IS THERE A GENETIC BASIS FOR STUTTERING?

John C. Harrison

IS THERE A GENETIC BASIS FOR STUTTERING?

John C. Harrison

IS THERE A GENETIC BASIS FOR STUTTERING?

Is there something buried deep in our chromosomes that lies at the root of stuttering–a stuttering gene, if you will, that affects us in the same way that some lurking genetic presence creates multiple sclerosis and cancer? If we don't turn to genetics, how can we possibly explain the fact that stuttering often runs in families?

I'd like to begin this exploration by making an improbable comparison and talking about something far removed from stuttering–the conflict in Kosovo.

Like many people, I was appalled when I first heard of the atrocities that took place in Kosovo during 1999. I was even more stunned to discover that the bad feelings

between Serbs and ethnic Albanians reach far back in history, as far back as the fourteenth century. At the Battle of Kosovo in 1389, the Serbs were defeated by the invading Ottoman Turks, and by the mid-fifteenth century, all of Serbia, including Kosovo, had fallen under Turkish rule. This initiated the beginning of a Serb migration northwards to Bosnia, and the replacement of Serbs by mostly Muslim Albanians who came to the fertile lands of Kosovo from the more arid, mountainous regions of Albania. To this day, Serbia still sees Kosovo as belonging to them, while the Albanian nationals in Kosovo continue to push for independence, and in this controversy over land, the animosity between Serbs and ethnic Albanians continues to fester.

But for *500 years?* There must be some explanation for the persistence of such rancor. How could these animosities be transmitted so effectively from one generation to another? Ah, I have it. At the heart of it, the Serbs must have a genetic predisposition to kill Albanians, while Albanians have a genetic predisposition to kill Serbs. *That* would explain it.

An absurd conclusion, of course. Genetic predispositions aren't the only determinants that can run in families for centuries. Other factors can be communicated from one generation to another. But these factors are never even considered when it comes to stuttering, because of the tendency we all have to enlist one mystery (in this case, genetics) to explain another.

Genetics is one of the catch-all answers that people turn to whenever they run out of ideas to explain the unknown. For example, if I've heard it once, I've heard it a hundred times–"Since my father (mother, uncle, aunt, brother, etc.) stuttered, there must be a genetic predisposition in my family to stutter."

Many researchers have taken the bait. There has been considerable research among scientists to find and isolate a stuttering gene, or at the very least, the key genetic factor that is the central cause of stuttering. Maybe it has to do with the timing of speech, they say, or perhaps it relates to a failure in the auditory feedback system. Whatever the cause, a growing number of people have had the inside of their skulls illuminated by a CAT scan, or had their blood drawn and analyzed by scientists looking for clues to their stuttering in the ghostly patterns made by their DNA on film.

I applaud their persistence, but I suspect that history will ultimately prove their efforts futile. There is a much simpler explanation to why stuttering often runs in families.

Yet, this explanation seems to elude researchers in speech pathology.

Why?

Because they suffer from a common malady. It is something we call *paradigm paralysis*.

Thinking "inside the box"

A paradigm is a model, a shared set of assumptions about how we perceive the world. Paradigms tell us what we need to pay attention to and what we can safely ignore. Paradigms are essential because without the ability to filter the important from the unimportant, we'd have to wrestle with too much data and too much sensory input.

But sometimes a paradigm can work against us. This happens when the paradigm filters out information that is really important–data, impressions, information that we *should* be noticing and dealing with. I propose that this is precisely what has happened with stuttering.

Some years ago, researchers decided that chronic stuttering was a unitary disorder primarily caused by a malfunction of the speech-making system. They weren't precisely sure what was malfunctioning. But they were sure that if they looked hard enough, the answer would be forthcoming, and that they'd find it somewhere in the dark recesses of the brain.

By totally embracing this belief, their thinking became paralyzed. By staying within the familiar paradigm, they limited their research for alternative answers. They kept their thinking "inside the box." In so doing, they allowed the probable cause of stuttering to slip between their fingers.

To better understand why their thinking went amiss, let's examine four key assumptions on which genetic research

into stuttering is founded.

• We can all agree on what we mean by "stuttering."

• We can accurately identify when a person is blocked.

• Only genetic factors can be transmitted from one generation to another.

• You can do meaningful research without having a clear idea of what you're looking for.

Assumption #1: We can all agree on what we mean by "stuttering."

In determining whether stuttering is genetic, researchers approach their investigations as if "stuttering" were something very specific. But is it? Let us say that you have four people who stutter. One suffers from a physical disorder such as Parkinson's, another is a young child still trying to master speech, a third is a person who tends to stumble when she's flustered, and a fourth is a person who frequently finds his speech locked up, rendering him unable to speak until the block is released. Because of the lack of useful words to set apart one speech pattern from another, the researcher is forced to call each type of disfluency "stuttering." This lumping together of different phenomena cannot help but cloud the perception of the researcher and make it virtually impossible to generate reliable data on stuttering because *what* is being studied (ie: stuttering) has not been clearly defined.

This situation is due in part to the paucity of language used to describe stuttering. Imagine if we were conducting a study on chameleons but instead of talking specifically about chameleons, we constantly referred to the subjects under observation only as reptiles. Of course, *we* would know what we were referring to, but someone else might be picturing another kind of reptile; snakes, for example, or iguanas. This couldn't help but be confusing. Yet, isn't that precisely what happens when we undertake investigations into the genetic cause of stuttering? We have four different kinds of stuttering, but we have only one word to distinguish between the four.

One way around this problem is to have separate words or phrases to differentiate the four kinds of disfluency. For example, I call first kind of stuttering *pathological disfluency* to identify the fractured speech that results when a person is suffering from a physical deficit such as a brain lesion or Parkinson's. The second is *developmental disfluency* which describes the speech of a child who is struggling to master the uncertainties of communication. The third kind of stuttering is *bobulating* (a coined word), the effortless, stumbling disfluency characteristic of the person who is emotionally upset or discombobulated. Finally, there is *blocking* where the person has locked up and is unable to speak.

By substituting these words for "stuttering," it is possible to be clear about the issue under observation. These distinctions, however, are usually not made when researchers conduct their studies, so when they say "We're

looking into the genetic cause of stuttering," it's really not clear how their investigative studies are being directed.

Yet another problem overlooked by researchers is that chronic stuttering is actually a compound problem. It is comprised of (1) the speech block and (2) what the individual does to break through or avoid the block (see fig. 1). Therefore, if you want to carry out meaningful research, you have to decide what part of the problem you're going to study; that is, you have to observe it in its most elemental form.

VARIETIES OF STUTTERING

PATHOLOGICAL DISFLUENCY

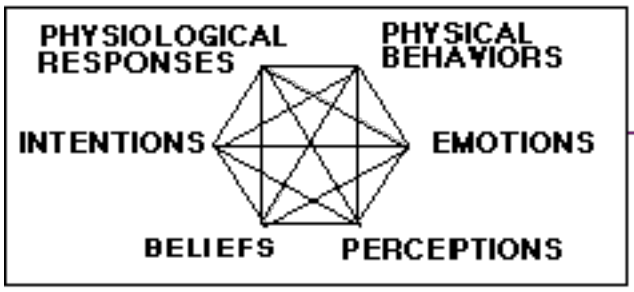
CAUSED BY AN INSULT TO THE BRAIN SUCH AS A HEAD INJURY OR PARKINSONS DISEASE

DEVELOPMENTAL DISFLUENCY

EFFORTLESS, UNSELFCONSCIOUS DISFLUENCY ASSOCIATED WITH YOUNG CHILDREN (USUALLY DISAPPEARS BY EARLY TEENS)

BOBULATING

EFFORTLESS DISFLUENCY DISPLAYED WHEN PEOPLE ARE UPSET, CONFUSED, OR DISCOMBOBULATED. ALSO CALLED "LINGUISTIC UNCERTAINTY."



SPEECH BLOCK

STRATEGIES TO BREAK THROUGH THE BLOCK

PUSH

BATTER

STRATEGIES TO AVOID THE BLOCK

REPEAT PREVIOUS WORD OR SOUND (STALLING)

BE SILENT UNTIL BLOCK PASSES

VISIBLE STRUGGLES

Fig. 1

To draw an analogy, let's say your car develops an intermittent problem when you start it up in the morning, and after several frustrating days, you finally bring it in to Gunnart, the local auto mechanic. Gunnart does a thorough examination and calls you the next day with a diagnosis.

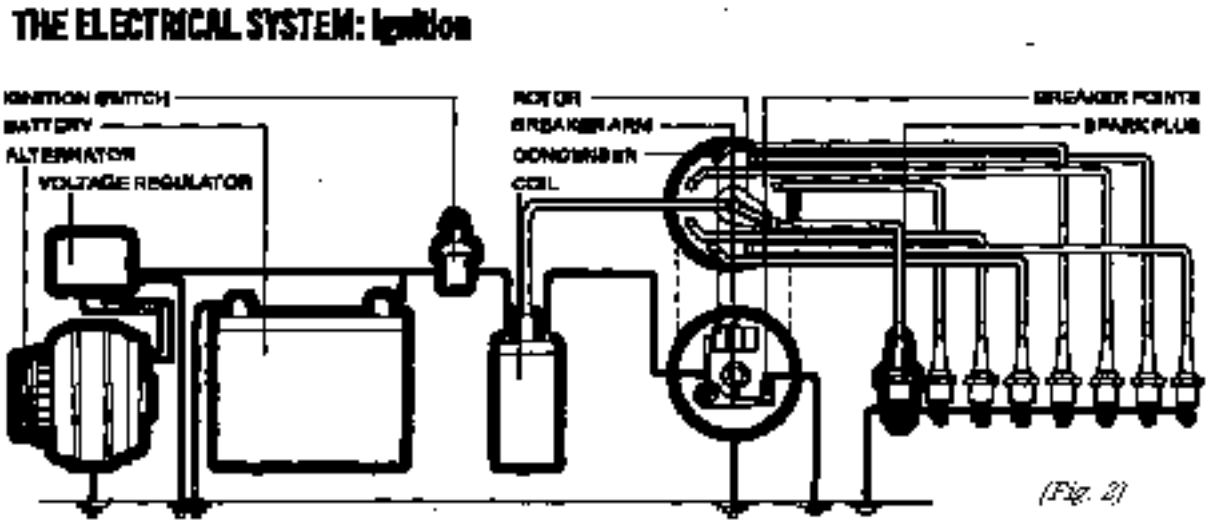
"Well...?" you ask.

"It's the electrical system," Gunnart reports.

Well, you think, that's a relief, Now we've solved the problem.

Or have you.

Here is a diagram of your car's electrical system.



(Fig. 2)

Because the electrical system is a *system,* it is, by definition, composed of more than one part. Thus, although you know *in general* where the problem lies, Gunnart has to do a lot more investigating before he can tell you *specifically* where the problem resides.

Yet, most research into the possible genetic causes of stuttering does not follow the same logical approach that Gunnart does in diagnosing your car. It does not break down chronic stuttering into its components but looks instead at the whole system as if it were a single unitary problem.

Assumption #2: We can accurately identify when a person is blocked.

Some time ago I mentioned to a speech pathologist that I had grown up with a stuttering problem, and that I'd struggled with it more or less for 30 years but now I was fully recovered. She nodded her head abstractly as I told her this, and then confided that she could tell I was a stutterer because she could still see traces of it in my occasional disfluencies.

That was news to me. My definition of chronic stuttering is pretty straightforward. If my speech is blocked so that I cannot *spontaneously* move beyond a particular word or sound, then I have a problem. (Not something I experience anymore.) If, on the other hand, my speech is occasionally stumbly but I'm feeling no resistance to speaking and am not even aware of these minor bobulations, then I don't

have a problem. The difference between my speech now and forty years ago is that I don't block.

No block, no problem.

Yet, most researchers fail to make this distinction, and therefore, they end up mixing apples and oranges. For example, a woman who says, I-I-I-I-I can't make it over tonight" and stumbles on the word "I" because she's upset about letting someone down may sound exactly like the woman who repeats the word "I" because she fears she won't be able to say the word "can't." In one case the speaker is uncertain, embarrassed, discombobulated; in the other instance, she's blocked. Yet the researcher will call both of these speech patterns "stuttering."

Or let's take another example. One person's fluent speech is totally spontaneous while the other individual is constantly substituting to avoid her blocks. Yet, the researcher will call both of those people "fluent."

Do researchers make these distinctions? Generally not. If someone manifests disfluencies, they stutter. If not, they don't. The researcher doesn't measure the person's subjective experience to find out what's really going on. Therefore, it is difficult to know the meaning of whatever findings come to light.

Assumption #3: Only genetic factors can be transmitted from one generation to another.

It is such a logical assumption. The stutterer purses his lips, but no sound emerges. He becomes caught in a repetitive cycle of "ra-ra-ra-ra-ra-ra" before "report" shoots from his mouth like an errant missile. Surely, there is some mechanical or nervous problem that is impeding his speech, and surely, that problem has deep genetic roots. How else could we explain that these behaviors are often present in families from one generation to another.

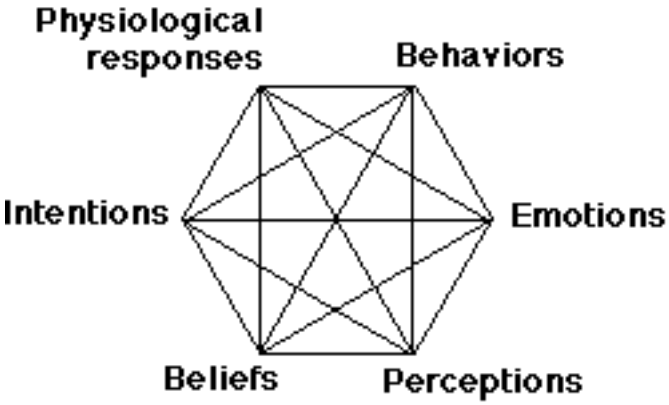
Yet, as we have seen in the long-standing animosity between Serbs and ethnic Albanians, attitudes and beliefs, too, can be passed from grandfather to father to son.

But what do they have to do with stuttering? And if attitudes and beliefs are in fact major contributing factors, why have we never considered them?

We have not considered them because our thinking has been paralyzed by an out-of-date paradigm. The old paradigm says that stuttering is a unitary problem driven by genetic factors. Thus, our perceptions and beliefs are seen only as outgrowths of our stuttering. They are not seen as causal agents.

In the new paradigm, chronic stuttering is not understood as a unitary problem but a *system* with six key components in a dynamic relationship in which *each point of the system affects and is affected by all the other points*. It is not the components by themselves that create the problem but the dynamic moment-by-moment interaction of these six components that brings to life the stuttering behavior.

THE STUTTERING HEXAGON



For example, when the person wants to ask a stranger on the street "Do you have the time" and gets caught on tuh-tuh-tuh-tuh-tuh-tuh, there are a number of forces at work. Let us look at some of the components that make up this system.

Let's begin by looking at the circumstance of stopping a stranger to ask a question. The stutterer is aware of totally obsessing on his fear of saying "time." He believes he will block, and the feared word is adding to his fight-or-flight reaction. But is this the only thing triggering his response? Not likely. There is also the issue of encountering a stranger. Who is this person? What does she look like? What is at risk? Is she pretty, and does he feel "worthy" enough to talk to her? How will he respond? What is he projecting into the encounter?

What about the momentary flick of her eye before she actually looks at him. Is she annoyed? Afraid? In a hurry? Does she resent being stopped by a perfect stranger? How does he interpret all this?

Does he feel he has to talk perfectly? Or be perfect? Does he have a preconceived idea of how he wants her to respond? Suppose she doesn't follow his script? Is it a trigger for more panic?

What about his response threshold? Is he highly sensitive? Is he quick to initiate a fight-or-flight response? Is he inclined to overreact? Is he having a good day emotionally? Is he feeling positive and confident, or insecure or dejected? The stutterer's beliefs and how he interprets his perceptions will have an enormous impact on his feelings.

Many of the forces that bring to bear on the moment have nothing to do with his fear of stuttering per se but with his response to the environment. However, because these forces usually operate outside of his awareness, the only thing he may be conscious of is his fear about his speech.

Assumption #4: You can do meaningful research without having a clear idea of what you're looking for.

All investigations into the genetic causes of stuttering seem to have one thing in common–the researchers don't have a credible theory behind their study. In other words, they don't specifically know what they're looking for. This curious situations arises because for many and perhaps most researchers, stuttering is an ill-defined speech anomaly whose very definition seems to be rooted in the unknown. But, says William Perkins, Professor Emeritus,

University of Southern California and former Director of the Stuttering Center at USC, if you want to do *meaningful* research, you must start out with a credible theory and then use a methodology to test predictions that are derived from that theory.

In the January/February 1997 issue of the NSA newsletter *Letting GO,* Perkins develops this point further:

Since the advancement of theories of stuttering is looked down upon as speculation, investigators are left in an odd position. Here they sit with all this high powered equipment. But they don't have any scientifically rational theory to use it on that predicts cause and effect. Instead, stuttering research has been akin to trawling a net behind a boat to see what you pick up. It's undertaken with the idea that if only you can just gather enough data, then the cause of stuttering will become apparent....

In fact, most research does not even focus on stuttering directly, but on conditions associated with stuttering. This is especially true of neurological studies, which have rarely been used to challenge theories. Like other research, it has only been used to find support for ideas.

One of the foundations of science is that a theory must account for all *defining characteristics, [and it is this fact that] is perhaps most responsible for perpetuating the belief that stuttering is unsolvable. Here's why.*

Virtually all research intended for understanding stuttering

involves groups of subjects–in fact, the larger the group the better. On the face of it, this seems sensible. With a single subject or small group, results cannot be applied with confidence to the stuttering population in general.

But in group studies, how a particular person speaks is ignored in favor of numerical group averages for pauses, disfluencies, etc. This means that group results probably do not describe any individual in a study. This would not pose a problem if all those who stutter were alike. But groups do not stutter. Individuals stutter. And the causes of their stuttering vary from one-person to the next.

It is only after individual causes are understood that group research can be productive in helping to find out how widespread these causes are among the stuttering population.

But to start with group research?

That's a guarantee that the core of stuttering will never be solved with this approach.

The only thing that researchers seem to be able to determine is that *something* is happening in certain parts of the brain when an individual stutters. But what it is, and what effect it may have in creating stuttering remains totally speculative. Yet, the fact that something *is* happening does not discourage researchers from confidently asserting that there are genetic factors that cause stuttering. Otherwise, they say, these responses

would not be present to a greater statistical degree in families that have a history of stuttering. But whether these are *causal* factors or by-products of other events associated with stuttering is not something they are able to determine.

Finally, since stuttering does *not* appear in the family histories of 75% of people who stutter, how can we claim that stuttering is genetically driven? What other maladies that *have* been proven to be genetically determined show such a low statistical presence within the families transmitting the problem?

Where does that leave us genetics-wise?

From my own experiences as a recovered stutterer, as well as from more than 22 years of active involvement in the stuttering self-help movement, I have observed that stuttering seems to be a problem in which six key elements–emotions, perceptions, beliefs, intentions, physiological responses, and physical behaviors–interact and form a self-perpetuating, behavioral system.

Several parts of the system can, in fact, be transmitted from parent to child–namely, ones perceptions and beliefs about life, proper behavior, and what one should expect from others. These are elements that travel effectively through time and undoubtedly contribute to the higher incidence of stuttering in certain families.

There is, however, a genetic component that I'm sure does play a role in stuttering. It does not relate to speech directly,

but to how the individual relates to stress. It is something that can be passed along in the genetic make-up of certain families. It has to do with the part of the brain which is most responsible for storing emotional memory.

A discovery

One summer while I was in college, I worked in the mail room of my father's advertising agency. In the mail room was a black phone that connected directly to the photostat house several blocks away. Twice, maybe three times a day, one of the art directors from upstairs would call down and ask me to phone up the stat house and ask for a pick-up.

I lived in terror of that phone since I invariably blocked on the letter "p." Usually, the first request for a pickup happened about mid-afternoon, after I'd had a good part of the day to worry about it, so when I finally had to make the call, my nerves were a mess. Even so, my natural stubbornness would fill me with resolve. This time, I'd say to myself, I'd say "pick-up" without resorting to a starter sound, like "um," or "ah," or starter words like, "Yeah, could you make a pick-up." And each time I'd chicken out when the gruff voice at the other end answered.

One morning I showed up at the office feeling especially good. At about 10 o'clock, the first call came down to order a pick-up. Since it came so early, I hadn't had much time to worry about it. I decided to really go for broke and say "pick-up" without any kind of "cheating." I picked up the phone.

The voice said, "Hello."

Hurling headlong into the experience, I took a deep breath and said, "Pick-up." I did not use a starter word. I did not block. I kept my throat and lips relaxed. At that moment I was startled by an enormous rush of feeling, a panic reaction such as I had not experienced before.

"Wow!" I thought as I replaced the handset. "Where did that come from?" I felt I had discovered what was behind the speech block, and what would happen if I *didn't* block or avoid the word. I discovered that I had been preventing myself from experiencing an overwhelming sense of panic.

I had never known that these feelings were lurking there until that moment. It turned out to be an enormously useful revelation. Fear of being overwhelmed by a sudden rush of feeling has gone a long way to explain to me what a speech block is all about, and my beliefs were validated by the years of work I did in personal growth programs. The more I became comfortable with my expressing my emotions, the less inclination there was to block.

This is not to say that chronic stuttering is *caused* by the suppression of unwanted feelings. But I *am* saying that a holding back of feeling seems to be an important component of the total stuttering system.

To gain a better understanding of this, it is useful to know something about the functioning of an important part of the brain called the amygdula.

The role of the amygdula

The amygdula is an almond-shaped cluster of interconnected structures perched above the brainstem, near the bottom of the limbic ring, that acts as a storehouse of emotional memory. It is one of the most primitive parts of the brain, evolving hundreds of thousands of years before the development of the cerebral cortex where rational thought takes place.

The original role of the amygdula was to ensure that animals would have particularly vivid memories of what threatens or pleases them. Like a neural tripwire, whenever the animal was threatened, the amygdula would send urgent messages to every major part of the brain to trigger the secretion of the body's fight-or-flight hormones, activate the cardiovascular system, and prepare the muscles for action. Thus, if primitive man heard a deep growl and a rustle of grass, the hair-trigger response of his amygdula would marshal him to take action before the marauding predator could attack him by surprise.

Though modern man has developed a highly evolved cerebral cortex capable of abstract thinking, the amygdula still occupies a favored position, and in the event of an emotional emergency, either physical *or* social, the amygdula will effectively hijack the rest of the brain, including the rational mind. Thus, if you've been unfortunate enough to have been hit by a race car while you

were a spectator at a TransAm event (as happened to my wife some years ago), the sound of screeching brakes on a city street is enough to trigger an instant fight-or-flight reaction. In fact, the amygdula can often trigger an emotional response before the cortical centers have fully understood what is happening–as if our emotions have a mind of their own which operates independently of our rational mind.

In the best selling book, *Emotional Intelligence,* by Daniel Goleman (must reading for anyone who stutters), the author notes that some people are born with a neurochemistry that makes this circuit easily aroused. For example, says Goleman, some children "may have inherited chronically high levels of norepinephrine or other brain chemicals that activate the amygdula and so create a new threshold of excitability, making the amygdula more easily triggered."

The relevance of this to chronic stuttering is underscored by a study done by Dr. Libby Oyler for her Ph.D. dissertation in speech pathology. In an article that ran in the April 1998 edition of *Letting GO,* Oyler reported that in a statistically significant number of cases, people who stutter show a higher level of sensitivity than do non-stutterers. Thus, a subtle change in tone of voice, a gesture, a momentary expression or other non-verbal form of communication would have a greater impact on the child who is at risk to develop the blocking behaviors that characterize chronic stuttering. Therefore, if one wishes to conduct research into the impact of genetic factors on chronic stuttering, it might be much more productive to

look into the individual's sensitivity to the environment and his or her responsiveness to threatening events than into the systems associated with speech making per se.

Summary

As the editor of the National Stuttering Association's monthly newsletter *Letting GO,* I regularly receive requests from researchers who ask that we run their announcement soliciting subjects for their latest study into the genetic cause of stuttering. I am always happy to oblige. But I cannot help but feel these investigations are destined to come up with findings that are inconclusive and of little practical value. Here's why I think so:

1. Researchers treat stuttering as if it were a specific, clearly defined phenomenon, whereas most of us don't even agree on what the word stuttering means, let alone what is actually going on when a person locks up and cannot move his speech forward.

2. Social scientists conducting genetic studies look at stuttering as if it were a unitary problem, whereas chronic stuttering is better defined as a combination of a speech block and the strategy to break through or avoid the block. Because it is a compound problem, researchers would be better served by studying the most elemental component, the speech block, rather than lumping the block together with the coping strategies.

3. Most researchers look only at the presence of superficial

disfluencies to determine whether or not a person is stuttering, but people who bobulate when excited are not necessarily having any conscious speech difficulty per se. A classic example of misidentifying chronic stuttering took place several years ago on the *Marilu* show on TV where the invited guests were either speech-language pathologists or people dealing with their own stuttering. Actor Gordon Clapp was also invited on the show to talk about his sympathetic portrayal of a character with a stuttering problem on the TV drama *NYPD Blue*. Clapp was even acknowledged as a hero by one NSA member in the audience for being a positive role model for stutterers. Lt. Medavoy, the New York detective played by Clapp, does in fact display occasional disfluencies, but there are none of the struggle behaviors or the self-consciousness normally associated with blocked speech. Nor did Clapp ever even consider stuttering as a problem when he was fleshing out his character. I've always felt that the actor was somewhat mystified by his presence on the show.

4. Researchers assume that only genetic factors can be transmitted intergenerationally, but attitudes and beliefs can also run in families. The reason why these have not been considered as contributing factors is the narrow paradigm used to define chronic stuttering. When you look at stuttering as something that is constructed of ordinary building blocks, however, then attitudes and beliefs become causal agents and you no longer have to resort to genetics to explain why chronic stuttering often runs in families.

Even so, there probably are genetic factors that relate to

chronic stuttering, but they do so indirectly. These have to do with the degree of sensitivity of the individual and the level of responsiveness in reacting to stress, factors that can be passed along from parent to child.

5. Finally, research into the genetic causes of stuttering traditionally involves "trawling" for answers. It is not based on a credible theory. The researcher often has no idea what he's looking for. He just hopes that something interesting shows up, but what it could means is highly speculative.

I'm well aware that this essay is likely to prove unpopular with anyone engaged in genetic research on stuttering. However, rather than establishing an adversarial relationship, I hope that the points I've raised are cause for reflection and perhaps a clearer definition of objectives.

I would be happy to create a dialogue with anyone in the field who would like to explore these matters further.