinn AB, Carraway R. Methyl 5-diazolevulinate intervention in chemically induced porphyria of rats. Biochem Med 2:289-306, 1969.

2. Marshall JS, Green AM, Pensky J, Williams, Zinn AB, Carlson DM. Measurement of circulating desialylated glycoproteins and correlation with hepatocellular damage. J Clin Invest 54:555-562, 1974.

3. Teng TL, Harpst JA, Lee JC, Zinn AB, Carlson DM. Composition and molecular weights of butyrylcholinesterase from horse serum. Arch Biochem Biophys 176:71-81, 1976.

4. Zinn AB, Marshall JS, Carlson DM. Preparation of glycopeptides and oligosaccharides from thyroxine-binding globulin. J Biol Chem 253:6761-6767, 1978.

5. Zinn AB, Marshall JS, Carlson DM. Carbohydrate structures of thyroxine-binding globulin and their effects on hepatocyte membrane binding. J Biol Chem 253:6768-6773, 1978.

6. Zinn AB, Hine DB, Mahoney MJ, Tanaka K. The stable isotope dilution method: A highly accurate approach to the prenatal diagnosis of methylmalonic acidemia. Pediatr Res 16:740-745, 1982.

7. Phipps S, Zinn AB. Psychological response to amniocentesis: 1. Mood state and adaptation to pregnancy. Am J Med Genet 25:131-142, 1986.

8. Phipps S, Zinn AB. Psychological response to amniocentesis: II. Effects of coping style. Am J Med Genet 25:143-148, 1986.

9. Zinn AB, Kerr DS, Hoppel CL. Fumarase deficiency: A new cause of mitochondrial encephalomyopathy. N Engl J Med 315:469-475, 1986.

10. Olsen MM, Caldamone AA, Jackson CL, Zinn AB. Gonadoblastoma in infancy: Indications for early gonadectomy in 46,XY Gonadal Dysgenesis. J Pediatr Surg 23:270-272,1988.

11. Cantrell MA, Bicknell JA, Pagon RA, Page DC, Walker DC, Saal HM, Zinn AB, Disteche CM. Molecular analysis of 46,XY females and regional assignment of a new Y-chromosome-specificprobe. Hum Genet 83:88-92, 1989.

12. Zurcher VL, Golden WL, Zinn AB. Distal deletion of the short arm of chromosome 6. Am J Med Genet 35:261-265, 1990.

13. Wallis GA, Starman BJ, Zinn AB, Byers PH. Variable expression of osteogenesis imperfecta in a nuclear family is explained by somatic mosaicism for a lethal point mutation in the α1(1) gene (COL1A1) of type I collagen in a parent. Am J Hum Genet 46:1034-1040, 1990.

14. Johnsen DC, Weissman BM, Murray GS, Zinn AB. Enamel defects: a developmental marker for hemifacial microsomia. Am J Med Genet 36: 444-448, 1990.

15. Rinaldo P, Welch RD, Schmidt-SommerfeldE, Gargus JJ, Previs SF, Zinn AB. Ethylmalonic/ adipic aciduria: Effect of oral carnitine and glycine on urinary excretion of organic acids, acylcarnitines and acylglycines. Pediatr Res 30:216-221, 1991.

16. Coppes MJ, Liefers GJ, Higuchi M, Zinn AB, Balfe JW, Williams BRG. Inherited WT1 mutation in Denys-Drash Syndrome. Cancer Res 52:6125-6128, 1992.

17. Mehlman MJ, Kodish ED, Whitehouse P, Zinn AB, Sollitto S, Berger J, Chiao EJ, Dosick MS, Cassidy SB. The need for anonymous genetic counseling and testing. Am J Hum Genet 58:393-397, 1996.

18. Putman EA, Cho M, Zinn AB, Towbin JA, Byers PH, Milewicz DM. Delineation of the Marfan phenotype associated with mutations in exons 23-32 of the FBN1 gene. Am J Med Genet 62:233-242, 1996.

19. Schwartz S, Depinet TW, Leana-Cox J, Isada NB, Karson EM, Pasztor LM, Sheppard LC, Wolff DJ, Zinn AB, Zurcher VL, Zackowski JL. Sex chromosome markers: characterization using fluorescence in situ hybridization and review of the literature. Am J Med Genet, 71:1-7, 1997.

20. Depinet TW, Zackowski JL, Earnshaw WC, Kaffe S, Sekhon GS, Stallard R, Sullivan BA, Vance GH, Van Dyke DL, Willard HF, Zinn AB, Schwartz S. Characterization of neo-centromeres in marker chromosomes lacking detectable alpha-satellite DNA. Hum Molec Genet 6:1195-1204, 1997.

21. Post SG, Whitehouse PJ, Binstock RH, Bird TD, Farrer LA, Fleck LM, Gaines AD, Juengst ET, Karlinsky H, Miles S, Murray TH, Quaid KA, Relkin NR, Roses AD, St. George-Hyslop PH, Sachs GA, Steinbock B, Truschke EF, Zinn AB. The clinical introduction of genetic testing for Alzheimer Disease: an ethical perspective. JAMA 277:832-836, 1997.

22. DiPiero AD, Lourie EM, Berman BM, Robin NH, Zinn AB, Hostoffer RW. Recurrent immunocytopenias in two patients with DiGeorge/velocardiofacial syndrome. J Pediatr 131:484-486, 1997.

23. Kori AA, Robin NH, Jacobs JB, Erchul DM, Zaidat O, Remler BF, Averbuch-Heller L, Dell'Osso LF, Leigh RJ, Zinn AB. Pendular nystagmus in a peroxisomal assembly disorder. Arch Neurol 55:554-558, 1998.

24. Morrow MJ, Zinn AB Tucker T, Leigh RJ. Maculopathy in spinocerebellar ataxia type 7 (NeuroImages). Neurology 53:244, 1999.

25. Riley D, Wiznitzer M, Schwartz S, Zinn A. A 13-year-oldboy with cognitive impairment, retinoblastoma and Wilson disease, Neurology 57:141-3,2001.

Thesis/Books:

1. Zinn AB. Ph.D. Dissertation, Case Western Reserve University. Studies on thyroxine-binding globulin (TBG): carbohydrate structure and liver binding, 1977.

Book Chapters:

1. Zinn AB, Plantner JJ, Carlson DM. Nature of linkages between protein core and oligosaccharides. In: The Glycoconjugates, Vol 1, Horowitz MI, and Pigman W (eds), Academic Press, New York, 1978, pp. 69-81.

2. Tanaka K, Zinn A, Hyman D, Hine D. Study of metabolism and prenatal diagnosis of inborn metabolic disorders using stable isotopes. In: Mass Spectrometry in the Health and Life Sciences, Burlingame AL, and Castagnoli N Jr (eds), Elvesior, Amsterdam, 1985, pp. 471-489.

3. Zinn AB. Genetics of sex reversal syndromes. In: Sex reversal syndromes, Caldamone AA (ed), Dialogues in Pediatric Urology, 1987, 9:2-8.

4. Zinn AB, Supinski G. Genetic diseases of the pulmonary parenchyma. In: Textbook of Pulmonary Medicine, 4th edition, Baum GL, and Wolinsky E (eds), Little, Brown, and Company, Boston, 1989, pp. 1521-1540.

5. Hoffmann G, Zinn AB. Aciduria, fumaric. In: Birth Defects Encyclopedia. The comprehensive, systematic, illustrated reference source €or the diagnosis, delineation, etiology, biodynamics, occurrence, prevention, and treatment of human anomalies of clinical relevance, 2nd edition, Buyse ML (ed), Alan R Liss, Inc, New York, 1990, pp. 28-29.

6. Ruggerie D, Zinn AB. Cardiomyopathy. In: A Practical Guide to Pediatric Intensive Care, 3rd edition. Blumer JB (ed) CV Mosby, St. Louis, 1990, pp. 410-416.

7. Zinn AB. Inborn errors of metabolism. In: Neonatal-Perinatal Medicine, 5th edition, Fanaroff AA, and Martin RJ (eds), WB Saunders, Philadelphia, 1992, pp. 1118-1151.

8. Zinn AB. Genetic diseases of the pulmonary parenchyma. In: Textbook of Pulmonary Medicine, 5th edition, Baum GL, and Wolinsky E (eds), Little, Brown, and Company, Boston, 1994, pp. 1785-1816.

9. Zinn AB. Genetic disorders that mimic child abuse or sudden infant death syndrome. In: Child Abuse: Medical diagnosis and management, Reece RM (ed), Lea & Febinger, 1994, pp. 404-429.

10. Kerr DS, Zinn AB. The pyruvate dehydrogenase complex and tricarboxylic acid cycle. In: Inborn Metabolic Diseases: Diagnosis and Treatment, 2nd edition, Fernandes J, Saudubray J-M, and van den Berghe G (eds), Springer-Verlag, Berlin, 1995, pp. 110-119.

11. Zinn AB. Inborn errors of metabolism. In: Neonatal-Perinatal Medicine, 6th edition. Fanaroff AA, Martin RJ, eds, St. Louis: WB Saunders, 1997, pp. 1390-1438.

12. Post SG, Whitehouse PJ, Zinn AB. Genetics, ethics, and Alzheimer's Disease. In: Post SG, Whitehouse, eds, Genetic Testing for Alzheimer Disease: Ethical and Clinical Issues. Baltimore, MD: The Johns Hopkins Press, 1998, pp. 1-13.

13. Kerr DS, Wexler I, Zinn AB. Defects of pyruvate metabolism and the citric acid cycle. In: Inborn Metabolic Diseases, 3rd edition, Fernandes J, Saudubray J-M, van den Berghe G, eds, Springer, Berlin, 2000, **pp.** 127-138.

14. Zinn AB. Inborn errors of metabolism. In: Neonatal-Perinatal Medicine: Diseases of the Fetus and Infant, 7th edition. Fanaroff AA, Martin RJ (eds), Mosby, St. Louis, 2001 **, pp.** 1468-1516.

Invited talkslworkshopslshort courses:

1. Maternal PKU: A Problem Born of Success. Ohio Department of Health and Ohio Genetic Services Network, 1983.

2. Medical Genetics and Birth Defects. Northern Ohio Pediatric Society Annual Postgraduate Course (A series of eight lectures), Cleveland, 1983 (organizer and speaker).

3. Medical Genetics and Birth Defects. Northern Ohio Pediatric Society Annual Postgraduate Course (A series of four lectures), Youngstown, 1983 (organizer and speaker).

4. The role of mitochondrial disorders in neuro-ophthalmology. Neuro-Ophthalmology Update 1991, Cleveland, OH, 1991.

5. Man's 25th Chromosome: Biology and clinical disorders of the mitochondrial genome. American Academy of Pediatrics national meeting, Fall, 1991 (plenary lecture).

6. ABC's of the New Genetics. American Academy of Pediatrics national meeting, Fall, 1991 (3-hour seminar).

7. Inborn errors of fatty acid oxidation. Visiting Professor, Department of Biological Chemistry, Chicago Medical School, 1992.

8. Disorders of the mitochondrial genome. American Academy of Pediatrics national meeting, Fall, 1993 (plenary lecture).

9. ABC's of the New Genetics. American Academy of Pediatrics national meeting, Fall, 1993 (3-hour seminar).

10. Krebs cycle defects. VI International Congress of Inborn Errors of Metabolism, Troina, Italy, June 2, 1994 (plenary lecture).

11. Disorders of the mitochondrial genome. American Academy of Pediatrics CME Program, Williarnsburg, VA, December 13, 1996.

12. Unstable repeat disorders: Fragile X syndrome and beyond. American Academy of Pediatrics CME Course, Williamsburg, VA, December 14, 1996.

13. Pandora's box revisited: Presymptomatic genetic testing. American Academy of Pediatrics CME Program, Williamsburg, VA, December 15, 1996.

14. Current status of genetic testing. American Academy of Pediatrics CME Course, Williamsburg, VA, December 13, 14 and 15, 1996.

15. Anonymous genetic testing. American College of Medical Genetics, Ft. Lauderdale, FL, March 2, 1997.

16. Mitochondrial diseases. Great Lakes Regional Genetics Group, Cleveland, OH, April 4, 1997.

17. Introduction to mitochondrial disease. Visiting Professor, Dept. of Medical and Molecular Genetics, Indiana University School of Medicine, Lecturer to 2nd year medical student class, and Workshop of Mitochondrial Disorders, Riley Children's Hospital, Indianapolis, IN, April 1,1998.

18. Triplet repeat disease: Fragile X syndrome and beyond. Practical Pediatrics CME Course, American Academy of Pediatrics, St. Petersburg, FL, September 11, 1998.

19. Mitochondrial disease: The great imitator. Practical Pediatrics CME Course, American Academy of Pediatrics, St. Petersburg, FL, September 12, 1998.

20. Pandora's box revisited: Presymptomatic genetic testing of children. Practical Pediatrics CME Course, American Academy of Pediatrics, St. Petersburg, FL, September 13, 1998.

21. The changing role of molecular diagnosis in clinical practice. Practical Pediatrics CME Course, American Academy of Pediatrics, St. Petersburg, FL, September 11,12,and 13, 1998

22. Genetic defects of the electron transport chain: A cause of myocardial dysfunction (Session co-chair: Genetic disorders of energy metabolism). Biomedical Engineering Society Annual national meeting, Cleveland, OH, October 12, 1998.

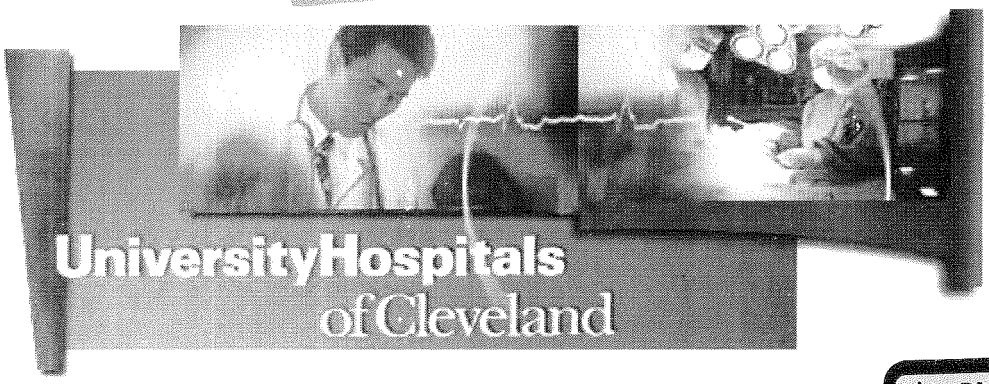
23. Nature versus nurture: Genetic determinism (Lecturer to Freshman Class Colloquium), Visiting Professor, John Carroll University, South Euclid, OH, November 6, 1998.

24. Mitochondrial genetics. United Mitochondrial Disease Foundation (Annual national meeting), Cleveland, OH, June 1,2000.

25. Case-oriented problem solving. United Mitochondrial Disease Foundation (Annual national meeting), Cleveland, OH, June 3,2000.

26. Mitochondrial genetics, and Thrombophilia: a complex trait. Visiting professor, Indiana University Medical School, August 23,2000.

UH - Genetics Department Roster



Center for Human Genetics

CLINICAL DIRECTOR:	**SUZANNE B. CASSIDY, MD** 216-844-7236
DIRECTOR:	**HUNTINGTON F. WILLARD, PhD** 216-368-1617
CONSULTATION/ REFERRAL PROCEDURE:	For information and appointments, call 216-844-3936
DESCRIPTION OF SERVICES:	The Center for Human Genetics maintains an active service focusing on clinical and laboratory diagnosis, management and genetic counseling of patients and family members for a wide range of pediatric, prenatal and adult genetic conditions. The Center also coordinates genetic counseling for individuals at risk for genetic conditions, and is actively involved in prograins offered in collaboration with specialty clinics for metabolic disorders, Prader-Willi syndrome, Marfan syndrome, bone disorders, craniofacial disorders and fainilial cancer.

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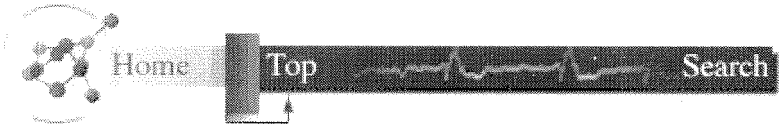

Wake Forest University School of Medicine
Department of Pediatrics / Section on Medical Genetics

Molecular Genetic Laboratory

Tel 336-716-4321 **Fax** 336-716-2554 Tuesday, November 7, 2000

Patient		**Sample**	
Name.....	Gwynne. Emily	Laboratory Number..	005640
Social Security #...		Date Received.....	11/3/00
Date of Birth.....	10/28/99	Date of Report.....	11/7/00
Hospital.....	Private outside doctor	Type of Specimen.,.	Blood
Hospital Unit #.....		Test Requested.....	LCHAD mutation testing

Physicians: Gunay-Aygin, Meral, Childrens Hospital Medical Ctr, 1 Perkins Square, Akron, OH 44308

Interpretation

Analysis indicates that Emily Gwynne is homozygous for the LCHAD G1528C (Glu474Gln) mutation which accounts for 55-60% of LCHAD alleles.

Laboratory Comments

A blood sample ~vasreceived from Emily Gwynne for LCHAD mutation testing. DNA was isolated by standard procedures and amplified by polymerase chain reaction. Following amplification, the product was digested with Pst I and analyzed by polyacrylamide gel electrophoresis. The G1528C mutation creates a Pst I site (a natural site occurs at bp 238 of the 270 bp product). A normal allele results in 238 and 32 bp fragments. A mutant allele results in 121, 117 and 32 bp fragments. (Reference: Ibdah JA et al., N Engl J Med 1999;340:1723-31)

Mark J. Pettenati, Ph.D., FACMG
Director, Molecular Genetics Laboratory

Wake Forest University School of Medicine
Department of Pediatrics /Section on Medical Genetics

Molecular Genetic Laboratory

Tel 336-716-4321 Fax 336-716-2554 **Tuesday, November 7,2000**

Patient		**Sample**	
Name.....	Gwynne, Eric	Laboratory Number..	005642
Social Security #..,		Date Received.....	11/3/00
Date of Birth.....	2/14/71	Date of Report..	11/7/00
Hospital.....	Private outside doctor	Type of Specimen.,.	Blood
Hospital Unit #.....		'Test Requested.....	LCHAD mutation testing

Physicians: Gunay-Aygin, Meral, Childrens Hospital Medical Ctr, 1 Perkins Square, Akron, OH 44308

Interpretation

Analysis indicates that Eric J. Gwynne is heterozygous for the LCHAD G1528C (Glu474Gln) mutation which accounts for 55-60% of LCHAD alleles. Genetic counseling is warranted.

Laboratory Comments

A blood sample was received from Eric J. Gwynne for LCHAD mutation testing. DNA was isolated by standard procedures and amplified by polymerase chain reaction. Following amplification, the product was digested with Psi I and analyzed by polyacrylamide gel electrophoresis. The G1528C mutation creates a Pst I site (a natural site occurs at bp 238 of the 270 bp product). A normal allele results in 238 and 32 bp fragments. A mutant allele results is 121, 117 and 32 bp fragments. (Reference: Ibdah JA et al., 1999;340:1723-31)

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Wake Forest University School of Medicine
Department of Pediatrics / Section on Medical Genetics

Molecular Genetic Laboratory

Tel 336-716-4321 Fax 336-716-2554

Tuesday, November 7, 2000

Patient		Sample	
Name	Gwynne, Kristie	Laboratory Number..	005641
Social Security #...		Date Received.....	11/3/00
Date of Birth.....	5/12/64	Date of Report.....	11/7/00
Hospital.....	Private outside doctor	Type of Specimen...	Blood
Hospital Unit #.....		Test Requested.....	LCHAD mutation testing

Physicians: Gunay-Aygin, Meral, Childrens Hospital Medical Ctr, 1 Perkins Square, Akron. OH 44305

Interpretation

Analysis indicates that Kristie L, Gwynne is heterozygous for the LCHAD G1528C (Glu474Gln) mutation which accounts for 55-60 % of LC'HAD alleles. Genetic counseling is warranted,

Laboratory Comments

A blood sample was received from Kristie L. Gwynne for LCHAD mutation testing. ~ ~ ~ isolated by standard procedures and amplified by polymerase chain reaction. Following amplification, the product was digested with Pst I and analyzed by polyacrylamide gel electrophoresis. The G1528C mutation creates a Pst I site (a natural site occurs at bp 238 of the 270 bp product). A normal allele results in 238 and 32 bp fragments. A mutant allele results in 121, 117 and 32 bp fragments. (Reference: Ibdah JA et al., N Engl J Med 1999;340:1723-31)

Mark J. Pettenati, Ph.D., FACMG
Director, Molecular Genetics Laboratory

Patient Name: GWYNNE, EMILY MDR #: 000656953
CHEMISTRY - BLOOD Chemistry - Blood - Sendout
10/31/2000 10:00:00

Carnitine Plasma
To Do Acycarnitine if enough specimen
Acyl to DUKE
Mayo#8802
Test Name ~~~~~
Acylcarnitine, Quantitative in Plasma
Patient Results ~~~~~

In the plasma sample, elevations of several long-chain acylcarnitines were detected. The prominent accumulation of 3-OH-acylcarnitine species is consistent with a diagnosis of long chain L-3-OH acyl-CoA dehydrogenase (LCHAD) deficiency. Molecular analysis is indicated.
Performed by:
MAYO MED LABS

Patient Name: GWYNNE, EMILY MDR #: 000656953
CHEMISTRY - BLOOD Chemistry - Blood - DIAGNOSTIC
10/30/2000 09:21:00

Organic Acid Screen, U~~~~~
In this urine sample a significant pattern of hypoketotic C6-C10 dicarboxylic acidria and C8-C14 dicarboxylic aciduria with prominent unsaturated aciduria. The profile is strongly suggestive of long chain L-3-OH acyl-CoA dehydrogenase (LCHAD) deficiency. Recommend molecular analysis.

Testing referred to Mayo Laboratories.

Interpretation ,..----

Testing referred to Mayo Laboratories.