

What Studies of Twins Tell Us About SLI

My thanks to Margaret for such a generous introduction. I'm really pleased to be here, and happy to share a little preview of what our panel is going to do today. And then we will launch right into our presentation.

So, that's my title. And we don't need those anymore. And... hm. I lost some slides. Come on guys. This isn't the, uh, hm. What happened? (Pause) This isn't; I saw one when I came up here to begin with. (Laughs) And so somehow this isn't, this isn't what I loaded on. (Laughs)

(She takes a minute to sort the slide problem out.)

Okay. Are we all awake now? (Laughs) Let us, uh... actually this still isn't, this isn't... hm. How, how interesting. I wonder what else is missing? (Laugh) Let's start, we'll start here, and then we'll go back and pick up that one. Somehow, somehow it's... left out of order. So bear with me while I, uh, as we move forward, kind of track our way through.

So our, our topic today is, that of scientific studies of SLI. So let me just briefly introduce the, the panelists here who are with me, who are going to walk us through various topics related to the current research on this, on this area of study. It's a little bit misleading in your program because of the way the ASHA convention has the electronic software set up the slots, so that under the Heritability of Specific Language Impairment and Sentence Diversity in Toddlers at Risk for SLI, are two different presentations; mine, to be followed by Pam Hadley. So you shouldn't burden Pam with my arguments for heritability. (Laugh) Nor should I be held responsible for Pam's expertise in toddlers. So, (Laugh) we're starting with kinda the big overview of the heritability and the environment genetics kinds of discussions, and some detail on scientific methods. That's what I'm going to do. And then we're going to move into toddlers, and work our way forward from there. And Larry Leonard is going to share with us his... legacy work, and his high level of expertise on roughly characterized as processing mechanisms that are in play with children with specific language impairment, and in this program of investigation he's going to talk about his work on word learning, vocabulary, and recall, memory as a mediator in... the performance of these youngsters. He of course, has done work on many other aspects of language and related abilities in children with SLI as well. And then Sean Redmond is going to walk us through his work on SLI and attention deficit hyperactivity disorder, ADHD. And in this talk he's also going to tell us how that interfaces with Social (Pragmatic) Communication Disorder, which is another timely topic area in our area of work. Suzanne Adlof is going to anchor us in the area of reading, and we all know that children with SLI are at risk for reading impairments, and Suzanne has done an extensive program of research for some time now on this topic area. So, I'm thrilled that they were able to make time on their commitment list to join us

today and share their work. Is somebody going to time me, because timing is going to get wild? Okay. Yeah.

Alright. So the content preview then that I'm going to start us with is... hm, what happened to that slide? There we go. The... let me, let me do a little preview. My previous slide is coming from recent comments by a renowned physicist who's the chair of the physics department at Harvard. And this is to cue this up as about the science that we've been involved with. By the way, you should all have a copy of the, the new brochure from NIDCD, and I'll mention later that NIDCD is the institute who is... has SLI listed as their scope of funding, so it's been a parent institute and served an important role in its line of investigation.

So back to the scientific studies of SLI. So Abraham Loeb's point is science is not about getting more life, we all like our, our media. And I have extracted one of his comments is, extraordinary group think, leads to extraordinary ignorance. And then I have here just the... the quote that I thought is relevant to what we're doing here today. "Physics is not supposed to be a recreational activity that makes us feel good about ourselves. History teaches us that groups of humans can feel happy in the company of each other while advocating the wrong ideas. Science is a learning experience about nature that holds the potential of showing us wrong, irrespective of our popularity status on Twitter. Physics is a dialogue with nature, not a monologue."

So the science that we're talkin' about here today is science that we have standards that apply across studies to help us know the extent to which we have confirmed the hypothesis that we tested, or the extent to which we should drop those hypotheses and start over again. Okay. So we're looking for confirmation and disconfirmation. Legacy we know that replicability and transparency are big issues in the world of science as we know it today.

So this is what I'm going to talk about in my... presentation. There's a long history of debating whether human language is inherited, or determined only by the environment. Studies of twins provide an elegant experimental design. Technical elements of twin studies, as well as the linguistic measures need to be understood to set the stage for digesting the meaning of those outcomes. And I will give you an overview of outcomes of 4 important recent twin studies. This is a program of research that's charting, plowing some uncharted territory in our history of studying twins. One of them is that twinning effects can complicate the identification of children with SLI, and we'll get into the details of that. Number 2, replicated findings of statistically significant inheritance has been obtained across various speech and language measures. Number 3, nonverbal IQ is not on the same causal pathway as specific language impairment or nonspecific language impairment. Number 4, implications for clinical diagnosis and treatment are to be gained from this kind of a program of work.

Now let's jump into the history of all this. Is there an inherited influence on language acquisition that is unique to language? So we're gonna give it a very quick historical scan with some highlights cued up here. One was Eric Lenneberg's work in 1967, a book

called *Biological Foundations of Language*, where he argued the predictions of a genetic causal pathway. He hypothesized that there was such a pathway, but he argued in that book, his conclusion was that it needed to be empirically tested. That the evidence wasn't sufficient at the time to come down on one side or the other.

We have the work of Noam Chomsky from 1957 to the current state of affairs, where he has proposed theoretical frameworks from linguistic studies in what is now called a "biolinguistics approach," hypothesized in that abstract knowledge of language is part of our human biological endowment.

Both of these two bullet points that I've been through are neither confirmed nor disconfirmed yet, but there matters a very active discussion among many disciplines around the world.

And then there's the issue, there is the method of twin studies. As I say, the method of twin studies is empirically driven. There's a logic to the data analytic methods that are being used to help us sort this out.

Now let's have a quick look at the history of studies of SLI. The notion of children with SLI appeared in Paula Menyuk's studies of children meeting this criterion to determine if SLI was characterized by deviant grammar, framed in Chomsky's claims about universal grammar. That work appeared in 1964. I've shared with another audience, that in 1964 at MIT where she did this work, they didn't even have toilets for women in the buildings. Paula Menyuk was a remarkable woman who did remarkable work, far ahead of her time.

Early on, a guy named Larry Leonard queried what is deviant language; a very important question that has cued up the subsequent literature. That work appeared in 1972, and was part of the inspiration for my returning out of clinical practice to the university to pursue a PhD.

Number 3, Stark and Tallal, very important leaders early on; in 1981, noted the issue of herit—heterogeneity in clinical samples, and developed a set a measurement criteria for studying children with SLI. There was at that time, as there is now, a general term called Developmental Language Disorders, which didn't mean what I'm going to tell you is the current definition. At that time, as of now, that term also means children who have any kind of a condition that may re—may be related to a language impairment. It's an umbrella arching term that among other things, has allowed us to compare subsets within that label.

So anyway, they noted it's, it leads to heterogeneity in clinical samples, and they explicitly excluded low nonverbal IQ, as were other conditions such as hearing loss and neurological disorders. This is a definition that has served us well since 1981.

NIDCD was established on October 28th, 1988 for the first time providing a home base for research on SLI. We were getting beat up before then by various other institutes that denied the existence of these kids. And NIH continues to highlight it is a, a... a

mechanism of funding for— NIDCD continues to highlight SLI as a target area for their funding.

There is a, a robust literature on this topic. The current SLI literature base estimates I've gotten from people who do these kinds of literature reviews and searches as part of their job definitions. There's 1,622 citations in the medical library of NIH in the search for Pub Med work. And that's, if it appeared in the title or abstract and there's 660 articles, abstracted titles from the ASHA publications themselves.

So we're talking about a tradition of research here that we're proud of, and that it has replicated, and we're going to learn more about what it tells us that's relevant to clinical practice. Let's now segue into the case of twins.

There's an increased rate of twin births. This is a timely topic. In the USA in 2009, one in 30 babies, or 3.3 % were twins, compared to 1980, 1 in 53 babies were 1.8%. And it's increasing, and continues to increase, and it's related to the third bullet here, that I will come to in a minute.

Twins of course are paired at birth. They are two babies that were ... conceived at the same time, and most of them are born on the same day. And if we don't jiggle the, the clock one minute before midnight and one minute after midnight, they generally are born at the same time. There's two kinds of pairs of twins, and this establishes the logic of, of all of the data analytics methods and the inferential assumptions that come out of it. There's monozygotic, or identical twins, which are cases of a single fertilized egg that, that splits into two. So they develop from one zygote, which splits and forms two embryos. In that case their DNA is almost identical. We do know now that it actually isn't completely identical but darn close. And then the, the second kind of twin pair are dizygotic or fraternal twins which develop from two different eggs, each fertilized from its own sperm cell. So the dizygotic twins, fraternal twins, I'm going to use in MZ's and DZ's from here on, and we'll walk through it. But the DZ's are no more like each other than another sibling, they just happen to be the same age. Now the reason there's an increase in twins is with assistive reproductive technology methods that actually generates many DZ twin possibilities, because these are separate fertilized eggs, and they're implanted at the same time and the birth is at the same time, that they are going to be DZ's and not MZ's, okay. So, and this is prevalent everywhere around the world. The twin studies that I'm going to tell you about that were conducted in Australia, were conducted, they were ascertained before ART was used as a method of conception in that area. So the data that I have are not confounded by the ART phenomenon, and it, there's also a sample as in every other naturally obtained sample, um... in the pre ART days, it's 1/3 MZ's and 2/3 DZ's. Nature doesn't go for them MZ option nearly as much as the DZ option.

This is the way these relationships are schematized in the, the traditional literature in this area. So here's a pair of identical twins, and they're girls. We can tell they're girls because their heads are round, and the little tent above their head has a line unifying the two slanted lines, indicating that they're MZ's. Here's another, uh... set of relationships.

These are DZ twins. And we see that because the little tent over the head doesn't have the unifying line, and these are also girls, but here we see there is a big black L in the middle of one of their heads, and then a small red L in the middle of the head of the other one. These L's represent a phenotype or the behavioral symptom that you're studying in genetic studies. I'm going to tell you about speech and language phenotypes. That's the part of the mental apparatus that we study as outcomes. The big L indicates robust or age expected, and the small red L indicates not up to age expectations, right. So these would be twins who are discrepant, on that phenotype, and... these would be twins who are concordant on that phenotype, okay.

So now here we go. Twins. That's, let's get into why study twins; what do they tell us that we can't get from single born children. There's a... formula, referred to as a behavioral genetics approach, which has been in play for a long time and very helpful. And basically it assumes that we're looking to account for all of the variants, and so our, uh... uh... end game is 1.0 and then it is carved up into 3 different ways in which children could vary within twin pairs. One way is, it's because there's inherited differences, and those are differences that would allow us to predict that the MZ twins are going to be more like each other than the DZ twins. Then there's environmental effects that are common to the twin pair. That's taking into account that these two twins live in the same household at the same time. And we can evaluate those influences because that means that the DZ twins should be as much alike each other as the MZ twins. And then there's environmental effects, which sometimes gets characterized as error as well, which is unique to each twin. So for example, if one of the twins has encephalitis and the other twin does not, that would be an environmental effect, although it's a biological effect. So you have to be thinking in terms of environmental effects of that kind, making a difference that hasn't anything to do with the birth status of the twins. So this comes under the rubric of behavioral genetics. There's a huge literature on it, and I will walk through some of the key studies.

So these methods are based on correlations, or quantitative estimates, and in today's word-- world, modeling approaches are used in which we can control for mother's education, we can control for family income, and all kinds of other related things. So we can develop separate estimates for these covariates, so maternal education and maternal age are shared by the twin pair. You can specify that in the model, and twin specific predictors like age at assessment, sex, birthweight, APGAR scores at birth; these kinds of things can also be included in the model and estimated.

I'm going to talk a lot about twinning effects. This refers to, it's been speculation out there, but never really confirmed in the way in which our study's been able to do, that twin children are later than single born children in language development. It's been a widespread assumption among pediatricians, but the evidence hasn't been available.

The point of this slide is just to repeat what I just said about concordance and heritability. So, the MZ's bein' alike, and the DZ's bein' less alike is consistent with heritability. Okay. And so we're, we're really interested in the phenotype and the phenotype's relationship within twin pairs.

Let's now unpack a little bit then what this means. So if we put it in a figure, it's Twin 1 on the Y axis, and twin 2 on the, uh... uh... X axis or Twin Y on the vertical and Twin 2 on the horizontal. Let's assume it's, uh, vocabulary. So Twin 1's PPDT score and Twin 2's PPDT score, if this is heritable, those two core, those two scores should be very much alike if they're MZ twins. And... if it's heritable, the DZ twins will not have that high a correlation. And that's what we're estimating in all of these quite involved statistics.

So how do heritability estimates differ from twinning effects? Correlations are for heritability, and level of ability are for twin effects. So let's unpack the difference between a correlation and the level of ability. So we all work with correlations. So now we're looking at that vertical axis is as being the vocabulary standard score, and let's correlate it with the reading standard score, okay. So if vocabulary and reading run together perfectly, you'll get that diagonal line with a correlation of .1, 1.0. And if it's not so perfectly associated, it's .8. And if it's what we usually see in our line of investigation, it's .3. Suzanne's work, and that of others, show this, that this relationship with reading and language is in the .5-ish range in most of the studies, okay.

So those are correlations. Now let's look at levels of performance. Now we have the same level of correlation, it's a .8, and it's, it's the same for the typical children and for the low performing children. But you can see the low performing children perform much lower in ability. The, the issue is, when you're looking at correlations, you're looking at the extent to one, one predicts the other, but that doesn't tell you about the level of performance. So the twinning effect is about the level of performance, the heritability estimates are about the correlations, okay. So the expected outcomes for a twinning effect would be that the MZ and DZ twins, if nothing else is operating in there, perform lower than the single twins.

So, here we go. Why don't we have more twin studies? You need large sample size for these kinds of statistical methods, and you need population based samples. Some time ago, twin studies were mostly done by recruiting out of twin clubs, and that's mostly MZ twins. So it's only on the population based that you get MZ's as well as DZ's. So the current state of results about twin language acquisition, there's multiple studies, mostly from one large study in England beginning about 2000. It's called the Twin's Early Development Study known as TEDS. And there also have been some important studies of reading, but I'm not going to get into those today. The heritability estimates that have been reported previously are in the range of .24 to .59. The generalizations are that there's increasing heritability with age, increasing heritability with lower levels of performance, and... in that existing database, before we started our work, there were limitations of language phenotypes, most of them are general in nature. Individual differences in the big TEDS study, was reduced to rank within the twin sample, because they were under increased, uh, enormous political pressure to test everything that anybody wanted to have tested in that sample. They were very... constrained by how much time they could spend in any one area of investigation. So for language measures, they chose not to give the standardized version of the language measures, so they could not compare their twin's performance levels to single born estimates. Instead they used

the rank within their sample. So within the twin sample, they have low performing and high performing. That is not the same thing as comparing to a full population sample of single born kids. They, most of these studies have overlooked the dimensions of grammar, and they've overlooked speech abilities. For both of those, it's been mostly because they're hard to measure, and the teams that were working with them either didn't know or didn't want to spend the time to go after it. The other thing, they have limited growth estimates in the big TEDS study, because they didn't use the same measures over time. The political pressures were such that if they got something in with their two year old cohort, they dropped that line of measurement, and then in their four year old cohort, they measured things that other people wanted to have done. So they met the, the political pressures on it. So there's little known about possible twinning effects, until what I'm going to share with you now.

So, this is a longitudinal study of twins at ages 2, 4, 6, 9 and 14 years. Data collection was in Western Australia, based on Perth, a beautiful, beautiful city, which happens to be demographically almost identical to Kansas City. Go figure. Um... it isn't anymore, because the Chinese are in there mining all their natural resources. So they now have a ver—a much higher Asian proportion than Kansas City does. But when we were there, they didn't.

The first report came out in 2014 on the 24 month twins, which is parent report data. And incidentally, these girls were with us all the way through. And, they, as adults, are just as adorable as they were when they were little. So, this, this first study is the population base sample with 470 twins or 964 children. Parent reports, the outcome as twins had lower average language scores than single born norms. The percent of words produced was only 26% relative to age expectation. And the late language emergence proportion are the kids who were behind the age expectation was 38%. There was lower performance for MZ twins, and for boys. So notice that twinning effect my God, it's there more for the MZ, and that has replicated. I'm going to tell you what happens to it over age. And, the biologists are quite confirmed that this is exactly what they would of expected, but we've never had this kind of information. Boys we're not so surprised about. Heritability, like other studies of children of this age, is relatively low, but it's there. .25 for vocabulary, grammar was .52, using the McArthur estimates for grammar, and that's replicating now around the world in other studies. Combining words, the heritability's .22, and again, I'm sharing the concordance rate because it turns out in this age span, the reason you get lower heritability is because both the MZ's and DZ's are quite a bit alike. Maybe we could say that's because environmental effects are stronger on children when they're younger, or we may say it's because our measurement systems aren't yet precise. Either one of those things could account for it. But this is a well-known phenomenon, and our finds are in line with others.

So Study 2 came out in 2018. Now we're looking at children at 4 and 6 years of age. Same children, slightly bigger sample because we added some as we were doing cohort recruitment into the study. So now we're at 1255 children, 628 twin pairs. You'll see that's roughly 1/3 MZ's and 2/3 DZ's, and it's equally distributed across males and females. We had 10 phenotypes. We have other things that I'm; we have lots of

phenotypes, but in this study there's 10 phenotypes that were done at 4 and at 6 years of age. Peabody Picture Vocabulary Test, the toll, spoken language managed syntax, Columbia Mental Maturity scales for nonverbal IQ. Incidentally, that thing is a psychometric work course. We see it in all kinds of ways. The Goldman Fristoe Test of Articulation for Speech, mean lengths of utterance, and then the Test of Early Grammatical Impairment, the TEGI test that I had developed in conjunction with Kim Wexler. So it's, looking at a particular property of grammar that there's multiple reasons to think that there's something going on there that's a little bit different from vocabulary learning for example.

The main outcomes were the expected patterns of correlation were evident, and they're getting higher. Higher for MZ than DZ twins, high for both types of twins, for within twin assessments at 4 and 6. So these are at the .45 to .68 range. This is a persistent twinning effect across various language and speech phenotypes. It's just there. And it's across multiple indicators of it. And these twinning effects decreased from 4 to 6 years. These MZ twins are sus—a significantly lower standard scores at 4, for 6 of the 10 phenotypes, but the zygosity differences are washed out by age 6. So whatever it is that nature is cueing up for, a, a later developmental sequence for MZ twins on average, seems to have worked itself through by 6 years of age. So the conclusion is that twinning effects are evident although reduced with age, and there's its resolution, and there are no twinning effects for nonverbal IQ.

So that sets up some interesting things for identifying kids with SLI.

The heritability estimates are higher than reported previously, in the range of .44 . 92 at 6 years. And that .92 is for the clinical grammar marker, and we're going to see that that level replicated in the PED study at a different age level on a different way of measuring into it.

So here's the figure summary then. DZ's and MZ's, this is an effect that's about 10 standard score points, so that the average for the wins is roughly at 90. It's a little bit less than 90. Roughly at 90, and the DZ's are better than the MZ's. At 2, the DZ's are better than the MZ's, those children are still about 10 standard score points, more than half a standard deviation below the mean, which again it's a very large effect size. And by 6 they're still below, although they're inching toward the singletons, but the DZ/MZ difference has washed out.

Study 3 then takes us into the heritability of SLI and NLI at ages 4 and 6 years, which is a paper that's under review. Same participant sample as Study 2, and now we're going after the question is there higher heritability for the kids at the bottom of the distribution than at, for the... typically or above kids. Same phenotypes. And now we've grouped 'em as SLI, the language levels below 85 standard score with no hearing loss or other developmental impairments, and a nonverbal IQ of 86 or above. We screened excessively in this sample. We do not have children who are exposed to English as another language. Like Kansas City at that time, Perth was almost uniformly monolingual, and we were quite diligent. We did not include twin pairs who were

attending a preschool where they were being instructed in a language other than English, so we really made sure that we didn't confound exposure to multiple languages with twinning effects. And we also did not include children who were carrying a neurological diagnosis, so that knocked out the kids with epilepsy and all that. We did let low birth weight float a little bit, and we've actually done the follow-up study to indicate that the risk indicators from all of our perinatal data do not differ from those for singleton kids.

And then we also in this study that I'm talkin' about now, we also identified the children who would of met the criteria for nonspecific language impairment. This is the impairment that Bruce Tomblin and his colleagues work with on a very important epidemiology longitudinal study based at the University of Iowa.

So we're interested in the percentage of twins with SLI at 4 and 6. The proportion at lower levels of performance, and the concordances and heritabilities for SLI compared to NLI. So the key outcomes are the twinning effects elevate the proportion of children who fall into SLI definition. Not surprising because of the twinning effect. So it's 28% of these kids that fall below singleton age norms for their age, whether it be 4 or at 6, compared to 7 to 85 in singleton population studies. Twinning effects change from age 4 to 6 somewhat, reducing 28% to 25%. We have analyzed some of the data from 9 and 14, so we know the rest of the story as well. But that, you see this shift beginning in this age range.

Children in the NLI group are 8% at 4, and 3 % at 6 years, compared to 2 to 3% expected for population studies of singletons. So we do get a little more movement into the NLI group, but most of the movement is into the SLI group. So, the caution that comes out of this work is that twinning effects should not be confused with SLI in singleton children. So we, we really have to be mindful that when twins come in for assessment, it's not the same situation as with singleton kids. And the effect works for differently for speech and language relative to nonverbal cognition.

This is what it looks like when you look at the full distribution of these samples at 4 and at 6. So on the upper right quadrant up there, are typical language, or, typical IQ and typical language areas, so the kind of a generic typically developing kids at 4 years, it's only 59% of the twin sample. The, when we shift down into SLI, the green at the bottom there picks up 28%. And then over in the low IQ low language, it's 8%. So be mindful when looking at these figures, the dotted line is the mean for the group, and the clinical definitions that we're using are the shaded part in the lower left corner, okay. So that's where you begin to see, you have to run those lines for -1 standard deviation across, across the... oh... let's see if I can make the, pointer work. So you have to see this line going all the way over here. Right? So what, why these things are important though, is to kinda lock onto what happens when this group of children shifts with age. The other thing is that we too, like other people, have found low IQ in typical language kids, even in this twin study where they're biased toward low language. And we picked up 4% of those kids when they were 4, and when they were at 6 it was down to 2%. But those kids are out there. We don't know very much about them. We would benefit from knowing a lot more about them I think, but they don't get picked up by speech pathologists for

obvious reasons, and they don't get followed up in longitudinal studies usually because they're not typical, and that they're not typical in nonverbal IQ. Okay.

So at 6 years you see the whole thing shifting diagonally up toward typical, and at 6 years, typical being defined by singleton norms at 71% of them who are there.

So the movement is dynamically out of the language impaired group, and we're... happily working on the report of what happens at, uh, 9 and 14 years after that.

So, SLI versus NLI over time in twins, as nonverbal IQ shows no twinning effect. The greatest consistency over time is for nonverbal IQ, the 85% or above are -1 standard deviation over 4 and 6. That's quite stable, meaning that if you calculate the percentage of kids who stayed in their quadrant. The language assessment's consistently above -1 standard deviation over 4 to 6 years of age, drops to 50 to 60%. So there's a lot of shift over time in language when we look at it that way on an individual child basis as well as these group means. Heritabilities were calculated at this -1 standard deviation criterion, and it increased from 4 to 6 years, perhaps again due to improved measurement, by reduced twinning effects, or perhaps because there's really something quite different happening in them. Heritability was significant for SLI and NLI in multiple language phenotypes, and it's greater at 6 years. We're, we're replicating the age effects on these estimates, .55 to .71. There were more technical difficulties in modeling the estimates for the NLI group, so for, we could use tolled only on doing that in, and note the tolled has to be psychometrically very robust in order to hold up under these modeling methods. And... the tolled under these modeling methods showed high heritability of .74 to .85. There was no double hit for nonverbal IQ and language impairments in this sample, these big samples of twins. Meaning, that it was, language scores were not the lowest for the low, low group. And, I think that has to do with how this twinning effect plays out. But it is very interesting. And their heri—uh, in other work out of the TEDS sample, their conclusion was that they definitely felt that there was a double hit, but I think that's because they were working with within twin definition, within twin group definitions of what constituted low.

The heritability for nonverbal IQ is inconsistent across age, and does not show congruence with speech and language measures outcomes. So we're not the only ones finding that nonverbal IQ can be inconsistent across ages.

Finiteness marking is the only heritable phenotype theoretically and empirically linked to adult grammar outcomes. So it is a, a meaningful measure if we're doing biolinguistics for example. Well that's interesting, because we have a way of referencing exactly what they should know, and relative to what they don't know in the adult grammar. It doesn't mean the other phenotypes are less interesting, it means that we just need to work out better ideas of what the other phenotypes are actually measuring.

So there's a 4th study, and I'm sharing this because I was contacted by the people doing the TEDS study, saying, we see what you're doing with these grammaticality judgment tasks, and we're going to have to collect data from our twins at 16 years of age, and we're

going to have to do it over the phone. “Are you willing to share your... measures with us? Because we’re going to put it out over the phone, (Laughs) and who knows what’s gonna happen.” So I said sure. And we worked it out, and this was a study led by Philip Dale, and a lovely young woman named Carla Rimfield I think was, and Hayiou-Thomas is, Mary Emma Hayiou-Thomas is the lead geneticist on this.

There were 3 language phenotypes in that study of those kids at 16. One was a grammaticality judgment task for finiteness marking that is coming directly out of my lab. It’s a 20 item grammaticality task, administered on the phone. So basically it’s a set of items which are paired, they’re not presented as paired, but they’re paired for which elements are... provided in a sentence, and which elements are omitted. So “What does she like to drink” appeared somewhere, and then many items later would be, “What’s he like to drink.” The task for the person receiving it is to indicate is that good or not so good. And again, they ran it on the phone. They also had a vocabulary measure, and a figurative language measure in the study. So this is just to show you what this, this task looks like in the work coming out of my lab. We have a lot more data now, and it still looks like that. So basically it’s a task that reliably separates the affected kids from the unaffected kids, and this persists for a long period a time up into adulthood we now know.

So the validity of the GJ marker for language impairment. Some reviewer wanted us to convince them that this wasn’t just some esoteric task that had nothing to do with other language things. So they were able to use other variables in their database to show that the mothers who had the lowest performance on this task had significantly lower levels of education. Incidentally, low levels of education is not a disconfirming reality for the diagnosis of specific language impairment. It’s an outcome that we are studying in trying to learn more about how this, how this is a risk for these kids. The... the... sample scored significantly lower on TEDS tests, the vocabulary and figurative language at age 16. So we were able to link performance on the GJ task to performance on vocabulary and figurative language tasks. And these kids even had significantly lower scores in school administered English and mathematics performance indicators, which kinda goes along with the lower education of the mothers. We have lots of reasons to know why these kids are cued up for low levels of education (inaudible word).

So the twin outcomes in this study, these are the concordances. So we’re starting there, and then I’ll show you the heritabilities.

So if we mark it off as the lowest 10% of the sample, and again, 10, the lowest 10% of this sample does mean the same thing as the lowest 10% in the Australia sample. Right? This is the lowest 10% of the sample of twins. And so we can see that the MZ kids were concordant at .28, and the DZ’s at .19. So, that’s not outstanding. The lowest 7% is .4, compared to .24, and the lowest 5% is .43, compared to .15, which just tells us that within their way of measuring low performance relative to the others, it gets better when you go farther down, and they get, they get bigger numbers, so it’s more stable. When we do that comparing it to... singleton standard scores, then we get into the instability range because of the, the fewer that are down there. The outcomes of Study 3 then, the

heritabilities for the grammaticality judgment marker at varying levels of language impairment, I can tell you that this is the highest level of heritability they've found for anything that they've measured in that big TEDS study. So at that lowest 10% it was .36, the lowest 7% was .47, and the lowest 5% is .74. So, this is... the terminology is the substantial level of heritability, and that leads us to some emerging conclusions. I think these are emerging. And, there's all kinds of reasons why we need more samples like this to be studied in this way.

Twins lag behind single born in language acquisition. This twinning effect that seems to resolve around 6 years. It matters which speech and language phenotypes are used for estimating heritabilities. There's a persistent pattern of highest heritability in these little grammar markers. On the other hand, there is evidence of heritability across different speech and language measures. The speech measures kinda drop out because we don't get a speech phenotype after 6 years. And even at 6 years, it's not as robust as, to use as, as it is at 4 in these kinds of studies with the test that we used. But, there's a lot there, and we don't have very good ways to quantify these apparent differences in strength of heritability. This kind of mathematical calculation doesn't lend itself well to (sounds like T Tests) for example. Although environment plays a role in language acquisition, I wish to highlight my firm conviction that environment does play a role in language acquisition. Individual differences include differences in inherited abilities, especially for children with SLI, and probably other forms of language impairment. My lab's involved in a lot of work on this now, and we have lots of pedigree showing that this runs in the families, and we're chasing down what some of that might be on the DNA level. But environment plays a very big role, and especially when you have parents who had SLI as a child themselves, and are also at the bottom of the expected norms for their age range. And that interfered with their ability to read well. And then they have offspring that looks just like they were when they were children. And they say to us, "He's just like I was, and I'd do anything to help him out." And then they go to school and visit with the speech pathologist who tells them that they just need to read to their child more, when they're speaking to a woman that doesn't read very well herself. So that's what, that's what we have to be able to sort out here as a consequence of this work.

Back to the science part. The implications for causal pathways. We need to know the likely linkages between clinical markers of grammar and other language measures and individual differences in genetic mechanisms. We're nowhere near knowing that. It'll be a long program of research. We need to know what are the bio—underlying biological controls of child development that may be implicated in twinning effects. The biologists who work on twinning effects are very interested in this. And there's other timed elements of language acquisition. Why does language acquisition start at the time it does? Why doesn't it start earlier or later? And why does it show such rapid acceleration until it hits preadolescence, when it levels off in rate?

Back to Lenneberg. I found myself after rereading his book, wondering if he would have reason to be vindicated or frustrated. It's been a long time since he wrote that book, and suggested that we get on with the empirical work, and we haven't arrived at a definitive answer yet. The implications for clinical diagnosis of SLI is that an independent measure

of nonverbal cognition would be helpful at arriving at more precise information about causal pathways and planning intervention. We would not have found out what we know now about this twin sample, if we had just lumped everybody together as a language disorder. We wouldn't have found that out.

Twins with language scores below 85 who are younger than 6 years of age bear watching. Twinning effects elevate the proportion of children who fall into this SLI definition, this 28%, compared to 7 to 8% in singleton population studies. So we have to qualify what we think is a real delay here, or how to operationally work with it, and how to understand what it is, and understand the developmental trajectory of it. Is it always gonna be this way? Well, we know already, no it isn't. How in the heck do they outgrow it; we have no idea.

The children in the NLI group were 8% at 4, 3% at 6 years, compared to, to 2 to 3% expected for population studies of singletons. So that's another way of understanding there's something different going on in this language system than in the nonverbal EQ one.

So, just to nail it down, language and nonverbal cognition and IQ are not the same thing. They have different developmental trajectories. Significant inherited component in speech and language, and to speech and language impairments during early childhood, and in, in, uh, adolescence. Some properties of grammar, but not all, I am quite confident this would not be true of all. I've heard the most ridiculous generalizations that everything about grammar is inherited, and so forth, that, uh, we have no reason to believe that's true. Twin children can be slower than singleton children in early speech and language, yet they catch up between 6 and 9, and we have no idea how they did it.

Let me end. I'm looping back to my opening slide now. "Extraordinary claims require extraordinary evidence." And this is from Carl Sagan, another prominent scientist who went on to do a great deal of public advocacy for science. And in my case, I believe that continued studies of children with SLI can confirm or disconfirm long standing, but wrong assumptions about the sources of children's effortless language acquisition, or poorly understood reasons for persisting language impairments, how to diagnose this, then, and how to treat them. Most of all, I want you to understand firmly that ... children with SLI don't have language impairments because they aren't very smart. I've been told that by prominent pediatricians. And I've been happy to work for years to convince them that they were wrong. And children with SLI whose parents have low SES status, are not producing children whose language impairment is caused by poverty. Those, both of those assumptions bring a great deal of shame to the people that we study.

So I'm happy to be here to share the work that my group of very talented people has been doing. We've been doing it with the good fortune of support from NIDCD of various kinds over the years. My collaborators include geneticist Shelly Smith and Javier Gayan and others in England. The primary investigators in Australia are Cate Taylor and Steve Zubrick, my brilliant statistician is Lesa Hoffman. I'm very grateful for all the ways in which they make me better at what I do. These are their photos, and this is some of the

Australians who look like a lot like us, don't they; um-hm. And, there's references here; if you want any a these references, I'll be happy to talk more. For those of you that are real longs, we have a lovely paper that appeared in a pediatric journal on perinatal predictors of late language emergence at 24 months. Thank you very much. (Applause)